Chapter 2 : Gentaur Products List

• The PAX5 gene is a member of the paired box PAX family of transcription factors The central feature of this gene family is a novel highly conserved DNA binding motif known as the paired box The
• The protein encoded by TRIM3 is a member of the tripartite motif TRIM family also called the RING B box coiled coil RBCC subgroup of RING finger proteins The TRIM motif includes three zinc bin
• ZMYND11 was first identified by its ability to bind the adenovirus E1A protein The protein localizes to the nucleus It functions as a transcriptional repressor and expression of E1A inhibits this r
• LRRC14 belongs to the PRAME family and contains 6 LRR leucine rich repeats
• At the nuclear envelope the nuclear lamina and heterochromatin are adjacent to the inner nuclear membrane CBX3 binds DNA and is a component of heterochromatin CBX3 also can bind lamin B receptor a
• PRR7 encodes a proline rich membrane protein which is involved in modulating neural activities via interactions with the NMDA receptor and PSD 95 and PSD core formation
• KEAP1 contains KELCH 1 like domains as well as a BTB POZ domain Kelch like ECH associated protein 1 interacts with NF E2 related factor 2 in a redox sensitive manner and the dissociation of the prot
• RIMS3 belongs to a family of synaptic proteins that are essential for normal neurotransmitter release
• The protein encoded byTRIM14 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region
• BTBD3 is a protein containing BTB POZ domain Its function has not been determined yet
• The protein encoded by TRIM39 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region
• TRIM34 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region Expression of this ge
• RNF36 is specifically expressed in germ cells at round spermatid stages during spermatogenesis The protein encoded by this gene contains an N terminal RING finger motif a B box and a C terminal B 3
• UHRF2 encodes a nuclear protein which is involved in cell cycle regulation The encoded protein is a ubiquitin ligase capable of ubiquinating PCNP PEST containing nuclear protein and together they
• CREM is a bZIP transcription factor that binds to the cAMP responsive element found in many viral and cellular promoters It is an important component of cAMP mediated signal transduction during the s
• The THRA gene encodes a protein that is a nuclear hormone receptor for triiodothyronine It is one of the several receptors for thyroid hormone and has been shown to mediate the biological activities
• The T cell specific transcription factor TCF7 activates genes involved in immune regulation and is a candidate locus for genetic susceptibility to type 1 diabetes
• TRIM38 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region The function of this
• BRD9 is a member of protein family that contains bromodomain It is potentially related to cancer
• The gene encoding the hypothetical protein HS747E2A is located on chromosome 22
• Located on chromosome 19 this gene encodes for zinc finger protein 627
• ZNF627 is a new candidate transcription factor
• ZNF22 encodes a protein that is expressed in the epithelial component of the developing tooth organ during early bud and cap stages as well as in osteoblasts of craniofacial bone and the developing to
• BRD9 encodes a bromodomain containing 9 protein and is located on chromosome 5
• HSF2 binding protein HSF2BP associates with HSF2 The interaction occurs between the trimerization domain of HSF2 and the amino terminal hydrophilic region of HSF2BP that comprises two leucine zippe
• ZNF258 is part of a novel putative zinc binding motif MYM family which encodes proteins that maintain the repeats of the MYM motif
• ZNF614 is a new candidate transcription factor
• The protein encoded by TRIM9 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region
• ZNF396 acts as a DNA dependent transcriptional repressor
• ZNF568 is a candidate transcription factor
• The function of the Anti ZNF568 gene has not yet been determined
• In mice the death inducer obliterator 1 gene is upregulated by apoptotic signals and encodes a cytoplasmic protein that translocates to the nucleus upon apoptotic signal activation When overexpresse
• ZNF57 belongs to the krueppel C2H2 type zinc finger protein family It contains 13 C2H2 type zinc fingers and 1 KRAB domain ZNF57 may be involved in transcriptional regulation
• ZNF547 may be involved in transcriptional regulation
• The muscle acetylcholine receptor consiststs of 5 subunits of 4 different types 2 alpha isoforms and 1 each of beta gamma and delta subunits 2 CHRNA1 encodes an alpha subunit that plays a role in a
• The muscle acetylcholine receptor consiststs of 5 subunits of 4 different types 2 alpha isoforms and 1 each of beta gamma and delta subunits 2 The CHRNA1 gene encodes an alpha subunit that plays a
• Chloride channel Kb CLCNKB is a member of the CLC family of voltage gated chloride channels which comprises at least 9 mammalian chloride channels Each is believed to have 12 transmembrane domains
• When expressed in the bladder epithelium the protein encoded by the SCNN1B gene might be implicated in the mechanosensory transduction in the bladder afferent pathways thereby inducing detrusor inst
• CACNA2D1 encodes a member of the alpha 2 delta subunit family a protein in the voltage dependent calcium channel complex Calcium channels mediate the influx of calcium ions into the cell upon membra
• The protein encoded by CACNB1 belongs to the calcium channel beta subunit family It plays an important role in the calcium channel by modulating G protein inhibition increasing peak calcium current
• CACNB2 is a member of the ion channel gene superfamily Described as a Lambert Eaton myasthenic syndrome LEMS antigen in humans this gene is found close to a region that undergoes chromosome rearra
• CACNB4 is a member of the beta subunit family a protein in the voltage dependent calcium channel complex CACNB4 plays an important role in calcium channel function by modulating G protein inhibition
• Nicotinic acetylcholine receptors nAChRs such as CHRNA5 are members of a superfamily of ligand gated ion channels that mediate fast signal transmission at synapses The nAChRs are thought to be h
• Mutations in nAChRs are found in a rare form of nocturnal frontal lobe epilepsy Previously some nAChR mutations have been described that are associated with additional neurological features such as
• The gamma aminobutyric acid GABA A receptor is a multisubunit chloride channel that mediates the fastest inhibitory synaptic transmission in the central nervous system GABRB2 encodes GABA A recepto
• N methyl D aspartate NMDA receptors are a class of ionotropic glutamate receptors NMDA channel has been shown to be involved in long term potentiation an activity dependent increase in the efficie
• ACCN1 is a member of the degenerin epithelial sodium channel DEG ENaC superfamily ACCN1 may play a role in neurotransmission In addition a heteromeric association between ACCN1 and ACCN3 variant
• Chloride channels are a diverse group of proteins that regulate fundamental cellular processes including stabilization of cell membrane potential transepithelial transport maintenance of intracellul
• Chloride intracellular channel 1 is a member of the p64 family a diverse group of proteins that regulate fundamental cellular processes including stabilization of cell membrane potential transepithe
• Chloride intracellular channel 2 is a member of the p64 family a diverse group of proteins that regulate fundamental cellular processes including stabilization of cell membrane potential transepithe
• KCNK3 encodes one of the members of the superfamily of potassium channel proteins containing two pore forming P domains The gene product is an outwardly rectifying channel that is sensitive to change
• Action potentials in vertebrate neurons are followed by an afterhyperpolarization AHP that may persist for several seconds and may have profound consequences for the firing pattern of the neuron Ea
• P2RX1 belongs to the family of purinoceptors for ATP This receptor functions as a ligand gated ion channel with relatively high calcium permeability Binding to ATP mediates synaptic transmission bet
• The voltage dependent anion selective channel 1 VDAC1 functions as a channel in membranous structures for the outer mitochondrial membrane the cell membrane endosomes caveolae the sarcoplasmatic
• VDAC2 forms a channel through the mitochondrial outer membrane that allows diffusion of small hydrophilic molecules The channel adopts an open conformation at low or zero membrane potential and a clo
• Potassium channels represent the most complex class of voltage gated ion channels from both functional and structural standpoints Their diverse functions include regulating neurotransmitter release
• CUL5 encodes a protein that is involved in the regulation of cellular growth and promotes vif ubiquination
• CLCNKA is a member of the CLC family of voltage gated chloride channels It is predicted to have 12 transmembrane domains and requires a beta subunit called barttin to form a functional channel It i
• ACCN3 encodes a member of the degenerin epithelial sodium channel DEG ENaC superfamily The members of this family are amiloride sensitive sodium channels that contain intracellular N and C termini
• P2RXL1 encodes a protein in the family of P2X receptors which are ATP gated ion channels and mediate rapid and selective permeability to cations P2RXL1 is predominantly expressed in skeletal muscle
• Potassium channels represent the most complex class of voltage gated ion channels from both functional and structural standpoints Four sequence related potassium channel genes shaker shaw shab an
• The product of HTR3B belongs to the ligand gated ion channel receptor superfamily HTR3B encodes subunit B of the type 3 receptor for 5 hydroxytryptamine serotonin a biogenic hormone that functions
• FXYD5 is a member of a family of small membrane proteins that share a 35 amino acid signature sequence domain beginning with the sequence PFXYD and containing 7 invariant and 6 highly conserved amino
• TRPM5 is a voltage modulated Ca 2 activated monovalent cation channel VCAM that mediates a transient membrane depolarization and plays a central role in taste transduction It is activated by ar
• KCNIP1 is a member of the family of voltage gated potassium Kv channel interacting proteins KCNIPs which belong to the recoverin branch of the EF hand superfamily Members of the KCNIP family are
• The product of P2RX2 belongs to the family of purinoceptors for ATP This receptor functions as a ligand gated ion channel Binding to ATP mediates synaptic transmission between neurons and from neuro
• P2RX2 belongs to the family of purinoceptors for ATP This receptor functions as a ligand gated ion channel Binding to ATP mediates synaptic transmission between neurons and from neurons to smooth mu
• CLIon Channel5 specifically associates with the cytoskeleton of placenta microvilli CLIon Channel 5A has a role as a chloride channel in vitro and binds to cortical actin cytoskeleton
• Chloride intracellular channels are involved in chloride ion transport within various subcellular compartments CLIon Channel5 specifically associates with the cytoskeleton of placenta microvilli
• Chloride intracellular channels are involved in chloride ion transport within various subcellular compartments CLIon Channel5 specifically associates with the cytoskeleton of placenta microvilli Chlo
• The gamma aminobutyric acid GABA A receptor is a multisubunit chloride channel that mediates the fastest inhibitory synaptic transmission in the central nervous system This gene encodes GABA A rece
• NMUR2 encodes for one of two G protein coupled receptors for the neuropeptide neuromedin U This peptide is found in highest levels in the gut and genitourinary system where it potently contracts smo
• MCOLN1 encodes a protein that may be involved in calcium signaling and membrane trafficking in mucolipidosis IV
• KCNK10 is one of the members of the superfamily of potassium channel proteins containing two pore forming P domains The message for this gene is highly expressed in the kidney and pancreas This chan
• The CLCN family of voltage dependent chloride channel genes comprises nine members CLCN1 7 Ka and Kb which demonstrate quite diverse functional characteristics while sharing significant sequence ho

• GRIK2 encodes a subunit of a kainate glutamate receptor Glutamate receptors mediate the majority of excitatory neurotransmission in the brain This receptor may have a role in synaptic plasticity and
• This gene GRIK2 encodes a subunit of a kainate glutamate receptor Glutamate receptors mediate the majority of excitatory neurotransmission in the brain This receptor may have a role in synaptic pl
• GABRE belongs to the ligand gated ionic channel TC 1 A 9 family It encodes the gamma aminobutyric acid GABA A receptor which is a multisubunit chloride channel that mediates the fastest inhibitor
• KCNK13 encodes one of the members of the superfamily of potassium channel proteins containing two pore forming domains The product of this gene is an open channel that can be stimulated by arachidoni
• KCNK13 encodes one of the members of the superfamily of potassium channel proteins containing two pore forming domains The product of KCNK13 is an open channel that can be stimulated by arachidonic a
• KCTD10 a rat potassium channel tetramerisation domain containing 10 gene is a novel member of the polymerase delta interacting protein 1 PDIP1 gene family KCTD10 shares significant similarity in a
• CLIon Channel6 encodes a member of the chloride intracellular channel family of proteins The gene is part of a large triplicated region found on chromosomes 1 6 and 21
• KCNK10 encodes one of the members of the superfamily of potassium channel proteins containing two pore forming P domains The message for this gene is highly expressed in the kidney and pancreas This
• L type calcium channels are composed of five subunits The protein encoded by CACNG6 represents one of these subunits gamma and is one of several gamma subunit proteins It is an integral membrane p
• The protein encoded by CHRNA7 displays marked permeability to calcium ions and is a major component of brain nicotinic receptors that are blocked by and highly sensitive to alpha bungarotoxin Once
• The function of Anti KCTD18 has not yet been determined
• VMD2L2 is 1 of 3 VMD2 like genes which encode transmembrane spanning proteins that share a homology region with a high content of aromatic residues including an invariant arginine R phenylalanine
• KCTD6 is a domain of potassium channel
• Calcium ions play a primary role in the regulation of sperm motility This gene belongs to a family of putative cation channels that are specific to spermatozoa and localize to the flagellum The prot
• Calcium ions play a primary role in the regulation of sperm motility Anti CATSPER2 belongs to a family of putative cation channels that are specific to spermatozoa and localize to the flagellum The
• The M channel is a slowly activating and deactivating potassium channel that plays a critical role in the regulation of neuronal excitability The M channel is formed by the association of the protein
• This gene encodes a member of the potassium channel voltage gated subfamily H This member is a pore forming alpha subunit of a voltage gated non inactivating delayed rectifier potassium channel
• The KCNIP2 gene encodes a member of the family of voltage gated potassium Kv channel interacting proteins KCNIPs which belongs to the recoverin branch of the EF hand superfamily Members of the K
• KCNIP2 encodes a member of the family of voltage gated potassium Kv channel interacting proteins KCNIPs which belongs to the recoverin branch of the EF hand superfamily Members of the KCNIP fami
• CLCN3 plays an important role in the cell proliferation of vascular smooth muscle cells The PDZ binding isoform of CLCN3 resides in the Golgi where it co localizes with a small amount of CFTR cystic
• The GRIK2 gene encodes a subunit of a kainate glutamate receptor Glutamate receptors mediate the majority of excitatory neurotransmission in the brain This receptor may have a role in synaptic plast
• The product P2RX7 belongs to the family of purinoceptors for ATP This receptor functions as a ligand gated ion channel and is responsible for ATP dependent lysis of macrophages through the formation
• KCTD13 mRNA is expressed in 3T3 L1 adipocytes and THP 1 macrophages It is suggested that this gene provides a link between cytokine activation and DNA replication in liver as well as in other tissues
• KCNQ1 encodes a protein for a voltage gated potassium channel required for the repolarization phase of the cardiac action potential The gene product can form heteromultimers with two other potassium
• ACCN4 belongs to the superfamily of acid sensing ion channels which are proton gated amiloride sensitive sodium channels These channels have been implicated in synaptic transmission pain perceptio
• Human ADP ribose pyrophosphatase NUDT9 belongs to a superfamily of Nudix hydrolases that catabolize potentially toxic compounds in the cell NUDT9 alpha protein is targeted highly specifically to mito
• SCN5A is an integral membrane protein and tetrodotoxin resistant voltage gated sodium channel subunit The protein is found primarily in cardiac muscle and is responsible for the initial upstroke of t
• Gamma aminobutyric acid GABA the major inhibitory neurotransmitter in the brain mediates neuronal inhibition by binding to GABA receptors The type A GABA receptors are pentameric chloride channel
• KCNRG is a soluble protein with characteristics suggesting it forms heterotetramers with voltage gated K channels and inhibits their function KCNRG is a soluble protein with characteristics sugges
• CACNB2 is a subunit of voltage dependent calcium Ca2 channels expressed in the CNS It appears to serve an obligatory function
• TRPM3 encodes a protein that belongs to the family of transient receptor potential TRP channels TRP channels are cation selective channels important for cellular calcium signaling and homeostasis
• The KCTD11 gene encodes a protein that has been identified as a suppressor of Hedgehog signaling Its inactivation might lead to a deregulation of the tumor promoting Hedgehog pathway in medulloblasto
• The PITX2 gene encodes a member of the RIEG PITX homeobox family which is in the bicoid class of homeodomain proteins This protein acts as a transcription factor and regulates procollagen lysyl hydr
• THRB is a nuclear hormone receptor for triiodothyronine It is one of the several receptors for thyroid hormone and has been shown to mediate the biological activities of thyroid hormone Knockout st
• The C EBP family of transcription factors regulates viral and cellular CCAAT enhancer element mediated transcription C EBP proteins contain the bZIP region which is characterized by two motifs in th
• Extracellular HMGB1 is an activator of human tumor cell migration operating in concert with EGF HMGB1 encodes a protein that is potentially involved in the regulation of lipogenic and cholesterogenic
• SLC18A1 is an integral protein in the membrane of secretory vesicles of neuroendocrine and endocrine cells that allows the transport of biogenic monoamines such as serotonin from the cytoplasm into
• SMARCD3 is a member of the SWI SNF family of proteins whose members display helicase and ATPase activities and which are thought to regulate transcription of certain genes by altering the chromatin s
• ZNF84 a gene located on chromosome 12 encodes a zinc finger protein whose function is undescribed
• ZNF134 is a candidate transcription factor
• ZNF134 is a new candidate transcription factor
• Far upstream element binding protein activates the far upstream element FUSE of c myc and stimulates expression of c myc in undifferentiated cells Regulation of FUSE by FUBP occurs through single s
• AIP may play a positive role in AHR mediated signalling possibly by influencing its receptivity for ligand and or its nuclear targeting AIP is the cellular negative regulator of the HBV X protein AIP
• SOX14 encodes a member of the SOX SRY related HMG box family of transcription factors involved in the regulation of embryonic development and in the determination of the cell fate The encoded prote
• This intronless gene SOX14 encodes a member of the SOX SRY related HMG box family of transcription factors involved in the regulation of embryonic development and in the determination of the cell
• ZNF213 contains three C2H2 zinc fingers a Kruppel associated A box and a leucine rich motif LeR domain SCAN box strongly suggestive of a transcription factor C2H2 zinc finger proteins such as ZNF
• The mammalian Kruppel like transcription factor KLF4 is involved in preventing centrosome amplification following DNA damage caused by gamma irradiation It is both necessary and sufficient in preven
• The thyroid hormone T3 receptors TRs are hormone dependent transcription factors that regulate expression of a variety of specific target genes TRIP13 specifically interacts with the ligand bindi
• Nuclear factor of activated T cells NFAT is a transcription factor required for T cell expression of the interleukin 2 gene NFAT binds to a sequence in the interleukin 2 gene enhancer known as the
• The function of PIAS2 has not yet been determined
• PIAS2 may function as a co repressor of Stat4
• NMYC interactor NMI encodes a protein that interacts with NMYC and CMYC two members of the oncogene Myc family and other transcription factors containing a Zip HLH or HLH Zip motif The NMI pro
• MTA2 has been identified as a component of NuRD a nucleosome remodeling deacetylase complex identified in the nucleus of human cells It shows a very broad expression pattern and is strongly expresse
• The DLX6 gene encodes a member of a homeobox transcription factor gene family similiar to the Drosophila distal less gene This family is comprised of at least 6 different members that encode proteins
• The human HMG2a gene is transcribed mainly in the placenta HMG2a encodes a protein which is part of the high mobility group HMG family Members of this family are ubiquitously expressed and facilit
• The MYCL1 gene encodes a protein which is associated with lung cancer in idiopathic pulmoary fibrosis
• ZBTB22 contains 1 BTB POZ domain and 3 C2H2 type zinc fingers and belongs to the krueppel C2H2 type zinc finger protein family ZBTB22 may be involved in transcriptional regulation
• MOK2 proteins are DNA and RNA binding proteins that are mainly associated with nuclear RNP components including the nucleoli and extranucleolar structures Arranz et al 1997 PubMed 9121460
• MOK2 proteins are DNA and RNA binding proteins that are mainly associated with nuclear RNP components including the nucleoli and extranucleolar structures MOK2 proteins are DNA and RNA binding prot
• The CHST4 gene encodes a protein that is involved in enzymatic synthesis in vitro of the disulfated disaccharide unit of corneal keratan sulfate
• CHST4 encodes a protein involved in enzymatic synthesis in vitro of the disulfated disaccharide unit of corneal keratan sulfate
• POLR2I is a subunit of RNA polymerase II the polymerase responsible for synthesizing messenger RNA in eukaryotes This subunit in combination with two other polymerase subunits forms the DNA bindin
• Located on chromosome 19 ZNF230 is part of a ZNF gene cluster that encodes Kruppel associated box KRAB motifs
• ZNF271 may be involved in transcriptional regulation It plays a role in the EBV mediated transformation process
• ZNF271 may be involved in transcriptional regulation It plays a role in the EBV mediated transformation processZinc finger proteins interact with nucleic acids and have diverse functions The zinc fi
• Located on chromosome 19 ZNF266 is part of the Kruppel related zinc finger gene family
• HOXC9 belongs to the homeobox family The homeobox family is a highly conserved family of transcription factors that play an important role in morphogenesis in all multicellular organisms This gene be
• RXRG encodes a member of the retinoid X receptor RXR family of nuclear receptors which are involved in mediating the antiproliferative effects of retinoic acid RA This receptor forms dimers with
• ZIC2 is a member of the ZIC family of C2H2 type zinc finger proteins This protein functions as a transcriptional repressor and may regulate tissue specific expression of dopamine receptor D1 Mutatio
• The ZNF75 gene which is located on chromosome X is predicted to encode a zinc finger protein currently with unknown fucntion
• ZNF79 mapped to 9q34 centromeric to the ABL gene and between a constitutional chromosomal translocation on the centromeric side and the CML specific ABL translocation on the telomeric side
• ZNF175 expression in brain mononuclear phagocytes is a signature for advanced HIV 1 encephalitis As a transcriptional suppressor this protein plays a role in macrophage control of viral replication
• GTF3C3 subunit of human transcription factor TFIIIon Channel was identified as a DEDD and FLAME 3 specific interacting protein
• GTF3C5 is polypeptide 5 of general transcription factor IIIon Channel Human transcription factor IIIon Channel hTFIIIon Channel is a multisubunit complex that directly recognizes promoter elements
• The zona pellucida is an extracellular matrix that surrounds the oocyte and early embryo It is composed primarily of three or four glycoproteins with various functions during fertilization and preimp
• ZNF223 is part of a ZNF gene cluster located on human chromosome 19q13 2 which encodes Kruppel associated box KRAB motifs
• ZNF225 is a candidate transcription factor
• Matrix assisted laser desorption ionization time of flight analysis and examination of protein profiles from the SwissProt database revealed that the previously defined p97 repressor is ZNF224 a zinc
• Located on chromosome 11 the BTBD15 gene encodes a BTB POZ domain containing 15 protein with unknown

function
• ZNF232 is a new candidate transcription factor
• ZNF432 belongs to the krueppel C2H2 type zinc finger protein family It contains 16 C2H2 type zinc fingers and 1 KRAB domain ZNF432 may be involved in transcriptional regulation
• As a transcriptional repressor ETV7 binds to the DNA sequence 5 CCGGAAGT 3 But Isoform A and isoform C do not seem to have a repressor activity
• Located on chromosome X this gene encodes hypothetical protein LOC51402
• TRIP4 is a transcription coactivator of nuclear receptors which functions in conjunction with CBP p300 and SRC 1 and may play an important role in establishing distinct coactivator complexes under dif
• ZNF12 is a candidate transcription factor
• VGLL1 is a specific coactivator for the mammalian TEFs The mammalian TEF and the Drosophila scalloped genes belong to a conserved family of transcriptional factors that possesses a TEA ATTS DNA bindi
• PHF20 is a possible transcription factor
• ZNF416 may be involved in transcriptional regulation
• ZNF446 is a new candidate transcription factor
• JMJD2D belongs to JMJD2 family that is consist of four cancer associated genes
• ZNF358 a gene located on chromosome 19 encodes a protein involved in embryogenesis
• ZNF167 a gene located on chromosome 3 encodes for a zinc finger protein
• ZNF307 contains 1 SCAN box domain 1 KRAB domain and 7 C2H2 type zinc fingers It belongs to the Krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation
• LHX9 encodes a member of the LIM homeobox gene family of developmentally expressed transcription factors The encoded protein contains a homeodomain and two cysteine rich zinc binding LIM domains invo
• SBZF3 located on chromosome 1 encodes a zinc finger protein
• Located on chromosome 17 the ZNF286 encodes zinc finger protein 286 with unknown function
• The ZNF2 gene is part of a sub family of structurally related finger protein genes Members of this sub family are expressed as multiple transcripts in several cell lines
• The function remains unknown Western blots using two different antibodies against two unique regions of this protein target confirm the same apparent molecular weight in our tests
• CREBZF strongly activates transcription when bound to HCFC1 CREBZF suppresses the expression of HSV proteins in cells infected with the virus in a HCFC1 dependent manner It also suppresses the HCFC1
• ZNF350 belongs to the krueppel C2H2 type zinc finger protein family It contains 8 C2H2 type zinc fingers and 1 KRAB domain ZNF350 is a transcriptional repressor It binds to a specific sequence 5
• The function of the ZNF70 gene has not yet been determined
• EBF2 belongs to the conserved Olf EBF family of helix loop helix transcription factors
• The FLJ12644 gene encodes a protein may act as a transcriptional repressor in mitogen activated protein kinase signaling pathway to mediate cellular functions
• ZSCAN5 contains 1 SCAN box domain and may be involved in transcriptional regulation
• ZNF557 is a new candidate transcription factor
• LASS4 is a member of LASS family Members of this family regulate dihydro ceramide synthases responsible for production of sphingolipids containing different fatty acids
• ZNF322 contains four exons and spans 23 2kb in chromosome 6p22 1 region and transcribes a 2 7kb mRNA that encodes a protein with 402 amino acid residues Through northern blot analysis ZNF322 was sho
• The function of Anti ZNF419 has not yet been determined
• ZNF665 is a new candidate transcription factor Western blots using two different antibodies ARP36042 T200 and ARP36043 T200 against two unique regions of this protein target confirm the same appare
• The function of the Anti ZNF613 gene has not yet been determined
• ZNF435 contains 1 SCAN box domain and 4 C2H2 type zinc fingers It belongs to the kruppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation
• The ZNF323 gene encodes a protein that plays a role in early human embryonic development
• ZNF559 is a new candidate transcription factor
• ZNF577 is a candidate transcription factor
• ZNF607 contains 12 C2H2 type zinc fingers and may be involved in transcriptional regulation
• RAXL1 is a hypothetical protein
• CCNB3 belongs to the highly conserved cyclin family whose members are characterized by a dramatic periodicity in protein abundance through the cell cycle Cyclins function as regulators of CDK kinase
• Located on chromosome 1 this gene encodes for the DMRT like family B with proline rich C terminal 1 protein
• The HSFY1 gene encodes a member of the heat shock factor HSF family of transcriptional activators for heat shock proteins HSFY1 is a candidate gene for azoospermia since it localizes to a region o
• This gene ZNF670 which is located on chromosome 1 is predicted to encode a zinc finger protein currently with unknown fucntion
• The CREB3L1 gene encodes a protein with a transmembrane domain that is a transcriptional activator of the CREB ATF family
• Located on chromosome 17 this gene encodes for a coiled coil domain containing 16 protein with unknown function
• ZFP91 is a member of the zinc finger family of proteins The gene product contains C2H2 type domains which are the classical zinc finger domains found in numerous nucleic acid binding proteins In ad
• The protein encoded by ZFP91 is a member of the zinc finger family of proteins This protein contains C2H2 type domains which are the classical zinc finger domains found in numerous nucleic acid bind
• CTCFL is a paralog of CTCF and appears to be expressed primarily in the cytoplasm of spermatocytes CTCFL is normally expressed in a mutually exclusive pattern that correlates with resetting of methyl
• ZNF675 may be involved in transcriptional regulation It may play a role during osteoclast differentiation by modulating TRAF6 signaling activity
• Located on chromosome 19 ZNF551 encodes a protein that is part of the zinc finger protein family
• Located on chromosome 2 the LOC93349 gene encodes a hypothetical protein with unknown function
• TGIF2LY is a member of the TALE TGIF homeobox family of transcription factors This gene lies within the male specific region of chromosome Y in a block of sequence that is thought to be the result o
• This gene ZNF651 which is located on chromosome 3 is predicted to encode a zinc finger protein currently with unknown fucntion
• ZNF276 is a new candidate transcription factor
• The function of Anti ZNF276 has not yet been determined
• ZNF554 is a new candidate transcription factor
• ZNF441 contains 19 C2H2 type zinc fingers and belongs to the krueppel C2H2 type zinc finger protein family It may be involved in transcriptional regulation
• ZNF690 is a new candidate transcription factor
• The function of ZNF785 remains unknown
• ZNF645 contains 1 RING type zinc finger which is probably involved in mediating protein protein interactions
• ZNF709 may be involved in transcriptional regulation
• ZNF92 is a candidate transcription factor
• FERD3L is an evolutionarily conserved bHLH transcription factor that is identified in fly mouse and man Nervous system expression of this protein is detected in fly and mouse
• ZNF488 is a candidate transcription factor
• Gonadotropins are essential for ovarian follicular development and differentiation Gene expression of GIOT1 is restricted to the pituitary adrenal testis and ovary The kruppel associated box A do
• ESX1 is a homeobox gene related to the mouse Esx1 homeobox gene ESX1 is expressed during all stages of placental development and is localized to sparse areas of trophoblast in terminal villi in assoc
• HMG1L10 contains 2 HMG box DNA binding domains and belongs to the HMGB family It binds preferentially single stranded DNA and unwinds double stranded DNA
• ZNF610 is a new candidate transcription factor
• ZNF596 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation
• This gene ZNF683 which is located on chromosome 1 is predicted to encode a zinc finger protein currently with unknown fucntion
• ZFP42 is a transcription factors and a specific marker of undifferentiated embryonic stem ES cells
• HKR1 is one of the GLI Kruppel family of human genes Members of this family have similar H C links i e a conserved stretch of 9 amino acids connecting the C terminal histidine of one finger to the
• ZNF227 is a new candidate transcription factor
• Located on chromosome 16 the FLJ31875 gene encodes a hypothetical protein
• ZNF713 is a new candidate transcription factor
• ZNF605 may be involved in transcriptional regulation
• ZNF17 is a new candidate transcription factor
• AKAP7 encodes a member of the A kinase anchoring protein AKAP family a group of functionally related proteins that bind to a regulatory subunit RII of cAMP dependent protein kinase A PKA and ta
• AKAP7 is a member of the A kinase anchoring protein AKAP family a group of functionally related proteins that bind to a regulatory subunit RII of cAMP dependent protein kinase A PKA and target
• RECQL5 may have an important role in DNA metabolism
• CHD1L encodes a protein that interacts with ADP ribose ADP ribosylation of proteins is an important post translational modification that occurs in a variety of biological processes including DNA rep
• BAT1 is a member of the DEAD protein family of ATP dependent RNA helicases Members of this family are involved in a number of cellular functions including initiation of translation RNA splicing and
• DDX23 encodes a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of ce
• DDX42 is a member of the Asp Glu Ala Asp DEAD box protein family Members of this protein family are putative RNA helicases and are implicated in a number of cellular processes involving alteration
• DDX25 is a member of DEAD box proteins family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of cellular p
• DDX48 encodes a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of ce
• DDX47 encodes a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of ce
• Function of DDX19A has not been determined
• The function of Anti DDX49 has not yet been determined
• Werner s syndrome is a rare autosomal recessive disorder characterized by premature aging The protein encoded by this gene interacts with the N terminal portion of Werner protein containing the exonu
• DDX55 is a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of cellula
• Anti DDX55 encodes a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number
• MOV10 may be an helicase with an important function in development and or control of cell proliferation
• TMEM108 s function has not been determined yet
• DDX54 is a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of cellula
• The protein encoded by this gene is a DNA helicase A dominant negative mutant of this protein blocks chromosomal DNA replication and suggests that its function is required for S phase entry
• The piggyBac family of proteins found in diverse animals are transposases related to the transposase of the canonical piggyBac transposon from the moth Trichoplusia ni This family also includes ge
• AES is similar in sequence to the amino terminus of Drosophila enhancer of split groucho a protein involved in neurogenesis during embryonic development The protein which belongs to the groucho TLE
• Nucleobindin 2 is a calcium binding EF hand protein
• ANXA1 encodes a protein that belongs to a family of Ca 2 dependent phospholipid binding proteins which have a molecular weight of approximately 35 000 to 40 000 and are preferentially located on th
• The protein encoded by ANXA5 belongs to the annexin family of calcium dependent phospholipid binding proteins some of which have been implicated in membrane related events along exocytotic and endocyt
• ANXA8L2 is a member of the annexin family of evolutionarily conserved Ca2 and phospholipid binding proteins ANXA8L2 may function as an anticoagulant that indirectly inhibits the thromboplastin sp
• The ANXA3 gene encodes a member of the annexin family Members of this calcium dependent phospholipid binding protein family play a role in the regulation of cellular growth and in signal transduction
• This gene ANXA3 encodes a member of the annexin family Members of this calcium dependent phospholipid binding protein family play a role in the regulation of cellular growth and in signal transduct
• Annexin VI belongs to a family of calcium dependent membrane and phospholipid binding proteins Although their functions are still not clearly defined several members of the

annexin family have been
• ANXA7 is a member of the annexin family of calcium dependent phospholipid binding proteins The Annexin VII gene contains 14 exons and spans approximately 34 kb of DNA An alternatively spliced casset
• Annexin A7 is a member of the annexin family of calcium dependent phospholipid binding proteins The Annexin A7 gene contains 14 exons and spans approximately 34 kb of DNA Structural analysis of the
• ANXA11 is a member of the annexin family a group of calcium dependent phospholipid binding proteins Annexins have unique N terminal domains and conserved C terminal domains which contain the calciu
• ANXA2 encodes a member of the annexin family Members of this calcium dependent phospholipid binding protein family play a role in the regulation of cellular growth and in signal transduction pathways
• The ANXA13 gene encodes a member of the annexin family Members of this calcium dependent phospholipid binding protein family play a role in the regulation of cellular growth and in signal transductio
• ANXA13 encodes a member of the annexin family Members of this calcium dependent phospholipid binding protein family play a role in the regulation of cellular growth and in signal transduction pathway
• Gap junction protein alpha 1 is a member of the connexin gene family and a component of gap junctions Gap junctions are composed of arrays of intercellular channels and provide a route for the diffu
• This gene encodes for a Connexin 32 protein A large Charcot Marie Tooth disease family has been identified with a novel mutation in the Cx32 P2 promoter region at position 526bp Cx32 mutants that a
• Gap junctions were first characterized by electron microscopy as regionally specialized structures on plasma membranes of contacting adherent cells These structures were shown to consist of cell to c
• GJA5 belongs to the connexin family alpha type subfamily It is an integral membrane protein that forms transmembrane channels and may function in small molecule transport muscle contraction and ce
• CX31 9 is a member of the large family of connexins that are required for the formation of gap junctions Six connexin monomers form a hemichannel or connexon on the cell surface This connexon can
• Connexins are homologous four transmembrane domain proteins and major components of gap junctions The GJB4 gene encodes connexin 30 3 Cx30 3 A mutation in connexin 30 3 is causally involved in eryt
• Part of the connexin genes the CX40 1 gene appears to have evolved to different expression patterns and presumably to different functions compared to its orthologue in the mouse genome
• GJE1 is a protein component of GAP junction
• The protein encoded by MCM2 is one of the highly conserved mini chromosome maintenance proteins MCM that are involved in the initiation of eukaryotic genome replication The hexameric protein comple
• The protein encoded by MCM4 is one of the highly conserved mini chromosome maintenance proteins MCM that are essential for the initiation of eukaryotic genome replication The hexameric protein comp
• The protein encoded by the MCM6 gene is one of the highly conserved mini chromosome maintenance proteins MCM that are essential for the initiation of eukaryotic genome replication The hexameric pro
• MCM7 encodes a protein that is one of the highly conserved mini chromosome maintenance proteins MCM that are essential for the initiation of eukaryotic genome replication The hexameric protein comp
• The protein encoded by MCM5 is structurally very similar to the CDC46 protein from S cerevisiae a protein involved in the initiation of DNA replication The encoded protein is a member of the MCM fa
• MCM8 is one of the highly conserved mini chromosome maintenance proteins MCM that are essential for the initiation of eukaryotic genome replication The hexameric protein complex formed by the MCM p
• The SPP1 gene encodes an acidic matrix protein mainly expressed in mineralized tissues kidney and atherosclerotic vessels This protein also contributes to several steps in the process of prostate c
• RUNX2 is a member of the RUNX family of transcription factors and encodes a nuclear protein with an Runt DNA binding domain This protein is essential for osteoblastic differentiation and skeletal mor
• The RUNX2 gene is a member of the RUNX family of transcription factors and encodes a nuclear protein with an Runt DNA binding domain This protein is essential for osteoblastic differentiation and ske
• The protein encoded by the IBSP gene is a major structural protein of the bone matrix It constitutes approximately 12 of the noncollagenous proteins in human bone and is synthesized by skeletal asso
• GJA9 also called connexin 36 CX36 is a member of the connexin gene family that is expressed predominantly in mammalian neurons Connexins associate in groups of 6 and are organized radially around
• Annexin A4 ANXA4 belongs to the annexin family of calcium dependent phospholipid binding proteins Although their functions are still not clearly defined several members of the annexin family have
• ANXA5 belongs to the annexin family of calcium dependent phospholipid binding proteins some of which have been implicated in membrane related events along exocytotic and endocytotic pathways Annexin
• ALX4 may be rendered functionally haploinsufficient by a position effect
• ZBP1 encodes a Z DNA binding protein Z DNA formation is a dynamic process largely controlled by the amount of supercoiling
• SNAPC2 is a component of the snRNA activating protein complex SNAPc SNAPC2 is exceptionally proline rich interacts strongly with TBP and other SNAPC components is required for both RNA polymera
• TPTE encodes a putative transmembrane tyrosine phosphatase that may be involved in signal transduction pathways of the endocrine or spermatogenetic function of the testis
• CACNB2 is a subunit of voltage dependent calcium channel
• The product of the TRPM3 gene belongs to the family of transient receptor potential TRP channels TRP channels are cation selective channels important for cellular calcium signaling and homeostasis
• TRPM3 belongs to the family of transient receptor potential TRP channels TRP channels are cation selective channels important for cellular calcium signaling and homeostasis The protein encoded by
• ARX Aristaless related genes a subset of the Paired related homeobox genes is a group of regulators of essential events during vertebrate embryogenesis Arx was highly conserved between mouse and
• Bapx1 acts as a negative regulator of chondrocyte maturation The constitutive RelA activation mediated by Bapx1 controls chondrocyte viability
• C2ta is essential for transcriptional activity of the HLA class II promoter activation is via the proximal promoter No DNA binding of in vitro translated CIITA was detected C2ta may act in a coacti
• DLX4 is a member of the family of Dlx genes that are involved in early vertebrate morphogenesis notably of the head
• Elf1 belongs to the ETS family Elf1 is a transcription factor that activates the LYN and BLK promoters Elf1 may interact with other transcription factors in order to regulate specific genes Elf1 ca
• Elf3 is a transcriptional activator that binds and transactivates ETS sequences containing the consensus nucleotide core sequence GGA AT It acts synergistically with POU2F3 to transactivate the SPRR
• Foxh1 is a transcriptional activator Foxh1 can recognizes and binds to the DNA sequence 5 TGT GT GT ATT 3 and is required for induction of the goosecoid GSC promoter by TGF beta or activin signa
• GATA5 is induced at an early stage of endothelial endocardial differentiation prior to expression of such early endocardial markers as Tie2 and ErbB3 It is required for differentiation of cardiogenic
• Foxd1 is transcription factor required for formation of positional identity in the developing retina regionalization of the optic chiasm and morphogenesis of the kidney Mice lacking Foxd1 show disru
• FOXD1 belongs to the forkhead family of transcription factors which is characterized by a distinct forkhead domain FOXD1 specifically activate the 1b promoter of the RI alpha gene in testicular Sert
• HOXB1 belongs to the homeobox family The homeobox genes encode a highly conserved family of transcription factors that play an important role in morphogenesis in all multicellular organisms Mammals
• HOXB13 belongs to the homeobox family The homeobox genes encode a highly conserved family of transcription factors that play an important role in morphogenesis in all multicellular organisms Mammals
• HSF1 mediates the stress induced expression of heat shock proteins
• IRX3 is a member of the Iroquois homeobox gene family that encodes a protein known for its essential role in spinal cord development
• This is mouse homolog of ASCL2 It is a member of the basic helix loop helix BHLH family of transcription factors
• Mouse ASCL2 is a member of the basic helix loop helix BHLH family of transcription factors It is the first transcription factor shown to play a critical part in the development of the mammalian tro
• MEF2b is involved in regulation of fast Myosin heavy chain transcription
• Somite boundaries formed during embryogenesis at the interface between Notch1 activated and repressed domains Genetic and biochemical studies indicate that this interface is generated by suppressio
• Pias2 functions as an E3 type small ubiquitin like modifier SUMO ligase stabilizing the interaction between UBE2I and the substrate and as a SUMO tethering factor It plays a crucial role as a tra
• In mouse pausing of elongation complexes is mediated by the transcription termination factor TTF I bound to the Sal box terminator downstream of the rDNA transcription unit PTRF is a Pol I and tra
• RelB is a member of the Rel nuclear factor NF kappa B family of transcription factors defects in RelB affects antigen presenting cells and the formation of lymphoid organs Targeted disruption of t
• Sin3B is one of the Sin3 co repressors act as a protein scaffold to recruit transcription factors via its four highly homologous paired amphipathic helix PAH domains
• Stat1 is a member of the STAT family It can form homodimers to modulate the transcriptional repression of PPARgamma2 in adipocytes
• TFAM a mitochondrial transcription factor that is a key activator of mitochondrial transcription as well as a participant in mitochondrial genome replication Studies in mice have demonstrated that th
• Thrb is high affinity receptor for triiodothyronine
• Tob1 is a member of antiproliferative family proteins and acts as a bone morphogenic protein inhibitor as well as a suppressor of proliferation in T cells which have been implicated in postmenopausal
• Vax1 is required for axon guidance and major tract formation in the developing forebrain It may contribute to the differentiation of the neuroretina pigmented epithelium and optic stalk
• Zfpm1 is a transcription regulator that plays an essential role in erythroid and megakaryocytic cell differentiation It is essential cofactor that acts via the formation of a heterodimer with transcr
• Zfyl is a mouse Y chromosomal linked zinc finger protein which is thought to have some function during spermatogenesis
• Atf4 binds to asymmetric cAMP response elements CRE as a heterodimer and to palindromic CRE s as a homodimer
• SMYD1 is a histone methyltransferases and plays a critical role in myofibril organization during myofiber maturation
• CAMK4 plays key roles in the function and development of the cerebellu
• FOXf1a belongs to the forkhead family of transcription factors which is characterized by a distinct forkhead domain Foxf proteins are mesenchymal factors that control epithelial proliferation and sur
• Hmg20b is the component of a BHC histone deacetylase complex that contains HDAC1 HDAC2 HMG20B BRAF35 AOF2 LSD1 RCOR1 CoREST and PHF21A BHC80 Hmg20b is required for correct progression through G2
• Hoxa11 belongs to the Abd B homeobox family and contains 1 homeobox DNA binding domain Hoxa11 is a sequence specific transcription factor which is part of a developmental regulatory system that provi
• Hoxd3 is a sequence specific transcription factor which is part of a developmental regulatory system that provides cells with specific positional identities on the anterior posterior axis
• MSX1 belongs to the MSX family MSX and DLX are members of the Antennapedia class of non Hox homeodomain transcription factors that regulate gene expression and influence development of the craniofaci
• NFATC2 is a member of The NF AT family of potent transcription factors that are essential for T cell activation
• Khdrbs1 functions as a transcriptional repressor and may link cellular signaling pathways with components of the transcriptional machinery It shuttles between the nucleus and the cytoplasm in seconda
• SOX7 belongs to the SOX family of transcription factors bind PS4A and differentially modulate transcription It is a potent activator of Fgf 3 transcription
• Srebf1 is transcriptional activator that binds to the sterol regulatory element 1 SRE 1 5 ATCACCCCAC 3 Srebf1 also binds to an E box motif 5 ATCACGTGA 3 Srebf1 regulates the transcription
• Stat5 is a member of the STAT family It mediates signaling of many cytokines and growth factors
• TCEA1 participates in gene specific transcriptional activation in vivo despite the fact that it directly binds RNA polymerase II and does not recognize specific DNA sequences
• Tgfb1 is a multifunctional protein that control proliferation

differentiation and other functions in many cell types Many cells synthesize TGFB1 and essentially all of them have specific receptors
• Trp53 is a protein found in elevated levels in a great variety of transformed cells
• MTA2 modulates the enzymatic activity of the histone deacetylase core complex
• Sitpec functions as an essential component in two important signal transduction pathways and establishes a novel role for Ecsit as a cofactor for Smad proteins in the bone morphogenetic protein signal
• The activation of Toll like receptors TLRs is central to innate and adaptive immunity All TLRs use the adaptor MyD88 for signaling IRF 5 is generally involved downstream of the TLR MyD88 signallin
• MEF2a belongs to MEF2 family Members of MEF2 family of transcription factors bind a conserved A T rich sequence in the control regions of numerous muscle specific genes
• Recently it has been shown that genetic polymorphisms can result in diminished expression of CCL5 which results in increased susceptibility to and progression of infectious diseases CCL5 together
• SP1 is a transcription factor binds to GC box
• AIP may play a positive role in AHR mediated signalling possibly by influencing its receptivity for ligand and or its nuclear targeting AIP is the cellular negative regulator of the HBV X protein
• Mybbp1a may activate or repress transcription via interactions with sequence specific DNA binding proteins Repression may be mediated at least in part by histone deacetylase activity
• PDLIM1 belongs to the family of Enigma proteins Enigma proteins are a family of cytoplasmic proteins that contain an N terminal PDZ domain and a series of C terminal LIM domains
• TEF thyrotroph embryonic factor is a member of the PAR bZip proline and acidic amino acid rich basic leucine zipper transcription factor family It accumulates with robust circadian rhythms in tis
• Cited4 is a member of The CITED family proteins that bind to CBP p300 transcriptional integrators through their conserved C terminal acidic domain and function as coactivators It also interacts with
• Pus1 converts specific uridines to PSI in a number of tRNA substrates Pus1 acts on positions 27 28 in the anticodon stem and also positions 34 and 36 in the anticodon of an intron containing tRNA Pu
• RUNX3 belongs to The RUNX family of transcription factors that are important regulators of linage specific gene expression in major developmental pathways Runx3 is highly expressed in developing cran
• RBM14 is a RNA binding protein RBP RBP contribute to gene expression by regulating the form abundance and stability of both coding and non coding RNA In the vertebrate brain RBPs account for ma
• TAF1b belongs to the family of TATA box binding protein Tbp associated factors Promoter selectivity for all three classes of eukaryotic RNA polymerases is brought about by multimeric protein comple
• NIX1 holds two copies of the LXXLL motif NIX1 displays a neuronal specific expression pattern It selectively interacts with distinct nuclear receptors of the RAR and TR subfamily but does not bind
• TSG101 has a direct role in the control of growth and differentiation in primary epithelial cells It is required for normal cell function of embryonic and adult tissues but that this gene is not a tu
• IRX6 is one Iroquois Irx proteins comprise a family of homeodomain containing transcription factors involved in patterning and regionalization of embryonic tissues in both vertebrates and invertebra
• Pycard is a cytosolic soluble protein that forms insoluble aggregates and enhances etoposide induced apoptosis
• Nkx2 4 belongs to the NK 2 homeobox family and contains 1 homeobox DNA binding domain In the embryo it is detected in the posterior hypotalamus and later in the head In the adult it is detected only
• Elongator is a histone acetyltransferase complex that associates with the elongating form of RNA polymerase II The human homologue of yeast ELP4 is a component of enlogator and show that this gene is
• Mouse Foxi class genes may play important roles both during cranial placode specification and in later development of individual cranial sensory structures and other organs derived from the cranial e
• Tsc22d4 may be a transcriptional repressor
• Yaf2 binds to MYC and inhibits MYC mediated transactivation Yaf2 also binds to MYCN and enhances MYCN dependent transcriptional activation Yaf2 increases calpain 2 mediated proteolysis of YY1 in vit
• MEF2C is a transcription regulator of slow fiber
• cDNA library was prepared and sequenced in Mouse Genome Encyclopedia Project of Genome Exploration Research Group in Riken Genomic Sciences Center and Genome Science laboratory in RIKEN
• This protein was disclosed by the RIKEN Mouse Gene Encyclopaedia Project a systematic approach to determining the full coding potential of the mouse genome involves collection and sequencing of full
• Isl2 specifies RGC laterality by repressing an ipsilateral pathfinding program unique to VTC RGCs and involving Zic2 and EphB1 This genetic hierarchy controls binocular vision
• HSFY2 is Y linked hear shock transcription factor
• Dmrtc2 may be involved in sexual development
• BRF1 is one of the three subunits of the RNA polymerase III transcription factor complex This complex plays a central role in transcription initiation by RNA polymerase III on genes encoding tRNA 5S
• TTC19 s fucntion is not fully determined yet
• PARN is an oligomeric processive and cap interacting 3 exonuclease
• Foxp4 is a member of the murine forkhead family of transcription factors It is expressed exclusively in the epithelial cells of the developing intestine where in late development it is expressed i
• Rnf6 is involved in neuronal development High Rnf6 protein levels can be detected in developing axonal projections of motor and DRG neurons during mouse embryogenesis
• TAF7l belongs to the family of TATA box binding protein Tbp associated factors Promoter selectivity for all three classes of eukaryotic RNA polymerases is brought about by multimeric protein comple
• Spz1 a novel bHLH Zip protein is specifically expressed in testis
• TCFap4 belongs to the basic helix loop helix bHLH family are involved in various cell differentiation processes
• The alpha actinins are a multigene family of four actin binding proteins related to dystrophin The two skeletal muscle isoforms of alpha actinin ACTN2 and ACTN3 are major structural components of t
• TCF23 belongs to the basic helix loop helix bHLH family are involved in various cell differentiation processes
• Transcription factors containing a homeodomain play an important role in the organogenesis of vertebrates Otx3 Dmbx1 is a member of paired class homeodomain transcription factors Otx3 Dmbx1 represse
• ZFP287 is the product of Th2 specific gene It may play a role in selective regulation of cytokine genes in T cells
• PPARGC1b is a coactivator of nuclear receptors and other transcription factors that regulates several components of energy metabolism particularly certain aspects of adaptive thermogenesis in brown f
• Creb1 binds the cAMP response element CRE a sequence present in many viral and cellular promoters CREB stimulates transcription on binding to the CRE
• Nfat5 contains 1 RHD Rel like domain and plays a role in the inducible expression of genes Nfat5 regulates hypertonicity induced cellular accumulation of osmolytes Nfat5 might be capable of formi
• Mouse Ces6 is a member of carboxylesterases family Carboxylesterases are enzymes that catalyze the hydrolysis of a wide range of ester containing endogenous and xenobiotic compounds
• NR5A1 is an important regulator of steroidogeneisis which is present in human skin and its appendages It plays a role in regulating p450scc expression with TReP 132 and CBP p300
• DNAJC17 s function is not determined
• GATAD2A s function is not determined
• Obox6 belongs to the Obox family that represents a new family of tissue specific homeobox genes preferentially expressed in gonads
• DMRT2 is a protein disclosed by NIH MGC program
• EBF4 is a member of Olf 1 Ebf like gene family It is expressed in the neuronal and basal cell layers of olfactory epithelium and may interact with other O E family members to regulate gene expression
• MKL1 transduces cytoskeletal signals and induces smooth muscle cell differentiation from undifferentiated embryonic stem cells
• The homeobox gene superfamily is highly conserved during evolution These members act as transcription factors during several important developmental processes through their 60 amino acid homeodomains
• Mouse Cbfa2t2h associates with mSin3A N CoR and histone deacetylase 3 and that when tethered to DNA it acts as a transcriptional corepressor
• ZBTB38 physically associates with CtBP via a conserved CtBP binding motif PLDLR When heterologously targeted to DNA it represses transcription via two independent repression domains an N terminal
• Dmrta1 is a DM domain containing transcription factors DM dsx and mab 3 and may be involved in sexual development
• E2F7 is a E2F family member It can block the E2F dependent activation of a subset of E2F target genes as well as mitigate cellular proliferation of mouse embryo fibroblasts
• Homez and members of ZHX family of zinc finger homeodomain factors are delineated as a subset within the superfamily of homeobox containing proteins
• Murine Foxo6 is a member of the murine forkhead family of transcription factors This family consists of over 30 members the vast majority of which is important in embryonic development These forkhe
• Homeobox proteins are transcription factors notable for their ability to regulate embryogenesis The reproductive homeobox proteins Rhox are expressed in a cell type specific manner several are hor
• POU2F1 is a member of the POU family that represents the double stranded DNA binding proteins specifically binding to the block C region
• TRF3 is a nuclear protein that is present in all human and mouse tissues and cell lines examined
• Foxk1 is a transcriptional regulator that binds to the upstream enhancer region CCAC box of myoglobin gene It has a role in myogenic differentiation and in remodeling processes of adult muscles tha
• ZHX2 a zinc fingers and homeoboxes ZHX protein that is directly involved in the regulation of AFP synthesis It also controls AFP levels only indirectly e g by regulating the synthesis of a hormo
• NR1H5 belongs to nuclear receptors that are ligand modulated transcription factors
• The amino acid sequence of 1110033M05Rik is derived from an annotated genomic sequence NT 039589 using gene prediction method GNOMON supported by mRNA and EST evidence
• Cell surface heparan sulfate proteoglycans are composed of a membrane associated protein core substituted with a variable number of heparan sulfate chains Members of the glypican related integral mem
• L type calcium channels are composed of five subunits The protein encoded by this gene represents one of these subunits gamma and is one of several gamma subunit proteins This particular gamma sub
• The functions of Voltage gated potassium Kv channels include regulating neurotransmitter release heart rate insulin secretion neuronal excitability epithelial electrolyte transport smooth muscl
• KCNK5 is one of the members of the superfamily of potassium channel proteins containing two pore forming P domains KCNK5 is mainly expressed in the cortical distal tubules and collecting ducts of the
• ACCN5 belongs to the amiloride sensitive Na channel and degenerin NaC DEG family members of which have been identified in many animal species ranging from the nematode to human The amiloride sens
• SCN3B is one member of the sodium channel beta subunits of voltage gated sodium channels which are responsible for the generation and propagation of action potentials in neurons and muscle SCN3B inf
• KCTD6 is a domain of potassium channel
• G3BP is one of the DNA unwinding enzymes which prefers partially unwound 3 tailed substrates and can also unwind partial RNA DNA and RNA RNA duplexes in an ATP dependent fashion This enzyme is a mem
• DDX46 is a member of the DEAD box protein family DEAD box proteins characterized by the conserved motif Asp Glu Ala Asp DEAD are putative RNA helicases They are implicated in a number of cellula
• SSX2 belongs to the family of highly homologous synovial sarcoma X SSX breakpoint proteins These proteins may function as transcriptional repressors They are also capable of eliciting spontaneousl
• ZNF35 is a new candidate transcription factor
• EDF1 encodes a protein that may regulate endothelial cell differentiation It has been postulated that the protein functions as a bridging molecule that interconnects regulatory proteins and the basal
• LDB1 is a transcriptional activator that associates with the LIM homeoproteins and coordinate transcription LIM homeoproteins and LDBs are involved in a variety of developmental processes
• MED21 belongs to the Mediator complex subunit 21 family It is the component of the Mediator complex a coactivator involved in the regulated transcription of nearly all RNA polymerase II dependent ge
• MTA1 encodes a protein that was identified in a screen for genes expressed in metastatic cells specifically mammary adenocarcinoma cell lines Expression of this gene has been correlated with the me
• ZNF265 is a protein that has been shown to bind to the spliceosomal components U1 70K and U2AF35 and to direct alternative splicing Analysis of the structure reveals substantial similarity to known R
• The function of ZNF337 has not yet been determined

• The function of Anti ZFP589 has not yet been determined
• Two members of the human zinc finger Kruppel family ZNF 12 KOX 3 and ZNF 26 KOX 20 have been localized by somatic cell hybrid analysis and in situ chromosomal hybridization The presence of indi
• NSBP1 binds specifically to nuclear core particles and acts as a transcriptional activator
• The gene for human Kruppel related protein 4 gene symbol GLI4 a zinc finger protein of unknown function has been localized by fluorescence in situ hybridization to chromosome 8q24 3 distal to c
• Mouse Alx3 is a homeobox gene that is related to the Drosophila aristaless gene and to a group of vertebrate genes including Prx1 Prx2 Cart1 and Alx4 The protein encoded contains a diverged varian
• KLF2 Lung Kruppel like factor LKLF functions as a negative regulator of adipocyte differentiation KLF2 is highly expressed in adipose tissue and studies in cell lines and primary cells demonstrate
• PPARdelta is pivotal to control the program for fatty acid oxidation in the skeletal muscle thereby ameliorating obesity and insulin resistance through its activation in obese animals
• Sfpi1 binds to the PU box a purine rich DNA sequence 5 GAGGAA 3 that can act as a lymphoid specific enhancer Sfpi1 is a transcriptional activator that may be specifically involved in the differe
• Spic represents a subgroup within the Ets protein family It is a regulatory molecule during a specific phase of B lymphoid development
• Lmx1a acts as a transcriptional activator by binding to an A T rich sequence the FLAT element in the insulin gene promoter It is required for development of the roof plate and in turn for specifi
• FBN1 is a member of the fibrillin family FBN1 is a large extracellular matrix glycoprotein that serve as a structural component of 10 12 nm calcium binding microfibrils These microfibrils provide f
• MITF is a transcription factor that contains both basic helix loop helix and leucine zipper structural features It regulates the differentiation and development of melanocytes retinal pigment epithel
• SOX9 recognizes the sequence CCTTGAG along with other members of the HMG box class DNA binding proteins It acts during chondrocyte differentiation and with steroidogenic factor 1 regulates transcri
• RFX5 is the fifth member of the growing family of DNA binding proteins sharing a novel and highly characteristic DNA binding domain called the RFX motif RFX is a nuclear protein complex that binds to
• Basic helix loop helix bHLH transcription factors have been implicated in cell lineage determination and differentiation TWIST1 is a bHLH transcription factor and shares similarity with another bHLH
• BACH1 is a transcription factor that belongs to the cap n collar type of basic region leucine zipper factor family CNC bZip The encoded protein contains broad complex tramtrack bric a brac poxvir
• PRDM1 is a protein that acts as a repressor of beta interferon gene expression The protein binds specifically to the PRDI positive regulatory domain I element of the beta IFN gene promoter Transcr
• BTF3 forms a stable complex with RNA polymerase IIB and is required for transcriptional initiation Alternative splicing results in multiple transcript variants encoding different isoforms This gene
• LDB2 is a transcriptional activator that associates with the LIM homeoproteins and coordinate transcription LIM homeoproteins and LDBs are involved in a variety of developmental processes
• ENO1 is one of three enolase isoenzymes found in mammals its gene encodes alpha enolase a homodimeric soluble enzyme and also encodes a shorter monomeric structural lens protein tau crystallin Th
• FOXJ1 is a member of forkhead winged helix transcription factor family which play crucial roles during vertebrate development FOXJ1 may play an important role in cell fate determination during lung
• FOXO3A belongs to the forkhead family of transcription factors which are characterized by a distinct forkhead domain This gene likely functions as a trigger for apoptosis through expression of genes
• ATF4s a transcription factor that was originally identified as a widely expressed mammalian DNA binding protein that could bind a tax responsive enhancer element in the LTR of HTLV 1 The encoded prot
• Core binding factor CBF is a heterodimeric transcription factor that binds to the core element of many enhancers and promoters RUNX1 is the alpha subunit of CBF and is thought to be involved in the
• DR1 is a TBP TATA box binding protein associated phosphoprotein that represses both basal and activated levels of transcription DR1 is phosphorylated in vivo and this phosphorylation affects its i
• E2F3 is a member of the E2F family of transcription factors The E2F family plays a crucial role in the control of cell cycle and action of tumor suppressor proteins and is also a target of the transf
• The nerve growth factor induced clone C NGFI C EGR4 gene is a zinc finger transcription factor that is rapidly induced by nerve growth factor in rat pheochromocytoma PC12 cells and by seizure in bra
• FOXO1A belongs to the forkhead family of transcription factors which are characterized by a distinct forkhead domain The specific function of FOXO1A has not yet been determined however it may play
• Friend leukemia integration 1 Fli 1 is a member of the ETS family of transcriptional regulatory proteins that contain a highly conserved and structurally unique DNA binding ETS domain
• GATA4 is a member of the GATA family of zinc finger transcription factors Members of this family recognize the GATA motif which is present in the promoters of many genes GATA4is thought to regulate
• Human T cell leukemia virus enhancer factor HTLF may be a forkhead domain binding protein It may function in the transcriptional regulation of the human T cell leukemia virus long terminal repeat H
• Human T cell leukemia virus enhancer factor HTLF may be a forkhead domain binding protein It may function in the transcriptional regulation of the human T cell leukemia virus long terminal repeat
• IRF1 is interferon regulatory factor 1 a member of the interferon regulatory transcription factor IRF family IRF1 serves as an activator of interferons alpha and beta transcription and in mouse i
• IRF5 is a member of the interferon regulatory factor IRF family a group of transcription factors with diverse roles including virus mediated activation of interferon and modulation of cell growth
• JUN gene is the putative transforming gene of avian sarcoma virus 17 I JUN is highly similar to the viral protein and interacts directly with specific target DNA sequences to regulate gene expressio
• The c Jun proto oncogene was first identified as the cellular homolog of the avian sarcoma virus v Jun oncogene The c Jun protein along with c Fos is a component of the AP 1 transcriptional complex
• LMX1B is essential for the specification of dorsal limb fate at both the zeugopodal and autopodal levels Defects in LMX1B are the cause of nail patella syndrome NPS also knowan as Onychoosteodyspla
• Homeobox genes of which the most well characterized category is represented by the HOX genes play a crucial role in normal development In addition several homeoproteins are involved in neoplasia
• Cyclin dependent kinases CDKs which play an essential role in cell cycle control of eukaryotic cells are phosphorylated and thus activated by the CDK activating kinase CAK CAK is a multisubunit
• MSX2 is a member of the muscle segment homeobox gene family The protein is a transcriptional repressor whose normal activity may establish a balance between survival and apoptosis of neural crest der
• NPAS2 is a member of the basic helix loop helix bHLH PAS family of transcription factors A similar mouse protein may play a regulatory role in the acquisition of specific types of memory It also m
• PSMC2 is one of the ATPase subunits This subunit has been shown to interact with several of the basal transcription factors so in addition to participation in proteasome functions this subunit may
• The 26S proteasome is a multicatalytic proteinase complex with a highly ordered structure composed of 2 complexes a 20S core and a 19S regulator The 19S regulator is composed of a base which contai
• RCV1 is a member of the recoverin family of neuronal calcium sensors RCV1 contains three calcium binding EF hand domains and may prolong the termination of the phototransduction cascade in the retina
• RING1 can bind DNA and can act as a transcriptional repressor It is associated with the multimeric polycomb group protein complex The gene product interacts with the polycomb group proteins BMI1 ED
• SATB1 binds to DNA at special AT rich sequences at nuclear matrix or scaffold associated regions SATB1 was thought to recognize the sugar phosphate structure of double stranded DNA
• SMARCD2 is a member of the SWI SNF family of proteins whose members display helicase and ATPase activities and which are thought to regulate transcription of certain genes by altering the chromatin s
• SMARCE1 is part of the large ATP dependent chromatin remodeling complex SWI SNF which is required for transcriptional activation of genes normally repressed by chromatin The protein either alone or
• SNAPC1 is a part of the SNAPc complex required for the transcription of both RNA polymerase II and III small nuclear RNA genes It binds to the proximal sequence element PSE a non TATA box basal pr
• SOX2 is a member of the SRY related HMG box SOX family of transcription factors involved in the regulation of embryonic development and in the determination of cell fate The protein may act as a tr
• SOX11 is a member of the SOX SRY related HMG box family of transcription factors involved in the regulation of embryonic development and in the determination of the cell fate The protein may act as
• SSRP1 is a subunit of a heterodimer that along with SUPT16H forms chromatin transcriptional elongation factor FACT FACT interacts specifically with histones H2A H2B to effect nucleosome disassembly
• STAT6 is a member of the STAT family of transcription factors In response to cytokines and growth factors STAT family members are phosphorylated by the receptor associated kinases and then form hom
• SUPT6H may be functionally analogous to SPT6 and emb 5 and may therefore regulate transcription through establishment or maintenance of chromatin structure Spt6 may also participates in the regulatio
• TCF3 contains 1 basic helix loop helix bHLH domain Heterodimers between TCF3 and tissue specific basic helix loop helix bHLH proteins play major roles in determining tissue specific cell fate dur
• TCF12 encodes a protein that is a member of the basic helix loop helix bHLH E protein family which recognizes the consensus binding site E box CANNTG This encoded protein is expressed in many tis
• TEAD4 is a member of the transcriptional enhancer factor TEF family The family members contain the TEA ATTS DNA binding domain TEAD4 is preferentially expressed in skeletal muscle and binds to th
• YY1 is a ubiquitously distributed transcription factor belonging to the GLI Kruppel class of zinc finger proteins The protein is involved in repressing and activating a diverse number of promoters Y
• ZFP36 is a probable regulatory protein with a novel zinc finger structure involved in regulating the response to growth factors Knockdown of ZFP36 increased both cognate macrophage gene mRNAs and inf
• ZNF264 may be involved in transcriptional regulation
• ZNF76 belongs to the krueppel C2H2 type zinc finger protein family ZNF76 is a general transcription repressor targeting TATA binding protein TBP through a process regulated by sumoylation ZNF76 i
• ZNF124 belongs to the krueppel C2H2 type zinc finger protein family ZNF124 may be involved in transcriptional regulation
• ZNF177 contains 1 KRAB domain and 7 C2H2 type zinc fingers ZNF177 belongs to the krueppel C2H2 type zinc finger protein family And it may be involved in transcriptional regulation
• HMGA2 belongs to the non histone chromosomal high mobility group HMG protein family HMG proteins function as architectural factors and are essential components of the enhancesome This protein cont
• ZNF282 contains 1 KRAB domain and 5 C2H2 type zinc fingers It belongs to the krueppel C2H2 type zinc finger protein family and binds to the U5 repressive element U5RE of the human T cell leukemia v
• EED is a member of the Polycomb group PcG family PcG family members form multimeric protein complexes which are involved in maintaining the transcriptional repressive state of genes over successiv
• The adenovirus E1A protein both activates and represses gene expression to promote cellular proliferation and inhibit differentiation CREG1 antagonizes transcriptional activation and cellular transfo
• Histone acetylation plays a key role in the regulation of eukaryotic gene expression Histone acetylation and deacetylation are catalyzed by multisubunit complexes SAP30 is a component of the histone
• ZNF259 may be a signaling molecule that communicates mitogenic signals from the cytoplasm to the nucleus ZNF259 binds to the EGFR and is released from the receptor after activation ZNF259 is essentia
• BUD31 may be a nuclear regulator of transcription
• NFKB1 is a 105 kD protein which can undergo cotranslational processing by the 26S proteasome to produce a 50 kD protein The 105 kD protein is a Rel protein specific transcription inhibitor and the 50
• The TRAF tumor necrosis factor receptor associated factor family of proteins associate with and transduce signals from members of the tumor necrosis factor receptor superfamily TANK is found in the
• PTTG1 is a homolog of yeast securing proteins which

prevent separins from promoting sister chromatid separation It is an anaphase promoting complex APC substrate that associates with a separin unt

• he activation of gene transcription is a multistep process that is triggered by factors that recognize transcriptional enhancer sites in DNA These factors work with co activators to direct transcript

• RNF14 contains a RING zinc finger a motif known to be involved in protein protein interactions This protein interacts with androgen receptor AR and may function as a coactivator that induces AR ta

• EZH2 is a member of the Polycomb group PcG family PcG family members form multimeric protein complexes which are involved in maintaining the transcriptional repressive state of genes over successi

• HOXB7 is a member of the Antp homeobox family and is a protein with a homeobox DNA binding domain The nuclear protein functions as a sequence specific transcription factor that is involved in cell pr

• OVOL1 is a protein highly similar to Drosophila and mouse proteins In Drosophila the ovo protein plays a critical role in Drosophila oogenesis and cuticle formation In mice the ovo like protein is i

• TCF15 is found in the nucleus and may be involved in the early transcriptional regulation of patterning of the mesoderm The basic helix loop helix protein requires dimerization with another basic hel

• CDK5 is probably involved in the control of the cell cycle CDK5 can interact with d1 and d3 type G1 cyclins and phosphorylate histone H1 tau MAP2 and NF H and NF M CDK5 can also interacts with p35

• Histone acetylation and deacetylation catalyzed by multisubunit complexes play a key role in the regulation of eukaryotic gene expression HDAC1 belongs to the histone deacetylase acuc apha family a

• MDS1 is located at 3q26 170 400 kb upstream telomeric of EVI1 in the chromosomal region in which some of the breakpoints 5 of EVI1 have been mapped MDS1 has been identified as a single gene as wel

• NRF1 is a phosphorylated nuclear protein with a bZIP domain This protein homodimerizes and functions as a transcription factor that activates the expression of some key metabolic genes regulating cel

• XBP1 is a transcription factor that regulates MHC class II genes by binding to a promoter element referred to as an X box XBP1 is a bZIP protein which was also identified as a cellular transcription

• NR1D2 can interact with NCOA5 coactivator leading to a strong increase of transcription of target genes NR1D2 also binds to the sequences 5 AATGTAGGTCA 3 and 5 ATAACTAGGTCA 3 and acts as a poten

• DLX5 is a member of a homeobox transcription factor gene family similiar to the Drosophila distal less gene This protein may play a role in bone development and fracture healing Mutation in its gene

• FOXG1B belongs to the forkhead family of transcription factors which is characterized by a distinct forkhead domain The specific function of FOXG1B has not yet been determined however it may play a

• Goosecoidlike GSCL resides in the critical region for VCFS DGS on 22q11 Velocardiofacial syndrome VCFS is phenotypically related to DiGeorge syndrome DGS and both syndromes are associated with

• HCFC1 is a member of the host cell factor family with five Kelch repeats a fibronectin like motif and six HCF repeats each of which contains a highly specific cleavage signal This nuclear coactiva

• JUND is a member of the JUN family and a functional component of the AP1 transcription factor complex JUND has been proposed to protect cells from p53 dependent senescence and apoptosis Alternate t

• MAGEA9 is a member of the MAGEA gene family The members of this family encode proteins with 50 to 80 sequence identity to each other The promoters and first exons of the MAGEA genes show considerab

• MYB may be a transcriptional activator DNA binding protein that specifically recognize the sequence 5 YAAC GT G 3 It plays an important role in the control of proliferation and differentiation of

• NFIL3 is a basic leucine zipper bZIP transcription factor that represses or activates transcription in non osteoblastic cells NFIL3 play a role in attenuation of PTH target gene transcription in os

• TP53BP2 is a member of the ASPP apoptosis stimulating protein of p53 family of p53 interacting proteins TP53BP2 contains four ankyrin repeats and an SH3 domain involved in protein protein interacti

• TP73 belongs to the p53 family It participates in the apoptotic response to DNA damage When overproduced it activates transcription from p53 responsive promoters and induces apoptosis TP53 may be

• Initiation of transcription by RNA polymerase I requires the formation of a complex composed of the TATA binding protein TBP and three TBP associated factors TAFs specific for RNA polymerase I Th

• TSFM associates with the EF Tu GDP complex and induces the exchange of GDP to GTP It remains bound to the aminoacyl tRNA EF Tu GTP complex up to the GTP hydrolysis stage on the ribosome TSFM localiz

• The A kinase anchor proteins AKAPs are a group of structurally diverse proteins which have the common function of binding to the regulatory subunit of protein kinase A PKA and confining the holoen

• TRIM28 mediates transcriptional control by interaction with the Kruppel associated box repression domain found in many transcription factors TRIM28 localizes to the nucleus and is thought to associat

• Histone acetylation plays a key role in the regulation of eukaryotic gene expression Histone acetylation and deacetylation are catalyzed by multisubunit complexes SAP18 is a component of the histone

• SMAD2 belongs to the SMAD a family of proteins similar to the gene products of the Drosophila gene mothers against decapentaplegic Mad and the C elegans gene Sma SMAD proteins are signal transd

• MEOX2 may play a role in the regulation of vertebrate limb myogenesis Mutations in the related mouse protein may be associated with craniofacial and or skeletal abnormalities in addition to neurovas

• KBTBD10 contains 1 BTB POZ domain and is required for pseudopod elongation in transformed cells KBTBD10 mRNA is up regulated by less than two folds in the heart in human patients with HCM

• NFE2 MIM 601490 NFE2L1 MIM 163260 and NFE2L2 comprise a family of human genes encoding basic leucine zipper bZIP transcription factors They share highly conserved regions that are distinct fr

• NFYB is one subunit of a trimeric complex forming a highly conserved transcription factor that binds with high specificity to CCAAT motifs in the promoter regions in a variety of genes This gene pro

• PEG3 induces apoptosis in cooperation with SIAH1A and acts as a mediator between TP53 p53 and BAX in a neuronal death pathway that is activated by DNA damage PEG3 acts synergistically with TRAF2 and

• POU4F1 is a class IV POU domain containing transcription factor highly expressed in the developing sensory nervous system and in cells of the B and T lymphocytic lineages BRN3A POU4F1 is a class IV

• DPF2 is a member of the d4 domain family characterized by a zinc finger like structural motif DPF2 functions as a transcription factor which is necessary for the apoptotic response following depriva

• TSG101 belongs to a group of apparently inactive homologs of ubiquitin conjugating enzymes TSG101 contains a coiled coil domain that interacts with stathmin a cytosolic phosphoprotein implicated in

• HTATIP belongs to the MYST family of histone acetyl transferases HATs and was originally isolated as an HIV 1 TAT interactive protein HATs play important roles in regulating chromatin remodeling t

• This gene was identified as an estrogen and anti estrogen regulated gene in epithelial cells stably expressing estrogen receptor TRIM16 contains two B box domains and a coiled coiled region that are

• TFEB is a member of the basic Helix Loop Helix Zipper family of transcription factors TFEB can bind DNA as a homodimer or as a heterodimer with three closely related family members MITF TFE3 and TF

• CTCF is a transcriptional regulator protein with 11 highly conserved zinc finger ZF domains This nuclear protein is able to use different combinations of the ZF domains to bind different DNA target

• USP39 may play a role in mRNA splicing It is unsure if the protein really exhibits hydrolase activity It could be a competitor of ubiquitin C terminal hydrolases

• PLAGL1 is a C2H2 zinc finger protein with transactivation and DNA binding activity This gene has been shown to exhibit antiproliferative activities and is a tumor suppressor gene candidate This gene

• Leucine zipper like transcriptional regulator 1 LZTR1 is belived to be a DNA binding protein and transcriptional regulator based on its predicted structural characteristics The transcript is presen

• ZBTB33 is a transcriptional regulator with bimodal DNA binding specificity ZBTB33 binds to methylated CpG dinucleotides in the consensus sequence 5 CGCG 3 and also binds to the non methylated conse

• The human ATF7 ATFa proteins belong to the ATF CREB family of transcription factors They mediate the transcriptional activation by the largest E1a protein and can heterodimerize with members of the

• ZNF16 contains a C2H2 type of zinc finger and thus may function as a transcription factor The protein encoded by this gene contains a C2H2 type of zinc finger and thus may function as a transcriptio

• ZNF17 is a new candidate transcription factor

• CORO1A is a novel actin binding protein with a WD repeat and a leucine zipper motif CORO1A forms homodimers that the association is mediated by the leucine zipper structure in the C terminal region

• The LX receptors LXRs were originally identified as orphan members of the nuclear receptor superfamily because their ligands were unknown Like other receptors in the family LXRs heterodimerize wit

• ZNF195 contains 1 KRAB domain and 10 C2H2 type zinc fingers and belongs to the krueppel C2H2 type zinc finger protein family It may be involved in transcriptional regulation

• This gene was identified based on its homology to the gene encoding the RING3 protein a serine threonine kinase The gene localizes to 9q34 a region which contains several major histocompatibility c

• CIZ1 may regulate the subcellular localization of CIP WAF1

• The AATF is a protein that was identified on the basis of its interaction with MAP3K12 DLK a protein kinase known to be involved in the induction of cell apoptosis AATF contains a leucine zipper wh

• DUX5 an active gene of unusual structure is present in chromosome regions characterized by large amounts of heterochromatic repetitive DNA

• FOXB1 is a winged helix forkhead transcription factor FOXB1 is specifically expressed in the developing central nervous system CNS Early embryonic FOXB1 expression is restricted to the mammiliary

• Nuclear factor of activated T cells NFAT is a transcription factor required for T cell expression of interleukin 2 NFAT binds to a sequence in the IL2 enhancer known as the antigen receptor respons

• ILF3 may facilitate double stranded RNA regulated gene expression at the level of post transcription ILF3 can act as a translation inhibitory protein which binds to coding sequences of acid beta gluc

• RYBP contains 1 RanBP2 type zinc finger RYBP may be implicated in the regulation of the transcription as a repressor of the transcriptional activity of E4TF1 In tumor cell lines it may induce apopt

• HMG box containing protein 1 HBP1 is a member of the high mobility group HMG of chromosomal proteins It is a sequence specific HMG transcription factor Several features of HBP1 suggest an intrig

• MAFF is a basic leucine zipper bZIP transcription factor that lacks a transactivation domain It is known to bind the US 2 DNA element in the promoter of the oxytocin receptor OTR gene and most li

• MYST4 is a histone acetyltransferase which may be involved in both positive and negative regulation of transcription It is required for RUNX2 dependent transcriptional activation and May be involved

• GMEB2 is a member of KDWK family GMEB2 associates with GMEB1 protein and the complex is essential for parvovirus DNA replication Study of rat homolog implicates the role of this gene in modulation

• STAT5B is a member of the STAT family of transcription factors In response to cytokines andgrowth factors STAT family members are phosphorylated by the receptor associated kinases and then form hom

• IKZF3 is a member of the Ikaros family of zinc finger proteins It is a transcription factor that is important in the regulation of B lymphocyte proliferation and differentiation Both Ikaros and Aiol

• BRD7 is an activator of the Wnt signaling pathway in a DVL1 dependent manner by negatively regulating the GSK3B phosphotransferase activity It induces dephosphorylation of GSK3B at Tyr 216

• CNOT7 binds to an anti proliferative protein B cell translocation protein 1 which negatively regulates cell proliferation Binding of the two proteins which is driven by phosphorylation of the anti

• SMARCAL1 is a member of the SWI SNF family of proteins Members of this family have helicase and ATPase activities and are thought to regulate transcription of certain genes by altering the chromatin

• NFYC is one subunit of a trimeric complex forming a highly conserved transcription factor that binds with high specificity to CCAAT motifs in the promoter regions in a variety of genes NFYC forms a

• NR2E3 is part of a large family of nuclear receptor transcription factors involved in signaling pathways Nuclear receptors have been shown to regulate pathways involved in embryonic development as w

• CBX6 is involved in maintaining the transcriptionally repressive state of genes It modifies chromatin rendering it heritably changed in its expressibility

• BRD4 is homologous to the murine protein MCAP which associates with chromosomes during mitosis and to the

human RING3 protein a serine threonine kinase Each of these proteins contains two bromodom

• AKAP8L could play a role in constitutive transport element CTE mediated gene expression It does not seem to be implicated in the binding of regulatory subunit II of PKA It may be involved in nucle

• OTX1 is a member of the bicoid sub family of homeodomain containing transcription factors OTX1 acts as a transcription factor and may play a role in brain and sensory organ development A similar pro

• Proteins that contain a CXXC motif within their DNA binding domain such as CXXC1 recognize CpG sequences and regulate gene expression Proteins that contain a CXXC motif within their DNA binding doma

• SART3 is an RNA binding nuclear protein that is a tumor rejection antigen This antigen possesses tumor epitopes capable of inducing HLA A24 restricted and tumor specific cytotoxic T lymphocytes in ca

• KIAA0737 contains 1 HMG box DNA binding domain and the function remains unknows

• RNF10 contains a ring finger motif which is known to be involved in protein protein interactions The specific function of this protein has not yet been determined EST data suggests the existence of

• The members of the zinc fingers and homeoboxes gene family are nuclear homodimeric transcriptional repressors that interact with the A subunit of nuclear factor Y NF YA and contain two C2H2 type zin

• HIon Channel2 contains 5 C2H2 type zinc fingers and 1 BTB POZ domain It belongs to the krueppel C2H2 type zinc finger protein family Hic subfamily and is a transcriptional repressor

• POGZ appears to be a zinc finger protein containing a transposase domain at the C terminus This protein was found to interact with the transcription factor SP1 in a yeast two hybrid systemThe protein

• RCOR1 is a functional corepressor required for regulation of neural specific gene expression The RCOR gene encodes a functional corepressor required for regulation of neural specific gene expression T

• RCOR1 is a functional corepressor required for regulation of neural specific gene expression The RCOR gene encodes a functional corepressor required for regulation of neural specific gene expression

• PPARG is a regulator of adipocyte differentiation Additionally PPAR gamma has been implicated in the pathology of numerous diseases including obesity diabetes atherosclerosis and cancer The protei

• ZFR is critical for Staufen 2 isoform specific nucleocytoplasmic shuttling in neurons

• TLX2 contains 1 homeobox DNA binding domain and the function remains unknown

• ZNF580 may be involved in transcriptional regulation

• ERGIon Channel2 possibly play role in transport between endoplasmic reticulum and Golgi ERGIon Channel2 is downregulated in prostate cancer ERGIon Channel2 may play an important role in the growth a

• JMJD1B contains a highly conserved C terminus containing a zinc finger with the unique spacing Cys X2 Cys X7 His X2 Cys X2 Cys X4 Cys X2 Cys and a jmjC domain It plays an important role in histone de

• ZNF446 contains a KRAB and three C 2 H 2 zinc fingers ZNF446 is a transcription repressor when fused to GAL4 DNA binding domain and co transfected with VP 16 Overexpression of ZNF446 in COS 7 cel

• ZNF312 is a candidate transcription factor

• ZFP64 belongs to the krueppel C2H2 type zinc finger protein family It contains 9 C2H2 type zinc fingers and may be involved in transcriptional regulation

• The function of KLHL26 remains unknown

• ZNF444 has a domain structure and amino acid sequence similar to several zinc finger transcription factors which strongly suggested that ZNF444 is a transcriptional regulator ZNF444 has a domain stru

• ZNF286 may be involved in transcriptional regulation

• Hp66beta GATAD2B previously identified as a component of the human MeCP1 complex is a second member of a p66 gene family hp66alpha and hp66beta Both proteins interact with MBD2 and MBD3 functio

• NR2F2 an orphan member of the nuclear hormone receptor superfamily acts as a transcriptional repressor by antagonizing the functions of other nuclear hormone receptors and by actively silencing tran

• LZTS1 involved in the regulation of cell growth LZTS1 may stabilize the active CDC2 cyclin B1 complex and thereby contribute to the regulation of the cell cycle and the prevention of uncontrolled cel

• ZNF253 may function as a transcription factor Seem to have a transcriptional repression activity

• PRDM12 belongs to PR domain family which are involved in human cancers in an unusual yin yang fashion Two products are normally produced from a PR domain family member which differ by the presence or

• FOXA2 is a member of the forkhead class of DNA binding proteins These hepatocyte nuclear factors are transcriptional activators for liver specific transcripts such as albumin and transthyretin and t

• The function of the ZNF70 has not yet been determined

• FOXM1 contains 1 fork head DNA binding domain FOXM1 is a transcriptional activatory factor It may play a role in the control of cell proliferation

• TEAD1 is a transcriptional enhancer It interacts with a muscle specific cofactor to promote skeletal muscle gene expression The mutation in the TEAD1 gene is the cause of Sveinsson s chorioretinal a

• RXRB is a member of the retinoid X receptor RXR family of nuclear receptors which are involved in mediating the effects of retinoic acid RA This receptor forms homodimers with the retinoic acid

• HOXB4 is a nuclear protein with a homeobox DNA binding domain The protein functions as a sequence specific transcription factor that is involved in development Intracellular or ectopic expression of

• HOXB4 is a member of the Antp homeobox family and is a nuclear protein with a homeobox DNA binding domain The protein functions as a sequence specific transcription factor that is involved in develop

• ZNF655 is a zinc finger protein The zinc finger proteins are involved in DNA binding and protein protein interactions This gene encodes a zinc finger protein The zinc finger proteins are involved in

• HMBOX1 is located at the boundary of 8p12 3 and 8p21 1 HMBOX1 proteins are highly conserved in human mouse rat chicken and Xenopus laevis Functional HMBOX1 EGFP enhanced green fluorescent prote

• HSPBAP1 may play a role in cellular stress response

• The function of ZNF329 remains unknown

• ZNF419A contains 1 KRAB domain and 11 C2H2 type zinc fingers and belongs to the krueppel C2H2 type zinc finger protein family ZNF419A may be involved in transcriptional regulation

• ZNF408 may be involved in transcriptional regulation

• ZBTB3 contains 2 C2H2 type zinc fingers and 1 BTB POZ domain It may be involved in transcriptional regulation

• Epithelial cell cadherin is endocytosed as a consequence of tyrosine phosphorylation and ubiquitination CBLL1 is an E3 ubiquitin ligase that mediates ubiquitination of the CDH1 complex Epithelial cel

• ZSCAN16 may be involved in transcriptional regulation

• Mammalian mitochondrial ribosomal proteins help in protein synthesis within the mitochondrion Mitochondrial ribosomes mitoribosomes consist of a small 28S subunit and a large 39S subunit MRPS15 is

• OTP is a member of the homeodomain HD family HD family proteins are helix turn helix transcription factors that play key roles in the specification of cell fates This protein may function during b

• ZNF394 belongs to the krueppel C2H2 type zinc finger protein family It contains 7 C2H2 type zinc fingers 1 KRAB domain and 1 SCAN box domain ZNF394 may be involved in transcriptional regulation

• ZNF397 belongs to the krueppel C2H2 type zinc finger protein family This gene product isoform 3 acts as a DNA dependent transcriptional repressor

• PGBD1 belongs to the subfamily of piggyBac transposable element derived PGBD genes The PGBD proteins appear to be novel with no obvious relationship to other transposases or other known protein f

• The function of INSM2 remains unknown

• ZNF496 contains a SCAN box a KRAB A domain and four consensus C2H2 type zinc fingers preceded by a unique finger derivative referred to herein as the C2HR motif The C2HR motif functions to mediate

• MINA is a protein with a molecular weight of 53 kDa which is localized in the nucleus and with part of the protein concentrated in the nucleolus It is a direct target gene of Myc and is involved in

• MINA protein is directly involved in ribosome biogenesis most likely during the assembly process of preribosomal particles MINA is also involved in cell proliferation MINA may have a role in esopha

• ZNF514 belongs to the krueppel C2H2 type zinc finger protein family It contains 7 C2H2 type zinc fingers and 1 KRAB domain ZNF514 may be involved in transcriptional regulation

• Members of the C2H2 zinc finger transcription factor family such as ZNF382 play key roles in the regulation of cell proliferation differentiation and apoptosis in response to a variety of stimuli

• The function of ATOH8 remains unknown

• MLXIPL is a basic helix loop helix leucine zipper transcription factor of the Myc Max Mad superfamily This protein forms a heterodimeric complex and binds and activates in a glucose dependent manner

• ZNF682 contains 1 KRAB domain and 11 C2H2 type zinc fingers and belongs to the krueppel C2H2 type zinc finger protein family ZNF682 may be involved in transcriptional regulation

• MSL3L1 is a nuclear protein which is thought to play a similar function in chromatin remodeling and transcriptional regulation This gene has been found to undergo X inactivation

• FOXP4 belongs to subfamily P of the forkhead box FOX transcription factor family Forkhead box transcription factors play important roles in the regulation of tissue and cell type specific gene tra

• ZNF588 contains 1 KRAB domain and 9 C2H2 type zinc fingers It belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• ZNF564 may be involved in transcriptional regulation

• ZNF25 contains 1 KRAB domain and 12 C2H2 type zinc fingers ZNF25 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• HMGB4 contains two HMG box regions which is found in a variety of eukaryotic chromosomal proteins and transcription

• ZNF31contains 1 KRAB domain and 10 C2H2 type zinc fingers ZNF31 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• ZNF485 contains 1 KRAB domain and 11 C2H2 type zinc fingers It belongs to the krueppel C2H2 type zinc finger protein family and may be involved intranscriptional regulation

• ZSCAN21 belongs to the krueppel C2H2 type zinc finger protein family and is a strong transcriptional activator

• ZNF440 contains 1 KRAB domain and 12 C2H2 type zinc fingers ZNF440 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• ZNF597 contains 1 KRAB domain and 7 C2H2 type zinc fingers It belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• Nkx6 3 is a new member of the Nkx6 subfamily of homeodomain proteins Members of the Nkx family of homeodomain proteins are involved in a variety of developmental processes such as cell fate determina

• ZNF709 may be involved in transcriptional regulation

• GLIS3 is a member of the GLI similar zinc finger protein family and has five C2H2 type zinc finger domains GLIS3 functions as both a repressor and activator of transcription and is specifically invol

• ZBTB9 contains 1 BTB POZ domain and 2 C2H2 type zinc fingers It may be involved in transcriptional regulation

• ZNF555 belongs to the krueppel C2H2 type zinc finger protein family It contains 15 C2H2 type zinc fingers and 1 KRAB domain ZNF555 may be involved in transcriptional regulation

• HIF3A is the alpha 3 subunit of one of several alpha beta subunit heterodimeric transcription factors that regulate many adaptive responses to low oxygen tension hypoxia The alpha 3 subunit lacks t

• ZNF488 is a candidate transcription factor

• POU5F2 is a transcription factor that binds preferentially to the octamer motif 5 ATGTTAAT 3 It may exert a regulatory function in meiotic events that are required for terminal differentiation of

• ZNF550 contains 1 KRAB domain and 8 C2H2 type zinc fingers ZNF550 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• ZNF398 is a member of the Kruppel family of C2H2 type zinc finger transcription factor proteins This protein acts as a transcriptional activator This gene encodes a member of the Kruppel family of C2

• The function of ZNF57 remains unkonwn

• ZNF707 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• TFAP2E belongs to AP 2 family of transcription factors which play an important role in regulating gene expression during development and differentiation of multiple organs and tissues It may play an

• Gonadal dysgenesis in 9p deleted individuals might be due to combined hemizygosity of DMRT1 and DMRT2

• RNF8 contains a RING finger motif and a FHA domain This protein has been shown to interact with several class II ubiquitin conjugating enzymes E2 including UBE2E1 UBCH6 UBE2E2 and UBE2E3 and ma

• ZNF497 belongs to the krueppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• ZNF527 contains 1KRAB domain and 12 C2H2 type zinc

fingers It belongs to the kruppel C2H2 type zinc finger protein family and may be involved in transcriptional regulation

• NOBOX is a transcription factor which may play a role in oogenesis It binds preferentially to the DNA sequences 5 TAATTG 3 5 TAGTTG 3 and 5 TAATTA 3

• ZBTB38 contains 10 C2H2 type zinc fingers and 1 BTB POZ domain ZBTB38 acts as a transcriptional activator and may be involved in the differentiation and or survival of late postmitotic neurons

• The function remains unknows

• Ppargc1a is a transcriptional coactivator for steroid receptors and nuclear receptors Ppargc1a greatly increases the transcriptional activity of PPARG and thyroid hormone receptor on the uncoupling p

• Ehf is a transcriptional activator that may play a role in regulating epithelial cell differentiation and proliferation It may act as a repressor for a specific subset of ETS AP 1 responsive genes a

• Brf1 is a general activator of RNA polymerase which utilizes different TFIIIB complexes at structurally distinct promoters

• Tcfap2c is a sequence specific DNA binding protein that interacts with inducible viral and cellular enhancer elements to regulate transcription of selected genes Tcfap2c binds to the consensus sequen

• MBD2 belongs to a family of nuclear proteins related by the presence in each of a methyl CpG binding domain MBD Each of these proteins with the exception of MBD3 is capable of binding specificall

• HOMER1 is a member of the homer family of dendritic proteins Members of this family regulate group 1 metabotrophic glutamate receptor function This gene encodes a member of the homer family of dendri

• PRMT5 methylates specific arginine residues in the small nuclear ribonucleoproteins Sm D1 and Sm D3 to monomethylarginine and to symmetrical dimethylarginines sDMAs It methylates SUPT5H PRMT5 play

• PRMT8 probably methylates the guanidino nitrogens of arginyl residues in some proteins

• The protein arginine methyltransferases PRMTs include a family of proteins with related putative methyltransferase domains that modify chromatin and regulate cellular transcription PRMT2 inhibits N

• PRMT1 is a protein arginine methyltransferase that functions as a histone methyltransferase specific for H4 The HRMT1L2 gene encodes a protein arginine methyltransferase that functions as a histone me

• AIFM1 PDCD8 is a flavoprotein essential for nuclear disassembly in apoptotic cells that is found in the mitochondrial intermembrane space in healthy cells Induction of apoptosis results in the tran

• ANP32A is a novel potent heat stable inhibitor protein of protein phosphatase 2A It may play a key role in self renewing cell populations where it may act in the nucleus to limit their sensitivity to

• KRT18 Keratin 18 is type I intermediate filament chain Keratin 18 together with its filament partner keratin 8 are perhaps the most commonly found members of the intermediate filament gene family

• SMN1 localizes to both the cytoplasm and the nucleus Within the nucleus the protein localizes to subnuclear bodies called gems which are found near coiled bodies containing high concentrations of sm

• RPL9 is a ribosomal protein that is a component of the 60S subunit RPL9 belongs to the L6P family of ribosomal proteins It is located in the cytoplasm As is typical for genes encoding ribosomal pro

• RPL6 is a ribosomal protein that is a component of the 60S subunit RPL6 belongs to the L6E family of ribosomal proteins It is located in the cytoplasm The protein can bind specifically to domain C

• RPL32 is a ribosomal protein that is a component of the 60S subunit RPL32 belongs to the L32E family of ribosomal proteins It is located in the cytoplasm Although some studies have mapped this gene

• EXOSC10 contains 1 HRDC domain and 1 3 5 exonuclease domain Antibodies against PM SCL are found in patients with polymyositis and or scleroderma

• EXOSC3 is component of the exosome 3 5 exoribonuclease complex and is required for the 3 processing of the 7S pre RNA to the mature 5 8S rRNA

• RBMS3 is a member of a small family of proteins which bind single stranded DNA RNA These proteins are characterized by the presence of two sets of ribonucleoprotein consensus sequence RNP CS that c

• HNRPD belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are nucleic acid binding proteins and they complex with heterogeneous nuclear RNA

• Heterogeneous nuclear RNAs hnRNAs which include mRNA precursors and mature mRNAs are associated with specific proteins to form heterogenous ribonucleoprotein hnRNP complexes Heterogeneous nuclear

• This gene is a member of the DAZ gene family and is a candidate for the human Y chromosomal azoospermia factor AZF This RNA binding protein is important for spermatogenesis

• SF3B1 is the subunit 1 of the splicing factor 3b protein complex Splicing factor 3b together with splicing factor 3a and a 12S RNA unit forms the U2 small nuclear ribonucleoproteins complex U2 snR

• SF3B1 is subunit 1 of the splicing factor 3b protein complex Splicing factor 3b together with splicing factor 3a and a 12S RNA unit forms the U2 small nuclear ribonucleoproteins complex U2 snRNP

• DAZ2 is a member of the DAZ family and is a candidate for the human Y chromosomal azoospermia factor AZF This RNA binding protein is important for spermatogenesis This gene is a member of the DAZ g

• This gene is a member of the cytidine deaminase gene family It is one of seven related genes or pseudogenes found in a cluster thought to result from gene duplication on chromosome 22 Members of t

• RBMY1A1 is a protein containing an RNA binding motif in the N terminus and four SRGY serine arginine glycine tyrosine boxes in the C terminus The gene that encodes RBMY1A1 is Y linked RBMY1A1 m

• RP11 82K18 3 is an aminotransferase that transaminates kynurenine to form kynurenic acid Kynurenic acid is a metabolite of tryptophan

• SNRP70 contains 1 RRM RNA recognition motif domain and mediates the splicing of pre mRNA by binding to the loop I region of U1 snRNA Western blots using two different antibodies against two unique

• FUS binds both single stranded and double stranded DNA and promotes ATP independent annealing of complementary single stranded DNAs and D loop formation in superhelical double stranded DNA FUS may pl

• GTPBP9 belongs to the GTP1 OBG family and the function remains unknown

• U2 auxiliary factor U2AF comprised of a large and a small subunit is a non snRNP protein required for the binding of U2 snRNP to the pre mRNA branch site U2AF2 is the U2AF large subunit which con

• FXR1 is an RNA binding protein that interacts with the functionally similar proteins FMR1 and FXR2 These proteins shuttle between the nucleus and cytoplasm and associate with polyribosomes predomina

• RAE1 is a homolog of yeast Rae1 It contains four WD40 motifs and has been shown to localize to distinct foci in the nucleoplasm to the nuclear rim and to meshwork like structures throughout the cy

• SERBP1 may play a role in the regulation of mRNA stability It binds to the 3 most 134 nt of the SERPINE1 PAI1 mRNA a region which confers cyclic nucleotide regulation of message decay

• RARA contains 1 nuclear receptor DNA binding domain and belongs to the nuclear hormone receptor family NR1 subfamily It is a receptor for retinoic acid This metabolite has profound effects on verte

• RBM9 is a RNA binding protein that seems to act as a coregulatory factor of ER alpha This gene is one of several human genes similar to the C elegans gene Fox 1 This gene encodes an RNA binding prot

• RBM9 is a RNA binding protein that seems to act as a coregulatory factor of ER alpha

• ADARB1 is an enzyme responsible for pre mRNA editing of the glutamate receptor subunit B by site specific deamination of adenosines Studies in rat found that this enzyme acted on its own pre mRNA mol

• More than 99 of the vertebrate ocular lens is comprised of terminally differentiated lens fiber cells Two lens specific intermediate filament like proteins CP49 also known as phakinin and BFSP1

• CIRBP contains 1 RRM RNA recognition motif domain It seems to play an essential role in cold induced suppression of cell proliferation

• CSH1 is a member of the somatotropin prolactin family of hormones and plays an important role in growth control This particular family member is expressed mainly in the placenta and utilizes multiple

• IF4B contains 1 RRM RNA recognition motif domain and is required for the binding of mRNA to ribosomes Functions of EIF4B are in close association with EEF4 F and EIF4 A It binds near the 5 termin

• FBL is a component of a nucleolar small nuclear ribonucleoprotein snRNP particle thought to participate in the first step in processing preribosomal RNA It is associated with the U3 U8 and U13 sm

• The human alanyl tRNA synthetase AARS belongs to a family of tRNA synthases of the class II enzymes Class II tRNA synthases evolved early in evolution and are highly conserved This is reflected b

• AU specific RNA binding enoyl CoA hydratase AUH protein binds to the AU rich element ARE a common element found in the 3 UTR of rapidly decaying mRNA such as c fos c myc and granulocyte macrop

• EIF4A2 contains 1 helicase C terminal domain and 1 helicase ATP binding domain It belongs to the DEAD box helicase family eIF4A subfamily In the current model of translation initiation eIF4A unwin

• GRSF1 is a cellular protein that binds RNAs containing the G rich element Using indirect immunofluorescence microscopy this protein was found to be localized in the cytoplasm The protein encoded by t

• HNRPA1 belongs to the A B subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hn

• HNRPK belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hnRNA

• ACO1 also known as iron regulatory element binding protein 1 IREB1 is a cytosolic protein which binds to iron responsive elements IREs It plays a central role in cellular iron homeostasis It w

• ISG20 belongs to the the exonuclease superfamily It is exonuclease with specificity for single stranded RNA and to a lesser extent for DNA It degrades RNA at a rate that is approximately 35 fold hi

• NOVA2 may regulate RNA splicing or metabolism in a specific subset of developing neurons It binds single strand RNA

• PIGF is a protein involved in glycosylphosphatidylinositol GPI anchor biosynthesis The GPI anchor a glycolipid containing three mannose molecules in its core backbone is found on many blood cells

• RBM4 contains 1 CCHC type zinc finger and 2 RRM RNA recognition motif domains It may play a role in alternative splice site selection during pre mRNA processing RBM4 is down regulated in fetal Dow

• RBMS1 is a member of a small family of proteins which bind single stranded DNA RNA These proteins are characterized by the presence of two sets of ribonucleoprotein consensus sequence RNP CS that c

• RPN1 is a type I integral membrane protein found only in the rough endoplasmic reticulum It is part of an N oligosaccharyl transferase complex that links high mannose oligosaccharides to asparagine r

• SNRPF belongs to the snRNP Sm proteins family and is associated with snRNP U1 U2 U4 U6 and U5

• SRP14 belongs to the SRP14 family The signal recognition particle assembly has a crucial role in targeting secretory proteins to the rough endoplasmic reticulum membrane SRP9 together with SRP14 and

• SRP19 belongs to the SRP19 family It is signal recognition particle assembly and binds directly to 7S RNA and mediates binding of the 54 kDa subunit of the SRP

• SSB is involved in diverse aspects of RNA metabolism including binding and protecting 3 prime UUU OH elements of newly RNA polymerase III transcribed RNA processing 5 prime and 3 prime ends of pre

• HSP90B1 belongs to the heat shock protein 90 family It is molecular chaperone that functions in the processing and transport of secreted proteins

• XPO1 mediates leucine rich nuclear export signal NES dependent protein transport Exportin 1 specifically inhibits the nuclear export of Rev and U snRNAs It is involved in the control of several ce

• PRKRA contains 3 DRBM double stranded RNA binding domains It appears to have a pro apoptotic function that may be suppressed in the presence of growth factor and activates EIF2AK2 in absence of dou

• EIF3S9 binds to the 40S ribosome and promotes the binding of methionyl tRNAi and mRNA

• EIF3S4 contains 1 RRM RNA recognition motif domain It binds to the 40S ribosome and promotes the binding of methionyl tRNAi and mRNA This subunit binds to the 18S rRNA

• SFRS9 belongs to the splicing factor SR family It contains 2 RRM RNA recognition motif domains SFRS9 plays a role in constitutive splicing and can modulate the selection of alternative splice site

• WNT9B is a member of the WNT family They are secreted signaling proteins These proteins have been implicated in oncogenesis and in several developmental processes including regulation of cell fate

• Poly A binding proteins PABPs bind to the poly A tail present at the 3 prime ends of most eukaryotic mRNAs PABPC4 or IPABP inducible PABP was isolated as an activation induced T cell mRNA enc

• Eukaryotic translation initiation factor 2 EIF 2 functions in the early steps of protein synthesis by forming a ternary complex with GTP and initiator tRNA and binding to a 40S

ribosomal subunit EI
• Calcium dependent membrane binding proteins may regulate molecular events at the interface of the cell membrane and cytoplasm CPNE1 is a calcium dependent protein that also contains two N terminal ty
• The translation initiation factor eIF2 catalyzes the first regulated step of protein synthesis initiation promoting the binding of the initiator tRNA to 40S ribosomal subunits Binding occurs as a te
• SFRS10 contains 1 RRM RNA recognition motif domain and belongs to the splicing factor SR family It is a sequence specific RNA binding protein which participates in the control of pre mRNA splicing
• SNRPA binds stem loop II of U1 snRNA It is the first snRNP to interact with pre mRNA This interaction is required for the subsequent binding of U2 snRNP and the U4 U6 U5 tri snRNP In a snRNP free f
• As a RNA binding protein TROVE2 binds to several small cytoplasmic RNA molecules known as Y RNAs It may stabilize these RNAs from degradation
• TROVE2 belongs to the Ro 60 kDa family It is RNA binding protein that binds to several small cytoplasmic RNA molecules known as Y RNAs It may stabilize these RNAs from degradation
• STAU1 Staufen is a member of the family of double stranded RNA dsRNA binding proteins involved in the transport and or localization of mRNAs to different subcellular compartments and or organelles
• The removal of introns from nuclear pre mRNAs occurs on complexes called spliceosomes which are made up of 4 small nuclear ribonucleoprotein snRNP particles and an undefined number of transiently a
• RRP9 contains 7 WD repeats and belongs to the WD repeat RRP9 family It is a component of a nucleolar small nuclear ribonucleoprotein particle snoRNP thought to participate in the processing and mod
• SCYE1 is a cytokine that is specifically induced by apoptosis The release of this cytokine renders the tumor associated vasculature sensitive to tumor necrosis factor The precursor of SCYE1 pro SCY
• EIF4E2 belongs to the eukaryotic initiation factor 4E family It recognizes and binds the 7 methylguanosine containing mRNA cap during an early step in the initiation of protein synthesis and facilita
• HNRPF belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hnRNA
• PCBP2 appears to be multifunctional It along with PCBP 1 and hnRNPK corresponds to the major cellular poly rC binding proteins This protein together with PCBP 1 also functions as translational coa
• SFPQ is DNA and RNA binding protein It is essential pre mRNA splicing factor required early in spliceosome formation and for splicing catalytic step II probably as an heteromer with NONO It binds
• ZRSR2 is an essential splicing factor The protein associates with the U2 auxiliary factor heterodimer which is required for the recognition of a functional 3 splice site in pre mRNA splicing and m
• RBM8A is a protein with a conserved RNA binding motif The protein is found predominantly in the nucleus although it is also present in the cytoplasm It is preferentially associated with mRNAs produ
• THOC1 is part of the TREX transcription export complex which includes TEX1 THO2 ALY and UAP56 HPR1 is part of the TREX transcription export complex which includes TEX1 MIM 606929 THO2 MIM
• HDLBP is high density lipoprotein binding protein also known as vigilin is a 110 kD protein that specifically binds HDL molecules and may function in the removal of excess cellular cholesterol High
• Nucleolin NCL a eukaryotic nucleolar phosphoprotein is involved in the synthesis and maturation of ribosomes It is located mainly in dense fibrillar regions of the nucleolus Nucleolin NCL a eu
• Aminoacyl tRNA synthetases are a class of enzymes that charge tRNAs with their cognate amino acids Lysyl tRNA synthetase is a homodimer localized to the cytoplasm which belongs to the class II family
• GIPC1 belongs to the GIPC family It may be involved in G protein linked signaling
• SF3B4 is one of four subunits of the splicing factor 3B The protein cross links to a region in the pre mRNA immediately upstream of the branchpoint sequence in pre mRNA in the prespliceosomal complex
• SF3A1 is the subunit 1 of the splicing factor 3a protein complex The splicing factor 3a heterotrimer includes subunits 1 2 and 3 and is necessary for the in vitro conversion of 15S U2 snRNP into an
• HNRPM belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hnRNA
• GNB2L1 seems to bind protein kinase C acting as an intracellular receptor to anchor the activated PKC to the cytoskeleton
• PPIE is a member of the peptidyl prolyl cis trans isomerase PPIase family PPIases catalyze the cis trans isomerization of proline imidic peptide bonds in oligopeptides and accelerate the folding of
• The functions of NSF remain unknown
• PCBP1 appears to be multifunctional It along with PCBP 2 and hnRNPK corresponds to the major cellular poly rC binding protein It contains three K homologous KH domains which may be involved in R
• SFRS6 is involved in mRNA splicing and may play a role in the determination of alternative splicing It belongs to the splicing factor SR family and has been shown to bind with and modulate another me
• RBM14 contains 2 RRM RNA recognition motif domains Isoform 1 may function as a nuclear receptor coactivator enhancing transcription through other coactivators such as NCOA6 and CITED1 Isoform 2
• EMG1 involved in 40S ribosomal subunit biogenesis It seems to play a role a methylation reaction in pre rRNA processing
• NXF1 is one member of a family of nuclear RNA export factor Common domain features of this family are a noncanonical RNP type RNA binding domain RBD 4 leucine rich repeats LRRs a nuclear transp
• Heterogenous nuclear ribonucleoprotein hnRNP implicated in mRNA processing mechanisms SYNCRIP may be involved in translationally coupled mRNA turnover It implicated with other RNA binding proteins
• Nop56p is a yeast nucleolar protein that is part of a complex with the nucleolar proteins Nop58p and fibrillarin Nop56p is required for assembly of the 60S ribosomal subunit and is involved in pre rR
• Of the multiple RNases H in mammals RNase HI is the major enzyme and shows increased activity during DNA replication It shows more homology to the RNase HII of Escherichia coli Of the multiple RNase
• PAIP1 interacts with poly A binding protein and with the cap binding complex eIF4A It is involved in translational initiation and protein biosynthesis Overexpression of this gene in COS7 cells sti
• SARS belongs to the class II amino acyl tRNA family The enzyme catalyzes the transfer of L serine to tRNA Ser and is related to bacterial and yeast counterparts This gene belongs to the class II am
• SLBP binds to the stem loop structure in replication dependent histone mRNAs Histone mRNAs do not contain introns or polyadenylation signals and are processed by endonucleolytic cleavage The stem l
• As a RNA binding protein KHDRBS3 plays a role in the regulation of alternative splicing and influences mRNA splice site selection and exon inclusion KHDRBS3 may play a role as a negative regulator o
• KHDRBS1 recruited and tyrosine phosphorylated by several receptor systems for example the T cell leptin and insulin receptors Once phosphorylated KHDRBS1 functions as an adapter protein in signal
• Aminoacyl tRNA synthetases are a class of enzymes that charge tRNAs with their cognate amino acids FARS2 is a phenylalanine tRNA synthetase PheRS localized to the mitochondrion which consists of a
• POP4 is a part of ribonuclease P a protein complex that generates mature tRNA molecules by cleaving their 5 ends It may function with RPP38 to coordinate the nucleolar targeting and or assembly of
• Inhibition of the nuclear export of poly A containing mRNAs caused by the influenza A virus NS1 protein requires its effector domain The NS1 effector domain functionally interacts with the cellular
• This gene is located in the surfeit gene cluster a group of very tightly linked genes that do not share sequence similarity The gene demonstrates features of a housekeeping gene being ubiquitously
• APOBEC2 belongs to the cytidine and deoxycytidylate deaminase family It is probable C to U editing enzyme whose physiological substrate is not yet known It does not display detectable apoB mRNA edit
• SF3A3 is the subunit 3 of the splicing factor 3a protein complex The splicing factor 3a heterotrimer includes subunits 1 2 and 3 and is necessary for the in vitro conversion of 15S U2 snRNP into an
• HNRPA0 belongs to the A B subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hn
• SFRS1 is a member of the arginine serine rich splicing factor protein family and functions in both constitutive and alternative pre mRNA splicing The protein binds to pre mRNA transcripts and compon
• SNRPD1 is a small nuclear ribonucleoprotein that belongs to the SNRNP core protein family The protein may act as a charged protein scaffold to promote SNRNP assembly or strengthen SNRNP SNRNP interac
• NUDT21 is one subunit of a cleavage factor required for 3 RNA cleavage and polyadenylation processing The interaction of the protein with the RNA is one of the earliest steps in the assembly of the
• CPSF6 is one subunit of a cleavage factor required for 3 RNA cleavage and polyadenylation processing The interaction of the protein with the RNA is one of the earliest steps in the assembly of the 3
• U1SNRNPBP is a homolog of U1 snRNP binding protein Its N terminal half contains a RNA recognition motif and the C terminal half is rich in Arg Asp and Arg Glu dipeptides a characteristic of a variet
• HNRPUL1 is a nuclear RNA binding protein of the heterogeneous nuclear ribonucleoprotein hnRNP family This protein binds specifically to adenovirus E1B 55kDa oncoprotein It may play an important ro
• IRAK3 contains 1 protein kinase domain and 1 death domain and belongs to the Ser Thr protein kinase family Pelle subfamily It inhibits dissociation of IRAK1 and IRAK4 from the Toll like receptor sig
• NONO is DNA and RNA binding protein involved in several nuclear processes It binds the conventional octamer sequence in double stranded DNA It also binds single stranded DNA and RNA at a site inde
• ADAT1 is a member of the ADAR adenosine deaminase acting on RNA family Using site specific adenosine modification the family participates in the pre mRNA editing of nuclear transcripts ADAT1 tRN
• HNRPH3 belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hnRNA
• PRPF6 appears to be involved in pre mRNA splicing possibly acting as a bridging factor between U5 and U4 U6 snRNPs in formation of the spliceosome PRPF6 also can bind androgen receptor providing a
• CPSF1 is a component of the cleavage and polyadenylation specificity factor CPSF complex that plays a key role in pre mRNA 3 end formation recognizing the AAUAAA signal sequence and interacting wi
• EWSR1 is a putative RNA binding protein Mutations in this gene specifically a t 11 22 q24 q12 translocation are known to cause Ewing sarcoma as well as neuroectodermal and various other tumors
• PUF60 is a Ro RNP binding protein It interacts with Ro RNPs and their interaction is thought to represent a gain of function for Ro RNPs This protein also forms a ternary complex with far upstream e
• CSDC2 is an RNA binding factor which binds specifically to the very 3 UTR ends of both histone H1 and H3 3 mRNAs encompassing the polyadenylation signal It might play a central role in the negative
• EXOSC7 belongs to the RNase PH family It is a component of the exosome 3 5 exoribonuclease complex and is required for the 3 processing of the 7S pre RNA to the mature 5 8S rRNA
• CSTF2T may play a significant role in AAUAAA independent mRNA polyadenylation in germ cells It is directly involved in the binding to pre mRNAs
• PUM2 is a sequence specific RNA binding protein that regulates translation and mRNA stability by binding the 3 UTR of mRNA targets Its interactions and tissue specificity suggest that it may be requ
• The exact function of CTA 126B4 3 remains unknown
• TRNT1 belongs to the tRNA nucleotidyltransferase poly A polymerase family It adds and repairs the conserved 3 CCA sequence necessary for the attachment of amino acids to the 3 terminus of tRNA mo
• SF3B14 is a 14 kDa protein subunit of the splicing factor 3b complex Splicing factor 3b associates with both the U2 and U11 U12 small nuclear ribonucleoprotein complexes U2 snRNP of spliceosomes T
• NIP7 contains 1PUA domain and belongs to the NIP7 family It may play a role in 60S ribosomal subunit synthesis
• CPSF3 belongs to the RNA metabolizing metallo beta lactamase like family CPSF3 subfamily It is component of the cleavage and polyadenylation specificity factor CPSF complex that play a key role in
• CPSF2 is a component of the cleavage and polyadenylation specificity factor CPSF complex that play a key role in pre mRNA 3 end formation recognizing the AAUAAA signal sequence and interacting wit
• RBM38 is a probable RNA binding protein
• CARF was first cloned as a novel binding partner of ARF from a yeast interactive screen CARF and ARF colocalize in the perinucleolar region and have a collaborative function In the nucleoplasm CARF

• TRSPAP1 is involved in the early steps of selenocysteine biosynthesis and tRNA Sec charging to the later steps resulting in the cotranslational incorporation of selenocysteine into selenoproteins I
• C19orf24 is a novel human non classical secreted protein which is encoded by the hypothetical gene C19orf24 chromosome 19 open reading frame 24 The exact function of C19orf24 remains unknown
• RBM22 may be involved in pre mRNA splicing
• RBM28 protein may be a specific nucleolar component of the spliceosomal snRNPs
• SRBD1 contains 1 S1 motif domain The exact function of SRBD1 remains unknown
• STRBP contains 2 DRBM double stranded RNA binding domains and 1 DZF domain STRBP is involved in spermatogenesis and sperm function It plays a role in regulation of cell growth
• MATR3 is localized in the nuclear matrix It may play a role in transcription or may interact with other nuclear matrix proteins to form the internal fibrogranular network The protein encoded by this
• In mammals the Y chromosome directs the development of the testes and plays an important role in spermatogenesis A high percentage of infertile men have deletions that map to regions of the Y chromo
• EXOSC4 belongs to the RNase PH family It is a component of the exosome 3 5 exoribonuclease complex and is required for the 3 processing of the 7S pre RNA to the mature 5 8S rRNA It has a 3 5 e
• PCBP4 is a member of the KH domain protein subfamily Proteins of this subfamily also referred to as alpha CPs bind to RNA with a specificity for C rich pyrimidine regions Alpha CPs play important
• MIF4GD is a protein which contains an MIF4G domain This gene encodes a protein which contains an MIF4G domain
• SF4 is a member of the SURP family of splicing factors SF4 is a member of the SURP family of splicing factors
• PTBP2 binds to the intronic cluster of RNA regulatory elements downstream control sequence DCS It is implicated in controlling the assembly of other splicing regulatory proteins This protein is v
• WBSCR1 is one of the translation initiation factors which functions to stimulate the initiation of protein synthesis at the level of mRNA utilization This gene encodes one of the translation initiati
• NOC4L plays a specific role in biogenesis of 40 S subunit of ribosome
• FAM121B belongs to the FAM121 family and the function remains unknown
• RBM42 belongs to the RRM RBM42 family and it contains 1 RRM RNA recognition motif domain The functions of RBM42 remain unknown
• MRM1 belongs to the RNA methyltransferase trmH family and probably methylates the ribose of guanosine G 2270 in the peptidyl transferase center of the mitochondrial large ribosomal RNA 21S
• BXDC5 may be required for ribosome biogenesis
• FIP1L1 is a component of the cleavage and polyadenylation specificity factor CPSF complex that plays a key role in pre mRNA 3 end formation recognizing the AAUAAA signal sequence and interacting w
• As a RNA binding protein C14orf156 acts as a nuclear receptor corepressor It probably acts by binding the SRA RNA and repressing the SRA mediated nuclear receptor coactivation C14orf156 binds the
• HNRNPA2B1 belongs to the A B subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA
• This gene belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are produced by RNA polymerase II and are components of the heterogeneous nuc
• HNRPC belongs to the subfamily of ubiquitously expressed heterogeneous nuclear ribonucleoproteins hnRNPs The hnRNPs are RNA binding proteins and they complex with heterogeneous nuclear RNA hnRNA
• RBM4B contains 1 CCHC type zinc finger and 2 RRM RNA recognition motif domains It may play a role in alternative splice site selection during pre mRNA processing
• EIF2A is a 65 kD protein that catalyzes the formation of puromycin sensitive 80S preinitiation complexes EIF2A is a 65 kD protein that catalyzes the formation of puromycin sensitive 80S preinitiation
• The exact functions of DKFZP564O0523 remain unknown
• THOC3 is part of the TREX transcription export complex which includes THO2 HPR1 ALY and UAP56 TEX1 is part of the TREX transcription export complex which includes THO2 MIM 300395 HPR1 MIM
• NXF5 is one member of a family of nuclear RNA export factors Common domain features of this family are a noncanonical RNP type RNA binding domain RBD 4 leucine rich repeats LRRs a nuclear trans
• PNPT1 is a subunit of the exosome complex which is involved in 3 prime to 5 prime exoribonuclease activity for RNA processing and degradation PNPT1 is a subunit of the exosome complex which is invol
• LARP1 belongs to the LARP family and it contains 1 HTH La type RNA binding domain The exact function of LARP1 remains unknown
• FUSIP1 is a member of the serine arginine SR family of proteins which is involved in constitutive and regulated RNA splicing Members of this family are characterized by N terminal RNP1 and RNP2 mo
• EXOSC6 constitutes one of the subunits of the multisubunit particle called exosome which mediates mRNA degradation The composition of human exosome is similar to its yeast counterpart This protein
• RAVER1 contains 3 RRM RNA recognition motif domains It cooperates with PTBP1 to modulate regulated alternative splicing events and promotes exon skipping It cooperates with PTBP1 to modulate switc
• HNRPLL contains 3 RRM RNA recognition motif domains and may bind RNA and plays a role in mRNA processing
• MSI2 contains two conserved tandem RNA recognition motifs Similar proteins in other species function as RNA binding proteins and play central roles in posttranscriptional gene regulation This gene en
• ADAD2 belongs to the ADAD family and contains 1 A to I editase domain and 1 DRBM double stranded RNA binding domain The exact functions of ADAD2 remain unknown
• JAKMIP1 associates with microtubules and may play a role in the microtubule dependent transport of the GABA B receptor It may play a role in JAK1 signaling and regulate microtubule cytoskeleton rearr
• RG9MTD3 belongs to the RNA methyltransferase trmD family TRM10 subfamily It is a probable RNA methyltransferase
• DCP2 is necessary for the degradation of mRNAs both in normal mRNA turnover and in nonsense mediated mRNA decay DCP2 removes the 7 methyl guanine cap structure from mRNA molecules yielding a 5 pho
• DRB1 is RNA binding protein with binding specificity for poly C and may play an important role in neural development
• RBM45 is a RNA binding protein with binding specificity for poly C It May play an important role in neural development RBM45 contains 3 RRM RNA recognition motif domains
• THEX1 contains 1 SAP domain and 1 exonuclease domain It is an RNA exonuclease that binds to the 3 end of histone mRNAs and probably degrades them suggesting that it plays an essential role in histo
• SLA LP converts O phosphoseryl tRNA Sec to selenocysteinyl tRNA Sec required for selenoprotein biosynthesis
• EIF4E3 belongs to the EIF4E family of translational initiation factors that interact with the 5 prime cap structure of mRNA and recruit mRNA to the ribosome
• RALYL belongs to the RRM HNRPC family RALY subfamily It contains 1 RRM RNA recognition motif domain The functions of RALYL remain unknown
• CPEB2 is highly similar to cytoplasmic polyadenylation element binding protein CPEB an mRNA binding protein that regulates cytoplasmic polyadenylation of mRNA as a trans factor in oogenesis and spe
• NSUN6 may have S adenosyl L methionine dependent methyl transferase activity Potential NSUN6 belongs to the methyltransferase superfamily RsmB NOP family It contains 1 PUA domain Western blots us
• HNRPA3 plays a role in cytoplasmic trafficking of RNA It binds to the cis acting response element A2RE and may be involved in pre mRNA splicing
• HNRPA3 contains 2 RRM RNA recognition motif domains and plays a role in cytoplasmic trafficking of RNA It binds to the cis acting response element A2RE and may be involved in pre mRNA splicing
• DND1 contains 2 RRM RNA recognition motif domains It may play a role during primordial germ cell PGC development However DND1 does not seem to be essential for PGC migration
• SF1 contains 1 KH domain and 1 CCHC type zinc finger It is necessary for the ATP dependent first step of spliceosome assembly and binds to the intron branch point sequence BPS 5 UACUAAC 3 of the
• MBNL1 contains 4 C3H1 type zinc fingers and binds to CUG triplet repeat expansion dsRNA
• Members of the frizzled gene family encode 7 transmembrane domain proteins that are receptors for Wnt signaling proteins The FZD5 protein is believed to be the receptor for the Wnt5A ligand Members
• FZD7 is 7 transmembrane domain protein that is receptor for Wnt signaling proteins The FZD7 protein contains an N terminal signal sequence 10 cysteine residues typical of the cysteine rich extracell
• WNT2B is a member of the wingless type MMTV integration site WNT family of highly conserved secreted signaling factors WNT family members function in a variety of developmental processes including
• GIPC1 GIPC GIPC2 and GIPC3 are a family of central PDZ domain proteins GIPC2 might play important roles in human gastric cancer through modulation of growth factor signaling or cell adhesion GIPC1
• The WNT gene family consists of structurally related genes which encode secreted signaling proteins These proteins have been implicated in oncogenesis and in several developmental processes includin
• In the mouse Nkd is a Dishevelled binding protein that functions as a negative regulator of the Wnt beta catenin Tcf signaling pathway In the mouse Nkd is a Dishevelled see DVL1 MIM 601365 bind
• In the mouse Nkd is a Dishevelled binding protein that functions as a negative regulator of the Wnt beta catenin Tcf signaling pathway
• WDR13 is a member of the WD repeat protein family WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by gly his and trp asp GH WD which may facilitate f
• WDR6 is a member of the WD repeat protein family WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by gly his and trp asp GH WD which may facilitate fo
• WDR12 is a member of the WD repeat protein family WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by gly his and trp asp GH WD which may facilitate f
• GNB1L is a G protein beta subunit like polypeptide which is a member of the WD repeat protein family WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by
• CDC25B is a member of the CDC25 family of phosphatases CDC25B activates the cyclin dependent kinase CDC2 by removing two phosphate groups and it is required for entry into mitosis CDC25B shuttles be
• MSH2 was identified as a locus frequently mutated in hereditary nonpolyposis colon cancer HNPCC When cloned it was discovered to be a human homolog of the E coli mismatch repair gene mutS consis
• ASS catalyzes the penultimate step of the arginine biosynthetic pathway The protein encoded by this gene catalyzes the penultimate step of the arginine biosynthetic pathway There are approximately 10
• KHK is a ketohexokinase that catalyzes conversion of fructose to fructose 1 phosphate The product of this gene is the first enzyme with a specialized pathway that catabolizes dietary fructose KHK enc
• The sodium potassium chloride cotransporter isoform 2 is kidney specific and is found on the apical membrane of the thick ascending limb of Henle s loop and the macula densa It accounts for most of t
• TPM1 is a member of the tropomyosin family of highly conserved widely distributed actin binding proteins involved in the contractile system of striated and smooth muscles and the cytoskeleton of non
• MAT1A catalyzes the formation of S adenosylmethionine from methionine and ATP Methionine adenosyltransferase deficiency is caused by recessive and dominant mutations the latter identified in autosom
• MAT1A catalyzes a two step reaction that involves the transfer of the adenosyl moiety of ATP to methionine to form S adenosylmethionine and tripolyphosphate which is subsequently cleaved to PPi and P
• PON1 hydrolyzes the toxic metabolites of a variety of organophosphorus insecticides It is capable of hydrolyzing a broad spectrum of organophosphate substrates and a number of aromatic carboxylic aci
• Fructose 1 6 bisphosphatase 1 a gluconeogenesis regulatory enzyme catalyzes the hydrolysis of fructose 1 6 bisphosphate to fructose 6 phosphate and inorganic phosphate Fructose 1 6 diphosphatase de
• Renin catalyzes the first step in the activation pathway of angiotensinogen a cascade that can result in aldosterone release vasoconstriction and increase in blood pressure Renin an aspartyl prote
• CYP2E1 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol
• DDC catalyzes the decarboxylation of L 3 4 dihydroxyphenylalanine DOPA to dopamine L 5 hydroxytryptophan to serotonin and L tryptophan to tryptamine
• CLDN18 belongs to the large claudin family of proteins which form tight junction strands in epithelial cells CLDN18 belongs to the large claudin family of proteins which form tight junction strands
• AK3L1 is a member of the adenylate kinase family of

enzymes The protein is localized to the mitochondrial matrix Adenylate kinases regulate the adenine and guanine nucleotide compositions within a c

• Mammalian sodium dicarboxylate cotransporters transport succinate and other Krebs cycle intermediates They fall into 2 categories based on their substrate affinity low affinity and high affinity Bo

• MUC1 is a membrane bound glycosylated phosphoprotein The protein is anchored to the apical surface of many epithelia by a transmembrane domain with the degree of glycosylation varying with cell typ

• Dihydropyrimidinase catalyzes the conversion of 5 6 dihydrouracil to 3 ureidopropionate in pyrimidine metabolism Dihydropyrimidinase is expressed at a high level in liver and kidney as a major 2 5 kb

• Epithelial membrane protein 2 EMP2 is a member of the four transmembrane superfamily TM4SF and is thought to mediate trafficking of diverse proteins such as alpha6beta1 integrin and MHC class I to

• BHMT is a cytosolic enzyme that catalyzes the conversion of betaine and homocysteine to dimethylglycine and methionine respectively Defects in its gene could lead to hyperhomocyst e inemia but suc

• DES is a muscle specific class III intermediate filament Homopolymers of this protein form a stable intracytoplasmic filamentous network connecting myofibrils to each other and to the plasma membrane

• FN1 is a glycoprotein present in a soluble dimeric form in plasma and in a dimeric or multimeric form at the cell surface and in extracellular matrix Fibronectin is involved in cell adhesion and mig

• GPX3 belongs to the glutathione peroxidase family which functions in the detoxification of hydrogen peroxide It contains a selenocysteine Sec residue at its active site The selenocysteine is enco

• Many breast pancreatic colonic and non small cell lung carcinoma lines express CEACAM6 and CEACAM5 and antibodies to both can affect tumor cell growth in vitro and in vivo CEACAM6 expression is el

• Polyspecific organic cation transporters in the liver kidney intestine and other organs are critical for elimination of many endogenous small organic cations as well as a wide array of drugs and en

• HSD17B6 has both oxidoreductase and epimerase activities and is involved in androgen catabolism The oxidoreductase activity can convert 3 alpha adiol to dihydrotestosterone while the epimerase activ

• GPCR5A is a member of the type 3 G protein coupling receptor family characterized by the signature 7 transmembrane domain motif The protein may be involved in interaction between retinoid acid and G

• LBP is involved in the acute phase immunologic response to gram negative bacterial infections Gram negative bacteria contain a glycolipid lipopolysaccharide LPS on their outer cell wall Together

• ADAMTS4 is a member of the ADAMTS a disintegrin and metalloproteinase with thrombospondin motifs protein family Members of the family share several distinct protein modules including a propeptide

• HMGCS2 condenses acetyl CoA with acetoacetyl CoA to form HMG CoA which is the substrate for HMG CoA reductase

• FTCD is a bifunctional enzyme that channels 1 carbon units from formiminoglutamate a metabolite of the histidine degradation pathway to the folate pool FTCD is a bifunctional enzyme that channels 1

• LDB3 may function as an adapter in striated muscle to couple protein kinase C mediated signaling via its LIM domains to the cytoskeleton

• T cell activation by APCs is positively and negatively regulated by members of the B7 family VSIG4 is a strong negative regulator of murine and human T cell proliferation and IL 2 production

• C11orf54 exhibits ester hydrolase activity on the substrate p nitrophenyl acetate

• SMPX plays a role in the regulatory network through which muscle cells coordinate their structural and functional states during growth adaptation and repair

• The anaphase promoting complex APC consists of at least 8 protein subunits including APC5 CDC27 APC3 CDC16 APC6 and CDC23 APC8 The anaphase promoting complex APC consists of at least 8 p

• CHST7 belongs to the sulfotransferase family Sulfotransferases generate sulfated glycosaminoglycan GAG moities during chondroitin sulfate biosynthesis They create considerable structural diversity

• PRODH2 is similar to proline dehydrogenase oxidase 1 a mitochondrial enzyme which catalyzes the first step in proline catabolism The function of this protein has not been determined The protein en

• Gamma glutamyl transpeptidase is a membrane bound protein that is important in the metabolism of glutathione The protein is similar in sequence to several members of the gamma glutamyl transpeptidase

• AGER mediates interactions of advanced glycosylation end products AGE These are nonenzymatically glycosylated proteins which accumulate in vascular tissue in aging and at an accelerated rate in dia

• The ALAD enzyme is composed of 8 identical subunits and catalyzes the condensation of 2 molecules of delta aminolevulinate to form porphobilinogen a precursor of heme cytochromes and other hemoprote

• ASL is a member of the lyase 1 family The protein forms a cytosolic homotetramer and primarily catalyzes the reversible hydrolytic cleavage of argininosuccinate into arginine and fumarate an essenti

• CYP2D6 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol

• FAH is the last enzyme in the tyrosine catabolism pathway FAH deficiency is associated with Type 1 hereditary tyrosinemia This gene encodes the last enzyme in the tyrosine catabolism pathway FAH def

• Ferrochelatase is localized to the mitochondrion where it catalyzes the insertion of the ferrous form of iron into protoporphyrin IX in the heme synthesis pathway Defects in ferrochelatase are associ

• GAMT is a methyltransferase that converts guanidoacetate to creatine using S adenosylmethionine as the methyl donor Defects in its gene have been implicated in neurologic syndromes and muscular hypo

• The deficiency of HMGCL is related to an autosomal recessive branched chain organic aciduria

• LCAT is an extracellular cholesterol esterifying enzyme lecithin cholesterol acyltransferase The esterification of cholesterol is required for cholesterol transport Mutations in its gene have been

• PKLR is a pyruvate kinase that catalyzes the production of phohsphoenolpyruvate from pyruvate and ATP Defects in this enzyme due to gene mutations or genetic variations are the common cause of chro

• Protein C is a vitamin K dependent serine protease that regulates blood coagulation by inactivating factors Va and VIIIa in the presence of calcium ions and phospholipids

• The Rh blood group antigens are associated with human erythrocyte membrane proteins of approximately 30 kD the so called Rh30 polypeptides Heterogeneously glycosylated membrane proteins of 50 and 45

• Glucose transporter 2 isoform is an integral plasma membrane glycoprotein of the liver islet beta cells intestine and kidney epithelium It mediates facilitated bidirectional glucose transport Bec

• TSHR is receptor for thyrothropin It plays a central role in controlling thyroid cell metabolism The activity of this receptor is mediated by G proteins which activate adenylate cyclase It also act

• UROD is the fifth enzyme of the heme biosynthetic pathway This enzyme is responsible for catalyzing the conversion of uroporphyrinogen to coproporphyrinogen through the removal of four carboxymethyl

• HSD17B1 is favors the reduction of estrogens and androgens It also has 20 alpha HSD activity It uses preferentially NADH

• KRT17 is type I intermediate filament chain keratin 17 expressed in nail bed hair follicle sebaceous glands and other epidermal appendages Mutations in its gene lead to Jackson Lawler type pachyo

• KRT2A is a member of the keratin gene family The type II cytokeratins consist of basic or neutral proteins which are arranged in pairs of heterotypic keratin chains coexpressed during differentiation

• LOR is a major protein component of the cornified cell envelope found in terminally differentiated epidermal cells Mutations in this gene are associated with Vohwinkel s syndrome and progressive symm

• STK11is a member of the serine threonine kinase family regulates cell polarity and functions as a tumor suppressor Mutations in its gene have been associated with Peutz Jeghers syndrome an autosoma

• This locus has a highly complex imprinted expression pattern It gives rise to maternally paternally and biallelically expressed transcripts that are derived from four alternative promoters and 5 e

• OTC is a mitochondrial matrix enzyme Missense nonsense and frameshift mutations in this enzyme lead to ornithine transcarbamylase deficiency which causes hyperammonemia Since the gene for this en

• Cytosolic and membrane bound forms of glutathione S transferase are two distinct supergene families At present eight distinct classes of the soluble cytoplasmic mammalian glutathione S transferases

• BDKRB2 is a receptor for bradykinin The 9 aa bradykinin peptide elicits many responses including vasodilation edema smooth muscle spasm and pain fiber stimulation This receptor associates with G p

• ADH6 is class V alcohol dehydrogenase which is a member of the alcohol dehydrogenase family Members of this family metabolize a wide variety of substrates including ethanol retinol other aliphati

• CYP3A7 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol

• CYP2A13 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholestero

• CYP2C9 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol

• At present eight distinct classes of the soluble cytoplasmic mammalian glutathione S transferases have been identified alpha kappa mu omega pi sigma theta and zeta GSTM2 is a glutathione S tr

• Prostaglandin endoperoxide synthase PTGS also known as cyclooxygenase is the key enzyme in prostaglandin biosynthesis and acts both as a dioxygenase and as a peroxidase There are two isozymes of

• GHRHR is a receptor for growth hormone releasing hormone Binding of this hormone to the receptor leads to synthesis and release of growth hormone Mutations in its gene have been associated with isol

• CYP4A22 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholestero

• DHODH catalyzes the fourth enzymatic step the ubiquinone mediated oxidation of dihydroorotate to orotate in de novo pyrimidine biosynthesis This protein is a mitochondrial protein located on the ou

• CES1 is one of the enzymes responsible for the hydrolysis of ester and amide bond containing drugs such as cocaine and heroin They also hydrolize long chain fatty acid esters and thioesters This en

• PSG6 may have a role in modulation of the innate immune system

• RHOBTB1 belongs to the Rho family of the small GTPase superfamily It contains a GTPase domain a proline rich region a tandem of 2 BTB broad complex tramtrack and bric a brac domains and a cons

• ALAS2 specifies an erythroid specific mitochondrially located enzyme The protein catalyzes the first step in the heme biosynthetic pathway Defects in its gene cause X linked pyridoxine responsive si

• ALOX15B is a member of the lipoxygenase family of structurally related nonheme iron dioxygenases involved in the production of fatty acid hydroperoxides The protein converts arachidonic acid exclusiv

• The down regulation of C9orf127 may be closely associated with tumorigenesis and metastasis of colorectal carcinoma However it may not contribute to the development and progression of gastric carcin

• Overexpression of C9orf127 can influence the distribution of the cell cycle in NPC cells and it also plays a role in cell adhesion modulation in NPC cells

• ABP1 is a membrane glycoprotein that is expressed in many epithelium rich and or hematopoietic tissues and oxidatively deaminates putrescine and histamine The protein may play a role in controlling t

• CD40 is a member of the TNF receptor superfamily This receptor has been found to be essential in mediating a broad variety of immune and inflammatory responses including T cell dependent immunoglobul

• CHGA is a member of the chromogranin secretogranin family of neuroendocrine secretory proteins It is found in secretory vesicles of neurons and endocrine cells Its gene s product is a precursor to t

• Members of the immunoglobulin Ig superfamily which includes IGSF1 have a variety of functions but all appear to play a role in cell recognition and the regulation of cell behavior Members of the

• There are at least four distinct but related alkaline phosphatases intestinal placental placental like and liver bone kidney tissue non specific The first three are located together on chromoso

• APCS is a glycoprotein belonging to the pentraxin family of proteins which has a characteristic pentameric organization These family members have considerable sequence homology which is thought to

• The cystatin superfamily encompasses proteins that contain multiple cystatin like sequences Some of the members are active cysteine protease inhibitors while others have lost or perhaps never acquir

• DLAT is dihydrolipoamide acetyltransferase the E2 subunit

of the mammalian pyruvate dehydrogenase complex of the inner mitochondrial membrane Patients with primary biliary cirrhosis show autoantibod
• GCG is actually a preproprotein that is cleaved into four distinct mature peptides One of these glucagon is a pancreatic hormone that counteracts the glucose lowering action of insulin by stimulati
• GH2 is a member of the somatotropin prolactin family of hormones which play an important role in growth control Mutations in its gene lead to placental growth hormone lactogen deficiency The protein
• KRT8 together with KRT19 help to link the contractile apparatus to dystrophin at the costameres of striated muscle
• KRT13 is a member of the keratin gene family The keratins are intermediate filament proteins responsible for the structural integrity of epithelial cells and are subdivided into cytokeratins and hair
• As a tumor suppressor it blocks the growth invasion and metastatic properties of mammary tumors As it does not undergo the S stressed to R relaxed conformational transition characteristic of a
• PLIN coats lipid storage droplets in adipocytes thereby protecting them until they can be broken down by hormone sensitive lipase The protein is the major cAMP dependent protein kinase substrate in
• The pregnancy specific beta 1 glycoprotein PSG is a group of heterogeneous proteins produced in large amounts by the human syncytiotrophoblast They belong to the carcinoembryonic antigen CEA fami
• RBP1 is the carrier protein involved in the transport of retinol vitamin A alcohol from the liver storage site to peripheral tissue Vitamin A is a fat soluble vitamin necessary for growth reproduc
• S100A3 is a member of the S100 family of proteins containing 2 EF hand calcium binding motifs S100 proteins are localized in the cytoplasm and or nucleus of a wide range of cells and involved in the
• SEMG1 is the predominant protein in semen The secreted protein is involved in the formation of a gel matrix that encases ejaculated spermatozoa The prostate specific antigen PSA protease processes
• SLC2A5 is a cytochalasin B sensitive carrier It seems to function primarily as a fructose transporter
• Squalene epoxidase catalyzes the first oxygenation step in sterol biosynthesis and is thought to be one of the rate limiting enzymes in this pathway
• LEFTY2 is a member of the TGF beta family of proteins The protein is secreted and plays a role in left right asymmetry determination of organ systems during development The protein may also play a r
• Troponin proteins associate with tropomyosin and regulate the calcium sensitivity of the myofibril contractile apparatus of striated muscles Each troponin tropomyosin complex contains 1 molecule from
• TST is a mitochondrial matrix enzyme that is encoded by the nucleus It may play roles in cyanide detoxification the formation of iron sulfur proteins and the modification of sulfur containing enzym
• PPFIBP1 is a member of the LAR protein tyrosine phosphatase interacting protein liprin family Liprins interact with members of LAR family of transmembrane protein tyrosine phosphatases which are k
• PPAP2A is a member of the phosphatidic acid phosphatase PAP family PAPs convert phosphatidic acid to diacylglycerol and function in de novo synthesis of glycerolipids as well as in receptor activa
• SOCS1 is a member of the STAT induced STAT inhibitor SSI also known as suppressor of cytokine signaling SOCS family SSI family members are cytokine inducible negative regulators of cytokine sig
• MTMR1 is a member of the myotubularin related family of proteins Members of this family contain the consensus sequence for the active site of protein tyrosine phosphatases This gene encodes a member
• IL18RAP is an accessory subunit of the heterodimeric receptor for IL18 This protein enhances the IL18 binding activity of IL18R1 IL1RRP a ligand binding subunit of IL18 receptor The coexpression
• Carboxylesterase 2 is a member of a large multigene family The enzymes are responsible for the hydrolysis of ester and amide bond containing drugs such as cocaine and heroin They also hydrolize lon
• BAG proteins compete with Hip for binding to the Hsc70 Hsp70 ATPase domain and promote substrate release All the BAG proteins have an approximately 45 amino acid BAG domain near the C terminus but di
• ACP1 belongs to the phosphotyrosine protein phosphatase family of proteins It functions as an acid phosphatase and a protein tyrosine phosphatase by hydrolyzing protein tyrosine phosphate to protein
• ITGBL1 may be linked in some way with the evolution of the integrin beta subunits
• Alpha actinins belong to the spectrin superfamily which represents a diverse group of cytoskeletal proteins including the alpha and beta spectrins and dystrophins Alpha actinin is an actin binding p
• Alpha actinins belong to the spectrin superfamily which is a diverse group of cytoskeletal proteins including the alpha and beta spectrins and dystrophins Alpha actinin is an actin binding protein w
• The CD8 antigen is a cell surface glycoprotein found on most cytotoxic T lymphocytes that mediates efficient cell cell interactions within the immune system The CD8 antigen acting as a coreceptor a
• RAD23A is one of two human homologs of Saccharomyces cerevisiae Rad23 a protein involved in nucleotide excision repair NER This protein was shown to interact with and elevate the nucleotide excis
• Haptoglobin combines with free plasma hemoglobin preventing loss of iron through the kidneys and protecting the kidneys from damage by hemoglobin while making the hemoglobin accessible to degradativ
• GEM belongs to the RAD GEM family of GTP binding proteins It is associated with the inner face of the plasma membrane and could play a role as a regulatory protein in receptor mediated signal transdu
• Zeta globin is an alpha like hemoglobin The zeta globin polypeptide is synthesized in the yolk sac of the early embryo while alpha globin is produced throughout fetal and adult like The zeta globin
• MAGEA8 is a member of the MAGEA family The members of this family with 50 to 80 sequence identity to each other They have been implicated in some hereditary disorders such as dyskeratosis congenit
• SLC1A5 has a broad substrate specificity a preference for zwitterionic amino acids and a sodium dependence It accepts as substrates all neutral amino acids including glutamine asparagine and bra
• SLC6A8 is required for the uptake of creatine in muscles and brain
• PBEF1 catalyzes the condensation of nicotinamide with 5 phosphoribosyl 1 pyrophosphate to yield nicotinamide mononucleotide one step in the biosynthesis of nicotinamide adenine dinucleotide The prot
• CTDSPL may function as a phosphatase involved in the regulation of cell growth and differentiation
• N methylation is one method by which drug and other xenobiotic compounds are metabolized by the liver NNMT responsible for this enzymatic activity which uses S adenosyl methionine as the methyl donor
• Follistatin is a single chain gonadal protein that specifically inhibits follicle stimulating hormone release Follistatin is a single chain gonadal protein that specifically inhibits follicle stimulat
• AGR2 and hAG 3 human homologues of genes involved in differentiation are associated with oestrogen receptor positive breast tumours and interact with metastasis gene C4 4a and dystroglycan Increase
• SLC17A3 may be involved in actively transporting phosphate into cells via Na cotransport
• SLC22A7 is involved in the sodium independent transport and excretion of organic anions some of which are potentially toxic The protein is an integral membrane protein and appears to be localized to
• As a sodium dependent amino acid proton antiporter SLC38A3 mediates electrogenic cotransport of glutamine and sodium ions in exchange for protons It also recognizes histidine asparagine and alanine
• ZNF19 contains a zinc finger a nucleic acid binding domain present in many transcription factors The protein encoded by this gene contains a zinc finger a nucleic acid binding domain present in many
• The function remains known
• When FAM107A is transfected into cell lines in which it is not expressed it suppresses cell growth It may play a role in tumor development
• GPR161 is orphan receptor
• ITGB1BP2 may play a role during maturation and or organization of muscles cells
• Protein phosphatase 2 formerly named type 2A is one of the four major Ser Thr phosphatases and is implicated in the negative control of cell growth and division Protein phosphatase 2 holoenzymes ar
• CLEC4M is a type II integral membrane protein that is 77 identical to CD209 antigen a HIV gp120 binding protein This protein like CD209 efficiently binds both intercellular adhesion molecule 3 I
• PSD3 is a guanine nucleotide exchange factor for ARF6
• NELF influences outgrowth of olfactory axons and migration of LHRH neurons
• GAPVD1 acts both as a GTPase activating protein GAP and a guanine nucleotide exchange factor GEF and participates in various processes such as endocytosis insulin receptor internalization or LC2
• DCXR is an enzyme that has both diacetyl reductase and L xylulose reductase activities DCXR is an enzyme that has both diacetyl reductase EC 1 1 1 5 and L xylulose reductase EC 1 1 1 10 activities
• UPB1 is a protein that belongs to the CN hydrolase family Beta ureidopropionase catalyzes the last step in the pyrimidine degradation pathway The pyrimidine bases uracil and thymine are degraded via
• PIPOX metabolizes sarcosine L pipecolic acid and L proline
• PLUNC is the human homolog of murine plunc and is specifically expressed in the upper airways and nasopharyngeal regions The exact biological function of this protein is not known however it has be
• Subcellular location of HAO1 is the peroxisome Specifically HAO1 is expressed primarily in liver and pancreas and is most active on glycolate a two carbon substrate The protein is also active on 2
• BMP2K is the human homolog of mouse BMP 2 inducible kinase Bone morphogenic proteins BMPs play a key role in skeletal development and patterning BMP2K is thought to be a protein kinase with a puta
• ASPN belongs to a family of leucine rich repeat LRR proteins associated with the cartilage matrix The name asporin reflects the unique aspartate rich N terminus and the overall similarity to decori
• EPS8L1 a protein that is related to epidermal growth factor receptor pathway substrate 8 EPS8 a substrate for the epidermal growth factor receptor The function of this protein is unknown This gene
• SLC38A4 is found predominantly in liver and transports both cationic and neutral amino acids The transport of cationic amino acids by SLC38A4 is Na and pH independent while the transport of neut
• The function remains unknown The modification of proteins with ubiquitin is an important cellular mechanism for targeting abnormal or short lived proteins for degradation Ubiquitination involves at l
• KRT20 is a member of the keratin family The keratins are intermediate filament proteins responsible for the structural integrity of epithelial cells and are subdivided into cytokeratins and hair kera
• The preB cell receptor is found on the surface of proB and preB cells where it is involved in transduction of signals for cellular proliferation differentiation from the proB cell to the preB cell s
• C21orf7 plays a critical role in the TGF beta signaling transduction pathway
• AKR1B10 is a member of the aldo keto reductase superfamily which consists of more than 40 known enzymes and proteins This member can efficiently reduce aliphatic and aromatic aldehydes and it is le
• ANKRD2 may play an important role in skeletal muscle hypertrophy
• Ankyrins are a family of proteins that are believed to link the integral membrane proteins to the underlying spectrin actin cytoskeleton and play key roles in activities such as cell motility activat
• DCUN1D1 may contribute to neddylation of cullin components of SCF type E3 ubiquitin ligase complexes Neddylation of cullins play an essential role in the regulation of SCF type complexes activity
• CYP4F11 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholestero
• EDG8 is a receptor for the lysosphingolipid sphingosine 1 phosphate S1P S1P is a bioactive lysophospholipid that elicits diverse physiological effect on most types of cells and tissues It Is coupl
• There are at least four distinct but related alkaline phosphatases intestinal placental placental like and liver bone kidney tissue non specific The exact physiological function of the alkaline
• This enzyme is probably necessary for target cell lysis in cell mediated immune responses
• FAM129A is expressed in subsets of thyroid tumors and Hashimoto s thyroiditis and it is a novel tumor marker
• RSAD2 is a potential antiviral effector Western blots using two different antibodies against two unique regions of this protein target confirm the same apparent molecular weight in our tests
• PANX2 belongs to the innexin family Innexin family members are the structural components of gap junctions This protein and pannexin 1 are abundantly expressed in central nerve system CNS and are c
• PANX1 belongs to the innexin family Innexin family members are the structural components of gap junctions This protein and pannexin 2 are abundantly expressed in central nerve system CNS and are c
• Cytochrome c oxidase COX is the terminal enzyme of the

mitochondrial respiratory chain It is a multi subunit enzyme complex that couples the transfer of electrons from cytochrome c to molecular oxy
• RGS8 is a member of the regulator of G protein signaling RGS family and is a protein with a single RGS domain Regulator of G protein signaling RGS proteins are regulatory and structural component
• RGS18 is a member of the regulator of G protein signaling family This protein is contains a conserved 120 amino acid motif called the RGS domain The protein attenuates the signaling activity of G p
• Forkhead box FOX transcription factors are implicated in carcinogenesis through gene amplification retroviral integration or chromosomal translocation Human FOXN6 mRNA was expressed in breast can
• FOXB2 may act as a transcription factor
• One gap junction consists of a cluster of closely packed pairs of transmembrane channels the connexons through which materials of low MW diffuse from one cell to a neighboring cell
• Rgs2 inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits thereby driving them into their inactive GDP bound form It may play a role in leukemogenesis
• Rgs7 inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits thereby driving them into their inactive GDP bound form Activity on G o alpha is specifically enhance
• Anxa6 may associate with CD21 and may regulate the release of Ca 2 from intracellular stores
• Rgs1 inhibits signal transduction by increasing the GTPase activity of G protein alpha subunits thereby driving them into their inactive GDP bound form
• Members of the claudin protein family such as CLDN2 are expressed in an organ specific manner and regulate the tissue specific physiologic properties of tight junctions Members of the claudin protei
• Protein kinase C PKC zeta is a member of the PKC family of serine threonine kinases which are involved in a variety of cellular processes such as proliferation differentiation and secretion Unlike
• OCLN is an integral membrane protein which is located at tight junctions This protein may be involved in the formation and maintenance of the tight junction The possibility of several alternatively
• Angiomotin is a protein that binds angiostatin a circulating inhibitor of the formation of new blood vessels angiogenesis Angiomotin mediates angiostatin inhibition of endothelial cell migration a
• Autocrine motility factor is a tumor motility stimulating protein secreted by tumor cells AMFR is a glycosylated transmembrane protein and a receptor for autocrine motility factor The receptor whic
• UBE2L3 ubiquitin conjugating enzyme E2L 3 Antibody The modification of proteins with ubiquitin is an important cellular mechanism for targeting abnormal or short lived proteins for degradation Ubiq
• FKBP6 is a member of the immunophilin protein family which play a role in immunoregulation and basic cellular processes involving protein folding and trafficking FKBP6 is a cis trans prolyl isomeras
• CDC23 shares strong similarity with Saccharomyces cerevisiae Cdc23 a protein essential for cell cycle progression through the G2 M transition This protein is a component of anaphase promoting comple
• RAPSN belongs to a family of proteins that are receptor associated proteins of the synapse It contains a conserved cAMP dependent protein kinase phosphorylation site It is believed to play some role
• HIP2 belongs to the ubiquitin conjugating enzyme family It binds selectively to a large region at the N terminus of huntingtin This interaction is not influenced by the length of the huntingtin poly
• ARIH1 might act as an E3 ubiquitin protein ligase or as part of the E3 complex which accepts ubiquitin from specific E2 ubiquitin conjugating enzymes such as UBE2L3 UBCM4 and then transfers it to
• THOC4 is a heat stable nuclear protein and functions as a molecular chaperone It is thought to regulate dimerization DNA binding and transcriptional activity of basic region leucine zipper bZIP
• C20orf18 is similar to mouse UIP28 UbcM4 interacting protein The protein encoded by this gene is similar to mouse UIP28 UbcM4 interacting protein Alternative splicing has been observed at this locus
• FBXL5 is a member of the F box protein family which is characterized by an approximately 40 amino acid motif the F box The F box proteins constitute one of the four subunits of ubiquitin protein lig
• FBXO3 is a member of the F box protein family which is characterized by an approximately 40 amino acid motif The F box proteins constitute one of the four subunits of the ubiquitin protein ligase com
• FBXL7 is a member of the F box protein family which is characterized by an approximately 40 amino acid motif the F box The F box proteins constitute one of the four subunits of ubiquitin protein lig
• The Makorin ring finger protein 1 gene MKRN1 is a highly transcribed intron containing source for a family of intronless mammalian genes encoding a novel class of zinc finger proteins Phylogenetic
• The Makorin ring finger protein 1 MKRN1 is a novel class of zinc finger proteins Phylogenetic analyses indicate that the MKRN1 gene is the ancestral founder of this gene family
• PRPF19 plays a role in DNA double strand break DSB repair and pre mRNA splicing reaction It binds double stranded DNA in a sequence nonspecific manner PRPF19 acts as a structural component of the
• RNF40 contains a RING finger a motif known to be involved in protein protein and protein DNA interactions This protein was reported to interact with the tumor suppressor protein RB1 Studies of the
• RNF111 contains a RING finger domain a motif known to be involved in protein protein and protein DNA interactions The mouse counterpart of this gene Rnf111 arkadia has been shown to genetically in
• KIAA1333 is a probable E3 ubiquitin protein ligase which accepts ubiquitin from an E2 ubiquitin conjugating enzyme in the form of a thioester and then directly transfers the ubiquitin to targeted subs
• The protein contains a RING finger a motif present in a variety of functionally distinct proteins and known to be involved in protein protein and protein DNA interactions The protein encoded by this
• RAD18 is highly similar to S cerevisiae DNA damage repair protein Rad18 Yeast Rad18 functions through its interaction with Rad6 which is an ubiquitin conjugating enzyme required for post replicatio
• DTX2 is a regulator of Notch signaling a signaling pathway involved in cell cell communications that regulates a broad spectrum of cell fate determinations DTX2 probably acts both as a positive and
• RNF25 contains a RING finger motif The mouse counterpart of this protein has been shown to interact with Rela the p65 subunit of NF kappaB NFKB and modulate NFKB mediated transcription activity
• RNF38 is a protein with a coiled coil motif and a RING H2 motif C3H2C2 at its carboxy terminus The RING motif is a zinc binding domain found in a large set of proteins playing roles in diverse cell
• RNF128 is a type I transmembrane protein that localizes to the endocytic pathway This protein contains a RING zinc finger motif and has been shown to possess E3 ubiquitin ligase activity Expression
• Its gene lies within the major histocompatibility complex class I region on chromosome 6 Studies of a similar rat protein suggest that RNF39 plays a role in an early phase of synaptic plasticity Its
• RNF32 contains two RING ring finger motifs RING finger motifs are present in a variety of functionally distinct proteins and are known to be involved in protein DNA or protein protein interactions I
• SYVN1 is a protein involved in endoplasmic reticulum ER associated degradation The protein removes unfolded proteins accumulated during ER stress by retrograde transport to the cytosol from the E
• LNX1 is a membrane bound protein that is involved in signal transduction and protein interactions LNX1 is an E3 ubiquitin protein ligase which mediates ubiquitination and subsequent proteasomal degr
• NSMCE1 is a probable component of the SMC5 SMC6 complex a complex involved in DNA double strand breaks by homologous recombination The complex may promote sister chromatid homologous recombination b
• RNF165 is encoded in regions involved in pericentric inversions in patients with bipolar affective disorder Encoded in regions involved in pericentric inversions in patients with bipolar affective dis
• TRIM42 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains namely a RING a B box type 1 and a B box type 2 and a coiled coil region This gene encod
• UBE2E2 catalyzes the covalent attachment of ubiquitin to other proteins
• AS an endosomal ferrireductase STEAP3 is required for efficient transferrin dependent iron uptake in erythroid cells It participates in erythroid iron homeostasis by reducing Fe 3 to Fe 2 and
• M6PR is a receptor for mannose 6 phosphate groups on lysosomal enzymes The receptor forms a homodimer or homotetramer for intracellular targeting of lysosomal enzymes and export of newly synthesized
• This enzyme catalyzes O glycan branch synthesis of the core 2 and core 4 type in mucins and controls expression of core 2 branched oligosaccharides and I antigens on the cell surface This enzyme catal
• AADAT is a protein that is highly similar to mouse and rat kynurenine aminotransferase II The rat protein is a homodimer with two transaminase activities One activity is the transamination of alpha
• GCDH belongs to the acyl CoA dehydrogenase family It catalyzes the oxidative decarboxylation of glutaryl CoA to crotonyl CoA and CO 2 in the degradative pathway of L lysine L hydroxylysine and L
• ECH1 is a member of the hydratase isomerase superfamily ECH1 shows high sequence similarity to enoyl coenzyme A CoA hydratases of several species particularly within a conserved domain characteris
• GLS2 is a mitochondrial phosphate activated glutaminase that catalyzes the hydrolysis of glutamine to stoichiometric amounts of glutamate and ammonia This protein is functionally similar to the kidne
• DECR2 is auxiliary enzyme of beta oxidation It participates in the degradation of unsaturated fatty enoyl CoA esters having double bonds in both even and odd numbered positions in peroxisome It cat
• Human proteins MECP2 MBD1 MBD2 MBD3 and MBD4 comprise a family of nuclear proteins related by the presence in each of a methyl CpG binding domain MBD Each of these proteins with the exception
• SP1 belongs to the Sp1 C2H2 type zinc finger protein family It contains 3 C2H2 type zinc fingers SP1 binds to GC box promoters elements and selectively activates mRNA synthesis from genes that conta
• RELB neither associates with DNA nor with RELA p65 or REL It stimulates promoter activity in the presence of NFKB2 p49
• MTA2 modulates the enzymatic activity of the histone deacetylase core complex The protein encoded by this gene is a member of the immunophilin protein family which play a role in immunoregulation and
• IRF6 is a member of the interferon regulatory transcription factor IRF family Family members share a highly conserved N terminal helix turn helix DNA binding domain and a less conserved C terminal
• As an immediate early protein NOV is likely to play a role in cell growth regulation
• MTP encodes the large subunit of the heterodimeric microsomal triglyceride transfer protein Protein disulfide isomerase PDI completes the heterodimeric microsomal triglyceride transfer protein whi
• DLG2 is a member of the membrane associated guanylate kinase MAGUK family The protein forms a heterodimer with a related family member that may interact at postsynaptic sites to form a multimeric s
• ABCG2 is included in the superfamily of ATP binding cassette ABC transporters ABC proteins transport various molecules across extra and intra cellular membranes ABC genes are divided into seven d
• SLC11A1 is a member of the solute carrier family 11 proton coupled divalent metal ion transporters family and is a multi pass membrane protein The protein functions as a divalent transition metal
• SLC10A5 is a new member of Solute Carrier Family 10 SLC10 and the function remains unknown
• SLC25A29 belongs to the mitochondrial carrier family and it may has palmitoylcarnitine transporting activity
• By moving chloride into epithelial cells the Na K Cl cotransporter SLC12A2 aids transcellular movement of chloride across both secretory and absorptive epithelia By moving chloride into epithelial ce
• Amino acid transporters play essential roles in the uptake of nutrients production of energy chemical metabolism detoxification and neurotransmitter cycling SLC38A1 is an important transporter of
• SLC29A2 mediates equilibrative transport of purine pyrimidine nucleosides and the purine base hypoxanthine It is less sensitive than SLC29A1 to inhibition by nitrobenzylthioinosine NBMPR dipyrida
• SLC1A4 is a transporter for alanine serine cysteine and threonine It exhibits sodium dependence
• SLC7A1 is a high affinity low capacity permease involved in the transport of the cationic amino acids arginine lysine and ornithine in non hepatic tissues It may also function as an ecotropic ret
• SLC16A1 is a monocarboxylate transporter MCT1 that mediates the movement of lactate and pyruvate across cell membranes Import and export of these substrates by tissues such as erythrocytes muscle
• The vesicular monoamine transporter acts to accumulate cytosolic monoamines into synaptic vesicles using the proton gradient maintained across the synaptic vesicular membrane Its proper function is
• Mitochondrial uncoupling proteins UCP are members of the larger family of mitochondrial anion carrier proteins MACP

UCPs separate oxidative phosphorylation from ATP synthesis with energy dissipat
• SLC7A4 is involved in the transport of the cationic amino acids arginine lysine and ornithine
• SLCO1A2 is a sodium independent transporter which mediates cellular uptake of organic ions in the liver Its substrates include bile acids bromosulphophthalein and some steroidal compounds The prot
• As a Na PO4 cotransporter SLC17A4 may be important to the regulation of Li transport and its therapeutic effects
• SLC35B1 belongs to the nucleotide sugar transporter family and it is probable sugar transporter
• SLC35A3 is a uridine diphosphate N acetylglucosamine transporter in the Golgi apparatus
• SLC7A8 is sodium independent high affinity transport of large neutral amino acids It has higher affinity for L phenylalanine than LAT1 but lower affinity for glutamine and serine L alanine is trans
• SLC17A5 is a membrane transporter that exports free sialic acids that have been cleaved off of cell surface lipids and proteins from lysosomes Mutations in SLC17A5 gene cause sialic acid storage dise
• SLC16A8 is proton linked monocarboxylate transporter It catalyzes the rapid transport across the plasma membrane of many monocarboxylates such as lactate pyruvate branched chain oxo acids derived f
• SLC25A24 is a calcium dependent mitochondrial solute carrier Mitochondrial solute carriers shuttle metabolites nucleotides and cofactors through the mitochondrial inner membrane It may act as a AT
• SLC43A3 belongs to the SLC43A transporter family and is a putative transporter
• SLC5A4 belongs to the sodium solute symporter family and is a sodium dependent glucose transporter
• SLC25A39 is a member of the solute carrier family 25 and is known to transport molecules over the mitochondrial membrane
• SLC35F2 is a putative solute transporter
• Hexose transport into mammalian cells is catalyzed by a family of membrane proteins including SLC2A6 which contains 12 transmembrane domains and a number of critical conserved residues Hexose transp
• SLC25A38 is a members of the solute carrier family 25 SLC25 which is known to transport molecules over the mitochondrial membrane
• SLC35C1 is involved in GDP fucose import from the cytoplasm into the Golgi lumen
• SLC30A1 may be involved in zinc transport out of the cell
• SLC26A1 is a member of sulfate anion transporter family Family members are well conserved in their protein aa length among species structures but have markedly different tissue expression patterns
• The SLC25 family is mitochondrial carriers that transport a variety of metabolites across the inner mitochondrial membrane SLC25A22 also known as GC1 is 1 of the 2 mitochondrial glutamate H sympor
• SLC35F5 is a putative solute transporter
• SLC2A10 is a member of the facilitative glucose transporter family which plays a significant role in maintaining glucose homeostasis SLC2A10 is a member of the facilitative glucose transporter family
• SLC25A32 transports folate across the inner membranes of mitochondria
• SLC41A2 acts as a plasma membrane magnesium transporter
• Organic ion transporters such as SLC22A16 transport various medically and physiologically important compounds including pharmaceuticals toxins hormones neurotransmitters and cellular metabolite
• Cotransporters such as SLC5A11 represent a major class of proteins that make use of ion gradients to drive active transport for the cellular accumulation of nutrients neurotransmitters osmolytes
• SLC26A8 is one member of a family of sulfate anion transporters Family members are well conserved in their protein aa length among species structures yet have markedly different tissue expression p
• SLC25A46 is a members of the solute carrier family 25 SLC25 which is known to transport molecules over the mitochondrial membrane
• SLC43A2 is a Sodium chloride pH independent high affinity transport of large neutral amino acids
• SLC39A12 may act as a zinc influx transporter and also may be partly involved in the outbreak of schizophrenia
• SLC9A9 may act in electroneutral exchange of protons for Na across membranes
• Transport of folate compounds into mammalian cells can occur via receptor mediated or carrier mediated mechanisms A functional coordination between these 2 mechanisms has been proposed to be the meth
• SLC26A5 is a member of the SLC26A SulP transporter family SLC26A5 is specifically expressed in outer hair cells OHCs of the cochlea and is essential in auditory processing Intracellular anions are
• As a proton linked monocarboxylate transporter SLC16A12 catalyzes the rapid transport across the plasma membrane of many monocarboxylates
• NAT2 is a N acetyltransferase 2 arylamine N acetyltransferase 2 This enzyme functions to both activate and deactivate arylamine and hydrazine drugs and carcinogens Polymorphisms in its gene are re
• Mutant alleles at the BCHE locus are responsible for suxamethonium sensitivity Homozygous persons sustain prolonged apnea after administration of the muscle relaxant suxamethonium in connection with
• C8 beta is one of the three subunits that comprise the component 8 C8 of the complement system C8 participates in the formation of Membrane Attack Complex that results in the lysis of cells Patien
• GUSB plays an important role in the degradation of dermatan and keratan sulfates
• SHH is a protein that is instrumental in patterning the early embryo It has been implicated as the key inductive signal in patterning of the ventral neural tube the anterior posterior limb axis and
• Gamma sarcoglycan is one of several sarcolemmal transmembrane glycoproteins that interact with dystrophin probably to provide a link between the membrane associated cytoskeleton and the extracellular
• MICA is expressed on the cell surface although unlike canonical class I molecules does not seem to associate with beta 2 microglobulin It is thought that MICA functions as a stress induced antigen t
• PTCH1 is a member of the patched gene family The protein is the receptor for sonic hedgehog a secreted molecule implicated in the formation of embryonic structures and in tumorigenesis It functions
• TGFBI Binds to type I II and IV collagens This adhesion protein may play an important role in cell collagen interactions In cartilage may be involved in endochondral bone formation
• TYR is a copper containing oxidase that functions in the formation of pigments such as melanins and other polyphenolic compounds It catalyzes the rate limiting conversions of tyrosine to DOPA DOPA t
• Aldehyde dehydrogenase isozymes are thought to play a major role in the detoxification of aldehydes generated by alcohol metabolism and lipid peroxidation ALDH3A2 catalyzes the oxidation of long chai
• HFE is a membrane protein that is similar to MHC class I type proteins and associates with beta2 microglobulin beta2M It is thought that this protein functions to regulate iron absorption by regula
• NEU1 is a lysosomal enzyme which cleaves terminal sialic acid residues from substrates such as glycoproteins and glycolipids In the lysosome this enzyme is part of a heterotrimeric complex together
• Alzheimer s disease AD patients with an inherited form of the disease carry mutations in the presenilin proteins PSEN1 or PSEN2 or the amyloid precursor protein APP These disease linked mutatio
• C1QB is a major constituent of the human complement subcomponent C1q C1q associates with C1r and C1s in order to yield the first component of the serum complement system Deficiency of C1q has been a
• FGG is the gamma component of fibrinogen a blood borne glycoprotein comprised of three pairs of nonidentical polypeptide chains Following vascular injury fibrinogen is cleaved by thrombin to form f
• CSF1 is a cytokine that controls the production differentiation and function of macrophages The active form of the protein is found extracellularly as a disulfide linked homodimer and is thought t
• POR is an endoplasmic reticulum membrane oxidoreductase with an FAD binding domain and a flavodoxin like domain The protein binds two cofactors FAD and FMN which allow it to donate electrons direct
• PPIB is a cyclosporine binding protein and is mainly located within the endoplasmic reticulum It is associated with the secretory pathway and released in biological fluids This protein can bind to c
• ZDHHC13 may be involved in the NF kappa B signaling pathway
• ATP1B1 belongs to the family of Na K and H K ATPases beta chain proteins and to the subfamily of Na K ATPases Na K ATPase is an integral membrane protein responsible for establishing and m
• MTUS1 contains a C terminal domain and is able to interact with the angiotension II AT2 receptor and a large coiled coil region allowing dimerization One of the isoforms has been shown to be a mito
• ZFYVE27 may be associated with the neuronal intracellular trafficking in the corticospinal tract which is consistent with the pathology of HSP
• FMO3 is involved in the oxidative metabolism of a variety of xenobiotics such as drugs and pesticides It N oxygenates primary aliphatic alkylamines as well as secondary and tertiary amines It acts o
• GPNMB is a type I transmembrane glycoprotein which shows homology to the pMEL17 precursor a melanocyte specific protein GPNMB shows expression in the lowly metastatic human melanoma cell lines and x
• Microsomal arylacetamide deacetylase competes against the activity of cytosolic arylamine N acetyltransferase which catalyzes one of the initial biotransformation pathways for arylamine and heterocyc
• Activin receptors are all transmembrane proteins composed of a ligand binding extracellular domain with cysteine rich region a transmembrane domain and a cytoplasmic domain with predicted serine th
• TETRAN
• IL10RA is a receptor for interleukin 10 This protein is structurally related to interferon receptors It has been shown to mediate the immunosuppressive signal of interleukin 10 and thus inhibits th
• Butyrophilin is the major protein associated with fat droplets in the milk It is a member of the immunoglobulin superfamily It may have a cell surface receptor function The human butyrophilin gene
• CDH3 is a classical cadherin from the cadherin superfamily The protein is a calcium dependent cell cell adhesion glycoprotein comprised of five extracellular cadherin repeats a transmembrane region
• CDH8 is a type II classical cadherin from the cadherin superfamily integral membrane proteins that mediate calcium dependent cell cell adhesion Mature cadherin proteins are composed of a large N ter
• GZMK is a member of a group of related serine proteases from the cytoplasmic granules of cytotoxic lymphocytes Cytolytic T lymphocytes CTL and natural killer NK cells share the remarkable ability
• IHH is an intercellular signal essential for a variety of patterning events during development It binds to the patched PTC receptor which functions in association with smoothened SMO to activat
• LRPAP1 interacts with LRP1 alpha 2 macroglobulin receptor and glycoprotein 330
• LSAMP is a neuronal surface glycoprotein found in cortical and subcortical regions of the limbic system During development of the limbic system this protein is found on the surface of axonal membran
• The MAPEG Membrane Associated Proteins in Eicosanoid and Glutathione metabolism family consists of six human proteins several of which are involved in the production of leukotrienes and prostagland
• NDUFB5 is a subunit of the multisubunit NADH ubiquinone oxidoreductase complex I Mammalian complex I is composed of 45 different subunits It locates at the mitochondrial inner membrane This prote
• OLR1 is a receptor protein which belongs to the C type lectin superfamily OLR1 binds internalizes and degrades oxidized low density lipoprotein This protein may be involved in the regulation of Fas
• PLP2 may play a role in cell differentiation in the intestinal epithelium
• PTN is a heparin binding mitogenic protein It has neurite extension activity
• PTPN1 is the founding member of the protein tyrosine phosphatase PTP family PTPs catalyze the hydrolysis of the phosphate monoesters specifically on tyrosine residues Members of the PTP family sha
• PTPN2 is a member of the protein tyrosine phosphatase PTP family Members of the PTP family share a highly conserved catalytic motif which is essential for the catalytic activity PTPs are known to
• PTPRN is a member of the protein tyrosine phosphatase PTP family PTPs are known to be signaling molecules that regulate a variety of cellular processes including cell growth differentiation mitot
• PTPRR is a member of the protein tyrosine phosphatase PTP family PTPs are known to be signaling molecules that regulate a variety of cellular processes including cell growth differentiation mitot
• RPN2 is a type I integral membrane protein found only in the rough endoplasmic reticulum The protein is part of an N oligosaccharyl transferase complex that links high mannose oligosaccharides to asp
• SMPD2 converts sphingomyelin to ceramide Hydrolyze 1 acyl 2 lyso sn glycero 3 phosphocholine lyso PC and 1 O alkyl 2 lyso sn glycero 3 phosphocholine lyso platelet activating factor The physiolo
• The signal sequence receptor SSR is a glycosylated endoplasmic reticulum ER membrane receptor associated with protein translocation across the ER membrane The SSR consists of 2 subunits a 34 kD
• TAPBP is a transmembrane glycoprotein which mediates

interaction between newly assembled major histocompatibility complex MHC class I molecules and the transporter associated with antigen processing
• CHST1 catalyzes the transfer of sulfate to position 6 of galactose Gal residues of keratan It has a preference for sulfating keratan sulfate but it also transfers sulfate to the unsulfated polymer
• The function of LOC728864 remains unkonwn
• NR3C2 is a receptor for both mineralocorticoids MC such as aldosterone and glucocorticoids GC such as corticosterone or cortisol It binds to mineralocorticoid response elements MRE and transact
• RARB is a retinoic acid receptor beta a member of the thyroid steroid hormone receptor superfamily of nuclear transcriptional regulators This receptor localizes to the cytoplasm and to subnuclear co
• The protein encoded by RORA is a member of the NR1 subfamily of nuclear hormone receptors It can bind as a monomer or as a homodimer to hormone response elements upstream of several genes to enhance
• NR4A3 is a member of the steroid thyroid hormone retinoid receptor superfamily NR4A3 may act as a transcriptional activator The protein can efficiently bind the NGFI B Response Element NBRE Three
• OSBPL9 is a member of the oxysterol binding protein OSBP family a group of intracellular lipid receptors Most members contain an N terminal pleckstrin homology domain and a highly conserved C term
• Arginase catalyzes the hydrolysis of arginine to ornithine and urea The type I isoform of ARG1 is a cytosolic enzyme and expressed predominantly in the liver as a component of the urea cycle Inheri
• Plasma prekallikrein is a glycoprotein that participates in the surface dependent activation of blood coagulation fibrinolysis kinin generation and inflammation It is synthesized in the liver and s
• Kininogens are inhibitors of thiol proteases HMW kininogen plays an important role in blood coagulation by helping to position optimally prekallikrein and factor XI next to factor XII and inhibits th
• Carbamoyl phosphate synthetase I is the rate limiting enzyme that catalyzes the first committed step of the hepatic urea cycle The mitochondrial isozyme is designated CPS I and the cytoplasmic enzyme
• ALDH4A1 belongs to the aldehyde dehydrogenase family of proteins This enzyme is a mitochondrial matrix NAD dependent dehydrogenase which catalyzes the second step of the proline degradation pathway
• ECHS1 functions in the second step of the mitochondrial fatty acid beta oxidation pathway It catalyzes the hydration of 2 trans enoyl coenzyme A CoA intermediates to L 3 hydroxyacyl CoAs ECHS1 is
• GPT participates in cellular nitrogen metabolism and also in liver gluconeogenesis starting with precursors transported from skeletal muscles
• SBDS is a member of a highly conserved protein family that exists from archaea to vertebrates and plants The protein may function in RNA metabolism Mutations within its gene are associated with Shwa
• In vitro SGPP2 has high phosphohydrolase activity against dihydrosphingosine 1 phosphate and sphingosine 1 phosphate S1P
• CBS is involved in the transsulfuration pathway The first step of this pathway from homocysteine to cystathionine is catalyzed by this protein CBS deficiency can cause homocystinuria which affects
• CSTB is a stefin that functions as an intracellular thiol protease inhibitor The protein is able to form a dimer stabilized by noncovalent forces inhibiting papain and cathepsins l h and b The pro
• SOD1 binds copper and zinc ions and is one of two isozymes responsible for destroying free superoxide radicals in the body This isozyme is a soluble cytoplasmic protein acting as a homodimer to conv
• PDE9A catalyzes the hydrolysis of cAMP and cGMP to their corresponding monophosphates The protein plays a role in signal transduction by regulating the intracellular concentration of these cyclic nuc
• SH3BGR gene maps to the DS CHD region and is a potential candidate for the pathogenesis of CHD
• USP16 is a deubiquitinating enzyme that is phosphorylated at the onset of mitosis and then dephosphorylated at the metaphase anaphase transition It can deubiquitinate H2A one of two major ubiquitina
• Phosphofructokinase PFK is a tetrameric enzyme that catalyzes a key step in glycolysis namely the conversion of D fructose 6 phosphate to D fructose 1 6 bisphosphate PFK from muscle is a homotetra
• Carbonyl reductase is one of several monomeric NADPH dependent oxidoreductases having wide specificity for carbonyl compounds This enzyme is widely distributed in human tissues Carbonyl reductase is
• COL6A2 is one of the three alpha chains of type VI collagen a beaded filament collagen found in most connective tissues The protein contains several domains similar to von Willebrand Factor type A d
• RRP1 is the putative homolog of the yeast ribosomal RNA processing protein RRP1 The protein is involved in the late stages of nucleologenesis at the end of mitosis and may be required for the genera
• C21orf33 inhibits both estrogen and heregulin beta1 stimulated growth of breast cancer cells and further suggests that growth inhibition induced in these cells by all trans retinoic acid occurs via
• Chromatin assembly factor I CAF I is required for the assembly of histone octamers onto newly replicated DNA CAF I is composed of three protein subunits p50 p60 and p150 CHAF1B corresponds to t
• As a molecular chaperone CCT8 assists the folding of proteins upon ATP hydrolysis It is known to play a role in vitro in the folding of actin and tubulin
• POFUT2 catalyzes the reaction that attaches fucose through an O glycosidic linkage to a conserved serine or threonine residue in thrombospondin type 1 repeats
• This gene lies downstream of the SON gene and spans 10 kb on chromosome 21 The function of this gene is unknown This gene lies downstream of the SON gene and spans 10 kb on chromosome 21 The functio
• TCP10L is a human leucine zipper protein with transcription inhibition activity
• SNF1LK play a transient role during the earliest stages of myocardial cell differentiation and or primitive chamber formation and may also be important for the earliest stages of skeletal muscle growt
• The replication of DNA in eukaryotic cells is carried out by a complex chromosomal replication apparatus in which DNA polymerase alpha and primase are two key enzymatic components Primase which is
• ACTR2 is known to be a major constituent of the ARP2 3 complex This complex is located at the cell surface and is essential to cell shape and motility through lamellipodial actin assembly and protrus
• DUT is an essential enzyme of nucleotide metabolism This protein forms a ubiquitous homotetrameric enzyme that hydrolyzes dUTP to dUMP and pyrophosphate This reaction serves two cellular purposes
• RRM1 is one of two non identical subunits that constitute ribonucleoside diphosphate reductase an enzyme essential for the production of deoxyribonucleotides prior to DNA synthesis in S phase of divi
• Kynureninase is a pyridoxal 5 phosphate pyridoxal P dependent enzyme that catalyzes the cleavage of L kynurenine and L 3 hydroxykynurenine into anthranilic and 3 hydroxyanthranilic acids respectiv
• MTHFD2 is a nuclear encoded mitochondrial bifunctional enzyme with methylenetetrahydrofolate dehydrogenase and methenyltetrahydrofolate cyclohydrolase activities The enzyme functions as a homodimer a
• Centromeres are the differentiated chromosomal domains that specify the mitotic behavior of chromosomes CENPA is a centromere protein which contains a histone H3 related histone fold domain that is r
• eIF 2 functions in the early steps of protein synthesis by forming a ternary complex with GTP and initiator tRNA This complex binds to a 40S ribosomal subunit followed by mRNA binding to form a 43S
• Adenylate kinases are involved in regulating the adenine nucleotide composition within a cell by catalyzing the reversible transfer of phosphate groups among adenine nucleotides Three isozymes of ade
• BLK may function in a signal transduction pathway that is restricted to B lymphoid cells
• The catalytic conversion of UTP to CTP is accomplished by the enzyme cytidine 5 prime triphosphate synthetase The enzyme is important in the biosynthesis of phospholipids and nucleic acids and plays
• PPAT is a member of the purine pyrimidine phosphoribosyltransferase family This protein is a regulatory allosteric enzyme that catalyzes the first step of de novo purine nucleotide biosynthesis Its
• The elongation of primed DNA templates by DNA polymerase delta and DNA polymerase epsilon requires the accessory proteins proliferating cell nuclear antigen PCNA and replication factor C RFC RFC
• PEX3 is involved in peroxisome biosynthesis and integrity It assembles membrane vesicles before the matrix proteins are translocated As a docking factor for PEX19 it is necessary for the import of
• PAGE1 belongs to a family that are expressed in a variety of tumors but not in normal tissues except for the testis Nothing is presently known about the function of this protein This gene belongs to
• ATIC is a bifunctional protein requiring dimerization for transformylase activity
• FEN1 removes 5 overhanging flaps in DNA repair and processes the 5 ends of Okazaki fragments in lagging strand DNA synthesis Direct physical interaction between this protein and AP endonuclease 1 d
• Hearing impairment is a heterogeneous condition with over 40 loci described DFNA5 is expressed in fetal cochlea however its function is not known Nonsyndromic hearing impairment is associated with
• Related to nucleologenesis NOLC1 may play a role in the maintenance of the fundamental structure of the fibrillar center and dense fibrillar component in the nucleolus It has intrinsic GTPase and AT
• APOBEC3B is a member of the cytidine deaminase family It is thought that these proteins may be RNA editing enzymes and have roles in growth or cell cycle control This gene is a member of the cytidine
• SHMT2 plays a role in interconversion of serine and glycine
• PPIF is a member of the peptidyl prolyl cis trans isomerase PPIase family PPIases catalyze the cis trans isomerization of proline imidic peptide bonds in oligopeptides and accelerate the folding of
• The 26S proteasome is a multicatalytic proteinase complex with a highly ordered structure composed of 2 complexes a 20S core and a 19S regulator Proteasomes are distributed throughout eukaryotic cel
• STIP1 mediates the association of the molecular chaperones HSC70 and HSP90 HSPCA and HSPCB
• TPX2 binding decreases the size and accessibility of a hydrophobic pocket adjacent to the ATP site to inhibitors
• NCAPD2 is the regulatory subunit of the condensin complex a complex required for conversion of interphase chromatin into mitotic like condense chromosomes The condensin complex probably introduces p
• Phosphatidylinositol 3 4 5 trisphosphate dependent guanine nucleotide exchange factor GEF which independently of RAS transduces signals from tyrosine kinase receptors to RAC SWAP70 also mediates
• SEPT6 is a member of the septin family of GTPases Members of this family are required for cytokinesis This gene is a member of the septin family of GTPases Members of this family are required for cy
• SEPT6 is a member of the septin family of GTPases Members of this family are required for cytokinesis
• MRTO4 is a protein sharing a low level of sequence similarity with ribosomal protein P0 While the precise function of the protein is currently unknown it appears to be involved in mRNA turnover and
• ASF1B is a member of the H3 H4 family of histone chaperone proteins and is similar to the anti silencing function 1 gene in yeast The encoded protein is the substrate of the tousled like kinase famil
• The GINS complex plays an essential role in the initiation of DNA replication
• Mitochondrial ribosomes mitoribosomes consist of a small 28S subunit and a large 39S subunit MRPS12 is the 28S subunit protein that belongs to the ribosomal protein S12P family The protein is a ke
• PSAT1 is likely a phosphoserine aminotransferase based on similarity to proteins in mouse rabbit and Drosophila The protein encoded by this gene is likely a phosphoserine aminotransferase based on
• Biosynthesis of coenzyme A CoA from pantothenic acid vitamin B5 is an essential universal pathway in prokaryotes and eukaryotes PPCDC one of the last enzymes in this pathway converts phosphopan
• Proteins that contain formin homology FH domains such as FHOD3 play a role in regulation of the actin cytoskeleton
• CDT1 cooperates with CDC6 to promote the loading of the mini chromosome maintenance complex onto chromatin to form the pre replication complex necessary to initiate DNA replication It binds DNA in a
• Posttranslational glycosylphosphatidylinositol GPI anchor attachment serves as a general mechanism for linking proteins to the cell surface membrane GPAA1 presumably functions in GPI anchoring at t
• Ganglioside GM3 is known to participate in the induction of cell differentiation modulation of cell proliferation maintenance of fibroblast morphology signal transduction and integrin mediated cel
• FCGRT binds to the Fc region of monomeric immunoglobulins gamma It mediates the uptake of IgG from milk It plays a possible role in transfer of immunoglobulin G from mother to fetus
• TMPRSS11D is a trypsin like serine protease released from the submucosal serous glands onto mucous membrane It is a type II integral membrane protein and has 29 38 identity in the sequence of the ca
• CDH7 is a calcium dependent cell cell adhesion glycoprotein comprised of five extracellular cadherin repeats a transmembrane region and a highly conserved cytoplasmic tail Type II atypical cadheri

• Calmegin is a testis specific endoplasmic reticulum chaperone protein CLGN may play a role in spermatogeneisis and infertility Calmegin is a testis specific endoplasmic reticulum chaperone protein C
• Cytochrome c oxidase COX the terminal component of the mitochondrial respiratory chain catalyzes the electron transfer from reduced cytochrome c to oxygen This component is a heteromeric complex
• Retinoids exert biologic effects such as potent growth inhibitory and cell differentiation activities and are used in the treatment of hyperproliferative dermatological diseases These effects are med
• CCPG1 acts as an assembly platform for Rho protein signaling complexes It limits guanine nucleotide exchange activity of MCF2L toward RHOA which results in an inhibition of both its transcriptional
• KCNC1 belongs to the delayed rectifier class of channel proteins and is an integral membrane protein that mediates the voltage dependent potassium ion permeability of excitable membranes The Shaker ge
• DDOST is a component of the oligosaccharyltransferase complex which catalyzes the transfer of high mannose oligosaccharides to asparagine residues on nascent polypeptides in the lumen of the rough end
• LMAN1 is a type I integral membrane protein localized in the intermediate region between the endoplasmic reticulum and the Golgi presumably recycling between the two compartments The protein is a ma
• This gene belongs to the family of reticulon encoding genes Reticulons are associated with the endoplasmic reticulum and are involved in neuroendocrine secretion or in membrane trafficking in neuroe
• UST catalyzes the transfer of sulfate to the position 2 of uronyl residues UST has mainly activity toward iduronyl residues in dermatan sulfate and weaker activity toward glucuronyl residues of chon
• TSPAN5 is a member of the transmembrane 4 superfamily also known as the tetraspanin family Most of these members are cell surface proteins that are characterized by the presence of four hydrophobic
• ZMPSTE24 is a member of the peptidase M48A family This protein is a zinc metalloproteinase involved in the two step post translational proteolytic cleavage of carboxy terminal residues of farnesylate
• ST8SIA2 is a type II membrane protein that is thought to catalyze the transfer of sialic acid from CMP sialic acid to N linked oligosaccharides and glycoproteins ST8SIA2 may be found in the Golgi app
• NELL2 is a cytoplasmic protein that contains epidermal growth factor EGF like repeats Heterotrimeric protein may be involved in cell growth regulation and differentiation A similar protein in rod
• Synthesis of alpha 2 3 linked sialic acid to Gal beta 1 3 GalNAc is mediated by at least 3 distinct beta galactoside alpha 2 3 sialyltransferases EC 2 4 99 4 including ST3GAL4 In contrast only a
• ST3GAL3 is a type II membrane protein that catalyzes the transfer of sialic acid from CMP sialic acid to galactose containing substrates ST3GAL3 is normally found in the Golgi apparatus but can be pr
• EBP is an integral membrane protein of the endoplasmic reticulum It is a high affinity binding protein for the antiischemic phenylalkylamine Ca2 antagonist 3H emopamil and the photoaffinity label
• CORIN is a member of the type II transmembrane serine protease class of the trypsin superfamily Members of this family are composed of multiple structurally distinct domains CORIN converts pro atria
• Synaptobrevins VAMPs syntaxins and the 25 kD synaptosomal associated protein are the main components of a protein complex involved in the docking and or fusion of vesicles and cell membranes VAMP5
• Progesterone binding protein is a putative steroid membrane receptor The protein is expressed predominantly in the liver and kidney Progesterone binding protein is a putative steroid membrane recepto
• Sterol C4 mehtyl oxidase like protein was isolated based on its similarity to the yeast ERG25 protein It contains a set of putative metal binding motifs with similarity to that seen in a family of me
• LMAN2 plays a role as an intracellular lectin in the early secretory pathway It interacts with N acetyl D galactosamine and high mannose type glycans and may also bind to O linked glycans It is invo
• ST3GAL2 is a type II membrane protein that catalyzes the transfer of sialic acid from CMP sialic acid to galactose containing substrates The protein is normally found in the Golgi but can be proteoly
• SILV could be a melanogenic enzyme It could represent an oncofetal self antigen that is normally expressed at low levels in quiescent adult melanocytes but overexpressed by proliferating neonatal mel
• MMP23B is a member of the matrix metalloproteinase MMP family and it is part of a duplicated region of chromosome 1p36 3 Proteins of the matrix metalloproteinase MMP family are involved in the b
• RTN4 belongs to the family of reticulons Reticulons are associated with the endoplasmic reticulum and are involved in neuroendocrine secretion or in membrane trafficking in neuroendocrine cells RTN
• Voltage gated proton hydrogen channels play an important role in cellular defense against acidic stress They are unique among ion channels with respect to their extremely high selectivity marked t
• UGT1A4 is an enzyme of the glucuronidation pathway that transforms small lipophilic molecules such as steroids bilirubin hormones and drugs into water soluble excretable metabolites This enzyme
• The Sec61 complex is the central component of the protein translocation apparatus of the endoplasmic reticulum ER membrane SEC63 and SEC62 protein are found to be associated with ribosome free SEC6
• TMEM59L is a predicted type I membrane glycoprotein It may play a role in functioning of the central nervous system The protein encoded by this gene is thought be associated with an intracellular org
• CHIC2 is a member of the CHIC family of proteins This protein contains a cysteine rich hydrophobic CHIC motif and is localized to vesicular structures and the plasma membrane CHIC2 gene is associ
• FKBP8 is a member of the immunophilin protein family which play a role in immunoregulation and basic cellular processes involving protein folding and trafficking Unlike the other members of the fami
• The LETM1 is a factor of the mitochondrial K homeostasis with a potential role in the Wolf Hirschhorn syndrome
• TSPAN12 is a member of the transmembrane 4 superfamily also known as the tetraspanin family Most of these members are cell surface proteins that are characterized by the presence of four hydrophobic
• TSPAN15 is a member of the transmembrane 4 superfamily also known as the tetraspanin family Most of these members are cell surface proteins that are characterized by the presence of four hydrophobic
• POMT2 is an integral membrane protein of the endoplasmic reticulum ER that shares significant sequence similarity with a family of protein O mannosyltransferases of S cerevisiae
• Cell signaling pathways rely on a dynamic interaction between activating and inhibiting processes SHP 1 mediated dephosphorylation of protein tyrosine residues is central to the regulation of several
• ST6GALNAC6 belongs to a family of sialyltransferases that modify proteins and ceramides on the cell surface to alter cell cell or cell extracellular matrix interactions ST6GALNAC6 belongs to a family
• Neuregulin 1 NRG1 interacts with the NEU ERBB2 receptor tyrosine kinase to increase its phosphorylation on tyrosine residues Neuregulin 1 NRG1 was originally identified as a 44 kD glycoprotein tha
• MTCH1 is a potential mitochondrial transporter It may play a role in apoptosis
• FJX1 is the human ortholog of mouse and Drosophila four jointed gene product The Drosophila protein is important for growth and differentiation of legs and wings and for proper development of the ey
• SIGLEC9 is a putative adhesion molecule that mediates sialic acid dependent binding to cells It preferentially binds to alpha2 3 or 2 6 linked sialic acid The sialic acid recognition site may be ma
• TOR1B may serve as a molecular chaperone assisting in the proper folding of secreted and or membrane proteins
• LAPTM4A is a protein that has four predicted transmembrane domains The function of its gene has not yet been determined however studies in the mouse homolog suggest a role in the transport of small
• The function of GADD45B is involved in the regulation of growth and apoptosis
• KIAA0319 has been strongly associated with developmental dyslexia
• SV2A plays a role in the control of regulated secretion in neural and endocrine cells enhancing selectively low frequency neurotransmission It positively regulates vesicle fusion by maintaining the
• NPDC1 suppresses oncogenic transformation in neural and non neural cells and down regulates neural cell proliferation It might be involved in transcriptional regulation
• NSDHL is localized in the endoplasmic reticulum and is involved in cholesterol biosynthesis Mutations in NSDHL gene are associated with CHILD syndrome which is a X linked dominant disorder of lipid
• ERGIC3 plays a possible role in transport between endoplasmic reticulum and Golgi
• SDF4 may regulate calcium dependent activities in the endoplasmic reticulum lumen or post ER compartment
• GCNT4 is a glycosyltransferase that mediates core 2 O glycan branching an important step in mucin type biosynthesis It does not have core 4 O glycan or I branching enzyme activity
• GDE1 has glycerophosphoinositol phosphodiesterase activity It has little or no activity towards glycerophosphocholine GDE1 activity can be modulated by G protein signaling pathways
• RHBDL2 is a member of the rhomboid family of integral membrane proteins This family contains proteins that are related to Drosophila rhomboid protein Members of this family are found in both prokary
• Glycosylphosphatidylinositol GPI is a complex glycolipid that anchors many proteins to the cell surface The biosynthetic pathway of GPI is mediated by sequential addition of sugars and other compon
• MLSTD1 catalyzes the reduction of fatty acyl CoA to fatty alcohols The preferred substrates are C16 C18 C18 1 and C18 2 but low activity can be observed with C10 C14 substrates
• FBXW7 is a member of the F box protein family which is characterized by an approximately 40 amino acid motif the F box The F box proteins constitute one of the four subunits of ubiquitin protein lig
• CSGALNACT1 is a transfers 1 4 N acetylgalactosamine GalNAc from UDP GalNAc to the non reducing end of glucuronic acid GlcUA It is required for addition of the first GalNAc to the core tetrasaccha
• Receptor protein tyrosine kinases like STYK1 play important roles in diverse cellular and developmental processes such as cell proliferation differentiation and survival
• SERPINE1 acts as bait for tissue plasminogen activator urokinase and protein C Its rapid interaction with TPA may function as a major control point in the regulation of fibrinolysis
• Transglutaminases are enzymes that catalyze the crosslinking of proteins by epsilon gamma glutamyl lysine isopeptide bonds While the primary structure of transglutaminases is not conserved they all
• LHX8 is a member of the LIM homeobox family Members of this family share common structural features They all contain 2 tandemly repeated cysteine rich double zinc finger motifs called LIM domains
• ZSCAN12 may be involved in transcriptional regulation
• UNCX is a transcription factor involved in somitogenesis and neurogenesis It is required for the maintenance and differentiation of particular elements of the axial skeleton It may act upstream of P
• The developmentally regulated expression of the globin genes depends on upstream regulatory elements termed locus control regions LCRs LCRs are associated with powerful enhancer activity that is me
• ZNF131 may be involved in transcriptional regulation It plays a role during development and organogenesis as well as in the function of the adult central nervous system
• ZKSCAN1 may be involved in transcriptional regulation
• SMC3 belongs to the SMC3 subfamily of SMC proteins SMC3 occurs in certain cell types as either an intracellular nuclear protein or a secreted protein The nuclear form known as structural maintenan
• PA2G4 is an RNA binding protein that is involved in growth regulation This protein is present in pre ribosomal ribonucleoprotein complexes and may be involved in ribosome assembly and the regulation
• Huntington s disease HD a neurodegenerative disorder characterized by loss of striatal neurons is caused by an expansion of a polyglutamine tract in the HD protein huntingtin SETD2 is a protein b
• UBXD2 is an integral membrane protein of the endoplasmic reticulum ER that binds valosin containing protein and promotes ER associated protein degradation
• SEPN1 is a selenoprotein which contains a selenocysteine Sec residue at its active site Mutations in SEPN1 gene cause the classical phenotype of multiminicore disease and congenital muscular dystr
• IRX1 is a member of the Iroquois homeobox gene family Members of this family appear to play multiple roles during pattern formation of vertebrate embryos IRX1 is a member of the Iroquois homeobox gen
• MPND is a probable protease
• C3orf31 may be involved in the translocation of transit peptide containing proteins across the mitochondrial inner membrane
• Deficiency of human glycerate kinase leads to D glycerate acidemia D glyceric aciduria
• TBX15 is a probable transcriptional regulator involved in developmental processes
• ZNF169 may be involved in transcriptional regulation
• The function remain unknown
• NPNT is a functional ligand of integrin alpha 8 beta 1 in

kidney development It regulates with integrin alpha 8 beta 1 the expression of GDNF which is essential for kidney development It may also pl

• Cad regulates embryonic abdominal segment formation by zygotically activating expression of knirps kni and giant gt It plays a role in the establishment of the hindgut and in the invagination of

• Hb is a gap class segmentation protein that controls development of head structures

• Eve may play a role in determining neuronal identity It may be directly involved in specifying identity of individual neurons It is a pair rule protein required for segmentation involved in transfo

• Prd is a pair rule protein expressed in a segmentally repeating pattern to define the polarity of embryonic segments

• lab is a sequence specific transcription factor which is part of a developmental regulatory system that provides cells with specific positional identities on the anterior posterior axis It is require

• Pb is a sequence specific transcription factor which is part of a developmental regulatory system that provides cells with specific positional identities on the anterior posterior axis It controls de

• Antp is a sequence specific transcription factor which is part of a developmental regulatory system that regulates segmental identity in the mesothorax It provides cells with specific positional iden

• As a transcription factor exd acts with the selector homeodomain proteins altering the regulation of downstream target genes such as wingless teashirt and decapentaplegic Thus exd affects segmental

• DISC1 is a protein with multiple coiled coil motifs which is located in the nucleus cytoplasm and mitochondria The protein is involved in neurite outgrowth and cortical development through its inter

• DOT1L is a histone methyltransferase It methylates Lys 79 of histone H3 Nucleosomes are preferred as substrate compared to free histones

• UXT is a novel protein which is highly conserved in mouse It interacts with the N terminus of the androgen receptor and plays a role in facilitating receptor induced transcriptional activation It is

• NDRG1 is a member of the N myc downregulated protein family which belongs to the alpha beta hydrolase superfamily NDRG1 is a cytoplasmic protein involved in stress responses hormone responses cell

• NANOG is a new marker for testicular carcinoma in situ and germ cell tumors Gene knockdown of Nanog promotes differentiation thereby demonstrating a role for these factors in human embryonic stem ce

• CTAGE5 is a tumor associated antigen

• Members of arrestin beta arrestin protein family are thought to participate in agonist mediated desensitization of G protein coupled receptors and cause specific dampening of cellular responses to sti

• DKK1 is a protein that is a member of the dickkopf family It is a secreted protein with two cysteine rich regions and is involved in embryonic development through its inhibition of the WNT signaling

• The SMN complex plays an essential role in spliceosomal snRNP assembly in the cytoplasm and is required for pre mRNA splicing in the nucleus STRAP may play a role in the cellular distribution of the

• YWHAZ belongs to the 14 3 3 family of proteins which mediate signal transduction by binding to phosphoserine containing proteins This highly conserved protein family is found in both plants and mamma

• ST14 is an epithelial derived integral membrane serine protease This protease forms a complex with the Kunitz type serine protease inhibitor HAI 1 and is found to be activated by sphingosine 1 pho

• PAX8 is a member of the paired box PAX family of transcription factors Members of this family typically contain a paired box domain an octapeptide and a paired type homeodomain This nuclear prot

• MAPT is differentially expressed in the nervous system depending on stage of neuronal maturation and neuron type The mutations in the gene have been associated with several neurodegenerative disorde

• SLC14A1 is a specialized low affinity urea transporter It mediates urea transport in erythrocytes

• CD36 is the fourth major glycoprotein of the platelet surface and serves as a receptor for thrombospondin in platelets and various cell lines Since thrombospondins are widely distributed proteins inv

• ALDOA is a glycolytic enzyme that catalyzes the reversible conversion of fructose 1 6 bisphosphate to glyceraldehyde 3 phosphate and dihydroxyacetone phosphate Aldolase A is found in the developing e

• HADHB is the beta subunit of the mitochondrial trifunctional protein which catalyzes the last three steps of mitochondrial beta oxidation of long chain fatty acids The mitochondrial membrane bound h

• The pyruvate dehydrogenase complex is a nuclear encoded mitochondrial matrix multienzyme complex that provides the primary link between glycolysis and the tricarboxylic acid TCA cycle by catalyzing

• PGAM2 is the interconversion of 3 and 2 phosphoglycerate with 2 3 bisphosphoglycerate as the primer of the reaction It can also catalyze the reaction of EC 5 4 2 4 synthase and EC 3 1 3 13 phosph

• PGK1 is a glycolytic enzyme that catalyzes the conversion of 1 3 diphosphoglycerate to 3 phosphoglycerate The protein may also act as a cofactor for polymerase alpha The protein encoded by this gene

• P4HB is the beta subunit of prolyl 4 hydroxylase a highly abundant multifunctional enzyme that belongs to the protein disulfide isomerase family When present as a tetramer consisting of two alpha an

• PTGDS is a glutathione independent prostaglandin D synthase that catalyzes the conversion of prostaglandin H2 PGH2 to postaglandin D2 PGD2 PGD2 functions as a neuromodulator as well as a trophic

• EEF1A1 is an isoform of the alpha subunit of the elongation factor 1 complex which is responsible for the enzymatic delivery of aminoacyl tRNAs to the ribosome This isoform alpha 1 is expressed in

• EEF1G is a subunit of the elongation factor 1 complex which is responsible for the enzymatic delivery of aminoacyl tRNAs to the ribosome This subunit contains an N terminal glutathione transferase d

• IGFBP7 contains 1 Ig like C2 type immunoglobulin like domain 1 IGFBP N terminal domain and 1 Kazal like domain It binds IGF I and IGF II with a relatively low affinity IGFBP7 stimulates prostacyc

• AKR1B1 is a member of the aldo keto reductase superfamily which consists of more than 40 known enzymes and proteins This member catalyzes the reduction of a number of aldehydes including the aldehy

• ATP5B is a subunit of mitochondrial ATP synthase Mitochondrial ATP synthase catalyzes ATP synthesis utilizing an electrochemical gradient of protons across the inner membrane during oxidative phosph

• Alpha crystallins are composed of alpha A and alpha B for acidic and basic respectively They act as molecular chaperones although they do not renature proteins and release them in the fashion of a

• ENO3 is one of the three enolase isoenzymes found in mammals This isoenzyme a homodimer is found in skeletal muscle cells in the adult A switch from alpha enolase to beta enolase occurs in muscle

• Glutamic oxaloacetic transaminase is a pyridoxal phosphate dependent enzyme which exists in cytoplasmic and mitochondrial forms GOT1 and GOT2 respectively GOT plays a role in amino acid metabolism

• LDHB belongs to the LDH MDH superfamily LDH family Defects in LDHB are a cause of hereditary LDHB deficiency LDHB may also have roles in progression of medulloblastoma

• PEBP1 binds ATP opioids and phosphatidylethanolamine It has lower affinity for phosphatidylinositol and phosphatidylcholine It is also a serine protease inhibitor which inhibits thrombin neuropsin

• TPM2 is beta tropomyosin an isoform of tropomyosin that is mainly expressed in slow type 1 muscle fibers The TPM2 gene encodes beta tropomyosin an isoform of tropomyosin that is mainly expressed in

• Sarcomere assembly is regulated by the muscle protein titin Titin is a giant elastic protein with kinase activity that extends half the length of a sarcomere It serves as a scaffold to which myofibr

• RAB11B possesses GTPase activity

• PRDX6 is a member of the thiol specific antioxidant protein family This protein is a bifunctional enzyme with two distinct active sites It is involved in redox regulation of the cell it can reduce

• PFN1 is a ubiquitous actin monomer binding protein belonging to the profilin family It is thought to regulate actin polymerization in response to extracellular signals Deletion of this gene is assoc

• ALDOC gene is a member of the class I fructose biphosphate aldolase gene family ALDOC is a glycolytic enzyme that catalyzes the reversible aldol cleavage of fructose 1 6 biphosphate and fructose 1 ph

• Heterotrimeric guanine nucleotide binding proteins G proteins which integrate signals between receptors and effector proteins are composed of an alpha a beta and a gamma subunit These subunits

• Malate dehydrogenase catalyzes the reversible oxidation of malate to oxaloacetate utilizing the NAD NADH cofactor system in the citric acid cycle MDH1 is localized to the cytoplasm and may play pivo

• Malate dehydrogenase catalyzes the reversible oxidation of malate to oxaloacetate utilizing the NAD NADH cofactor system in the citric acid cycle MDH2 is localized to the mitochondria and may play p

• EIF3M is a broadly expressed protein containing putative membrane fusion domains that acts as a receptor or coreceptor for entry of herpes simplex virus HSV HFLB5 encodes a broadly expressed protein

• UGP2 is an important intermediary in mammalian carbohydrate interconversions It transfers a glucose moiety from glucose 1 phosphate to MgUTP and forms UDP glucose and MgPPi In liver and muscle tissu

• DSTN belongs to the actin binding proteins ADF family This family of proteins is responsible for enhancing the turnover rate of actin in vivo It is the actin depolymerizing protein that severs actin

• NDUFV1 is the 51 kD subunit of complex I NADH ubiquinone oxidoreductase of the mitochondrial respiratory chain The NDUFV1 gene encodes the 51 kD subunit of complex I NADH ubiquinone oxidoreductase

• PHB2 acts as a mediator of transcriptional repression by nuclear hormone receptors via recruitment of histone deacetylases By similarity It functions as an estrogen receptor ER selective coregula

• RSU1 is a protein that is involved in the Ras signal transduction pathway growth inhibition and nerve growth factor induced differentiation processes as determined in mouse and human cell line stud

• The function of GADD45B is involved in the regulation of growth and apoptosis This gene is a member of a group of genes whose transcript levels are increased following stressful growth arrest conditio

• Pseudouridine is an abundant component of rRNAs and tRNAs and is enzymatically generated by isomerization of uridine by pseudouridine synthase Pseudouridine is an abundant component of rRNAs and tRNAs

• GMPR2 catalyzes the irreversible NADPH dependent deamination of GMP to IMP It functions in the conversion of nucleobase nucleoside and nucleotide derivatives of G to A nucleotides and in maintainin

• PPA1 is a member of the inorganic pyrophosphatase PPase family PPases catalyze the hydrolysis of pyrophosphate to inorganic phosphate which is important for the phosphate metabolism of cells Stud

• NUDT16L1 is a probable adapter protein which may link syndecan 4 SDC4 and paxilin TGFB1I1 and PXN receptors

• HSPB6 is associated with actin and modulates smooth muscle relaxation HSPB6 is associated with actin see MIM 102540 and modulates smooth muscle relaxation Tessier et al 2003 PubMed 12842460

• SEPT10 is a member of the septin family of cytoskeletal proteins with GTPase activity This protein localizes to the cytoplasm and nucleus and displays GTP binding and GTPase activity This gene encode

• FUT6 is a Golgi stack membrane protein that is involved in the creation of sialyl Lewis X an E selectin ligand Mutations in this gene are a cause of fucosyltransferase 6 deficiency The protein enco

• Galactose 1 phosphate uridyl transferase GALT catalyzes the second step of the Leloir pathway of galactose metabolism namely the conversion of UDP glucose galactose 1 phosphate to glucose 1 phosp

• OXCT1 is a member of the 3 oxoacid CoA transferase gene family It is a homodimeric mitochondrial matrix enzyme that plays a central role in extrahepatic ketone body catabolism by catalyzing the rever

• CT1 is a member of the 3 oxoacid CoA transferase gene family It is a homodimeric mitochondrial matrix enzyme that plays a central role in extrahepatic ketone body catabolism by catalyzing the reversi

• The functions of TTC6 remain unknown

• NUDT17 belongs to the Nudix hydrolase family It probably mediates the hydrolysis of some nucleoside diphosphate derivatives

• Prenyltransferases attach either a farnesyl group or a geranylgeranyl group in thioether linkage to the cysteine residue of protein s with a C terminal CAAX box CAAX geranylgeranyltransferase and CAA

• The exact function of LOC390110 remains unknown

• GGPS1 is a member of the prenyltransferase family with geranylgeranyl diphosphate GGPP synthase activity The enzyme catalyzes the synthesis of GGPP from farnesyl diphosphate and isopentenyl diphosp

• TYMS catalyzes the methylation of deoxyuridylate to deoxythymidylate using 5 10 methylenetetrahydrofolate methylene THF as a cofactor This function maintains the dTMP thymidine 5 prime monophospha

• MFNG is one of the evolutionarily conserved secreted proteins that act in the Notch receptor pathway to demarcate boundaries during embryonic development Western blots using two different antibodies

• FDFT1 is a membrane associated enzyme located at a branch point in the mevalonate pathway The protein is the

first specific enzyme in cholesterol biosynthesis catalyzing the dimerization of two mole

• CDY1 containing a chromodomain and a histone acetyltransferase catalytic domain Chromodomain proteins are components of heterochromatin like complexes and can act as gene repressors Histone hyperace

• METTL1 contains a conserved S adenosylmethionine binding motif and is inactivated by phosphorylation This gene is similar in sequence to the S cerevisiae YDL201w gene The gene product contains a con

• Three classes of protein carboxyl methyltransferases distinguished by their methyl acceptor substrate specificity have been found in prokaryotic and eukaryotic cells The type II enzyme catalyzes th

• Involucrin a component of the keratinocyte crosslinked envelope is found in the cytoplasm and crosslinked to membrane proteins by transglutaminase Involucrin a component of the keratinocyte crossli

• MAP3K or MEK kinase is a serine threonine kinase that occupies a pivotal role in a network of phosphorylating enzymes integrating cellular responses to a number of mitogenic and metabolic stimuli i

• POLR2H is one of the essential subunits of RNA polymerase II that is shared by the other two eukaryotic DNA directed RNA polymerases I and III This gene encodes one of the essential subunits of RNA p

• Protein kinase activation is a frequent response of cells to treatment with growth factors chemicals heat shock or apoptosis inducing agents This protein kinase activation presumably allows cells

• LIAS belongs to the biotin and lipoic acid synthetases family It localizes in mitochondrion and plays an important role in alpha lipoic acid synthesis It may also function in the sulfur inserti

• POLS is a DNA polymerase that is likely involved in DNA repair In addition the protein may be required for sister chromatid adhesion The protein encoded by this gene is a DNA polymerase that is like

• STK38 belongs to the protein kinase superfamily AGC Ser Thr protein kinase family It contains 1 AGC kinase C terminal domain and 1 protein kinase domain NDR driven centrosome duplication requires C

• NAT6 belongs to the acetyltransferase family It contains 1 N acetyltransferase domain NAT6 seems to be involved in N acetylation Acts on peptides with a N terminal Met followed by Asp Glu Asn It m

• SEPHS1 is an enzyme that synthesizes selenophosphate from selenide and ATP Selenophosphate is the selenium donor used to synthesize selenocysteine which is co translationally incorporated into selen

• STK39 is a serine threonine kinase that is thought to function in the cellular stress response pathway The kinase is activated in response to hypotonic stress leading to phosphorylation of several c

• PGDS is a sigma class glutathione S transferase family member The enzyme catalyzes the conversion of PGH2 to PGD2 and plays a role in the production of prostanoids in the immune system and mast cells

• AASDHPPT is similar to Saccharomyces cerevisiae LYS5 which is required for the activation of the alpha aminoadipate dehydrogenase in the biosynthetic pathway of lysine Yeast alpha aminoadipate dehyd

• NUDT13 contains 1 nudix hydrolase domain The exact function of NUDT13 remains unknown

• The process of transferring lipoic acid to proteins is a two step process The first step is the activation of lipoic acid by lipoate activating enzyme to form lipoyl AMP For the second step LIPT1 t

• TRMU is a member of the trmU family It is a mitochondria specific tRNA modifying enzyme that is required for the 2 thio modification of 5 taurinomethyl 2 thiouridine tRNA Lys on the wobble position o

• POLR3B belongs to the RNA polymerase beta chain family DNA dependent RNA polymerase catalyzes the transcription of DNA into RNA using the four ribonucleoside triphosphates as substrates POLR3B is th

• METTL2B is a member of a family of methyltransferases that share homology with but are distinct from the UbiE family of methyltransferases This gene is a member of a family of methyltransferases tha

• CSGlcA T belongs to the chondroitin N acetylgalactosaminyltransferase family It transfers glucuronic acid GlcUA from UDP GlcUA to N acetylgalactosamine residues on the non reducing end of the elong

• PRTFDC1 belongs to the purine pyrimidine phosphoribosyltransferase family Epigenetic silencing of PRTFDC1 by hypermethylation of the CpG islands leads to a loss of PRTFDC1 function which might be in

• Carnitine octanoyltransferase is a carnitine acyltransferase that catalyzes the reversible transfer of fatty acyl groups between CoA and carnitine This provides a crucial step in the transport of med

• GMPPB is a GDP mannose pyrophosphorylase This enzyme catalyzes the reaction which converts mannose 1 phosphate and GTP to GDP mannose which is involved in the production of N linked oligosaccharides

• NSUN3 belongs to the methyltransferase superfamily RsmB NOP family It may have S adenosyl L methionine dependent methyl transferase activity

• OXCT2 is a testis specific succinyl CoA 3 oxoacid CoA transferase EC 2 8 3 5 which catalyzes the reversible transferof CoA from succinyl CoA to acetoacetate in the first step of ketone body utiliza

• The coenzyme NAD and its derivatives are involved in hundreds of metabolic redox reactions and are utilized in protein ADP ribosylation histone deacetylation and in some Ca 2 signaling pathways

• PGS1 functions in the biosynthesis of the anionic phospholipids phosphatidylglycerol and cardiolipin

• NARG1L may belong to a complex displaying N terminal acetyltransferase activity

• CXorf34 is a putative methyltransferase

• NAT13 is a probable catalytic component of the ARD1A NARG1 complex which displays alpha N terminal acetyltransferase activity

• PNPLA3 is a triacylglycerol lipase that mediates triacylglycerol hydrolysis in adipocytes The protein which appears to be membrane bound may be involved in the balance of energy usage storage in ad

• SETD7 is a histone methyltransferase that specifically monomethylates Lys 4 of histone H3 H3 Lys 4 methylation represents a specific tag for epigenetic transcriptional activation It plays a cent

• MAP2K2 is a dual specificity protein kinase that belongs to the MAP kinase kinase family This kinase is known to play a critical role in mitogen growth factor signal transduction It phosphorylates a

• Nucleotides are involved in numerous biochemical reactions and pathways within the cell as substrates cofactors and effectors Nudix hydrolases such as NUDT12 regulate the concentrations of indivi

• NSUN5C gene shares high sequence similarity with several genes in the Williams Beuren Syndrome critical region and its deletion is associated with this disorder This gene shares high sequence similari

• PHACS does not catalyze the synthesis of 1 aminocyclopropane 1 carboxylate but is capable of catalyzing the deamination of L vinylglycine

• ALG2 is a member of the glycosyltransferase 1 family It acts as an alpha 1 3 mannosyltransferase mannosylating Man 2 GlcNAc 2 dolichol diphosphate and Man 1 GlcNAc 2 dolichol diphosphate to fo

• Transglutaminases TGM EC 2 3 2 13 are a family of structurally and functionally related enzymes that stabilize protein assemblies through the formation of gamma glutamyl epsilon

• CHST14 belongs to the sulfotransferase 2 family CHST14 catalyzes the transfer of sulfate to position 4 of the N acetylgalactosamine GalNAc residue of dermatan sulfate It transfers sulfate to the C

• TTC5 is an adapter protein involved in p53 TP53 response that acts by regulating and mediating the assembly of multi protein complexes It is required to facilitate the interaction between JMY and p30

• TIGD3 belongs to the tigger subfamily of the pogo superfamily of DNA mediated transposons in humans These proteins are related to DNA transposons found in fungi and nematodes and more distantly to t

• The MAPEG Membrane Associated Proteins in Eicosanoid and Glutathione metabolism family includes a number of human proteins several of which are involved the production of leukotrienes LTC4S is an

• GSTZ1 is a member of the glutathione S transferase GSTs super family which are important in the detoxification of electrophilic molecules including carcinogens mutagens and several therapeutic dr

• CDYL2 contains 1 chromo domain The exact function of CDYL2 remains unknown

• TMTC2 is a multi pass membrane protein which contains 10 TPR repeats It belongs to the TMTC family The exact function of TMTC2 remains unknown

• METTL7B belongs to the methyltransferase superfamily It is a probable methyltransferase

• LYK5 belongs to the protein kinase superfamily STE Ser Thr protein kinase family STE20 subfamily It contains 1 protein kinase domain LYK5 is a pseudokinase which in complex with CAB39 binds to a

• HS3ST5 is the rate limiting enzyme for synthesis of HSact It performs the crucial step modification in the biosynthesis of anticoagulant heparan sulfate HSact that is to complete the structure of t

• GSTA5 belongs to the GST superfamily alpha family It contains 1 GST C terminal domain and 1 GST N terminal domain The exact functions of GSTA5 remain unknown

• The exact function of C3orf64 remains unknown

• ST6GALNAC4 is a type II membrane protein that catalyzes the transfer of sialic acid from CMP sialic acid to galactose containing substrates ST6GALNAC4 prefers glycoproteins rather than glycolipids as

• FTSJ1 is a member of the S adenosylmethionine binding protein family It is a nucleolar protein and may be involved in the processing and modification of rRNA The protein encoded by this gene is a mem

• Glycosylphosphatidylinositol GPI is a complex glycolipid that anchors many proteins to the cell surface PIGW acts in the third step of GPI biosynthesis and acylates the inositol ring of phosphatidy

• RABGGTA belongs to the protein prenyltransferase subunit alpha family and contains 6 PFTA repeats RABGGTA catalyzes the transfer of a geranyl geranyl moiety from geranyl geranyl pyrophosphate to both

• TGM6 belongs to the transglutaminase superfamily and catalyzes the cross linking of proteins and the conjugation of polyamines to proteins

• The sequence of LOC650515 is derived from an annotated genomic sequence NW 922073 using gene prediction method GNOMON supported by EST evidence The exact function of LOC650515 remains unknown

• The sequence of LOC652559 is derived from an annotated genomic sequence NW 922352 using gene prediction method GNOMON The exact function of LOC652559 remains unknown

• TEX2 is a multi pass membrane protein The exact function of TEX2 remains unknown

• This gene is a member of the protocadherin alpha gene cluster one of three related gene clusters tandemly linked on chromosome five that demonstrate an unusual genomic organization similar to that of

• C20orf3 may play a role in adipocyte differentiation

• C19orf15 is a single pass type I membrane protein The exact function of C19orf15 remains unknown

• CDH22 is a member of the cadherin superfamily CDH22 is composed of five cadherin repeat domains and a cytoplasmic tail similar to the highly conserved cytoplasmic region of classical cadherins

• ADAM30 is a member of the ADAM a disintegrin and metalloprotease domain family Members of this family are membrane anchored proteins structurally related to snake venom disintegrins and have been

• SLC22A23 belongs to a large family of transmembrane proteins that function as uniporters symporters and antiporters to transport organic ions across cell membranes SLC22A23 belongs to a large family

• LMBR1 is a member of the LMBR1 like membrane protein family Another member of this protein family has been shown to be a lipocalin transmembrane receptor A highly conserved cis acting regulatory mo

• Sulfate groups in carbohydrates confer highly specific functions on glycoproteins glycolipids and proteoglycans and are critical for cell cell interaction signal transduction and embryonic develop

• LPPR2 is a multi pass membrane protein and belongs to the PA phosphatase related phosphoesterase family LPPR2 shows dynamic expression regulation during brain development and neuronal excitation

• FNDC3B may be a positive regulator of adipogenesis

• The function of MGC4172 remains unknown

• Metalloreductase has the ability to reduce both Fe 3 to Fe 2 and Cu 2 to Cu 1 It uses NAD as acceptor

• UGT2A3 belongs to the UDP glycosyltransferase family UDP glucuronosyltransferases catalyze phase II biotransformation reactions in which lipophilic substrates are conjugated with glucuronic acid to i

• TREML2 is a single pass type I membrane protein and it contains 1 Ig like V type immunoglobulin like domain TREML2 is a cell surface receptor that may play a role in the innate and adaptive immune

• UBA5 is a member of the E1 like ubiquitin activating enzyme family UBA5 activates ubiquitin fold modifier 1 a ubiquitin like post translational modifier protein via the formation of a high energy t

• BTNL8 is a single pass type I membrane protein It belongs to the immunoglobulin superfamily BTN MOG family BTNL8 contains 1 B30 2 SPRY domain and 1 Ig like V type domain The exact function of BTNL

• ELOVL7 could be implicated in synthesis of very long chain fatty acids and sphingolipids

• LRRC8 is a multi pass membrane protein It contains 13 LRR leucine rich repeats LRRC8 involved in B cell development belongs to a novel family of leucine rich repeat proteins

• PQLC1 is a multi pass membrane protein It contains 2 PQ loop domains The exact function of PQLC1 remains unknown

• UNC84A is a a nuclear nuclear envelope protein with an

Unc84 SUN domain The protein is involved in nuclear anchorage and migration This gene is a member of the unc 84 homolog family and encodes a n

• ADAM33 is a member of the ADAM a disintegrin and metalloprotease domain family Members of this family are membrane anchored proteins structurally related to snake venom disintegrins and have been

• TRAF3IP3 stimulated cell growth by modulating the c Jun N terminal kinase JNK pathway TRAF3 interacts with Smac DIABLO via TRAF domain leading to an increased proapoptotic effect of Smac DIABLO in

• TRAF3IP3 may function as an adapter molecule that regulates TRAF3 mediated JNK activation

• REEP4 belongs to the DP1 family It may enhance the cell surface expression of odorant receptors

• TMPRSS5 belongs to the serine protease family Serine proteases are known to be involved in many physiological and pathological processes TMPRSS5 may play a role in hearing

• UNC93B1 is a protein with similarity to the C elegans unc93 protein The Unc93 protein is involved in the regulation or coordination of muscle contraction in the worm This gene encodes a protein with

• ST6GALNAC5 belongs to a family of sialyltransferases that modify proteins and ceramides on the cell surface to alter cell cell or cell extracellular matrix interactions ST6GALNAC5 belongs to a family

• CRISPLD2 is a novel NSCLP candidate gene

• MLSTD2 catalyzes the reduction of saturated fatty acyl CoA with chain length C16 or C18 to fatty alcohols

• ZDHHC16 may be involved in apoptosis regulation

• PPAPDC1B is a multi pass membrane protein and it belongs to the PA phosphatase related phosphoesterase family It may be a metastatic suppressor for hepatocellular carcinoma

• ORAI1 belongs to the Orai family ORAI1 CRACM1 is a plasma membrane protein essential for store operated calcium entry Defects in ORAI1 are a cause of severe combined immunodeficiency with CRAC cha

• TMEM118 is a multi pass membrane protein and contains 1 RING type zinc finger The function of TMEM118 remains unknown

• PCDH15 is a member of the cadherin superfamily Family members encode integral membrane proteins that mediate calcium dependent cell cell adhesion PCDH15 consists of a signal peptide 11 extracellula

• Acid phosphatases are enzymes capable of hydrolyzing orthophosphoric acid esters in an acid medium This gene is up regulated by androgens and is down regulated by estrogens in the prostate cancer cel

• Sialic acid binding immunoglobulin like lectins SIGLECs are a family of cell surface proteins belonging to the immunoglobulin superfamily They mediate protein carbohydrate interactions by selective

• FCRL1 may function as an activating coreceptor in B cells and it may function in B cells activation and differentiation

• TMC2 is considered a member of transmembrane proteins family The specific function of this gene is unknown however expression in the inner ear suggests that it may be crucial for normal auditory fu

• GIMAP1 belonging to the GTP binding superfamily and to the immuno associated nucleotide IAN subfamily of nucleotide binding proteins This gene encodes a protein belonging to the GTP binding superfam

• ZNF121 belongs to the krueppel C2H2 type zinc finger protein family It contains 11 C2H2 type zinc fingers ZNF121 may be involved in transcriptional regulation

• ZIK1 may be a transcriptional repressor

• ZNF763 may be involved in transcriptional regulation

• The exact function ofLOC401898 remains unknown

• HELT belongs to the HEY family It contains 1 basic helix loop helix bHLH domain and 1orange domain HELT is a transcriptional repressor which binds preferentially to the canonical E box sequence 5

• The exact function of DKFZp779O175 remains unknown

• SHC1 is a signaling adapter that couples activated growth factor receptors to signaling pathway Isoform p46Shc and isoform p52Shc once phosphorylated couple activated receptor tyrosine kinases to R

• EIF3H belongs to the peptidase M67 family It contains 1 MPN JAB Mov34 domain EIF3H binds to the 40S ribosome and promotes the binding of methionyl tRNAi and mRNA It associates with the p170 subun

• ING1 is a tumor suppressor protein that can induce cell growth arrest and apoptosis The protein is a nuclear protein that physically interacts with the tumor suppressor protein TP53 and is a componen

• SPRY3 contains 1 B30 2 SPRY domain Sprouty Caveolin 1 interaction modulates signaling in a growth factor and Sprouty isoform specific manner Spry 1 2 and 3 expression was observed in placental tis

• SPRY2 is a protein belonging to the sprouty family It contains a carboxyl terminal cysteine rich domain essential for the inhibitory activity on receptor tyrosine kinase signaling proteins and is req

• APC2 belongs to the adenomatous polyposis coli APC family It contains 6 ARM repeats APC2 promotes rapid degradation of CTNNB1 and may function as a tumor suppressor It may function in Wnt signali

• C1D is a DNA binding and apoptosis inducing protein and is localized in the nucleus It is also a Rac3 interacting protein which acts as a corepressor for the thyroid hormone receptor C1D is thought

• ZNHIT1 belongs to the ZNHIT1 family It contains 1 HIT type zinc finger ZNHIT1 may play a role in p53 mediated apoptosis induction

• PWP1 belongs to the WD repeat PWP1 family It contains 5 WD repeats PWP1 may play an important role in cell growth and or transcription

• CASP8AP2 is highly similar to FLASH a mouse apoptotic protein identified by its interaction with the death effector domain DED of caspase 8 Studies of FLASH protein suggested that this protein may

• PYGO1 contains 1 PHD type zinc finger PYGO1 is involved in signal transduction through the Wnt pathway

• TIPIN is required for normal progression of S phase It is important for cell survival after DNA damage or replication stress and may be specifically required for the ATR CHK1 pathway in the replicati

• TIPIN belongs to the CSM3 family TIPIN is required for normal progression of S phase It is important for cell survival after DNA damage or replication stress TIPIN may be specifically required for

• The function of this gene is not known but its existence is supported by mRNAs and EST data A similar gene in mice was found to be expressed preferentially in hematopoietic progenitors and mature bl

• ZC3H15 belongs to the ZC3H15 TMA46 family It contains 2 C3H1 type zinc fingers ZC3H15 protects DRG1 from proteolytic degradation

• The exact functions of NRIP3 remain unknown

• SMYD3 is a histone methyltransferase that plays a role in transcriptional regulation as a member of an RNA polymerase complex SMYD3 is a histone methyltransferase that plays a role in transcriptional

• This gene produces alternative transcripts encoding two distinct proteins One protein is a transcriptional repressor while the other isoform is a major component of specialized synapses known as syn

• OCEL1 belongs to the ELL occludin family The exact functions of OCEL1 remain unknown

• ZNF385D contains 3 matrin type zinc fingers The exact function of ZNF385D remains unknown

• ZBTB46 contains 1 BTB POZ domain and 2 C2H2 type zinc fingers ZBTB46 may be involved in transcriptional regulation

• CCNL2 belongs to the cyclin family Cyclin L subfamily CCNL2 is a transcriptional regulator which participates in regulating the pre mRNA splicing process CCNL2 also modulates the expression of crit

• NRIP2 down regulates transcriptional activation by nuclear receptors such as NR1F2

• Over expression of PDCD2L attenutates TNF alpha release in Daudi cells PDCD2L over expression restrained proliferation of HEK293T cells DNA flow cytometry analysis showed that the over expression of

• CIP29 physically and functionally associates with DDX39 suggesting their cooperation in the RNA metabolism CIP29 appears to be a new cytokine regulated protein involved in normal and cancer cell pro

• ZCCHC3 contains 3 CCHC type zinc fingers The exact function of ZCCHC3 remains unknown

• DNTTIP1 binds DNA and enhances the activity of terminal deoxynucleotidyltransferase TDT or DNTT a DNA polymerase that catalyzes the polymerization of DNA in the absence of a DNA template

• TCEAL2 is a member of the transcription elongation factor A SII like TCEAL gene family Members of this family contain TFA domains and may function as nuclear phosphoproteins that modulate transcr

• The predicted protein ZNF280A is similar to Drosophila suppressor of hairy wing protein a leucine zipper protein which represses the function of transcriptional enhancers of the gypsy retrotransposon

• ZNF618 belongs to the krueppel C2H2 type zinc finger protein family It contains 4 C2H2 type zinc fingers ZNF618 may be involved in transcriptional regulation

• DNER is an activator of the NOTCH1 pathway It may mediate neuron glia interaction during astrocytogenesis

• ZMAT3 is a protein containing three zinc finger domains and a nuclear localization signal The mRNA and the protein of this gene are upregulated by wildtype p53 and overexpression of this gene inhibit

• C1orf83 contains 1 TFIIS central domain and 1 TFIIS N terminal domain The exact functions of C1orf83 remain unknown

• VGLL2 belongs to the vestigial family It may act as a specific coactivator for the mammalian TEFs VGLL2 may play a role in the development of skeletal muscles

• LIN9 belongs to the lin 9 family and acts as a tumor suppressor It inhibits DNA synthesis Its ability to inhibit oncogenic transformation is mediated through its association with RB1 LIN9 plays a r

• WDFY3 is a protein which contains WD repeats and an FYVE domain WDFY3 might target cytosolic protein aggregates for autophagic degradation This gene encodes a protein which contains WD repeats and an

• Putative function of SPATA12 is to maintain the cell in a differentiated state and or to suppress cell proliferation in the human testis

• MDFIC is a member of a family of proteins characterized by a specific cysteine rich C terminal domain which is involved in transcriptional regulation of viral genome expression Alternative translati

• SP6 belongs to a family of transcription factors that contain 3 classical zinc finger DNA binding domains consisting of a zinc atom tetrahedrally coordinated by 2 cysteines and 2 histidines C2H2 moti

• The exact function of LOC727817 remains unknown

• The exact function of LOC728566 remains unknown

• The exact function of LOC730950 remains unknown

• The exact function of hCG 1646157 remains unknown

• The exact function of LOC731673 remains unknown

• The exact function of LOC729745 remains unknown

• The exact function of LOC649137 remains unknown

• The exact function of LOC732440 remains unknown

• The exact function of hCG 1660138 remains unknown

• ZFP92 belongs to the krueppel C2H2 type zinc finger protein family It contains 8 C2H2 type zinc fingers and 1 KRAB domain ZFP92 may be involved in transcriptional regulation

• The exact function of LOC344065 remains unknown

• The exact function of LOC390738 remains unknown

• The exact function of LOC391766 remains unknown

• The exact function of hCG 1982709 remains unknown

• The sequence of LOC442049 is derived from an annotated genomic sequence NW 921562 using gene prediction method GNOMON The exact function of LOC442049 remains unknown

• The exact function of LOC642623 remains unknown

• The functions of LOC653428 remain unknown

• The exact function of LOC653135 remains unknown

• The sequence of LOC641765 is derived from an annotated genomic sequence NT 079593 using gene prediction method GNOMON supported by EST evidence The exact function of LOC641765 remains unknown

• The exact function of LOC641765 remains unknown

• PRDM6 contains 4 C2H2 type zinc fingers and 1 SET domain PRDM6 may be involved in transcriptional regulation

• The exact function of hCG 2042202 remains unknown

• The exact function of ZNF389 remains unknown

• Apolipoprotein H has been implicated in a variety of physiologic pathways including lipoprotein metabolism coagulation and the production of antiphospholipid autoantibodies APOH may be a required c

• cdr2 normally sequesters c Myc in the neuronal cytoplasm thereby down regulating c Myc activity and suggest a mechanism whereby inhibition of cdr2 function by autoantibodies in PCD may contribute to

• NAGS is a mitochondrial enzyme that catalyzes the formation of N acetylglutamate NAG from glutamate and acetyl coenzyme A NAG is a cofactor of carbamyl phosphate synthetase I CPSI the first enzy

• The structure of MAS1 indicates that it belongs to the class of receptors that are coupled to GTP binding proteins and share a conserved structural motif which is described as a 7 transmembrane segm

• In vitro study revealed that RSRC2 might play a role in cell proliferation RSRC2 may be a novel tumor suppressor of esophageal cancer cell growth

• OLFM4 is a member of the olfactomedin related protein family The exact function of its gene has not yet been determined This gene was originally cloned from human myeloblasts and found to be selectiv

• Variation in NEDD9 is associated wih susceptibility to late onset Alzheimer and Parkinson diseases Changes in expression of the scaffold protein HEF1 CAS L NEDD9 were found to be a potent prometastat

• C3orf10 is involved in regulation of actin and microtubule organization It is a part of a WAVE complex that activates the Arp2 3 complex

• PHF6 is a member of the plant homeodomain PHD like finger PHF family PHF6 is a protein with two PHD type zinc finger domains indicating a potential role in transcriptional regulation that locali

• CACYBP is a calcyclin binding protein It may be involved in calcium dependent ubiquitination and subsequent proteosomal degradation of target proteins It probably serves as a molecular bridge in ubi
• Troponin I is the inhibitory subunit of troponin the thin filament regulatory complex which confers calcium sensitivity to striated muscle actomyosin ATPase activity
• Glycoprotein Ib GP Ib is a platelet surface membrane glycoprotein composed of a heterodimer an alpha chain and a beta chain that are linked by disulfide bonds The Gp Ib functions as a receptor fo
• PPIA is a member of the peptidyl prolyl cis trans isomerase PPIase family PPIases catalyze the cis trans isomerization of proline imidic peptide bonds in oligopeptides and accelerate the folding of
• GBX 1 contains 1 homeobox DNA binding domain The exact functions of GBX 1 remain unknown
• ZNF14 contains a zinc finger and a Kruppel associated box KRAB domain KRAB domain is known to be involved in the transcriptional repression of a number of zinc finger proteins The protein encoded b
• ZNF33A belongs to the krueppel C2H2 type zinc finger protein family It contains 16 C2H2 type zinc fingers and 1 KRAB domain ZNF33A may be involved in transcriptional regulation
• TRIP6 is a member of the zyxin family and encodes a protein with three LIM zinc binding domains This protein localizes to focal adhesion sites and along actin stress fibers Recruitment of TRIP6 to t
• CBX2 Contains 1 A T hook DNA binding domain and 1 chromo domain CBX2 is a component of the Polycomb group PcG multiprotein PRC1 complex a complex required to maintain the transcriptionally repress
• MBNL2 is a C3H type zinc finger protein which is similar to the Drosophila melanogaster muscleblind B protein MBNL2 is a RNA binding protein that binds to 5 ACACCC 3 core sequence It binds to CUG t
• PAXIP1 functions as a key component of the DNA damage response and phosphoserine or phosphothreonine specific binding modules PAXIP1 recognizes substrates phosphorylated by the kinases ataxia telang
• The exact functions of HSU79252 remain unknown
• SERTAD2 contains 1 SERTA domain It acts at E2F responsive promoters to integrate signals provided by PHD and or bromodomain containing transcription factors
• ZNF562 belongs to the krueppel C2H2 type zinc finger protein family It contains 8 C2H2 type zinc fingers and 1 KRAB domain ZNF562 may be involved in transcriptional regulation
• FHL5 is coordinately expressed with activator of cAMP responsive element modulator CREM It is associated with CREM and confers a powerful transcriptional activation function CREM acts as a transcr
• NUFIP2 binds RNA It is phosphorylated upon DNA damage probably by ATM or ATR The exact functions of NUFIP2 remain unknown
• TRIM48 belongs to the TRIM RBCC family It contains 1 B box type zinc finger 1 B30 2 SPRY domain and 1 RING type zinc finger The exact function of TRIM48 remains unknown
• MICALL1 contains 1 CH calponin homology domain and 1 LIM zinc binding domain It may be a cytoskeletal regulator
• ZNF486 belongs to the krueppel C2H2 type zinc finger protein family It contains 2 C2H2 type zinc fingers and 1 KRAB domain ZNF486 may be involved in transcriptional regulation
• ZNF681 belongs to the krueppel C2H2 type zinc finger protein family It Contains 16 C2H2 type zinc fingers ZNF681 may be involved in transcriptional regulation
• TP53I13 may act as a tumor suppressor It inhibits tumor cell growth when overexpressed TP53I13 was up regulated by genotoxic stresses of adriamycin and or ultraviolet beam irradiation in a p53 depe
• MFSD3 belongs to the major facilitator superfamily The exact functions of MFSD3 remain unknown
• The exact function of DEPDC7 remains unknown
• ZNF664 belongs to the krueppel C2H2 type zinc finger protein family It contains 9 C2H2 type zinc fingers ZNF664 may be involved in transcriptional regulation
• SSX8 belongs to the family of highly homologous synovial sarcoma X SSX breakpoint proteins These proteins may function as transcriptional repressors They are also capable of eliciting spontaneousl
• The exact functions of C8orf77 remain unknown
• The exact function of TPRXL remains unknown
• EBF4 belongs to the COE family and contains 1 IPT TIG domain EBF4 is a transcriptional factor which recognizes variations of the palindromic sequence 5 ATTCCCNNGGGAATT 3
• CROCCL2 belongs to the rootletin family The exact functions of CROCCL2 remain unknown
• The exact functions of DKFZp686E2433 remain unknown
• The exact functions of TNRC18 remain unknown
• ZNF90 belongs to the krueppel C2H2 type zinc finger protein family It contains at least 2 C2H2 type zinc fingers and 1 KRAB domain ZNF90 may be involved in transcriptional regulation
• Septin 9 is a member of the septin family which contains cytoplasmic cytoskeletal filament forming proteins that have a conserved GTP binding domain Septin 9 is a member of the septin family which c
• CYP1B1 is a member of the cytochrome P450 superfamily of enzymes The cytochrome P450 proteins are monooxygenases which catalyze many reactions involved in drug metabolism and synthesis of cholesterol
• C14orf174 contains 1 SAM sterile alpha motif domain The exact functions of C14orf174 remain unknown
• LIPJ belongs to the AB hydrolase superfamily lipase family The exact function of LIPJ remains unknown
• ROPN1B belongs to the ropporin family and contains 1 RIIa domain ROPN1B interacts with RHPN1 and AKAP3 It may interact with SPA17
• ASB11 is a member of the ankyrin repeat and SOCS box containing ASB family of proteins They contain ankyrin repeat sequence and SOCS box domain The SOCS box serves to couple suppressor of cytokine
• This gene Gcom1 is part of a complex transcript unit that includes the gene for glutamate receptor ionotropic N methyl D aspartate like 1A GRINL1A Transcription of this gene occurs at an upstre
• LRRC17 contains 6 LRR leucine rich repeats The exact function of LRRC17 remains unknown
• CHN2 is a protein with a phorbol ester DAG type zinc finger a Rho GAP domain and an SH2 domain This protein has GTPase activating protein activity that is regulated by phospholipid binding and bindi
• The functions of LOC202134 remain unknown
• CPN2 contains 13 LRR leucine rich repeats The 83 kDa subunit binds and stabilizes the catalytic subunit at 37 degrees Celsius and keeps it in circulation Under some circumstances it may be an allo
• In eukaryotic cells protein transport between the endoplasmic reticulum and Golgi compartments is mediated in part by non clathrin coated vesicular coat proteins COPs Seven coat proteins have been
• C4orf33 belongs to the UPF0462 family The exact function of C4orf33 remains unknown
• NHEDC1 is a sodium hydrogen exchanger and transmembrane protein Highly conserved orthologs of this gene have been found in other mammalian species The expression of NHEDC1 may be limited to testis
• Adaptins are important components of clathrin coated vesicles transporting ligand receptor complexes from the plasma membrane or from the trans Golgi network to lysosomes The adaptin family of protei
• Chondroadherin is a cartilage matrix protein thought to mediate adhesion of isolated chondrocytes CHAD contains 11 leucine rich repeats flanked by cysteine rich regions The chondroadherin messenger
• CHI3L1 may play an important role in the capacity of cells to respond to and cope with changes in their environment CHI3L1 may play a role in tissue remodeling and defense against pathogens It belon
• CSNK1G2 belongs to the protein kinase superfamily CK1 Ser Thr protein kinase family casein kinase I subfamily It contains 1 protein kinase domain Casein kinases are operationally defined by their
• CTF1 induces cardiac myocyte hypertrophy in vitro It binds to and activates the ILST gp130 receptor It belongs to the IL 6 superfamily
• Diphthamide is a unique posttranslationally modified histidine found only in translation elongation factor 2 EEF2 MIM 130610 This modification is conserved from archaebacteria to humans and serves
• This gene is one of two human genes similar to the yeast gene dph2 The yeast gene was identified by its ability to complement a diphthamide mutant strain and thus probably functions in diphthamide b
• CHN1 contains 1 phorbol ester DAG type zinc finger 1 Rho GAP domain and 1 SH2 domain CHN1 is a GTPase activating protein for p21 rac and a phorbol ester receptor It may play an important role in ne
• DNTT is a template independent DNA polymerase that catalyzes the addition of deoxynucleotides to the 3 hydroxyl terminus of oligonucleotide primers In vivo DNTT is expressed in a restricted populat
• CNN2 which can bind actin calmodulin troponin C and tropomyosin may function in the structural organization of actin filaments CNN2 could play a role in smooth muscle contraction and cell adhesi
• Casein kinases are operationally defined by their preferential utilization of acidic proteins such as caseins as substrates CSNK1G3 can phosphorylate a large number of proteins It participates in Wn
• DYNC1I1 belongs to the dynein intermediate chain family It contains 7 WD repeats The intermediate chains seem to help dynein bind to dynactin 150 kDa component DYNC1I1 may play a role in mediating
• ARPC3 is one of seven subunits of the human Arp2 3 protein complex The Arp2 3 protein complex has been implicated in the control of actin polymerization in cells and has been conserved through evolut
• ACTR1B is a 42 3 kD subunit of dynactin a macromolecular complex consisting of 10 subunits ranging in size from 22 to 150 kD Dynactin binds to both microtubules and cytoplasmic dynein and is involve
• BCKDK belongs to the PDK BCKDK protein kinase family It contains 1 histidine kinase domain BCKDK catalyzes the phosphorylation and inactivation of the branched chain alpha ketoacid dehydrogenase com
• This gene is deleted in the primary tumor of hepatocellular carcinoma It maps to 8p22 p21 3 a region frequently deleted in solid tumors It is suggested that this gene is a candidate tumor suppresso
• APPBP2 interacts with microtubules and is functionally associated with beta amyloid precursor protein transport and or processing The beta amyloid precursor protein is a cell surface protein with sig
• NudC was first identified as a regulator of nuclear movement in the asexual reproductive cycle of the filamentous fungus Aspergillus nidulans Human NUDC is a nuclear movement protein that associates
• RFPL2 contains 1 B30 2 SPRY domain and 1 RING type zinc finger The human RFPL2 gene has a role in neocortex development
• HBS1L belongs to the GTP binding elongation factor family The HBS1L MYB intergenic region on chromosome 6q23 3 influences erythrocyte platelet and monocyte counts HBS1L related genetic variants pl
• WASF3 is a member of the Wiskott Aldrich syndrome protein family It is a protein that forms a multiprotein complex that links receptor kinases and actin Binding to actin occurs through a C terminal
• METAP2 is a member of the methionyl aminopeptidase family and encodes a protein that binds 2 cobalt or manganese ions This protein functions both by protecting the alpha subunit of eukaryotic initiat
• The nuclear pore complex is a massive structure that extends across the nuclear envelope forming a gateway that regulates the flow of macromolecules between the nucleus and the cytoplasm Nucleoporin
• PADI2 encodes a member of the peptidyl arginine deiminase family of enzymes which catalyze the post translational deimination of proteins by converting arginine residues into citrullines in the prese
• ALDH1L1 catalyzes the conversion of 10 formyltetrahydrofolate NADP and water to tetrahydrofolate NADPH and carbon dioxide ALDH1L1 belongs to the aldehyde dehydrogenase family and is responsible f
• PRDX3 is a protein with antioxidant function and is localized in the mitochondrion Expression of this gene product in E coli deficient in the C22 subunit gene rescued resistance of the bacteria to a
• The coatomer is a cytosolic protein complex that binds to dilysine motifs and reversibly associates with Golgi non clathrin coated vesicles which further mediate biosynthetic protein transport from t
• ECT2 is a transforming protein that is related to Rho specific exchange factors and yeast cell cycle regulators The expression of ECT2 is elevated with the onset of DNA synthesis and remains elevated
• EGLN3 catalyzes the post translational formation of 4 hydroxyproline in hypoxia inducible factor HIF alpha proteins It hydroxylates HIF 1 alpha at Pro 564 and HIF 2 alpha EGLN3 functions as a c
• TOE1 belongs to the CAF1 family and 1 C3H1 type zinc finger TOE1 inhibits cell growth rate and cell cycle It induces CDKN1A expression as well as TGF beta expression and mediates the inhibitory grow
• COL4A3BP is a kinase that specifically phosphorylates the N terminal region of the non collagenous domain of the alpha 3 chain of type IV collagen known as the Goodpasture antigen Goodpasture diseas
• CC2D1B belongs to the CC2D1 family It contains 1 C2 domain A function of Cc2d1b Cc2d1a and their Drosophila homologue l 2 gd in D melanogaster in Notch trafficking have been reported
• LACTB belongs to the peptidase S12 family LACTB is a protein from the large 39S subunit of the mitochondrial ribosome mitoribosome It has some sequence similarity to prokaryotic beta lactamases bu
• VPS26B belongs to the VPS26 family VPS26B is probable component of the retromer complex a complex required to retrieve lysosomal enzyme receptors IGF2R and M6PR from endosomes to the trans Golgi n
• GBP4 belongs to the GBP family It binds GTP GDP and GMP Hydrolyzes GTP very efficiently GDP rather than GMP

is the major reaction product GBP4 plays a role in erythroid differentiation

• The exact function of C8orf34 remains unknown

• ACTRT2 belongs to the actin family Studies have shown that this protein may be involved in cytoskeletal organization similar to other cytoplasmic actin related protein ARP subfamily members Antibo

• Cerebellin is a sixteen aa peptide found mainly in the adrenal medulla where it has been shown to have a neuromodulatory function Cerebellin is derived from precerebellin a protein with sequence si

• EXOD1 belongs to the EXOD1 family It contains 1 exonuclease domain EXOD1 is a member of a new subclass of exonucleases called the 3 hExo ERI 1 subfamily of DEDDh nucleases

• DYNLL2 belongs to the dynein light chain family DYNLL2 may be involved in some aspects of dynein related intracellular transport and motility It may play a role in changing or maintaining the spatia

• UBE2F belongs to the ubiquitin conjugating enzyme family UBE2F accepts the ubiquitin like protein NEDD8 from the UBA3 NAE1 E1 complex and catalyzes its covalent attachment to other proteins

• The exact function of C20orf141 remains unknown

• C20orf141 is a single pass membrane protein The exact function of C20orf141 remains unknown

• NEURL2 contains 1 NHR neuralized homology repeat domain and 1 SOCS box domain NEURL2 plays an important role in the process of myofiber differentiation and maturation NEURL2 is the probable substr

• DUSP19 belongs to the protein tyrosine phosphatase family non receptor class dual specificity subfamily It contains 1 tyrosine protein phosphatase domain DUSP19 has a dual specificity toward Ser Th

• RAB40A belongs to the small GTPase superfamily Rab family It contains 1 SOCS box domain RAB40A may be a substrate recognition component of a SCF like ECS Elongin Cullin SOCS box protein E3 ubiqui

• Calcium dependent membrane binding proteins may regulate molecular events at the interface of the cell membrane and cytoplasm This gene is one of several genes that encode a calcium dependent protein

• ANKS3 contains 1 SAM sterile alpha motif domain and 6 ANK repeats The exact function of ANKS3 remains unknown

• EMID2 contains 1 EMI domain and 2 collagen like domains The exact function of ZCCHC3 remains unknown

• BMPER contains 1 TIL trypsin inhibitory like domain 5 VWFC domains and 1 VWFD domain BMPER is the inhibitor of bone morphogenetic protein BMP functions It may regulate BMP responsiveness of oste

• HUS1Bis most closely related to HUS1 a component of a cell cycle checkpoint protein complex involved in cell cycle arrest in response to DNA damage HUS1B can interact with the check point protein RA

• C15orf27 is a multi pass membrane protein The exact function of C15orf27 remains unknown

• C1orf55 is phosphorylated upon DNA damage probably by ATM or ATR The exact function of C1orf55 remains unknown

• FAM71A belongs to the FAM71 family The exact function of C15orf27 remains unknown

• KLHDC1 contains 6 Kelch repeats KLHDC1 and KLHDC2 have differential localization and activity in cultured mammalian cells The exact function of KLHDC1 remains unknown

• FBXO16 is a member of the F box protein family members of which are characterized by an approximately 40 amino acid motif the F box The F box proteins constitute one of the four subunits of ubiquit

• DCUN1D3 contains 1 DCUN1 domain The exact function of DCUN1D3 remains unknown

• The exact function of CCDC78 remains unknown

• FUNDC1 belongs to the FUN14 family The exact function of FUNDC1 remains unknown

• ZCCHC12 contains 1 CCHC type zinc finger ZCCHC12 is the transcriptional coactivator in the bone morphogenetic protein BMP signaling pathway It positively modulates BMP signaling by interacting wit

• CCDC50 is a soluble cytoplasmic tyrosine phosphorylated protein with multiple ubiquitin interacting domains Mutations in this gene cause nonsyndromic postlingual progressive sensorineural DFNA44

• TIP39 is related to parathyroid hormone PTH and PTH related protein PTHRP and is a ligand for PTH receptor 2 TIP39 is related to parathyroid hormone PTH MIM 168450 and PTH related protein PTHR

• LRRC50 contains 6 LRR leucine rich repeats It is proposed that LRRC50 to be a novel candidate gene for human cystic kidney disease involved in regulation of microtubule based cilia and actin based

• The exact function of CCDC60 remains unknown

• The exact function of C10orf83 remains unknown

• Sodium hydrogen antiporters such as NHEDC2 convert the proton motive force established by the respiratory chain or the F1F0 mitochondrial ATPase into sodium gradients that drive other energy requiri

• LAYN contains 1 C type lectin domain It is the receptor for hyaluronate

• LCA5 is a protein that is thought to be involved in centrosomal or ciliary functions Mutations in this gene cause Leber congenital amaurosis type V This gene encodes a protein that is thought to be

• SERPINA5 belongs to the serpin family It Inhibits activated protein C as well as plasminogen activators

• IL13RA2 is closely related to Il13RA1 a subuint of the interleukin 13 receptor complex This protein binds IL13 with high affinity but lacks cytoplasmic domain and does not appear to function as a

• IL13RA2 is closely related to Il13RA1 a subuint of the interleukin 13 receptor complex IL13RA2 binds IL13 with high affinity but lacks cytoplasmic domain and does not appear to function as a signa

• Carnitine acetyltransferase CRAT is a key enzyme in the metabolic pathway in mitochondria peroxisomes and endoplasmic reticulum CRAT catalyzes the reversible transfer of acyl groups from an acyl C

• PDE1A belongs to the cyclic nucleotide phosphodiesterase family It has a higher affinity for cGMP than for cAMP The exact function of PDE1A remains unknown

• GRK4 is a member of the guanine nucleotide binding protein G protein coupled receptor kinase subfamily of the Ser Thr protein kinase family The protein phosphorylates the activated forms of G prote

• AP2B1is one of two large chain components of the assembly protein complex 2 which serves to link clathrin to receptors in coated vesicles AP2B1 is found on the cytoplasmic face of coated vesicles in

• AP2B1 is one of two large chain components of the assembly protein complex 2 which serves to link clathrin to receptors in coated vesicles AP2B1 is found on the cytoplasmic face of coated vesicles i

• Fertilization consists of a sequence of specific cell cell interactions culminating in the fusion of the sperm and egg plasma membranes Recognition binding and fusion occur through the interaction

• The major pathway for the biosynthesis of phosphatidylcholine occurs via the CDP choline pathway CHKA is the initial enzyme in the sequence and may play a regulatory role It also catalyzes the phosp

• ADAM2 is a member of the ADAM a disintegrin and metalloprotease domain family Members of this family are membrane anchored proteins structurally related to snake venom disintegrins and have been i

• Lactate dehydrogenase C catalyzes the conversion of L lactate and NAD to pyruvate and NADH in the final step of anaerobic glycolysis LDHC is testis specific and belongs to the lactate dehydrogenase f

• The outer dense fibers are cytoskeletal structures that surround the axoneme in the middle piece and principal piece of the sperm tail The fibers function in maintaining the elastic structure and rec

• TNP1 is a spermatid specific product of the haploid genome which replaces histone and is itself replaced in the mature sperm by the protamines Transition protein 1 is a spermatid specific product of t

• PDXK phosphorylates vitamin B6 a step required for the conversion of vitamin B6 to pyridoxal 5 phosphate an important cofactor in intermediary metabolism PDXK is cytoplasmic and probably acts as a

• The ADAM family is composed of zinc binding proteins that can function as adhesion proteins and or endopeptidases They are involved in a number of biologic processes including fertilization neuroge

• Nucleoporins are the main components of the nuclear pore complex NPC of eukaryotic cells They are involved in the bidirectional trafficking of molecules especially mRNAs and proteins between the

• COIL is an integral component of Cajal bodies also called coiled bodies Cajal bodies are nuclear suborganelles of varying number and composition that are involved in the post transcriptional modifi

• Metallothionein proteins are highly conserved low molecular weight cysteine rich proteins that are induced by and bind to heavy metal ions and have no enzymatic activity They may play a central role

• CDKN3 belongs to the dual specificity protein phosphatase family It was identified as a cyclin dependent kinase inhibitor and has been shown to interact with and dephosphorylate CDK2 kinase thus p

• The nuclear pore complex NPC is comprised of approximately 50 unique proteins collectively known as nucleoporins The 98 kD nucleoporin is localized to the nucleoplasmic side of the NPC Rat studies

• HSPA1L is a 70kDa heat shock protein In conjunction with other heat shock proteins this protein stabilizes existing proteins against aggregation and mediates the folding of newly translated proteins

• CCIN is a basic protein of the sperm head cytoskeleton This protein contains kelch repeats and a BTB POZ domain and is necessary for normal morphology during sperm differentiation The protein encoded

• LGALS8 is a member of the galectin family Galectins are beta galactoside binding animal lectins with conserved carbohydrate recognition domains The galectins have been implicated in many essential f

• CCT6B is a molecular chaperone that is a member of the chaperonin containing TCP1 complex CCT also known as the TCP1 ring complex TRiC This complex consists of two identical stacked rings each

• STAG3 is a meiosis specific component of cohesin complex The cohesin complex is required for the cohesion of sister chromatids after DNA replication The cohesin complex apparently forms a large prot

• TEKT2 belongs to the tektin family of proteins Tektins comprise a family of filament forming proteins that are coassembled with tubulins to form ciliary and flagellar microtubules TEKT2 expresses in

• FEM1B is a component of an E3 ubiquitin protein ligase complex in which it may act as a substrate recognition subunit It involved in apoptosis by acting as a death receptor associated protein that m

• Sterile alpha motifs SAMs in proteins such as SAMD4A are part of an RNA binding domain that functions as a posttranscriptional regulator by binding to an RNA sequence motif known as the Smaug recogn

• Myoinositol the most common naturally occurring form of inositol is a component of plasma membrane phospholipids and functions as a cell signaling molecule ISYNA1 EC 5 5 1 4 or IPS is a rate li

• SPAG11B is one of several androgen dependent epididymis specific secretory proteins The specific functions of these proteins have not been determined but they are thought to be involved in sperm ma

• UBQLN3 is an ubiquitin like protein ubiquilin that shares high degree of similarity with related products in yeast rat and frog Ubiquilins contain a N terminal ubiquitin like domain and a C termin

• FAM46C belongs to the FAM46 family The exact function of FAM46C remains unknown

• TTC12 contains 3 TPR repeats Hypermethylation of TTC12 gene may play a role in acute lymphoblastic leukemia Haplotypic variants in DRD2 ANKK1 TTC12 and NCAM1 are associated with comorbid alcohol

• SPATA6 belongs to the SPATA6 family SPATA6 may play a role in spermatid maturation or sperm function

• LYZL6 belongs to the glycosyl hydrolase 22 family The exact function of LYZL6 remains unknown

• TSKS may play a role in testicular physiology spermatogenesis or spermiogenesis Expression of TSKS is highest in the testis and down regulated in testicular cancer The gene is localized to the regi

• TSKS may play a role in testicular physiology spermatogenesis or spermiogenesis Expression of the TSKS is highest in the testis and down regulated in testicular cancer The gene encoded TSKS is loca

• AGBL5 belongs to the peptidase M14 family The exact function of AGBL5 remains unknown

• CLMN is a single pass type IV membrane protein It contains 1 actin binding domain and 2 CH calponin homology domains The exact function of CLMN remains unknown

• GSG1 belongs to the GSG1 family GSG1 may cause the redistribution of PAPOLB from the cytosol to the endoplasmic reticulum

• Mitochondrial ADP ATP carriers such as SLC25A31 are nuclear coded mitochondrial proteins that catalyze the exchange of ATP generated in mitochondria by ATP synthase see MIM 108729 against ADP prod

• GK2 belongs to the FGGY kinase family GK2 is a key enzyme in the regulation of glycerol uptake and metabolism

• IFT122 is a member of the WD repeat protein family WD repeats are minimally conserved regions of approximately 40 amino acids typically bracketed by gly his and trp asp GH WD which may facilitate

• PGK2 is a testis specific form of phosphoglycerate kinase EC 2 7 2 3 which catalyzes the reversible conversion of 1 3 diphosphoglycerate to 3 phosphoglycerate during glycolysis generating one mole

• TEX14 belongs to the protein kinase superfamily It contains 3 ANK repeats and 1 protein kinase domain TEX14 is required for spermatogenesis and male fertility It may be required for normal structur

• C17orf81 belongs to the ELP5 family C17orf81 may be involved in TP53 mediated transcriptional regulation

• PRKAA1 belongs to the ser thr protein kinase family It is

the catalytic subunit of the 5 prime AMP activated protein kinase AMPK AMPK is a cellular energy sensor conserved in all eukaryotic cells
• ZC3H14 belongs to the ZC3H14 family It contains 2 C3H1 type zinc fingers ZC3H14 binds the polyadenosine RNA oligonucleotides
• ATG5 is required for autophagy It conjugates to ATG12 and associates with isolation membrane to form cup shaped isolation membrane and autophagosome The conjugate detaches from the membrane immediat
• Autophagy is the major intracellular degradation system delivering cytoplasmic components to lysosomes and it accounts for degradation of most long lived proteins and some organelles Cytoplasmic con
• Autophagy is a process for the bulk degradation of cytosolic compartments by lysosomes ATG10 is an E2 like enzyme involved in 2 ubiquitin like modifications essential for autophagosome formation ATG
• The exact function of C10orf132 remains unknown
• ETFB is the electron transfer flavoprotein beta polypeptide which shuttles electrons between primary flavoprotein dehydrogenases involved in mitochondrial fatty acid and amino acid catabolism and th
• HAGH belongs to the metallo beta lactamase superfamily glyoxalase II family It is classified as a thiolesterase and is responsible for the hydrolysis of S lactoyl glutathione to reduced glutathione
• PCDH17 contains six extracellular cadherin domains a transmembrane domain and a cytoplasmic tail differing from those of the classical cadherins It may play a role in the establishment and function
• LCK is a member of the Src family of protein tyrosine kinases PTKs It is a key signaling molecule in the selection and maturation of developing T cells It contains N terminal sites for myristylati
• MMD2 contains 1 COMM domain The exact function of MMD2 remains unknown
• Cdk4 is probably involved in the control of the cell cycle
• The protein encoded by KRT15 is a member of the keratin gene family The keratins are intermediate filament proteins responsible for the structural integrity of epithelial cells and are subdivided int
• Suppression of the expression of TRAP1 in mitochondria might play an important role in the induction of apoptosis caused via formation of ROS
• Alpha 1 antitrypsin is a protease inhibitor deficiency of which is associated with emphysema and liver disease The protein is encoded by a gene PI located on the distal long arm of chromosome 14
• In C elegans Ced4 binds and activates Ced3 an apoptotic initiator caspase via caspase associated recruitment domains CARDs Human Ced4 homologs include APAF1 NOD1 and NOD2 These proteins have
• CASP3 is a protein which is a member of the cysteine aspartic acid protease caspase family Sequential activation of caspases plays a central role in the execution phase of cell apoptosis Caspases
• CFLAR might contribute to the carcinogenesis and aggressiveness of endometrial carcinoma and might be a useful prognostic factor in the tumor CFLAR is specially overexpressed in colon cancers and it
• TNFRSF11B is a member of the TNF receptor superfamily This protein is an osteoblast secreted decoy receptor that functions as a negative regulator of bone resorption This protein specifically binds
• The BCL2 gene encodes an integral outer mitochondrial membrane protein that blocks the apoptotic death of some cells such as lymphocytes Constitutive expression of BCL2 such as in the case of transl
• Phospho cdk2 Thr160 probably involved in the control of the cell cycle It interacts with cyclins A D or E Activity of cdk2 is maximal during S phase and G2
• Histone H3 along with histone H4 plays a central role in nucleosome formation
• The protein encoded by FADD is an adaptor molecule that interacts with various cell surface receptors and mediates cell apoptotic signals Through its C terminal death domain this protein can be recr
• GSK3alpha is a potential regulator of platelet function and improves insulin action and glucose metabolism in skeletal muscle
• CRKL has been shown to activate the RAS and JUN kinase signaling pathways and transform fibroblasts in a RAS dependent fashion It is a substrate of the BCR ABL tyrosine kinase and plays a role in fib
• The SEPT4 gene is a member of the septin gene family of nucleotide binding proteins originally described in yeast as cell division cycle regulatory proteins Septins are highly conserved in yeast Dr
• BRCA1 which functions as a tumor suppressor in human breast cancer cells is a nuclear phosphoprotein which associates with RNA polymerase II holoenzyme Mutations in BRCA1 are predicted to be respon
• BAX encodes a protein that belongs to the BCL2 protein family BCL2 family members form hetero or homodimers and act as anti or pro apoptotic regulators that are involved in a wide variety of cellul
• DIABLO is an inhibitor of apoptosis protein IAP binding protein The encoded mitochondrial protein enters the cytosol when cells undergo apoptosis and it moderates the caspase inhibition of IAPs M
• FAK overexpression in human tumors provides a survival signal function by binding to RIP and inhibiting its interaction with the death receptor complex
• RIPK1 interacts with the death domain of FAS and TRADD and initiates apoptosis It is recruited by TRADD to TNFR1 in a TNF dependent process Required for TNFR1 activation of NF B
• TNFSF10 is a cytokine that belongs to the tumor necrosis factor TNF ligand family This protein preferentially induces apoptosis in transformed and tumor cells but does not appear to kill normal ce
• YWHAB belongs to the 14 3 3 family members of which mediate signal transduction by binding to phosphoserine containing proteins YWHAB has been shown to interact with RAF1 and CDC25 phosphatases sug
• The protein encoded by TIAL1 is a member of a family of RNA binding proteins and possesses nucleolytic activity against cytotoxic lymphocyte target cells The gene product is a cytotoxic granule assoc
• The protein encoded by TNFRSF18 is a member of the TNF receptor superfamily This receptor has been shown to have increased expression upon T cell activation and it is thought to play a key role in d
• TNFSF6 is a cytokine that binds to TNFRSF6 FAS a receptor that transduces the apoptotic signal into cells May be involved in cytotoxic T cell mediated apoptosis and in T cell development TNFRSF6 FA
• TP53 acts as a tumor suppressor in many tumor types induces growth arrest or apoptosis depending on the physiological circumstances and cell type Involved in cell cycle regulation as a trans activat
• The YWHAH gene product belongs to the 14 3 3 family of proteins which mediate signal transduction by binding to phosphoserine containing proteins This highly conserved protein family is found in both
• YWHAQ belongs to the 14 3 3 family of proteins which mediate signal transduction by binding to phosphoserine containing proteins This highly conserved protein family is found in both plants and mamma
• This is a receptor for the cytotoxic ligand TNFSF10 TRAIL The adaptor molecule FADD recruits caspase 8 to the activated receptor The resulting death inducing signaling complex DISC performs caspas
• CDK9 is a member of the cyclin dependent protein kinase CDK family CDK family members are highly similar to the gene products of S cerevisiae cdc28 and S pombe cdc2 and known as important cell
• CDK6 is a member of the cyclin dependent protein kinase CDK family CDK family members are highly similar to the gene products of Saccharomyces cerevisiae cdc28 and Schizosaccharomyces pombe cdc2
• Cdk2 is probably involved in the control of the cell cycle It interacts with cyclins A D or E Activity of cdk2 is maximal during S phase and G2
• Cdk7 is the catalytic subunit of the CDK activating kinase CAK complex a serine threonine kinase CAK activates the cyclin associated kinases CDC2 CDK1 CDK2 CDK4 and CDK6 by threonine phosphoryla
• PCNA is an auxiliary protein of DNA polymerase delta and is involved in the control of eukaryotic DNA replication by increasing the polymerase s processibility during elongation of the leading strand
• The protein encoded by CDC2 is a member of the Ser Thr protein kinase family This protein is a catalytic subunit of the highly conserved protein kinase complex known as M phase promoting factor MPF
• Cyclin H regulates CDK7 the catalytic subunit of the CDK activating kinase CAK enzymatic complex CAK activates the cyclin associated kinases CDC2 CDK1 CDK2 CDK4 and CDK6 by threonine phosphoryl
• The eukaryotic cell cycle is governed by cyclin dependent protein kinases CDKs whose activities are regulated by cyclins and CDK inhibitors The 8 species of cyclins reported in mammals cyclins A t
• CCND3 encodes a protein that belongs to the highly conserved cyclin family whose members are characterized by a dramatic periodicity in protein abundance through the cell cycle Cyclins function as r
• ROC1 encodes an evolutionarily conserved protein that interacts with cullins The protein plays a unique role in the ubiquitination reaction by heterodimerizing with cullin 1 to catalyze ubiquitin pol
• Cyclin A is essential for the control of the cell cycle at the G1 S start and the G2 M mitosis transitions
• CDKN2B s gene lies adjacent to the tumor suppressor gene CDKN2A in a region that is frequently mutated and deleted in a wide variety of tumors CDKN2B is a cyclin dependent kinase inhibitor which for
• p15 interacts strongly with cdk4 and cdk6 It is a potent inhibitor and a potential effector of TGF beta induced cell cycle arrest
• Cul1 may contribute to catalysis through the positioning of the substrate and the ubiquitin conjugating enzyme
• The protein encoded by CDK8 is a member of the cyclin dependent protein kinase CDK family CDK family members are highly similar to the gene products of Saccharomyces cerevisiae cdc28 and Schizosac
• TOP2A is a DNA topoisomerase an enzyme that controls and alters the topologic states of DNA during transcription This nuclear enzyme is involved in processes such as chromosome condensation chromat
• MAPK14 encodes a protein which is a member of the MAP kinase family MAP kinases act as an integration point for multiple biochemical signals and are involved in a wide variety of cellular processes
• MAPK14 is a member of the MAP kinase family MAP kinases act as an integration point for multiple biochemical signals and are involved in a wide variety of cellular processes such as proliferation d
• MDM4 inhibits p53 and p73 mediated cell cycle arrest and apoptosis by binding its transcriptional activation domain It inhibits degradation of MDM2 It can reverse MDM2 targeted degradation of p53 w
• MDM4 inhibits p53 and p73 mediated cell cycle arrest and apoptosis by binding its transcriptional activation domain inhibits degradation of MDM2 It can reverse MDM2 targeted degradation of p53 whil
• AKT1 is a general protein kinase capable of phosphorylating several known proteins The serine threonine protein kinase encoded by the AKT1 gene is catalytically inactive in serum starved primary and i
• AKT1 is a general protein kinase capable of phosphorylating several known proteins
• Cytokines are a family of secreted proteins involved in immunoregulatory and inflammatory processes The CC cytokines are proteins characterized by two adjacent cysteines CCL13 displays chemotactic a
• CXCL14 belongs to the cytokine family which encode secreted proteins involved in immunoregulatory and inflammatory processes The protein encoded by this gene is structurally related to the CXC Cys X
• Stromal cell derived factors 1 alpha and 1 beta are small cytokines that belong to the intercrine family members of which activate leukocytes and are often induced by proinflammatory stimuli such as
• Chemokines are a group of small approximately 8 to 14 kD mostly basic structurally related molecules that regulate cell trafficking of various types of leukocytes through interactions with a subse
• CXCL3 may play a role in inflammation and exert its effects on endothelial cells in an autocrine fashion
• CCL7 encodes monocyte chemotactic protein 3 a secreted chemokine which attracts macrophages during inflammation and metastasis It is a member of the C C subfamily of chemokines which are characteriz
• CCL18 is one of several Cys Cys CC cytokine genes clustered on the q arm of chromosome 17 Cytokines are a family of secreted proteins involved in immunoregulatory and inflammatory processes The CC
• DOK1 is constitutively tyrosine phosphorylated in hematopoietic progenitors isolated from chronic myelogenous leukemia CML patients in the chronic phase It may be a critical substrate for p210 bcr
• Serine or cysteine proteinase inhibitor clade H heat shock protein 47 member 1 collagen binding protein 1
• RGS3 inhibits signal transduction by increasing the GTPASE activity of G protein alpha subunits thereby driving them into their inactive GDP bound form This gene encodes a member of the regulator of G
• RGS3 inhibits signal transduction by increasing the GTPASE activity of G protein alpha subunits thereby driving them into their inactive GDP bound form
• HSP40 is a new member of the hsp40 family exhibits similar expression profile to that of hsc70 in mammalian cells
• RGS10 inhibits signal transduction by increasing the GTPASE activity of G protein alpha subunits thereby driving them into their inactive GDP bound form It associates specifically with the activated
• HSP90AB1 is an isoform of the hsp90 family It is expressed in cancer cells but not associated with cell proliferation
• HSP90 beta is a molecular chaperone that has ATPASE activity by similarity
• The scaffolding protein is the main component of the

caveolae plasma membranes found in most cell types The protein links integrin subunits to the tyrosine kinase FYN an initiating step in coupling
• CAV3 is a caveolin family member which functions as a component of the caveolae plasma membranes found in most cell types Caveolin proteins are proposed to be scaffolding proteins for organizing and
• RGS9 is a member of the RGS family of signaling proteins that suppress the activity of G proteins by promoting their deactivation
• DOK2 is constitutively tyrosine phosphorylated in hematopoietic progenitors isolated from chronic myelogenous leukemia CML patients in the chronic phase It may be a critical substrate for p210 bcr
• GAB1 is a member of the IRS1 like multisubstrate docking protein family The encoded protein is an important mediator of branching tubulogenesis and plays a central role in cellular growth response t
• EIF2AK2 might play a role in ER stress induced apoptosis and in Alzheimer s disease Alzheimer cases show prominent EIF2AK2 activation in association with neuritic plaques and pyramidal neurons in the
• This gene encodes a component of the interferon regulated 2 5A system that functions in the antiviral and antiproliferative roles of interferons Mutations in this gene have been associated with predi
• SREBF2 is a ubiquitously expressed transcription factor that controls cholesterol homeostasis by stimulating transcription of sterol regulated genes This gene encodes a ubiquitously expressed transcr
• PDCD4 is a protein localized to the nucleus in proliferating cells Expression of its gene is modulated by cytokines in natural killer and T cells PDCD4 is thought to play a role in apoptosis but the
• PDCD4 is a protein localized to the nucleus in proliferating cells Expression of this gene is modulated by cytokines in natural killer and T cells The gene product is thought to play a role in apopt
• BCL2A1s a member of the BCL 2 protein family The proteins of this family form hetero or homodimers and act as anti and pro apoptotic regulators that are involved in a wide variety of cellular activ
• SREBF1 is a transcription factor that binds to the sterol regulatory element 1 SRE1 which is a decamer flanking the low density lipoprotein receptor gene and some genes involved in sterol biosynthe
• Secreted frizzled related protein 1 SFRP1 is a member of the SFRP family that contains a cysteine rich domain homologous to the putative Wnt binding site of Frizzled proteins SFRPs act as soluble m
• APPD Apoptosis inducing protein D phafin 1 is predicted through gene annotation and contains pleckstrin homology domain It may binds to two Zn ions through its FYVE zinc finger domain Biologica
• CCR8 is a member of the beta chemokine receptor family which is predicted to be a seven transmembrane protein similar to G protein coupled receptors Chemokines and their receptors are important for
• The glycine receptor is a neurotransmitter gated ion channel Binding of glycine to its receptor increases the chloride conductance and thus produces hyperpolarization inhibition of neuronal firing
• The product of the CHML gene supports geranylgeranylation of most Rab proteins and may substitute for REP 1 in tissues other than retina CHML is localized close to the gene for Usher syndrome type II
• The CHRNA3 subunit is expressed in the soma of the majority of pyramidal cells with the most alpha 3 immunoreactivity observed in CA2 4 and entorhinal cortex and relatively less in CA1 and subicular
• The nicotinic acetylcholine receptors nAChRs are members of a superfamily of ligand gated ion channels that mediate fast signal transmission at synapses The nAChRs are thought to be hetero pentamer
• CHRNA9 is a member of the ligand gated ionic channel family and nicotinic acetylcholine receptor superfamily CHRNA9 is a plasma membrane protein that forms homo or hetero oligomeric divalent cation
• CHRNB2 is a neuronal nicotinic acetylcholine receptor nAChR that belong to ligand gated ion channels composed of alpha and beta subunits with specific structural functional and pharmacological prop
• The nicotinic acetylcholine receptors are members of a superfamily of ligand gated ion channels that mediate fast signal transmission at synapses Mutations in neuronal nicotinic acetylcholine recepto
• Nicotinic acetylcholine receptors nAChRs are ligand gated ion channels nAChRs are pentameric structures that are made up of combinations of individual subunits CHRNB3 is one of the subunits of nACh
• The acetylcholine receptor of muscle has 5 subunits of 4 different types 2 alpha and 1 each of beta gamma and delta subunits After acetylcholine binding the receptor undergoes an extensive conform
• After binding acetylcholine the ACHR responds by an extensive change in conformation that affects all subunits and leads to opening of an ion conducting channel across the plasma membrane
• GABA the major inhibitory neurotransmitter in the vertebrate brain mediates neuronal inhibition by binding to the gaba benzodiazepine receptor and opening an integral chloride channel
• Gamma aminobutyric acid GABA is the major inhibitory neurotransmitter in the mammalian brain where it acts at GABA A receptors which are ligand gated chloride channels Chloride conductance of thes
• The gamma aminobutyric acid GABA A receptor is a multisubunit chloride channel that mediates the fastest inhibitory synaptic transmission in the central nervous system The subunit encoded GABRP is
• The gamma aminobutyric acid GABA A receptor is a multisubunit chloride channel that mediates the fastest inhibitory synaptic transmission in the central nervous system The subunit encoded by this g
• GDP dissociation inhibitors are proteins that regulate the GDP GTP exchange reaction of members of the rab family small GTP binding proteins of the ras superfamily that are involved in vesicular tra
• GRIA2 is one of the glutamate receptors which are the predominant excitatory neurotransmitter receptors in the mammalian brain and are activated in a variety of normal neurophysiologic processes The
• L glutamate acts as an excitatory neurotransmitter at many synapses in the central nervous system The postsynaptic actions of GLU are mediated by a variety of receptors that are named according to th
• This is one of the several different receptors for 5 hydroxytryptamine serotonin a biogenic hormone that functions as a neurotransmitter a hormone and a mitogen The activity of this receptor is
• The pathophysiology of depression remains enigmatic although abnormalities that involve serotonin signaling have been implicated The serotonin 1B receptor 5 hydroxytryptamine 5 HT1B receptor int
• This is one of the several different receptors for 5 hydroxytryptamine serotonin a biogenic hormone that functions as a neurotransmitter a hormone and a mitogen This receptor mediates its actio
• HTR3A belongs to the ligand gated ion channel receptor superfamily It is the subunit A of the type 3 receptor for 5 hydroxytryptamine serotonin a biogenic hormone that functions as a neurotransmit
• This is one of the several different receptors for 5 hydroxytryptamine serotonin a biogenic hormone that functions as a neurotransmitter a hormone and a mitogen This receptor is a ligand gated
• The SNARE hypothesis is a model explaining the process of docking and fusion of vesicles to their target membranes According to this model membrane proteins from the vesicle v SNAREs and protein
• PACSIN1 may play a role in vesicle formation and transport
• MCH may act as a neurotransmitter or neuromodulator in a broad array of neuronal functions directed toward the regulation of goal directed behavior such as food intake and general arousal May also
• RAB3A is involved in exocytosis by regulating a late step in synaptic vesicle fusion It could play a role in neurotransmitter release by regulating membrane flow in the nerve terminal
• As a protein transport RAB3D is probably involved in regulated exocytosis
• APBA1 is a member of the X11 protein family It is a neuronal adaptor protein that interacts with the Alzheimer s disease amyloid precursor protein APP It stabilises APP and inhibits production of
• Receptor for glutamate L glutamate acts as an excitatory neurotransmitter at many synapses in the central nervous system The postsynaptic actions of GLU are mediated by a variety of receptors that a
• HEME oxygenase cleaves the heme ring at the alpha methene bridge to form biliverdin Biliverdin is subsequently converted to bilirubin by biliverdin reductase Under physiological conditions the acti
• The 5 HT1F receptor is present in both human vascular and neuronal tissue This gene is localized at 3p12
• CHRNA1 encodes an alpha subunit that plays a role in acetlycholine binding channel gating The muscle acetylcholine receptor consiststs of 5 subunits of 4 different types 2 alpha isoforms and 1 each
• Snap25 contains 2 t SNARE coiled coil homology domains and belongs to the SNAP 25 family t SNARE involved in the molecular regulation of neurotransmitter release Snap25 may play an important role in
• TRHR is a receptor for thyrotropin releasing hormone This receptor is mediated by G proteins which activate a phosphatidylinositol calcium second messenger system
• Localized to biosynthetic and endosomal compartments Rab14 is involved in the biosynthetic recycling pathway between the Golgi and endosomal compartments
• TRH functions as a regulator of the biosynthesis of TSH in the anterior pituitary gland and as a neurotransmitter neuromodulator in the central and peripheral nervous systems
• CTNNB1 is involved in the regulation of cell adhesion and in signal transduction through the Wnt pathway
• KRT14 is a type I keratin which is usually found as a heterotetramer with two keratin 5 molecules a type II keratin Together they form the cytoskeleton of epithelial cells
• Anti M13 polyclonal antibody was derived from rabbit immunized with highly purified M13 bacterial phage particles Recombinant protein domains such as antibody fragments can be fused to M13 coat prote
• Anti IgY polyclonal antibody was derived from rabbit immunized with highly purified full length chicken IgY
• MMP 2 is involved in the cleavage of gelatin type I and collagen types IV V VII X
• The SERPINF1 gene may play a significant role in determining the balance of angiogenesis antiangiogenesis during atherogenesis A novel role of extracellular phosphorylation is shown to completely ch
• Receptors for the C C chemokine family include CKR I CKR 2A CKR 3 CKR 4 CKR 5 and the Duffy blood group antigen The C C receptors are important in the function of T cell chemotaxis and migration
• The MAPKAP kinases MAP kinase activated protein kinases are a group of MAP kinase substrates that also have kinase activity This family of catalytic proteins includes Rsk 1 also known as MAPKAP Ki
• CBFB encodes a protein that is the beta subunit of a heterodimeric core binding transcription factor belonging to the PEBP2 CBF transcription factor family which master regulates a host of genes speci
• ETS 1 is a member of the ets family of transcription factors which are key mediators of physiological and pathological processes
• IRF2 encodes interferon regulatory factor 2 a member of the interferon regulatory transcription factor IRF family IRF2 competitively inhibits the IRF1 mediated transcriptional activation of interf
• SMAD3 is a member of Smad family proteins SMAD family is known to be important cytoplasmic mediators of signals from the transforming growth factor beta TGF beta receptor serine threonine kinases
• TAL1 is a basic helix loop helix transcription factor with a critical role in the development of both blood and endothelium
• The tal 1 proto oncogene encodes a helix loop helix DNA binding protein that has been implicated in the formation of T cell acute lymphoblastic leukemia T ALL
• DEK was first identified in a fusion with the CAN nucleoporin protein in a specific subtype of acute myelogenous leukemia DEK has also been shown to be an autoantigen in patients with pauciarticular
• Overexpression of the bZIP protein ATF5 a transcriptional activator stimulates asparagine synthetase promoter reporter gene transcription via the nutrient sensing response unit ATF5 is also a new i
• Transcription Factor AREB6 transcription factor 8 zinc finger homeodomain enhancer binding protein inhibits interleukin 2 IL 2 gene expression May be responsible for transcriptional repression o
• CEBPZ is a member of the CCAAT binding transcription factor CTF nuclear factor I NF I group of cellular DNA binding proteins that recognizes the sequence GCCAAT and is implicated in eukaryotic tra
• PGR is a member of the steroid receptor superfamily The protein mediates the physiological effects of progesterone which plays a central role in reproductive events associated with the establishment
• Elongin A TCEB3 which is a subunit of the transcription factor B SIII complex The SIII complex is composed of elongins A A2 B and C It activates elongation by RNA polymerase II by suppressing
• Myb Related Protein B MYBL2 a member of the MYB family of transcription factor genes is a nuclear protein involved in cell cycle progression The encoded protein is phosphorylated by cyclin A cycl
• ETS2 contains an ETS DNA binding domain and belongs to the ETS family ETS2 is a target of protein kinase C and upregulates GM CSF Ets2 and its targets play essential roles in endothelial cell functi
• CREB5 belongs to the CRE cAMP response element binding protein family Members of this family contain zinc finger and bZIP DNA binding domains This protein specifically binds to CRE as a homodimer

• Glucocorticoid receptor beta
• HSF1 is a heat shock transcription factor Transcription of heat shock genes is rapidly induced after temperature stress Hsp90 by itself and or associated with multichaperone complexes is a major r
• TCF4 encodes transcription factor 4 a basic helix turn helix transcription factor The protein recognizes an Ephrussi box E box binding site CANNTG a motif first identified in immunoglobuli
• p65 is a subunit of the nuclear factor kappa B a second messenger which activates the transcription of a number of genes in multiple tissues The inhibitory effect of I kappa B upon NF kappa B in th
• ETV6 ETS related protein Tel1 ETS translocation variant 6 Tel is an ETS family transcription factor This protein contains two functional domains a N terminal pointed PNT domain that is involv
• Tel encodes an ETS family transcription factor The product of this gene contains two functional domains a N terminal pointed PNT domain that is involved in the protein protein interactions with it
• The TFEB gene fuses with an intronless gene in renal tumors harboring the t 6 11 p21 q13 chromosome translocation It encodes a protein that is a highly sensitive and specific diagnostic marker fo
• GTF2F1 is a general transcription initiation factor that binds to RNA polymerase II and helps to recruit it to the initiation complex in collaboration with TFIIB It promotes transcription elongation
• Transcription Factor IIH polypeptide 1
• The E2F transcription factor family regulates the expression of various cellular promoters particularly those involved in the cell cycle E2F factors bind to DNA as homodimers or heterodimers in asso
• EVX1 is a member of the vertebrate eve related homeo box family This protein acts as a transcriptional repressor A similar protein in mice is critical for early embryogenesis
• GA Binding Protein chain GABP subunit GABPA nuclear respiratory factor 2 subunit transcription factor E4TF1 60 is one of three GA binding protein transcription factor subunits which function
• GABPB2 is the GA binding protein transcription factor beta subunit It forms a tetrameric complex with the alpha subunit and stimulates transcription of target genes GABPB2 may be involved in activ
• GCM1 is a DNA binding protein with a gcm motif glial cell missing motif The encoded protein is a homolog of the Drosophila glial cells missing gene gcm This protein binds to the GCM motif A G C
• GCM1 is a DNA binding protein with a gcm motif glial cell missing motif GCM1 is a homolog of the Drosophila glial cells missing gene gcm This protein binds to the GCM motif A G CCCGCAT a novel
• H2 0 like homeo box 1 H2 0 Drosophilia like homeo box 1
• Interferon regulatory factor 8 IRF8 interferon consensus sequence binding protein Ion ChannelSBP is a transcription factor of the interferon IFN regulatory factor IRF family Proteins of this
• Interferon consensus sequence binding protein Ion ChannelSBP is a transcription factor of the interferon IFN regulatory factor IRF family Proteins of this family are composed of a conserved DNA
• ZNF187 may be involved in transcriptional regulation
• SRF is a ubiquitous nuclear protein that stimulates both cell proliferation and differentiation It is a member of the MADS MCM1 Agamous Deficiens and SRF box superfamily of transcription factors
• TFAP2A is a sequence specific DNA binding protein that interacts with inducible viral and cellular enhancer elements to regulate transcription of selected genes AP 2 factors bind to the consensus seq
• The early growth response protein 2 EGR2 is a transcription factor with three tandem C2H2 type zinc fingers Mutations in this gene are associated with the autosomal dominant Charcot Marie Tooth dis
• LHX2 is a protein belonging to a large protein family members of which carry the LIM domain a unique cysteine rich zinc binding domain The encoded protein may function as a transcriptional regulato
• The general transcription factor TFIIE has an essential role in eukaryotic transcription initiation together with RNA polymerase II and other general factors Human TFIIE consists of two subunits of r
• GTF2 is encoded by a gene that is part of a 500 kb inverted duplication on chromosome 5q13 This duplicated region contains at least four genes and repetitive elements which make it prone to rearrange
• Zinc finger protein 174
• POU3F4 brain specific homeobox POU domain protein 4 brain 4 Brn 4 transcription factor 4 is a transcription factor with a POU domain
• CEBPA is a bZIP transcription factor which can bind as a homodimer to certain promoters and enhancers It can also form heterodimers with the related proteins CEBP beta and CEBP gamma It has been sho
• Human DNA binding protein dbpA is a member of a Y box binding protein family containing a cold shock domain The increased expression of Y box binding proteins in somatic cells is associated with ce
• E2F5 is a member of the E2F family of transcription factors The E2F family plays a crucial role in the control of cell cycle and action of tumor suppressor proteins and is also a target of the transf
• GABPB2 is the GA binding protein transcription factor beta subunit It forms a tetrameric complex with the alpha subunit and stimulates transcription of target genes The encoded protein may be invo
• GABPB2 encodes the GA binding protein transcription factor beta subunit This protein forms a tetrameric complex with the alpha subunit and stimulates transcription of target genes The encoded prot
• HOXC6 belongs to the homeobox family members of which encode a highly conserved family of transcription factors that play an important role in morphogenesis in all multicellular organisms Mammals po
• NFKB1 or NFKB2 is bound to REL RELA or RELB to form the NFKB complex The NFKB complex is inhibited by I kappa B proteins NFKBIA or NFKBIB which inactivate NF kappa B by trapping it in the cytopl
• IRF2 is a member of the interferon regulatory transcription factor IRF family IRF2 competitively inhibits the IRF1 mediated transcriptional activation of interferons alpha and beta and presumably
• MBP 1 encodes one of three enolase isoenzymes found in mammals it encodes alpha enolase a homodimeric soluble enzyme and also encodes a shorter monomeric structural lens protein tau crystallin Th
• POU5F1 is a POU transcription factor expressed by early embryo cells and germ cells It determines paracrine growth factor signaling from stem cells to the trophectoderm
• PC4 is a transcriptional coactivator General transcription factor IIH protects promoters from PC4 mediated repression by relieving the topological constraint imposed by PC4 through the ERCC3 helicase
• PC4 activated RNA polymerase II transcription cofactor 4 is a transcriptional coactivator possessing the ability to suppress promoter driven as well as nonspecific transcription via its DNA binding
• TCEB1 is known as protein elongin C which is a subunit of the transcription factor B SIII complex The SIII complex is composed of elongins A A2 B and C It activates elongation by RNA polymerase
• The intronless gene for this transcription coactivator is located between the protocadherin beta and gamma gene clusters on chromosome 5 The protein encoded by this gene is a component of the TFIID p
• TAL 1 modulates the angiogenic response of endothelial cells by stimulating cell morphogenesis and by influencing their behavior in migration
• TAL1 is a basic helix loop helix transcription factor with a critical role in the development of both blood and endothelium
• Forkhead related protein FKHL2 brain factor 2 FOXG1A Forkhead box protein G1A HFK2 is a member of the fork head gene family of transcription factors
• Transcription initiation factor IIA and chains GTranscription Factor Antibodies2A1 Transcription Factor AntibodiesIIA p35 and p19 subunits Transcription Factor AntibodiesIIAL binding ability o
• An extracellular calcium binding protein of the mineralized matrix of bone Calnuc or RGD 620030 Also useful as a Golgi marker in immunohistochemistry 1 100 dilution
• Nucleobindin also known as calnuc participates in Ca2 storage in the Golgi as well as in other biological processes that involve DNA binding and protein protein interactions
• Calnuc or NUCB1 belongs to the nucleobindin family It is a major calcium binding protein of the Golgi and is a good Golgi marker It may be involved in calcium homeostasis Calnuc also plays roles in
• MYEF2 is the transcriptional repressor of the myelin basic protein gene MBP MYEF2 binds to the proximal MB1 element 5 TTGTCC 3 of the MBP promoter Its binding to MB1 and function are inhibited b
• The specific function of the protein remains unknown
• SIRT4 is a member of the sirtuin family of proteins homologs to the yeast Sir2 protein Members of the sirtuin family are characterized by a sirtuin core domain and grouped into four classes The fun
• TAF5L functions as a component of the PCAF complex The PCAF complex is capable of efficiently acetylating histones in a nucleosomal context The PCAF complex could be considered as the human version
• DPF2 may be a transcription factor required for the apoptosis response following survival factor withdrawal from myeloid cells DPF2 might also have a role in the development and maturation of lymphoi
• TFE3 a member of the helix loop helix family of transcription factors binds to the mu E3 motif of the immunoglobulin heavy chain enhancer and is expressed in many cell types TFE3 a member of the h
• CLDN18 plays a major role in tight junction specific obliteration of the intercellular space through calcium independent cell adhesion activity CLDN18 belongs to the large claudin family of proteins
• KIF12 is a member of the kinesin superfamily of microtubule associated molecular motors that play important roles in intracellular transport and cell division KIF12 is a member of the kinesin superfam
• The specific function of this protein remains unknown
• RARG is a receptor for retinoic acid This metabolite has profound effects on vertebrate development Retinoic acid is a morphogen and is a powerful teratogen RARG controls cell function by directly
• TRIM27 is a member of the tripartite motif TRIM family The TRIM motif includes three zinc binding domains a RING a B box type 1 and a B box type 2 and a coiled coil region This protein localize
• RBBP9 may play a role in the transformation process due to its capacity to confer resistance to the growth inhibitory effects of TGF beta1 through interaction with retinoblastoma and the subsequent di
• ZNF566 may be involved in transcriptional regulation
• APOBEC3F is a member of the cytidine deaminase gene family It is one of seven related genes or pseudogenes found in a cluster thought to result from gene duplication on chromosome 22 Members of th
• MIF4GD is a protein which contains an MIF4G domain This gene encodes a protein which contains an MIF4G domain
• The specific function of MPPED2 is not yet known
• The exact functions of RNF169 remain unknown
• The exact function of C20orf30 remains unknown
• Members of the parvin family including PARVB are actin binding proteins associated with focal contacts Members of the parvin family including PARVB are actin binding proteins associated with focal
• The specific function of PDIA4 is not yet known
• The exact function of LRRC8B remains unknown
• Defects in NP are the cause of nucleoside phosphorylase deficiency NP deficiency It leads to a severe T cell immunodeficiency with neurologic disorder in children The specific function of NP is no
• The exact function of C1orf184 remains unknown
• UBA3 is a catalytic subunit of the dimeric UBA3 NAE1 E1 enzyme E1 activates NEDD8 by first adenylating its C terminal glycine residue with ATP thereafter linking this residue to the side chain of th
• Crystallins are the dominant structural components of the vertebrate eye lens Crystallins are separated into two classes taxon specific or enzyme and ubiquitous The latter class constitutes the ma
• The specific function of RWDD2A is not yet known
• The specific function of CIB3 is not yet known This gene product shares a high degree of sequence similarity with DNA dependent protein kinase catalytic subunit interacting protein 2 in human and mous
• The exact functions of C22orf25 remain unknown
• The exact functions of CCDC128 remain unknown
• The exact functions of C13orf31 remain unknown
• The exact functions of CXorf20 remain unknown
• FBXL16 is a substrate recognition component of the SCF SKP1 CUL1 F box protein type E3 ubiquitin ligase complex Members of the F box protein family such as FBXL16 are characterized by an approxima
• The exact functions of LDHD remain unknown The protein encoded by this gene belongs to the D isomer specific 2 hydroxyacid dehydrogenase family The similar protein in yeast has both D lactate and D g
• The exact functions of CPNE9 remain unknown
• The exact functions of LIX1L remain unknown
• H1FOO may play a key role in the control of gene expression during oogenesis and early embryogenesis presumably through the perturbation of chromatin structure H1FOO is essential for meiotic maturat
• 2 PDE is an enzyme that cleaves 2 5 phosphodiester bond linking adenosines of the 5 triphosphorylated oligoadenylates triphosphorylated oligoadenylates referred as 2 5A modulates the 2 5A system
• The exact functions of LOC81691 remain unknown
• The specific function of AMD1 is not yet known This gene encodes an important intermediate enzyme in polyamine biosynthesis The polyamines spermine spermidine and putrescine are low molecular weigh
• The specific function of FLJ35848 is not yet known
• LCN1 could play a role in taste reception LCN1 could be necessary for the concentration and delivery of sapid molecules in the gustatory system LCN1 can bind various

ligands with chemical structure
• PI16 is a putative serine protease inhibitor PI16 may serve as a marker following prostatectomy for prostate cancer
• The specific function of CDRT4 is not yet known
• SFRS12IP1 is a possible splicing regulator involved in the control of cellular survival
• The exact functions of C6orf182 remain unknown
• The exact functions of ZADH2 remain unknown
• LRRTM1 may play a role during the development of specific forebrain structures by influencing neuronal differentiation and connectivity with a possible role in intracellular trafficking within axons
• MAOA catalyzes the oxidative deamination of biogenic and xenobiotic amines and has important functions in the metabolism of neuroactive and vasoactive amines in the central nervous system and peripher
• The branched chain alpha keto dehydrogenase complex catalyzes the overall conversion of alpha keto acids to acyl CoA and CO2 It contains multiple copies of three enzymatic components branched chain
• BLVRB catalyzes electron transfer from reduced pyridine nucleotides to flavins as well as methylene blue pyrroloquinoline quinone riboflavin or methemoglobin BLVRB has possible role in protecting
• EVX1 is a member of the vertebrate eve related homeo box family This protein acts as a transcriptional repressor A similar protein in mice is critical for early embryogenesis This gene encodes a me
• The exact functions of TPD52L3 remain unknown
• RBL1 is similar in sequence and possibly function to the product of the retinoblastoma 1 RB1 gene The RB1 gene product is a tumor suppressor protein that appears to be involved in cell cycle regula
• NFE2L1 activates erythroid specific globin gene expression This gene encodes a protein that is involved in globin gene expression in erythrocytes Confusion has occurred in bibliographic databases d
• SDCBP was initially identified as a molecule linking syndecan mediated signaling to the cytoskeleton The syntenin protein contains tandemly repeated PDZ domains that bind the cytoplasmic C terminal
• AGPAT2 is a member of the 1 acylglycerol 3 phosphate O acyltransferase family The protein is located within the endoplasmic reticulum membrane and converts lysophosphatidic acid to phosphatidic acid

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