Help and Manual



DAVID Bioinformatics Resources (2021 Update)

Laboratory of Human Retrovirology and Immunoinformatics (LHRI)

*** Welcome to DAVID (2021 Update) ***

*** If you are looking for <u>DAVID 6.8</u>, it is still accessible on <u>this server</u>. ***

Functional Annotation Table

Current Gene List: List_1

Current Background: Homo sapiens

47 DAVID IDs

47 record(s)

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AFF2 AF4/FMR2 family member 2(AFF2) Related Genes Homo sapiens

GOTERM BP DIRECT mRNA processing, brain development, learning or memory, RNA splicing, regulation of gene expression, negative regulation of gene

GOTERM_BP_DIRECT mRNA processing, brain development, learning or memory, RNA splicing, regulation of gene expression, negative regulation of gene expression, nuclear speck organization, regulation of RNA splicing,

GOTERM_CC_DIRECT <u>nuclear speck</u>,

GOTERM_MF_DIRECT G-quadruplex RNA binding,
INTERPRO Transcription factor AF4/FMR2,

OMIM_DISEASE <u>Intellectual developmental disorder, X-linked 109,</u>

UP_KW_BIOLOGICAL_PROCESS mRNA processing, mRNA splicing,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP_KW_DISEASE Mental retardati
UP_KW_MOLECULAR_FUNCTION RNA-binding,
UP_KW_PTM Phosphoprotein.

UP_SEQ_FEATURE COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, REGION:Disordered,

AMER1 APC membrane recruitment protein 1(AMER1) Related Genes Homo sapiens

GOTERM_BP_DIRECT

What signaling pathway, positive regulation of protein ubiquitination, bone development, adipose tissue development, regulation of canonical what signaling pathway, mesenchymal cell differentiation involved in kidney development, negative regulation of canonical what signaling pathway, mesenchymal cell differentiation involved in kidney development, negative regulation of canonical what signaling pathway, mesenchymal cell differentiation involved in kidney development.

canonical Wnt signaling pathway, mesenchymal cell differentiation involved in kidney development, negative regulation of canonical Wnt signaling pathway, positive regulation of canonical Wnt signaling pathway, positive regulation of cellular protein catabolic process,

beta-catenin destruction complex assembly, beta-catenin destruction complex disassembly,

GOTERM_CC_DIRECT cytosol, plasma membrane, nuclear body, intracellular membrane-bounded organelle,

GOTERM_MF_DIRECT protein binding, phosphatidylinositol-4,5-bisphosphate binding, beta-catenin binding, beta-catenin destruction complex binding,

INTERPRO Uncharacterised protein family FAM123,
OMIM_DISEASE Osteopathia striata with cranial sclerosis,

UP_KW_BIOLOGICAL_PROCESS Wnt signaling pathway,

UP_KW_CELLULAR_COMPONENT Membrane, Nucleus, Cytoplasm, Cell membrane,

UP_KW_LIGAND Lipid-binding,

UP_KW_PTM Acetylation, Phosphoprotein,

UP_SEQ_FEATURE COMPBIAS:Acidic residues, COMPBIAS:Polar residues, MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane

localization; when associated with A-54; A-58; A-79; A-166; A-181 and A-183., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-54; A-58; A-79; A-83; A-166 and A-181., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-54; A-58; A-79; A-83; A-166 and A-183., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-54; A-58; A-79; A-83; A-181 and A-183., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-54; A-58; A-83; A-166; A-181 and A-183., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-54; A-79; A-83; A-166; A-181 and A-183., MUTAGEN:K->A: Abolishes interaction with PtdIns(4,5)P2 and cell membrane localization; when associated with A-58; A-79; A-83; A-166; A-181 and A-183., REGION:Disordered, A-181 and A-183., A-166; A-181 and A-183., A-166; A-181 and A-183., REGION:Disordered, A-181 and A-183., A-166; A-181 and A-183., A-166;

ASXL1 ASXL transcriptional regulator 3(ASXL3) Related Genes Homo sapiens

GOTERM_BP_DIRECT transcription, DNA-templated, regulation of transcription, DNA-templated, animal organ morphogenesis, positive regulation of transcription from RNA polymerase II promoter, negative regulation of lipid biosynthetic process,

GOTERM_CC_DIRECT <u>nucleus</u>, <u>PR-DUB complex</u>,

GOTERM_MF_DIRECT DNA binding, chromatin binding, peroxisome proliferator activated receptor binding, metal ion binding,

INTERPRO DNA-directed RNA polymerase delta subunit, Polycomb protein ASX/ASX-like, ASX-like protein 3, Protein ASX-like, PHD domain,

OMIM_DISEASE Bainbridge-Ropers syndrome,
UP_KW_BIOLOGICAL_PROCESS Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP_KW_DOMAIN

UP_KW_LIGAND

UP_KW_MOLECULAR_FUNCTION

Repressor,

UP_SEQ_FEATURE COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:DEUBAD, DOMAIN:HTH HARE-type,

REGION:Disordered, ZN_FING:PHD-type; atypical,

ARID1A AT-rich interaction domain 1A(ARID1A) Related Genes Homo sapiens

BIOCARTA Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes, Control of Gene Expression by Vitamin D Receptor,

GOTERM_BP_DIRECT nucleosome disassembly, chromatin remodeling, regulation of transcription from RNA polymerase II promoter, nervous system

development, intracellular estrogen receptor signaling pathway, androgen receptor signaling pathway, nucleosome mobilization, glucocorticoid receptor signaling pathway, ATP-dependent chromatin remodeling, positive regulation of gene expression, epigenetic,

positive regulation of transcription, DNA-templated, chromatin-mediated maintenance of transcription,

GOTERM_CC_DIRECT chromatin, nucleus, nucleoplasm, SWI/SNF complex, brahma complex, npBAF complex, nBAF complex

GOTERM_MF_DIRECT DNA binding, transcription coactivator activity, protein binding, ligand-dependent nuclear receptor binding, nucleosome binding,

INTERPRO

ARID/BRIGHT DNA-binding domain, Armadillo-like helical, Armadillo-type fold, Protein of unknown function DUF3518,

KEGG_PATHWAY Thermogenesis, Hepatocellular carcinoma,

OMIM DISEASE Coffin-Siris syndrome 2,

SMART BRIGHT,

UP_KW_BIOLOGICAL_PROCESS Neurogenesis, Transcription, Transcription regulation,

UP KW CELLULAR COMPONENT Nucleus,

UP KW DISEASE

UP_KW_MOLECULAR_FUNCTION Chromatin regulator, DNA-binding,

UP KW PTM Acetylation, Methylation, Phosphoprotein, UP_SEQ_FEATURE

COMPBIAS:Acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:ARID, DOMAIN:BAF250_C, MOTIF:LXXLL, MOTIF:Nuclear localization signal, MUTAGEN:KR->TT: Displays nucleocytoplasmic localization and increased stability; when associated with T-1383, MUTAGEN:R->T: Displays nucleocytoplasmic localization and increased stability; when associated with 1370-T-T-1371., MUTAGEN:RRR->TTT: No effect on subcellular localization., MUTAGEN:W->A: Partial loss of DNA-binding activity. Complete loss of activity; when associated with A-1096., MUTAGEN:Y->A: Partial loss of DNA-binding activity. Complete loss of activity; when

associated with A-1073., REGION: Disordered,

ATM ATM serine/threonine kinase(ATM)

BIOCARTA ATM Signaling Pathway, Role of BRCA1, BRCA2 and ATR in Cancer Susceptibility, cdc25 and chk1 Regulatory Pathway in response to DNA damage, Apoptotic Signaling in Response to DNA Damage, Cell Cycle, Cell Cycle, Hypoxia and p53 in the Cardiovascular system, p53 Signaling Pathway, Regulation of cell cycle progression by Plk3, RB Tumor Suppressor/Checkpoint Signaling in response to DNA

GOTERM BP DIRECT

DNA damage checkpoint, macropexophagy, telomere maintenance, double-strand break repair via homologous recombination, <u>DNA</u> double-strand break processing, ovarian follicle development, response to hypoxia, somitogenesis, <u>pre-B cell allelic exclusion</u>, <u>DNA</u> replication, <u>DNA</u> repair, double-strand break repair, double-strand break repair via nonhomologous end joining, <u>protein</u> phosphorylation, cellular response to DNA damage stimulus, DNA damage induced protein phosphorylation, DNA damage response, signal transduction by p53 class mediator resulting in cell cycle arrest, cell cycle arrest, mitotic spindle assembly checkpoint, mitotic G2 DNA damage checkpoint, reciprocal meiotic recombination, male meiosis, female meiotic division, signal transduction, brain development, heart development, determination of adult lifespan, intrinsic apoptotic signaling pathway in response to DNA damage, post-embryonic development, response to ionizing radiation, regulation of autophagy, positive regulation of gene expression, histone phosphorylation, peptidyl-serine phosphorylation, positive regulation of cell migration, negative regulation of B cell proliferation, regulation of telomere maintenance via telomerase, positive regulation of telomerase, positive regulat histone phosphorylation, V(D)J recombination, multicellular organism growth, phosphatidylinositol-3-phosphate biosynthetic proces peptidyl-serine autophosphorylation, lipoprotein catabolic process, regulation of apoptotic process, positive regulation of apoptotic process, positive regulation of DNA damage response, signal transduction by p53 class mediator, positive regulation of neuron apoptotic process, meiotic telomere clustering, positive regulation of cell adhesion, positive regulation of transcription from RNA polymerase II promoter, protein autophosphorylation, thymus development, occyte development, neuron apoptotic process, regulation of cell cycle, histone mRNA catabolic process, cellular response to retinoic acid, cellular response to gamma radiation, cellular response to X-ray, cellular response to nitrosative stress, signal transduction involved in mitotic G2 DNA damage checkpoint, cellular senescence, replicative senescence, establishment of RNA localization to telomere, establishment of macromolecular complex localization to telomere, regulation of cellular response to heat, regulation of signal transduction by p53 class mediator, positive regulation of DNA catabolic process, regulation of microglial cell activation, negative regulation of TORC1 signaling, negative regulation of telomere capping, positive regulation of telomere maintenance via telomere lengthening, positive regulation of telomerase catalytic core complex assembly, regulation of cellular response to gamma radiation, positive regulation of response to DNA damage stimulus,

GOTERM_CC_DIRECT

chromosome, telomeric region, <u>nucleus</u>, nucleoplasm, <u>nucleolus</u>, <u>cytoplasm, peroxisomal matrix</u>, <u>centrosome</u>, <u>spindle</u>, <u>cytosol</u>, <u>cytoplasmic vesicle</u>, <u>intracellular membrane-bounded organelle</u>, <u>DNA repair complex</u>,

GOTERM MF DIRECT DNA binding, protein serine/threonine kinase activity, DNA-dependent protein kinase activity, protein serine/threonine/tyrosine kinase activity, protein binding, ATP binding, 1-phosphatidylinositol-3-kinase activity, identical protein binding, macromolecular complex

INTERPRO Phosphatidylinositol 3-/4-kinase, catalytic domain, PIK-related kinase, FAT, PIK-related kinase, FATC, Protein kinase-like domain, PIK-related kinase, ATM/Tel1, Armadillo-type fold, Phosphatidylinositol 3/4-kinase, conserved site, Telomere-length maintenance and DNA damage repair,

Platinum drug resistance, Homologous recombination, NF-kappa B signaling pathway, FoxO signaling pathway, Cell cycle, p53 signaling pathway, Apoptosis, Cellular senescence, Shigellosis, Human papillomavirus infection, Human T-cell leukemia virus 1 infection, Human immunodeficiency virus 1 infection, Transcriptional misregulation in cancer, MicroRNAs in cancer, **KEGG PATHWAY**

Breast cancer, susceptibility to, Ataxia-telangiectasia, Lymphoma, B-cell non-Hodgkin, somatic, Lymphoma, mantle cell, somatic, T-cell

OMIM DISEASE prolymphocytic leukemia, somatic,

PI3Kc, SM01342, SM01343, UP_KW_BIOLOGICAL_PROCESS Cell cycle, DNA damage,

UP_KW_CELLULAR_COMPONENT Cytoskeleton, Nucleus, Cytoplasm, Cytoplasmic vesicle, UP_KW_DISEASE Tumor suppressor, Disease variant, Neurodegeneration,

UP_KW_DOMAIN Coiled coil

UP_KW_LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION DNA-binding, Kinase, Serine/threonine-protein kinase, Transferase,

UP_KW_PTM Acetylation, Phosphoprotein,

UP SEQ FEATURE

DOMAIN:FAT, DOMAIN:FATC, DOMAIN:PI3K/PI4K, DOMAIN:TAN, MUTAGEN:D->A: Loss of kinase activity., MUTAGEN:D->N: Decreased phosphorylation of target proteins., MUTAGEN:E->K: Decreased phosphorylation of target proteins., MUTAGEN:K->E: Decreased phosphorylation of target proteins., MUTAGEN:K->Q: Mimics acetylation, preventing dephosphorylation and subsequent ATM deactivation during the late stage of DNA damage response., MUTAGEN:K->R: Loss of DNA damage-inducible acetylation. Retains constitutive kinase activity, but blocks DNA damage-induced kinase activation. Disrupts dimer and abolishes S-1981 constitutive kinase activity, but blocks DNA damage-induced kinase activation. Disrupts difficulties and abolishes 5-1961 autophosphorylation., MUTAGEN:K->R: Retains DNA damage-inducible acetylation and S-1981 autophosphorylation., MUTAGEN:L->P: Loss of phosphorylation of target proteins., MUTAGEN:N->K: Loss of kinase activity, MUTAGEN:P->L: Loss of phosphorylation of target proteins., MUTAGEN:S->A: Loss of IR-induced S-1893 autophosphorylation. Reduced correction of cell cycle checkpoint defects and DNA-repair activity. No effect on S-367 nor S-1981 autophosphorylation., MUTAGEN:S->A: Loss of IR-induced S-1981 autophosphorylation. Reduced correction of cell cycle checkpoint defects and DNA-repair activity. No effect on S-367 nor S-1893 autophosphorylation. No dimer disruption., MUTAGEN:S->A: Loss of IR-induced S-367 autophosphorylation. Reduced correction of cell cycle checkpoint defects and DNA-repair activity. No effect on S-1893 nor S-1981 autophosphorylation., MUTAGEN:S->D,E: Disrupts the dimer., MUTAGEN:S->L: Loss of phosphorylation of target proteins., MUTAGEN:S->T: No effect on phosphorylation of target proteins., MUTAGEN:Y->C: Loss of phosphorylation of target proteins., MUTAGEN:Y->C: Loss of phosphorylation of target proteins., REGION:Interaction with ABL1,

B-Raf proto-oncogene, serine/threonine kinase(BRAF) BRAF

BIOCARTA MAPKinase Signaling Pathway,

GOTERM BP DIRECT MAPK cascade, activation of MAPKK activity, myeloid progenitor cell differentiation, protein phosphorylation, signal transduction, epidermal growth factor receptor signaling pathway, visual learning, animal organ morphogenesis, positive regulation of gene expression, negative regulation of fibroblast migration, positive regulation of glucose transport, thyroid gland development, positive regulation of peptidyl-serine phosphorylation, somatic stem cell population maintenance, intracellular signal transduction, cellular response to drug, regulation of cell proliferation, negative regulation of apoptotic process, CD4-positive, alpha-beta T cell differentiation, CD4-positive or CD8-positive, alpha-beta T cell lineage commitment, response to peptide hormone, negative regulation of neuron apoptotic process, regulation of T cell differentiation, thymus development, positive regulation of axon regeneration, positive regulation of axonogenesis, T cell receptor signaling pathway, positive regulation of stress fiber assembly, response to cAMP, long-

<u>term synaptic potentiation, head morphogenesis, face development, positive regulation of ERK1 and ERK2 cascade, trehalos</u>

metabolism in response to stress, cellular response to calcium ion, establishment of protein localization to membrane, positive regulation of substrate adhesion-dependent cell spreading, cellular response to nerve growth factor stimulus, negative regulation of synaptic vesicle exocytosis, negative regulation of endothelial cell apoptotic process,

GOTERM CC DIRECT

<u>ndrion, cytosol, plasma membrane, neuron projection, intracellular membrane-bounded organelle, cell body,</u>

GOTERM MF DIRECT

protein kinase activity, protein serine/threonine kinase activity, MAP kinase kinase activity, MAP kinase kinase activity, protein tyrosine kinase activity, calcium ion binding, protein binding, ATP binding, small GTPase binding, mitogen-activated protein kinase kinase binding, identical protein binding, macromolecular complex binding, scaffold protein binding,

INTERPRO

Protein kinase, catalytic domain, Serine-threonine/tyrosine-protein kinase catalytic domain, Protein kinase C-like, phorbol ester/diacylglycerol binding, Raf-like Ras-binding, Serine/threonine-protein kinase, active site, Protein kinase-like domain, Protein kinase, active site, Protein kinase-like domain, Pro kinase, ATP binding site, Diacylglycerol/phorbol-ester binding, Tyrosine-protein kinase, catalytic domain,

KEGG PATHWAY

EGFR tyrosine kinase inhibitor resistance, Endocrine resistance, MAPK signaling pathway, ErbB signaling pathway, Rap1 signaling pathway, cAMP signaling pathway, Chemokine signaling pathway, FoxO signaling pathway, mTOR signaling pathway, Vascular smooth muscle contraction, Focal adhesion, Natural killer cell mediated cytotoxicity, Long-term potentiation, Neurotrophin signaling pathway, Serotonergic synapse, Long-term depression, Regulation of actin cytoskeleton, Insulin signaling pathway, Progesterone-mediated oocyte maturation, Parathyroid hormone synthesis, secretion and action, Cushing syndrome, Alzheimer disease, Pathways of neurodegeneration - multiple diseases, Alcoholism, Hepatitis C, Hepatitis B, Pathways in cancer, Proteoglycans in cancer, Chemical carcinogenesis - reactive oxygen species, Colorectal cancer, Renal cell carcinoma, Pancreatic cancer, Endometrial cancer, Glioma, Prostate cancer, Thyroid cancer, Melanoma, Bladder cancer, Chronic myeloid leukemia, Acute myeloid leukemia, Non-small cell lung cancer, Breast cancer, Hepatocellular carcinoma, Gastric cancer,

OMIM_DISEASE

Colorectal cancer, somatic, Cardiofaciocutaneous syndrome, Melanoma, malignant, somatic, Adenocarcinoma of lung, somatic, Nonsmall cell lung cancer, somatic, Noonan syndrome 7, LEOPARD syndrome 3,

SMART

C1, TyrKc, S TKc, RBD

UP_KW_CELLULAR_COMPONENT Membrane, Nucleus, Cytoplasm, Cell membrane,

Ectodermal dysplasia, Cardiomyopathy, Deafness, Disease variant, Proto-oncogene, Mental retardation,

UP KW DISEASE UP_KW_DOMAIN

Coiled coil, Zinc-finger,

UP KW LIGAND UP_KW_MOLECULAR_FUNCTION

Allosteric enzyme, Kinase, Serine/threonine-protein kinase, Transferase, Tyrosine-protein kinase,

UP KW PTM

Acetylation, Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond

ATP-binding, Metal-binding, Nucleotide-binding, Zinc,

UP SEQ FEATURE

ACT_SITE:Proton acceptor, BINDING:ATP, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in ubiquitin), DOMAIN:Phorbol-ester/DAG-type, DOMAIN:Protein kinase, DOMAIN:RBD, METAL:Zinc 1, METAL:Zinc 2, MUTAGEN:I->R: No effect on MAP2K1-mediated-BRAF-KSR1 dimerization, however loss of BRAF-mediated phosphorylation of MAP2K1., MUTAGEN:K->E: Reduces interaction with KSR1 and MAP2K1 and thus phosphorylation of MAP2K1., MUTAGEN:K->R: Blocks EGF-induced ubiquitination and ERK activation., MUTAGEN:K->S: Reduces kinase activity with MAP2K1., MUTAGEN:M->D: Reduces interaction with KSR1 and MAP2K1 and thus phosphorylation of MAP2K1., MUTAGEN:R->H: Loss of MAP2K1-mediated-BRAF-KSR1 dimerization., MUTAGEN:R->K: Increased kinase activity and stability in response to EGF treatment., NP_BIND:ATP, REGION:Disordered, SITE:Breakpoint for translocation to form KIAA1549-BRAF fusion protein, ZN_FING:Phorbol-

ester/DAG-type,

CREBBP

CREB binding protein(CREBBP)

BBID

 $139. Fig 1 CD_Eukaryotic_transcriptional_control, \ 140. Fig 2_assembly_model_transcription, \ 141. Fig 3_pre-assembly_model_initiation, \ 142. Fig 4 AB_signal-mediated_transfer_model, \ 143. Fig 4 CD_signal-mediated_transfer_model, \ 85. Nuc_Rec-Coact_Complex, \ 99. NF-coact_Complex, \ 99. NF-coact_Complex, \ 99. NF-coact_Coa$

BIOCARTA

CARM1 and Regulation of the Estrogen Receptor, Transcription Regulation by Methyltransferase of CARM1, Activation of Csk by cAMPdependent Protein Kinase Inhibits Signaling through the T Cell Receptor, Inhibition of Huntington's disease neurodegeneration by histone deacetylase inhibitors, IL-7 Signal Transduction, NFAT and Hypertrophy of the heart (Transcription in the broken heart), NFkB activation by Nontypeable Hemophilus influenzae, The information-processing pathway at the IFN-beta enhancer, Pelp1 Modulation of Estrogen Receptor Activity, Multi-step Regulation of Transcription by Pitx2, Regulation of transcriptional activity by PML, Mechanism of Gene Regulation by Peroxisome Proliferators via PPARa(alpha), Role of PPAR-gamma Coactivators in Obesity and Thermogenesis, Acetylation and Deacetylation of RelA in The Nucleus, Granzyme A mediated Apoptosis Pathway, TGF beta signaling pathway, Control of Gene Expression by Vitamin D Receptor, WNT Signaling Pathway,

GOTERM_BP_DIRECT

negative regulation of transcription from RNA polymerase II promoter, response to hypoxia, stimulatory C-type lectin receptor signaling pathway, regulation of transcription, DNA-templated, transcription initiation from RNA polymerase II promoter, protein acetylation, signal transduction, Notch signaling pathway, positive regulation of transcription of Notch receptor target, regulation of smoothened signaling pathway, viral process, histone acetylation, N-terminal peptidyl-lysine acetylation, regulation of lipid metabolic process, positive regulation of transforming growth factor beta receptor signaling pathway, protein destabilization, positive regulation of type I interferon production, cellular response to UV, homeostatic process, embryonic digit morphogenesis, regulation of apoptotic process, regulation of myeloid cell differentiation, positive regulation of Notch signaling pathway, positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, rhythmic process, regulation of transcription from RNA polymerase II promoter in response to hypoxia, macromolecular complex assembly, regulation of cellular response to heat, beta-catenin-TCF complex assembly, histone glutamine methylation,

GOTERM_CC_DIRECT GOTERM MF DIRECT

histone acetyltransferase complex, chromatin, nucleus, nucleoplasm, transcription factor complex, cytoplasm, nuclear body, RNA polymerase II transcription factor binding, RNA polymerase II activating transcription factor binding, transcription coactivator

binding, p53 binding, chromatin binding, damaged DNA binding, transcription cofactor activity, transcription coactivator activity, transcription corepressor activity, histone acetyltransferase activity, protein binding, transcription factor binding, zinc ion binding, acetyltransferase activity, chromatin DNA binding, peptide N-acetyltransferase activity, MRF binding, metal ion binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, peptide-lysine-N-acetyltransferase activity,

INTERPRO

Zinc finger, TAZ-type, Zinc finger, ZZ-type, Bromodomain, Coactivator CBP, KIX domain, Nuclear receptor coactivator, interlocking, Domain of unknown function DUF902, CREBbp, Zinc finger, FYVE/PHD-type, Zinc finger, RING/FYVE/PHD-type, Histone H3-K56 acetyltransferase, RTT109, Nuclear receptor coactivator, CREB-bp-like, interlocking, Bromodomain, conserved site,

KEGG PATHWAY

cAMP signaling pathway, HIF-1 signaling pathway, FoxO signaling pathway, Cell cycle, Wnt signaling pathway, Notch signaling pathway, TGF-beta signaling pathway, Adherens junction, JAK-STAT signaling pathway, Long-term potentiation, Melanogenesis, Thyroid hormone signaling pathway, Glucagon signaling pathway, Growth hormone synthesis, secretion and action, Huntington disease, Tuberculosis, Hepatitis B, Influenza A, Human papillomavirus infection, Human T-cell leukemia virus 1 infection, Kaposi sarcoma-associated herpesvirus infection, Pathways in cancer, Viral carcinogenesis, MicroRNAs in cancer, Renal cell carcinoma, Pathways in cancer, Viral carcinogenesis, MicroRNAs in cancer, Renal cell carcinoma,

OMIM DISEASE Rubinstein-Taybi syndrome 1, Menke-Hennekam syndrome 1,

ZnF_ZZ, BROMO, ZnF_TAZ, SM01250,

UP_KW_BIOLOGICAL_PROCESS Biological rhythms, Transcription, Transcription regulation, Host-virus interaction,

UP_KW_CELLULAR_COMPONENT Nucleus, Cytoplasm,

UP KW DISEASE Disease variant, Mental retardation, UP_KW_DOMAIN Bromodomain, Repeat, Zinc-finger,

UP_KW_LIGAND Metal-binding, Zinc,

UP_KW_MOLECULAR_FUNCTION Activator, Acyltransferase, Transferase,

UP_KW_PTM Acetylation, Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

BINDING:Acetyl-CoA, BINDING:Acetyl-CoA; via carbonyl oxygen, COMPBIAS:Basic and acidic residues, COMPBIAS:Basic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO1), DOMAIN:Bromo, DOMAIN:CBP/p300-type HAT, DOMAIN:KIX, DOMAIN:TAZ-type, DOMAIN:ZZ-type, METAL:Zinc 1, METAL:Zinc 2, UP SEQ FEATURE

METAL:Zinc 3, METAL:Zinc 4, METAL:Zinc 5, MUTAGEN:D->R: Impairs binding to acetylated histones., MUTAGEN:E->R: Abolishes interaction with ASF1A., MUTAGEN:F->A: Impairs binding to acetylated histones., MUTAGEN:K->E: Abolishes interaction with ASF1A., MUTAGEN:K->E: Impairs binding to acetylated histones., MUTAGEN:N->E,R: Abolishes interaction with ASF1A., MUTAGEN:S->I: Impairs interaction with ASF1A., MUTAGEN:W->A: Abolishes interaction with ASF1A., REGION:Acetyl-CoA binding, REGION:Disordered, REGION:Interaction with ASF1A, REGION:Interaction with TRERF1, REGION:Interaction with histone, SITE:Breakpoint for translocation to form KAT6A-CREBBP, SITE:Breakpoint for translocation to form KAT6B-CREBBP, ZN_FING:TAZ-type, ZN_FING:TAZ-type 1, ZN_FING:TAZ-type 2, ZN_FING:ZZ-type,

DNMT3A

Related Genes

Homo sapiens

GOTERM_BP_DIRECT

negative regulation of transcription from RNA polymerase II promoter, mitotic cell cycle, DNA methylation, methylation-dependent chromatin silencing, regulation of gene expression by genetic imprinting, protein methylation, spermatogenesis, aging, response to xenobiotic stimulus, response to toxic substance, response to ionizing radiation, response to lead ion, regulation of gene expression, positive regulation of cell death, neuron differentiation, response to estradiol, DNA methylation on cytosine, response to vitamin A, response to cocaine, response to drug, DNA methylation involved in embryo development, DNA methylation involved in gamete generation, negative regulation of gene expression, epigenetic, negative regulation of transcription, DNA-templated, cellular response to amino acid stimulus, cellular response to ethanol, cellular response to hypoxia, C-5 methylation of cytosine, hepatocyte apoptotic process, cellular response to bisphenol A,

GOTERM_CC_DIRECT

chromosome, centromeric region, euchromatin, heterochromatin, XY body, nucleus, nucleoplasm, cytoplasm, nuclear matrix, catalytic

GOTERM_MF_DIRECT

RNA polymerase II core promoter proximal region sequence-specific DNA binding, DNA binding, chromatin binding, transcription corepressor activity, DNA (cytosine-5-)-methyltransferase activity, protein binding, transcription factor binding, DNA-methyltransferase activity, identical protein binding, metal ion binding, RNA polymerase II sequence-specific DNA binding transcription factor binding,

INTERPRO

PWWP, C-5 cytosine methyltransferase, DNA methylase, C-5 cytosine-specific, active site, ADD domain,

KEGG PATHWAY

Cysteine and methionine metabolism, Metabolic pathways, MicroRNAs in cancer,

OMIM_DISEASE

Acute myeloid leukemia, somatic, Tatton-Brown-Rahman syndrome, Heyn-Sproul-Jackson syndrome,

SMART

PWWP,

UP_KW_CELLULAR_COMPONENT Chromosome, Nucleus, Cytoplasm,

FPHA4

UP KW DISEASE

Disease variant, Dwarfism, Mental retardation,

UP KW DOMAIN UP_KW_LIGAND

Zinc-finger, Metal-binding, Zinc, S-adenosyl-L-methionine,

UP_KW_MOLECULAR_FUNCTION

Chromatin regulator, DNA-binding, Methyltransferase, Repressor, Transferase,

UP KW PTM UP_SEQ_FEATURE Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond

BINDING:S-adenosyl-L-methionine, COMPBIAS:Basic and acidic residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), DOMAIN:ADD, DOMAIN:PHD-type, DOMAIN:PWWP, DOMAIN:SAM-dependent MTase C5-type, MUTAGEN:F->A: Loss of activity due to the incapacity to bind the regulatory subunit DNMT3L., REGION:Disordered, REGION:Interaction with DNMT1 and DNMT3B, REGION:Interaction with the PRC2/EED-EZH2 complex, REGION:S-adenosyl-L-

methionine binding, ZN_FING:GATA-type; atypical, ZN_FING:PHD-type; atypical, FPH receptor A4(FPHA4)

BIOCARTA

Eph Kinases and ephrins support platelet aggregation,

GOTERM_BP_DIRECT

cell adhesion, negative regulation of cell adhesion, transmembrane receptor protein tyrosine kinase signaling pathway, multicellular organism development, axon guidance, adult walking behavior, motor neuron axon guidance, positive regulation of cell proliferation, glial cell migration, negative regulation of epithelial to mesenchymal transition, negative regulation of neuron projection development, peptidyl-tyrosine phosphorylation, corticospinal tract morphogenesis, positive regulation of cell migration, negative regulation of cell migration, positive regulation of kinase activity, adherens junction organization, regulation of GTPase activity, positive regulation of JUN kinase activity, positive regulation of cell adhesion, protein autophosphorylation, ephrin receptor signaling pathway, negative regulation of axon regeneration, regulation of astrocyte differentiation, regulation of axonogenesis, positive regulation of dendrite morphogenesis, protein stabilization, regulation of dendritic spine morphogenesis, positive regulation of protein tyrosine kinase activity, negative regulation of ERK1 and ERK2 cascade, nephric duct morphogenesis, cochlea development, fasciculation of sensory neuron axon, fasciculation of motor neuron axon, neuron projection guidance, synapse disassembly, negative regulation of cellular response to hypoxia, negative regulation of long-term synaptic potentiation, positive regulation of beta-amyloid formation, positive regulation of aspartic-type endopeptidase activity involved in amyloid precursor protein catabolic process, negative regulation of proteolysis involved in cellular protein catabolic process, cellular response to beta-amyloid, regulation of modification of synaptic structure, positive regulation of Rho guanyl-nucleotide exchange factor activity,

GOTERM_CC_DIRECT

cytoplasm, mitochondrial outer membrane, endoplasmic reticulum, Golgi apparatus, plasma membrane, integral component of plasma membrane, adherens junction, cell surface, postsynaptic density, integral component of membrane, filopodium, axon, dendrite, neuromuscular junction, early endosome membrane, neuron projection, dendritic spine, dendritic shaft, perikaryon, receptor complex, axon terminus, axonal growth cone, Schaffer collateral - CA1 synapse, postsynaptic density membrane, glutamatergic synapse, integral component of postsynaptic membrane, integral component of presynaptic membrane

GOTERM MF DIRECT

beta-amyloid binding, protein kinase activity, protein serine/threonine/tyrosine kinase activity, transmembrane receptor protein tyrosine kinase activity, ephrin receptor activity, GPI-linked ephrin receptor activity, transmembrane-ephrin receptor activity, protein binding, ATP binding, kinase activity, PH domain binding, identical protein binding, ephrin receptor binding, DH domain binding, protein tyrosine kinase binding,

INTERPRO

Protein kinase, catalytic domain, Ephrin receptor ligand binding domain, Serine-threonine/tyrosine-protein kinase catalytic domain, Tyrosine-protein kinase, receptor class V, conserved site, Sterile alpha motif domain, Fibronectin, type III, Tyrosine-protein kinase, active site, Galactose-binding domain-like, Protein kinase-like domain, Sterile alpha motif/pointed domain, Immunoglobulin-like fold, Tyrosine-protein kinase, ephrin receptor, Protein kinase, ATP binding site, Tyrosine-protein kinase, catalytic domain,

KEGG PATHWAY

Axon guidance,

PIR_SUPERFAMILY tyrosine-protein kinase, ephrin receptor type,

UP_KW_BIOLOGICAL_PROCESS

FN3, TyrKc, S_TKc, SAM, EPH_lbd, Cell adhesion, Neurogenesis,

UP_KW_CELLULAR_COMPONENT Membrane, Postsynaptic cell membrane, Synapse, Cell junction, Cell projection, Endosome, Cell membrane,

UP KW DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix,

UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION

Developmental protein, Kinase, Receptor, Transferase, Tyrosine-protein kinase, Developmental protein,

UP KW PTM Glycoprotein, Phosphoprotein,

UP_SEQ_FEATURE

ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GlcNAc...) asparagine, DOMAIN:Eph LBD, DOMAIN:Fibronectin type-III, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 2, DOMAIN:Protein kinase, DOMAIN:SAM, MOTIF:PDZ-binding, MUTAGEN:E->A: 10-fold reduced affinity for EFNB2; when associated with A-40., MUTAGEN:Q->A: 10-fold reduced affinity for EFNB2; when associated with A-42., NP_BIND:ATP, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

FAT1

FAT atypical cadherin 1(FAT1) Related Genes Homo sapiens

GOTERM_BP_DIRECT

lens development in camera-type eye, epithelial cell morphogenesis, establishment of epithelial cell apical/basal polarity involved in camera-type eye morphogenesis, actin filament organization, cell adhesion, homophilic cell adhesion via plasma membrane adhesion molecules, establishment or maintenance of cell polarity, cell-cell signaling, anatomical structure morphogenesis, cell migration, cell-

GOTERM_CC_DIRECT

nucleus, plasma membrane, integral component of plasma membrane, cell-cell junction, focal adhesion, membrane, integral

Homo sapiens

component of membrane, apical plasma membrane, lamellipodium, cell junction, filopodium, perinuclear region of cytoplasm,

GOTERM_MF_DIRECT

calcium ion binding, protein binding,

INTERPRO

RRID

EGF-type aspartate/asparagine hydroxylation site, Epidermal growth factor-like domain, Laminin G domain, EGF-like calcium-binding, Cadherin, Concanavalin A-like lectin/glucanase, subgroup, Cadherin-like, EGF-like calcium-binding, conserved site, Cadherin conserved

SMART CA, EGF_CA, EGF, LamG,

UP KW BIOLOGICAL PROCESS

Cell adhesion

UP_KW_CELLULAR_COMPONENT Membrane, Nucleus, Cell membrane,

UP KW DOMAIN EGF-like domain, Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_LIGAND Calcium

UP_KW_PTM Glycoprotein, Disulfide bond,

UP_SEQ_FEATURE

CARBOHYD:N-linked (GlcNAc...) asparagine, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:Cadherin, DOMAIN:Cadherin 1, DOMAIN:Cadherin 10, DOMAIN:Cadherin 11, DOMAIN:Cadherin 12, DOMAIN:Cadherin 13, DOMAIN:Cadherin 14, DOMAIN:Cadherin 15, DOMAIN:Cadherin 16, DOMAIN:Cadherin 17, DOMAIN:Cadherin 18, DOMAIN:Cadherin 19, DOMAIN:Cadherin 20, DOMAIN:Cadherin 21, DOMAIN:Cadherin 22, DOMAIN:Cadherin 23, DOMAIN:Cadherin 24, DOMAIN:Cadherin 25, DOMAIN:Cadherin 26, DOMAIN:Cadherin 27, DOMAIN:Cadherin 28, DOMAIN:Cadherin 29, DOMAIN:Cadherin 3, DOMAIN:Cadherin 30, DOMAI

DOMAIN:LAM_G_DOMAIN, DOMAIN:Laminin G-like, MOTIF:Nuclear localization signal, MOTIF:PTB-like motif, REGION:Disordered, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

Related Genes

KRAS proto-oncogene, GTPase(KRAS)

KRAS

BIOCARTA Telomeres, Telomerase, Cellular Aging, and Immortality,

65.Integrin_affinity_modulation,

GOTERM_BP_DIRECT

MAPK cascade, liver development, positive regulation of protein phosphorylation, stimulatory C-type lectin receptor signaling pathway, signal transduction, Ras protein signal transduction, female pregnancy, positive regulation of cell proliferation, visual learning, positive regulation of gene expression, cytokine-mediated signaling pathway, forebrain astrocyte development, actin cytoskeleton organization, regulation of protein stability, regulation of synaptic transmission, GABAergic, positive regulation of Rac protein signal transduction, response to isolation stress, endocrine signaling, positive regulation of MAP kinase activity, negative regulation of neuron apoptotic process, negative regulation of cell differentiation, regulation of long-term neuronal synaptic plasticity, homeostasis of number of cells within a tissue, positive regulation of nitric-oxide synthase activity, positive regulation of NF-kappaB transcription factor activity, striated muscle cell differentiation, response to glucocorticoid, response to mineralocorticoid, epithelial tube branching involved in lung

GOTERM CC DIRECT GOTERM MF DIRECT Golgi membrane, cytoplasm, mitochondrion, mitochondrial outer membrane, endoplasmic reticulum membrane, cytosol, plasma membrane, focal adhesion, membrane, extrinsic component of cytoplasmic side of plasma membrane, membrane raft,

GTPase activity, protein binding, ATP binding, GTP binding, transferase activity, GMP binding, GDP binding, LRR domain binding, identical protein binding, macromolecular complex binding

morphogenesis, positive regulation of cellular senescence,

INTERPRO Ubiquitin-conjugating enzyme, E2, Small GTPase superfamily, Small GTP-binding protein domain, Ubiquitin-conjugating enzyme/RWD-

like, Small GTPase superfamily, Ras type, Ubiquitin-conjugating enzyme, active site, P-loop containing nucleoside triphosphate hydrolase,

KEGG_PATHWAY

EGFR tyrosine kinase inhibitor resistance, Endocrine resistance, MAPK signaling pathway, ErbB signaling pathway, Ras signaling pathway, Rap1 signaling pathway, Chemokine signaling pathway, FoxO signaling pathway, Sphingolipid signaling pathway, Phospholipase D signaling pathway, Mitophagy - animal, Autophagy - animal, mTOR signaling pathway, PI3K-Akt signaling pathway, Apoptosis, Longevity regulating pathway, Longevity regulating pathway - multiple species, Cellular senescence, Axon guidance, VEGF signaling pathway, Apelin signaling pathway, Gap junction, Signaling pathways regulating pluripotency of stem cells, C-type lectin receptor signaling pathway, Natural killer cell mediated cytotoxicity, T cell receptor signaling pathway, B cell receptor signaling pathway, Fc epsilon RI signaling pathway, Thermogenesis, Long-term potentiation, Neurotrophin signaling pathway, Cholinergic synapse, Serotonergic synapse, Long-term depression, Regulation of actin cytoskeleton, Insulin signaling pathway, GnRH signaling pathway, Progesterone-mediated oocyte maturation, Estrogen signaling pathway, Melanogenesis, Prolactin signaling pathway, Thyroid hormone signaling pathway, Oxytocin signaling pathway, Relaxin signaling pathway, GnRH secretion, AGE-RAGE signaling pathway in diabetic complications, Growth hormone synthesis, secretion and action, Aldosterone-regulated sodium reabsorption, Alzheimer disease, Pathways of neurodegeneration - multiple diseases, Alcoholism, Hepatitis C, Hepatitis B, Human cytomegalovirus infection, Human papillomavirus infection, Human T-cell leukemia virus 1 infection, Kaposi sarcoma-associated herpesvirus infection, Human immunodeficiency virus 1 infection, Pathways in cancer, Viral carcinogenesis, Proteoglycans in cancer, MicroRNAs in cancer, Chemical carcinogenesis - receptor activation, Chemical carcinogenesis - reactive oxygen species, Colorectal cancer, Renal cell carcinoma Pancreatic cancer, Endometrial cancer, Glioma, Prostate cancer, Thyroid cancer, Melanoma, Bladder cancer, Chronic myeloid leukemia, Acute myeloid leukemia, Non-small cell lung cancer, Breast cancer, Hepatocellular carcinoma, Gastric cancer, Central carbon metabolism in cancer, Choline metabolism in cancer, PD-L1 expression and PD-1 checkpoint pathway in cancer, Lipid and

OMIM_DISEASE

GOTERM_MF_DIRECT

Arteriovenous malformation of the brain, somatic, Bladder cancer, somatic, Breast cancer, somatic, Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, Lung cancer, somatic, Pancreatic carcinoma, somatic, Oculoectodermal syndrome, somatic, Leukemia, acute myeloid, somatic, Noonan syndrome 3, Gastric cancer, somatic, RAS-associated autoimmune leukoproliferative disorder, Cardiofaciocutaneous syndrome 2,

UP_KW_CELLULAR_COMPONENT Membrane, Cytoplasm, Cell membrane,

UP_KW_DISEASE Ectodermal dysplasia, Cardiomyopathy, Deafness, Disease variant, Proto-oncogene, Mental retardation,

UP KW LIGAND ATP-binding, GTP-binding, Nucleotide-binding,

atherosclerosis.

UP_KW_MOLECULAR_FUNCTION Hydrolase, Transferase

UP KW PTM Acetylation, Glycoprotein, Lipoprotein, Methylation, Palmitate, Prenylation, Ubl conjugation, Isopeptide bond,

UP SEQ FEATURE ACT_SITE:Glycyl thioester intermediate, CARBOHYD:(Microbial infection) O-linked (Glc) threonine; by P.sordellii toxin TcsL,

COMPBIAS: Basic and acidic residues, COMPBIAS: Basic residues, COMPBIAS: Polar residues, CROSSLNK: Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in ubiquitin), DOMAIN: UBC core, LIPID:S-farnesyl cysteine, LIPID:S-palmitoyl cysteine, MOTIF: Effector region, MUTAGEN:C->S: Abolished interaction with GPR131., NP_BIND:GTP, PROPEP:Removed in mature form, REGION:Disordered,

REGION:Hypervariable region,

MGA MAX dimerization protein MGA(MGA) Related Genes GOTERM BP DIRECT

cell fate specification, regulation of transcription from RNA polymerase II promoter, negative regulation of G0 to G1 transition, GOTERM CC DIRECT chromatin, nucleoplasm, MLL1 complex,

RNA polymerase II core promoter proximal region sequence-specific DNA binding, RNA polymerase II transcription factor activity,

sequence-specific DNA binding, DNA binding, transcription factor activity, sequence-specific DNA binding, protein binding, protein

dimerization activity,

INTERPRO Transcription factor, T-box, p53-like transcription factor, DNA-binding, Myc-type, basic helix-loop-helix (bHLH) domain, Transcription

factor, T-box, conserved site,

SMART HLH, TBOX,

UP_KW_BIOLOGICAL_PROCESS Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus,

Homo sapiens

UP KW DOMAIN Coiled coil,

UP_KW_MOLECULAR_FUNCTION DNA-binding, Repressor,

UP_KW_PTM Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

UP_SEQ_FEATURE COMPBIAS:Acidic residues, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), DNA_BIND:T-box, DOMAIN:BHLH, DOMAIN:bHLH, REGION:Disordered,

MET proto-oncogene, receptor tyrosine kinase(MET)

BIOCARTA CBL mediated ligand-induced downregulation of EGF receptors, Signaling of Hepatocyte Growth Factor Receptor,

GOTERM_BP_DIRECT MAPK cascade, endothelial cell morphogenesis, liver development, phagocytosis, signal transduction, cell surface receptor signaling

pathway, transmembrane receptor protein tyrosine kinase signaling pathway, multicellular organism development, nervous system development, negative regulation of autophagy, cell migration, peptidyl-tyrosine phosphorylation, neuron differentiation, pancreas development, positive regulation of microtubule polymerization, positive regulation of kinase activity, negative regulation of Rho protein signal transduction, entry of bacterium into host cell, positive regulation of transcription from RNA polymerase II promoter, hepatocyte growth factor receptor signaling pathway, branching morphogenesis of an epithelial tube, positive chemotaxis, negative regulation of stress fiber assembly, positive regulation of protein kinase B signaling, establishment of skin barrier, negative regulation of thrombin-activated receptor signaling pathway, semaphorin-plexin signaling pathway, negative regulation of hydrogen peroxide-mediated programmed cell death, negative regulation of guanyl-nucleotide exchange factor activity, positive regulation of endothelial

GOTERM_CC_DIRECT extracellular region, plasma membrane, integral component of plasma membrane, basal plasma membrane, cell surface, membrane, integral component of membrane, receptor complex,

GOTERM_MF_DIRECT protein kinase activity, protein serine/threonine/tyrosine kinase activity, protein tyrosine kinase activity, transmembrane receptor

protein tyrosine kinase activity, hepatocyte growth factor-activated receptor activity, protein binding, ATP binding, semaphorin receptor activity, protein phosphatase binding, identical protein binding,

INTERPRO Protein kinase, catalytic domain, Serine-threonine/tyrosine-protein kinase catalytic domain, Semaphorin/CD100 antigen, Plexin, Cell

surface receptor IPT/TIG, Tyrosine-protein kinase, active site, <u>Protein kinase-like domain</u>, Immunoglobulin-like fold, Immunoglobulin <u>E-set</u>, WD40/YVTN repeat-like-containing domain, <u>Plexin-like fold</u>, Tyrosine-protein kinase, HGF/MSP receptor, Protein kinase, ATP

binding site, Tyrosine-protein kinase, catalytic domain,

EGFR tyrosine kinase inhibitor resistance, MAPK signaling pathway, Ras signaling pathway, Rap1 signaling pathway, Calcium signaling pathway, PI3K-Akt signaling pathway, Axon guidance, Focal adhesion, Adherens junction, Bacterial invasion of epithelial cells, Epithelial **KEGG PATHWAY**

cell signaling in Helicobacter pylori infection, Malaria, Pathways in cancer, Transcriptional misregulation in cancer, Proteoglycans in cancer, MicroRNAs in cancer, Chemical carcinogenesis - reactive oxygen species, Renal cell carcinoma, Melanoma, Non-small cell lung cancer, Hepatocellular carcinoma, Gastric cancer, Central carbon metabolism in cancer,

Hepatocellular carcinoma, childhood type, somatic, Renal cell carcinoma, papillary, 1, familial and somatic, Osteofibrous dysplasia, susceptibility to, Deafness, autosomal recessive 97, OMIM_DISEASE

PIR SUPERFAMILY tyrosine-protein kinase, HGF/MSP receptor type,

SMART TyrKc, PSI, IPT, Sema, UP_KW_CELLULAR_COMPONENT Membrane, Secreted,

UP KW DISEASE Deafness, Disease variant, Proto-oncogene, Non-syndromic deafness,

UP_KW_DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix,

UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Kinase, Receptor, Transferase, Tyrosine-protein kinase, UP KW PTM

Glycoprotein, Phosphoprotein, Ubl conjugation, Disulfide bond

ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GlcNAc...) asparagine, DOMAIN:IPT/TIG, DOMAIN:IPT/TIG 1, DOMAIN:IPT/TIG 2, DOMAIN:IPT/TIG 3, DOMAIN:PK_Tyr_Ser-Thr, DOMAIN:Protein kinase, DOMAIN:Sema, MUTAGEN:Y->F: Complete loss of kinase activity and of ligand-induced ubiquitination. Alters interaction with PTPN1 and PTPN2. Loss of interaction with PTPN1 and PTPN2; when associated with F-1235., MUTAGEN:Y->F: Complete loss of kinase activity. Alters interaction with PTPN1 and PTPN2. Loss of interaction with PTPN1 and PTPN2. When associated with F-1234., MUTAGEN:Y->F: No effect on ligand-induced CBL-mediated UP_SEQ_FEATURE ubiquitination; when associated with F-1313, F-1349 and F-1356., MUTAGEN:Y->F: No effect on ligand-induced CBL-mediated ubiquitination; when associated with F-1313, F-1349 and F-1365., MUTAGEN:Y->F: No effect on ligand-induced CBL-mediated ubiquitination; when associated with F-1313, F-1349 and F-1365., MUTAGEN:Y->F: No effect on ligand-induced CBL-mediated

ubiquitination; when associated with F-1313, F-1359 and F-1365., MUTAGEN:Y->F: No effect on ligand-induced CBL-mediated ubiquitination; when associated with F-1313, F-1356 and F-1365., MP_BIND:ATP, REGION:Disordered, REGION:Interaction with MUC20, REGION:Interaction with RANBP9, SITE:Breakpoint for translocation to form TPR-MET oncogene, SITE:Cleavage, SITE:Required for ligand-induced CBL-mediated ubiquitination, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

RB1 RB transcriptional corepressor 1(RB1) Related Genes Homo sapiens

BBID 94.E2F_transcriptional_activity_cell_cycle, BIOCARTA

Tumor Suppressor Arf Inhibits Ribosomal Biogenesis, BTG family proteins and cell cycle regulation, Cyclins and Cell Cycle Regulation, FAS signaling pathway (CD95), Cyclin E Destruction Pathway, Cell Cycle, Human Cytomegalovirus and Map Kinase Pathways, HIV-I Nef, Regulation of p27 Phosphorylation during Cell Cycle Progression, p53 Signaling Pathway, Regulation of transcriptional activity by PML, Mechanism of Gene Regulation by Peroxisome Proliferators via PPARa(alpha), Influence of Ras and Rho proteins on G1 to S <u>Transition</u>, RB <u>Tumor Suppressor/Checkpoint Signaling in response to DNA damage</u>, <u>E2F1 Destruction Pathway</u>, <u>Telomeres</u>, <u>Telomerase</u>, <u>Cellular Aging</u>, and <u>Immortality</u>, <u>Overview of telomerase</u> RNA component gene hTerc <u>Transcriptional Regulation</u>, <u>Chaperones modulate</u>

interferon Signaling Pathway, TNFR1 Signaling Pathway, GOTERM BP DIRECT

G1/S transition of mitotic cell cycle, negative regulation of transcription from RNA polymerase II promoter, tissue homeostasis, chondrocyte differentiation, aortic valve morphogenesis, chromatin remodeling, regulation of transcription, DNA-templated, regulation of transcription from RNA polymerase II promoter, negative regulation of protein kinase activity, cell cycle, cell cycle arrest, chromosome condensation, Ras protein signal transduction, spermatogenesis, regulation of mitotic cell cycle, negative regulation of gene expression, viral process, cell differentiation, negative regulation of cell growth, sister chromatid biorientation, anaphasepromoting complex-dependent catabolic process, neuron projection development, heterochromatin assembly, cellular response to insulin stimulus, maintenance of mitotic sister chromatid cohesion, glial cell apoptotic process, skeletal muscle cell differentiation, neuron maturation, enucleate erythrocyte differentiation, negative regulation of sequence-specific DNA binding transcription factor activity, regulation of lipid kinase activity, myoblast differentiation, positive regulation of macrophage differentiation, negative regulation of cell cycle, positive regulation of mitotic metaphase/anaphase transition, negative regulation of smoothened signaling pathway, negative regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, digestive tract development, cell morphogenesis involved in neuron differentiation, negative regulation of epithelial cell proliferation, negative regulation of inflammatory response, striated muscle cell differentiation, cell division, neuron apoptotic process, regulation of cell cycle, protein localization to chromosome, centromeric region, cellular response to xenobiotic stimulus, negative regulation of protein serine/threonine kinase activity, regulation of cohesin loading, negative regulation of transcription involved in G1/S transition of mitotic cell cycle, regulation of centromere complex assembly, hepatocyte apoptotic process, negative regulation of tau-protein kinase activity, positive regulation of extracellular matrix organization, negative regulation of hepatocyte apoptotic process, positive regulation of collagen fibril organization, negative regulation of myofibroblast differentiation, negative regulation of G1/S transition of mitotic cell cycle, positive regulation of transcription regulatory region DNA binding, negative regulation of apoptotic signaling

GOTERM_CC_DIRECT

chromatin, nucleus, nucleoplasm, transcription factor complex, spindle, cytosol, SWI/SNF complex, PML body, Rb-E2F complex,

chromatin lock complex

GOTERM_MF_DIRECT

RNA polymerase II regulatory region sequence-specific DNA binding, RNA polymerase II activating transcription factor binding, transcription corepressor activity, protein binding, transcription factor binding, enzyme binding, kinase binding, ubiquitin protein ligase binding, identical protein binding, phosphoprotein binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, importin-alpha family protein binding, disordered domain specific binding,

INTERPRO

Retinoblastoma-associated protein, B-box, Retinoblastoma-associated protein, A-box, Cyclin-like, Rb C-terminal, Domain of unknown

function DUF3452, retinoblastoma-associated,

KEGG_PATHWAY Endocrine resistance, Cell cycle, Cellular senescence, Cushing syndrome, Hepatitis C, Hepatitis B, Human cytomegalovirus infection,

Human papillomavirus infection, Human T-cell leukemia virus 1 infection, Kaposi sarcoma-associated herpesvirus infection, Epstein-Barr virus infection, Pathways in cancer, Viral carcinogenesis, Chemical carcinogenesis - receptor activation, Pancreatic cancer, Glioma, Prostate cancer, Melanoma, Bladder cancer, Chronic myeloid leukemia, Small cell lung cancer, Non-small cell lung cancer, Breast

cancer, Hepatocellular carcinoma, Gastric cancer

OMIM_DISEASE Bladder cancer, somatic, Retinoblastoma, Retinoblastoma, trilateral, Small cell cancer of the lung, somatic, Osteosarcoma, somatic,

SMART CYCLIN, SM01367, SM01368, SM01369,

UP_KW_BIOLOGICAL_PROCESS Cell cycle, Transcription, Transcription regulation, Host-virus interaction,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP KW DISEASE Tumor suppressor, Disease variant,

UP_KW_DOMAIN Coiled coil.

UP_KW_MOLECULAR_FUNCTION Chromatin regulator, DNA-binding, Repressor, UP_KW_PTM Acetylation, Methylation, Phosphoprotein,

UP_SEQ_FEATURE

COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:CYCLIN, DOMAIN:DUF3452, DOMAIN:RB_A, DOMAIN:RB_B, DOMAIN:Rb_C, MOTIF:Bipartite nuclear localization signal, MUTAGEN:K->R: Abolishes monomethylation by SMYD2 and subsequent interaction with L3MBTL1., MUTAGEN:K->R: Does not affect the ability to be methylated by SMYD2; when associated with 873-R-R-874., MUTAGEN:KK->R: Does not affect the ability to be methylated by SMYD2; when associated with 873-R-R-874., MUTAGEN:KK->R: Does not affect the ability to be methylated by SMYD2; when associated with 873-R-R-874., MUTAGEN:KK->R: Does not after Rb localization in cycling cells, but mislocalizes to the cytoplasm during keratinocyte differentiation. Does not affect the ability to arrest cell growth. Probable loss of acetylation by PCAF., MUTAGEN:T-

>A: Abolishes interaction with Pocket domain; when associated with A-821., MUTAGEN:T->A: Abolishes interaction with Pocket domain; when associated with A-826., REGION:Disordered, REGION:Domain A, REGION:Domain B, REGION:Domain C; mediates interaction with E4F1, REGION:Interaction with LIMD1, REGION:Pocket; binds T and E1A, REGION:Spacer,

RNA binding motif protein 10(RBM10) RBM10 Homo sapiens

negative regulation of transcription from RNA polymerase II promoter, regulation of alternative mRNA splicing, via spliceosome, mRNA splicing, via spliceosome, negative regulation of cell proliferation, RNA splicing, positive regulation of smooth muscle cell apoptotic process, regulation of apoptotic process, negative regulation of mRNA splicing, via spliceosome, 3'-UTR-mediated mRNA stabilization, GOTERM BP DIRECT

GOTERM_CC_DIRECT nucleus, nuclear speck, macromolecular complex,

GOTERM_MF_DIRECT RNA binding, protein binding, miRNA binding, identical protein binding, metal ion binding,

INTERPRO G-patch domain, RNA recognition motif domain, Zinc finger, RanBP2-type, Nucleotide-binding, alpha-beta plait, Zinc finger C2H2-

type/integrase DNA-binding domain

OMIM_DISEASE TARP syndrome, SMART RRM, G_patch, ZnF_RBZ,

UP_KW_BIOLOGICAL_PROCESS mRNA processing, mRNA splicing,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP_KW_DOMAIN Coiled coil, Repeat, Zinc-finger,

UP KW LIGAND Metal-binding, Zinc, UP_KW_MOLECULAR_FUNCTION RNA-binding,

UP_KW_PTM Acetylation, Methylation, Phosphoprotein,

UP_SEQ_FEATURE COMPBIAS: Acidic residues, COMPBIAS: Basic and acidic residues, COMPBIAS: Polar residues, DOMAIN: C2H2-type, DOMAIN: G-patch,

DOMAIN:RRM, DOMAIN:RRM 1, DOMAIN:RRM 2, DOMAIN:RanBP2-type, REGION:Disordered, ZN_FING:C2H2-type; atypical, ZN_FING:RanBP2-type,

SETD2 SET domain containing 2, histone lysine methyltransferase(SETD2) Related Genes

GOTERM_BP_DIRECT

angiogenesis, morphogenesis of a branching structure, <u>neural tube closure</u>, <u>mismatch repair</u>, <u>regulation of transcription</u>, <u>DNA-templated</u>, <u>transcription elongation from RNA polymerase II promoter</u>, <u>regulation of double-strand break repair via homologous</u> recombination, regulation of mRNA export from nucleus, viral process, peptidyl-lysine trimethylation, peptidyl-lysine monomethylation, forebrain development, regulation of cytokinesis, positive regulation of interferon-alpha production, response to type I interferon, nucleosome organization, cell migration involved in vasculogenesis, endodermal cell differentiation, mesoderm morphogenesis, embryonic cranial skeleton morphogenesis, <u>stem cell differentiation</u>, <u>stem cell development</u>, <u>defense response to virus</u>, <u>pericardium development</u>, <u>embryonic placenta morphogenesis</u>, <u>coronary vasculature morphogenesis</u>, <u>histone H3-K36 trimethylation</u>, <u>histone H3-</u> K36 dimethylation, microtubule cytoskeleton organization involved in mitosis, regulation of protein localization to chromatin,

GOTERM CC DIRECT nucleus, nucleoplasm, chromosome,

GOTERM_MF_DIRECT protein binding, protein-lysine N-methyltransferase activity, histone-lysine N-methyltransferase activity, alpha-tubulin binding, metal ion binding, histone methyltransferase activity (H3-K36 specific),

INTERPRO WW domain, SET domain, Post-SET domain, AWS, Ferritin-like superfamily, SRI, Set2 Rpb1 interacting,

KEGG PATHWAY Lysine degradation, Metabolic pathways,

OMIM DISEASE Luscan-Lumish syndrome, SET, WW, PostSET, AWS

UP_KW_BIOLOGICAL_PROCESS Antiviral defense, Differentiation, DNA damage, DNA repair, Immunity, Innate immunity, Transcription, Transcription regulation, Host-

virus interaction,

UP_KW_CELLULAR_COMPONENT Chromosome, Nucleus,

UP_KW_DISEASE Tumor suppressor, Disease variant, Mental retardation, Autism spectrum disorder,

UP KW DOMAIN Coiled coil

UP_KW_LIGAND Metal-binding, Zinc, S-adenosyl-L-methionine,

UP_KW_MOLECULAR_FUNCTION Activator, Chromatin regulator, Developmental protein, Methyltransferase, Transferase, Developmental protein,

UP KW PTM Phosphoprotein, Ubl conjugation, Isopeptide bond,

UP SEQ FEATURE

BINDING:N-propyl sinefungin; inhibitor, BINDING:N-propyl sinefungin; inhibitor; via amide nitrogen, BINDING:S-adenosyl-L-methionine, BINDING:S-adenosyl-L-methionine; via amide nitrogen, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), DOMAIN:AWS, DOMAIN:Post-SET, DOMAIN:SET, DOMAIN:WW, METAL:Zinc 1, METAL:Zinc 2, METAL:Zinc 3, MUTAGEN:C->A: Does not affect methyltransferase activity., MUTAGEN:E->A: Increased methyltransferase activity., MUTAGEN:E->A: Increases interaction with hyperphosphorylated POLR2A; when associated with A-2528., MUTAGEN:E->A: Increases interaction with hyperphosphorylated POLR2A; when associated with A-2531., MUTAGEN:F->A: Does not affect interaction with hyperphosphorylated POLR2A., MUTAGEN:F->A: Does not affect interaction with hyperphosphorylated POLR2A., MUTAGEN:F->A: Does not affect interaction with hyperphosphorylated POLR2A., MUTAGEN:F->A: Does not affect interaction with hyperphosphorylated POLR2A.

POLRZA; when associated with A-Z531., MUTAGEN:F->A: Does not affect interaction with hyperphosphorylated POLRZA, MUTAGEN:F->A: Strongly reduced methyltransferase activity, MUTAGEN:G->A,T: Does not affect interaction with hyperphosphorylated POLRZA., MUTAGEN:H->A: Impairs interaction with hyperphosphorylated POLRZA., MUTAGEN:K->A: Does not affect interaction with hyperphosphorylated POLRZA., MUTAGEN:C->A: Does not affect interaction with hyperphosphorylated POLRZA., MUTAGEN:C->A: Does not affect interaction with hyperphosphorylated POLRZA., MUTAGEN:R->A: Loss of methyltransferase activity., MUTAGEN:R->A: Does not affect interaction with hyperphosphorylated POLRZA., MUTAGEN:R->H,G: Loss of methyltransferase activity. Abolishes ability to monomethylate STAT1., MUTAGEN:R->P,W,K,Q: Loss of methyltransferase activity., MUTAGEN:T->A: Increased methyltransferase

activity., MUTAGEN:V->A: Impairs interaction with hyperphosphorylated POLR2A., MUTAGEN:Y->A: Increased methyltransferase activity, MUTAGEN: V->A: Impairs interaction with hyperphosphorylated Policia, MUTAGEN: 1->A: Interaction with Policials activity, MUTAGEN: 1->A: Interaction with Policials activity, MUTAGEN: V->A: Interaction with Policials activity, REGION: Disordered, REGION: REGION: REGION: REGION: Interaction with TUBA1A, REGION: Low charge region, REGION: N-propyl sinefungin binding; inhibitor, REGION: S-adenosyl-Lmethionine binding.

SMAD4

SMAD family member 4(SMAD4)

Related Genes

Homo sapiens

BIOCARTA

ALK in cardiac myocytes, CTCF, Cell Cycle, NFkB activation by Nontypeable Hemophilus influenzae, TGF beta signaling pathway, Role of

Tob in T-cell activation, WNT Signaling Pathway,

GOTERM_BP_DIRECT

negative regulation of transcription from RNA polymerase II promoter, ovarian follicle development, osteoblast differentiation, branching involved in ureteric bud morphogenesis, response to hypoxia, in utero embryonic development, gastrulation with mouth forming second, outflow tract septum morphogenesis, cardiac conduction system development, atrioventricular valve formation, epithelial to mesenchymal transition involved in endocardial cushion formation, left ventricular cardiac muscle tissue morphogenesis, positive regulation of cell proliferation involved in heart valve morphogenesis, brainstem development, transcription, DNA-templated, regulation of transcription, DNA-templated, regulation of transcription from RNA polymerase II promoter, cellular iron ion homeostasis, transforming growth factor beta receptor signaling pathway, SMAD protein complex assembly, spermatogenesis, single fertilization, axon guidance, aging, cell proliferation, negative regulation of cell proliferation, anatomical structure morphogenesis, negative regulation of cardiac muscle hypertrophy, positive regulation of cardiac muscle cell apoptotic process, positive regulation of epithelial to mesenchymal transition, positive regulation of pathway-restricted SMAD protein phosphorylation, neural crest cell differentiation, protein deubiquitination, regulation of pathway-restricted SMAD protein protein incorporation, regulation of pathway, restricted SMAD protein incorporation, regulation of transforming growth factor beta receptor signaling pathway, cell differentiation, negative regulation of cell growth, adrenal gland development, BMP signaling pathway, positive regulation of transforming growth factor beta receptor signaling pathway, positive regulation of BMP signaling pathway, negative regulation of BMP signaling pathway, somite rostral/caudal axis specification, regulation of transforming growth factor beta2 production, positive regulation of luteinizing hormone secretion, somatic stem cell population maintenance, intracellular signal transduction, atrioventricular canal development, wound healing, endothelial cell activation, negative regulation of protein catabolic process, embryonic digit morphogenesis, negative regulation of transcription, DNA-templated, positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, positive regulation of follicle-stimulating hormone secretion, mesendoderm development, developmental growth, neuron fate commitment, sebaceous gland development, formation of anatomical boundary, regulation of binding, positive regulation of histone H3-K4 methylation, regulation of hair follicle development, uterus development, positive regulation of SMAD protein import into nucleus, SMAD protein signal transduction, ventricular septum morphogenesis, negative regulation of cell death, endocardial cell differentiation, female gonad morphogenesis, interleukin-6-mediated signaling pathway, ERK1 and ERK2 cascade, negative regulation of ERK1 and ERK2 cascade, cellular response to transforming growth factor beta, cellular response to BMP stimulus, metanephric mesenchyme morphogenesis, seminiferous tubule development, positive regulation of transcription from RNA polymerase II promoter involved in sellular proporter in chemical citizulus positive regulation of transcription from RNA polymerase II promoter involved in sellular proporter in chemical citizulus positive regulation of transcription from RNA polymerase II promoter involved in positive regulation of transcription from RNA polymerase II promoter involved in proportion from RNA polymerase II promoter involved in proportion from RNA polymerase II promoter involved in proportion from RNA polymerase II promoter involved in the control of the proportion from RNA polymerase II promoter involved in the control of the RNA polymerase II promoter involved in the control of the RNA polymerase II promoter involved in the RNA polymerase II promoter in cellular response to chemical stimulus, positive regulation of pri-miRNA transcription from RNA polymerase II promoter, negative regulation of cardiac myofibril assembly, positive regulation of histone H3-K9 acetylation,

GOTERM_CC_DIRECT

chromatin, nucleus, nucleoplasm, transcription factor complex, cytoplasm, centrosome, cytosol, activin responsive factor complex, SMAD protein complex, SMAD2-SMAD3 protein complex, RNA polymerase II transcription factor complex,

GOTERM MF DIRECT

transcription regulatory region sequence-specific DNA binding, RNA polymerase II core promoter proximal region sequence-specific DNA binding, RNA polymerase II transcription factor activity, sequence-specific DNA binding, RNA polymerase II transcription factor binding, transcription coactivator binding, transcriptional activator activity, RNA polymerase II transcription regulatory region sequence-specific binding, DNA binding, chromatin binding, transcription factor activity, sequence-specific DNA binding, protein binding, collagen binding, filamin binding, identical protein binding, protein homodimerization activity, sulfate binding, sequencespecific DNA binding, metal ion binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, I-SMAD binding, R-SMAD binding,

INTERPRO **KEGG PATHWAY** SMAD domain, Dwarfin-type, MAD homology 1, Dwarfin-type, SMAD/FHA domain, MAD homology, MH1, Dwarfin, SMAD domain-like, FoxO signaling pathway, Cell cycle, Wnt signaling pathway, TGF-beta signaling pathway, Apelin signaling pathway, Hippo signaling

pathway, Adherens junction, Signaling pathways regulating pluripotency of stem cells, Th17 cell differentiation, AGE-RAGE signaling pathway in diabetic complications, Hepatitis B, Human T-cell leukemia virus 1 infection, Pathways in cancer, Colorectal cancer, Pancreatic cancer, Chronic myeloid leukemia, Hepatocellular carcinoma, Gastric cancer

OMIM DISEASE

SMART

Myhre syndrome, Polyposis, juvenile intestinal, Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, Pancreatic cancer, somatic

DWA, DWB

UP_KW_BIOLOGICAL_PROCESS

Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus, Cytoplasm, UP_KW_DISEASE Disease variant UP KW DOMAIN Coiled coil, UP_KW_LIGAND Metal-binding, Zinc, UP_KW_MOLECULAR_FUNCTION DNA-binding,

UP KW PTM

Acetylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

UP_SEQ_FEATURE

COMPBIAS:Polar residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in ubiquitin), DOMAIN:MH1, DOMAIN:MH2, METAL:Zinc, MUTAGEN:K->R: Abolishes ubiquitination., MUTAGEN:R->S: No effect on heterotrimerization. Greatly reduced transcriptional activation., MUTAGEN:R->S: No effect on heterotrimerization. Partially diminished transcriptional activation., MUTAGEN:R->S: Reduced heterotrimerization., REGION:Disordered, REGION:Mediates interaction with ZBTB7A, REGION:SAD, SITE:Necessary for heterotrimerization,

SMARCA4

SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a, member 4(SMARCA4)

BIOCARTA

Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes, Control of Gene Expression by Vitamin D Receptor,

GOTERM BP DIRECT

negative regulation of transcription from RNA polymerase II promoter, RNA polymerase I transcriptional preinitiation complex assembly, neural retina development, chromatin organization, nucleosome disassembly, chromatin remodeling, regulation of transcription, DNA-templated, regulation of transcription from RNA polymerase II promoter, nervous system development, positive regulation of Wnt signaling pathway, negative regulation of cell growth, interleukin-7-mediated signaling pathway, ATP-dependent chromatin remodeling, positive regulation by host of viral transcription, negative regulation of transcription, DNA-templated, positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, positive regulation of sequence-specific DNA binding transcription factor activity, negative regulation of androgen receptor signaling pathway, positive regulation of transcription of nuclear large rRNA transcript from RNA polymerase I promoter, positive regulation of glucose mediated signaling pathway, positive regulation of pri-miRNA transcription from RNA polymerase II promoter, beta-catenin-TCF complex

GOTERM_CC_DIRECT

chromatin, extracellular space, nucleus, nucleoplasm, nucleolus, membrane, SWI/SNF complex, macromolecular complex, npBAF

complex, nBAF complex,

GOTERM_MF_DIRECT

RNA polymerase I CORE element sequence-specific DNA binding, transcription cofactor binding, p53 binding, DNA binding, transcription coactivator activity, transcription corepressor activity, RNA binding, helicase activity, protein binding, ATP binding, DNAdependent ATPase activity, transcription factor binding, hydrolase activity, ATPase activity, Tat protein binding, <u>nucleosomal DNA</u> binding, <u>histone binding</u>, protein N-terminus binding, androgen receptor binding, DNA polymerase binding, lysine-acetylated histone binding, nucleosome-dependent ATPase activity,

INTERPRO

SNF2-related, Bromodomain, Helicase, C-terminal, BRK domain, Helicase, superfamily 1/2, ATP-binding domain, Helicase/SANT-associated, DNA binding, Glutamine-Leucine-Glutamine, QLQ, Bromodomain, conserved site, P-loop containing nucleoside triphosphate

KEGG PATHWAY Thermogenesis, Hepatocellular carcinoma,

OMIM_DISEASE Rhabdoid tumor predisposition syndrome 2, Coffin-Siris syndrome 4, SMART BROMO, DEXDc, HELICc, HSA, BRK, SM00951, SM01314, UP KW BIOLOGICAL PROCESS Neurogenesis, Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP KW DISEASE Disease variant, Mental retardation, UP_KW_DOMAIN Bromodomain, Coiled coil UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Activator, Chromatin regulator, Helicase, Hydrolase, Repressor, RNA-binding,

UP KW PTM Acetylation, Phosphoprotein, Ubl conjugation, Isopeptide bond

UP_SEQ_FEATURE COMPBIAS: Acidic residues, COMPBIAS: Basic and acidic residues, COMPBIAS: Polar residues, COMPBIAS: Pro residues,

COMPBIAS:Action residues, COMPBIAS:Basic and action residues, COMPBIAS:Polar residues, Polar residues, COMPBIAS:Polar res

for binding to 'Lys-15'-acetylated histone 3,

Homo sapiens ADGRL 2 adhesion G protein-coupled receptor L2(ADGRL2) Related Genes

GOTERM_BP_DIRECT cell surface receptor signaling pathway, G-protein coupled receptor signaling pathway, adenylate cyclase-activating G-protein coupled receptor signaling pathway, brain development, response to bacterium, synapse organization, positive regulation of synapse assembly,

GOTERM CC DIRECT plasma membrane, integral component of plasma membrane, integral component of membrane, axon, neuron projection,

glutamatergic synapse, integral component of postsynaptic membrane,

GOTERM MF DIRECT G-protein coupled receptor activity, latrotoxin receptor activity, carbohydrate binding,

INTERPRO GPS domain, GPCR, family 2, secretin-like, D-galactoside/L-rhamnose binding SUEL lectin domain, GPCR, family 2, extracellular

hormone receptor domain, Olfactomedin-like, GPCR, family 2, latrophilin, C-terminal, GPCR, family 2, latrophilin, GPCR, family 2-like,

GPCR, family 2, secretin-like, conserved site,

SMART HormR, OLF, GPS, UP_KW_CELLULAR_COMPONENT Membrane

UP KW DOMAIN Signal, Transmembrane, Transmembrane helix,

UP KW LIGAND Lectin

GOTERM_BP_DIRECT

UP_KW_MOLECULAR_FUNCTION G-protein coupled receptor, Receptor, Transducer, UP_KW_PTM Glycoprotein, Phosphoprotein, Disulfide bond,

UP_SEQ_FEATURE

chromatin remodeling, regulation of transcription from RNA polymerase II promoter, modulation of age-related behavioral decline,

CARBOHYD:N-linked (GlcNAc...) asparagine, COMPBIAS:Polar residues, DOMAIN:GPS, DOMAIN:G_PROTEIN_RECEP_F2_3, DOMAIN:G_PROTEIN_RECEP_F2_4, DOMAIN:Latrophilin, DOMAIN:Olfactomedin-like, DOMAIN:SUEL-type lectin, REGION:Disordered, SITE:Cleavage, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical, TRANSMEM:Helical; Name=1, TRANSMEM:Helical; Name=2, TRANSMEM:Helical; Name=3, TRANSMEM:Helical; Name=4, TRANSMEM:Helical; Name=5, TRANSMEM:Helical; Name=6, TRANSMEM:Helical; Name=7,

BAZ2B bromodomain adjacent to zinc finger domain 2B(BAZ2B) Related Genes Homo sapiens

GOTERM CC DIRECT nucleus,

GOTERM_MF_DIRECT DNA binding, protein binding, metal ion binding,

INTERPRO Bromodomain, Methyl-CpG DNA binding, Zinc finger, PHD-type, Zinc finger, FYVE/PHD-type, Zinc finger, RING/FYVE/PHD-type, DNA-

binding, integrase-type, Bromodomain, conserved site, DDT domain superfamily, Zinc finger, PHD-finger,

SMART PHD, BROMO, MBD, DDT,

UP KW BIOLOGICAL PROCESS Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus,

UP KW DOMAIN Bromodomain, Coiled coil, Zinc-finger,

UP_KW_LIGAND Metal-binding, Zinc, UP_KW_MOLECULAR_FUNCTION **DNA-binding**

UP KW PTM Acetylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

COMPBIAS: Acidic residues, COMPBIAS: Basic and acidic residues, COMPBIAS: Polar residues, CROSSLNK: Glycyl lysine isopeptide (Lys-UP SEQ FEATURE

Gly) (interchain with G-Cter in SUMO2), DOMAIN:Bromo, DOMAIN:DDT, DOMAIN:MBD, REGION:Disordered, ZN_FING:PHD-type,

cadherin 12(CDH12) CDH12

GOTERM BP DIRECT cell morphogenesis, cell-cell junction assembly, homophilic cell adhesion via plasma membrane adhesion molecules, multicellular organism development, calcium-dependent cell-cell adhesion via plasma membrane cell adhesion molecules, adherens junction

organization, cell-cell adhesion via plasma-membrane adhesion molecules,

GOTERM CC DIRECT plasma membrane, adherens junction, integral component of membrane, catenin complex,

GOTERM_MF_DIRECT calcium ion binding, cadherin binding,

INTERPRO Cadherin, cytoplasmic domain, Cadherin, Cadherin-like, Cadherin conserved site, Catenin binding domain,

UP_KW_BIOLOGICAL_PROCESS Cell adhesion,

UP_KW_CELLULAR_COMPONENT Membrane, Cell membrane,

UP KW DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_LIGAND Calcium, Metal-binding,

UP_KW_PTM Cleavage on pair of basic residues, Glycoprotein, Phosphoprotein,

CARBOHYD:N-linked (GlcNac...) asparagine, DOMAIN:Cadherin 1, DOMAIN:Cadherin 2, DOMAIN:Cadherin 3, DOMAIN:Cadherin 4, DOMAIN:Cadherin 5, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical, UP_SEQ_FEATURE

catenin beta 1(CTNNB1) Homo sapiens

BIOCARTA ALK in cardiac myocytes, Cell to Cell Adhesion Signaling, Inactivation of Gsk3 by AKT causes accumulation of b-catenin in Alveolar Macrophages, Multi-step Regulation of Transcription by Pitx2, Presenilin action in Notch and Wnt signaling, Trefoil Factors Initiate Mucosal Healing, WNT Signaling Pathway,

GOTERM_BP_DIRECT negative regulation of transcription from RNA polymerase II promoter, protein polyubiquitination, embryonic axis specification, cell

morphogenesis involved in differentiation, branching involved in blood vessel morphogenesis, branching involved in ureteric bud morphogenesis, in utero embryonic development, gastrulation with mouth forming second, cell fate specification, endodermal cell fate commitment, neuron migration, epithelial to mesenchymal transition, neural plate development, positive regulation of neuroblast proliferation, positive regulation of mesenchymal cell proliferation, lens morphogenesis in camera-type eye, regulation of secondary heart field cardioblast proliferation, metanephros morphogenesis, negative regulation of mesenchymal to epithelial transition involved

in metanephros morphogenesis, cell adhesion, cell-matrix adhesion, Wnt signaling pathway, calcium modulating pathway, chemical synaptic transmission, ectoderm development, glial cell fate determination, negative regulation of cell proliferation, response to xenobiotic stimulus, anterior/posterior axis specification, dorsal/ventral axis specification, proximal/distal pattern formation, positive

regulation of gene expression, negative regulation of gene expression, positive regulation of epithelial to mesenchymal transition, positive regulation of heparan sulfate proteoglycan biosynthetic process, viral process, Wnt signaling pathway, negative regulation of angiogenesis, stem cell population maintenance, layer formation in cerebral cortex, central nervous system vasculogenesis, osteoclast differentiation, male genitalia development, hindbrain development, regulation of centriole-centriole cohesion, pancreas development, hair follicle morphogenesis, regulation of myelination, positive regulation of telomere maintenance via telomerase, negative regulation of chondrocyte differentiation, response to estradiol, positive regulation of type I interferon production, positive regulation of transcription elongation from RNA polymerase II promoter, T cell differentiation in thymus, negative regulation of protein sumoylation, adherens junction organization, adherens junction assembly, protein localization to cell surface, cellular protein localization, embryonic heart tube development, genitalia morphogenesis, embryonic forelimb morphogenesis, embryonic hindlimb morphogenesis, hair cell differentiation, entry of bacterium into host cell, detection of muscle stretch, embryonic skeletal limb joint morphogenesis, astrocytedopaminergic neuron signaling, regulation of T cell proliferation, odontogenesis of dentin-containing tooth, response to drug, embryonic digit morphogenesis, positive regulation of apoptotic process, negative regulation of apoptotic process, positive regulation of I-kappaB kinase/NF-kappaB signaling, proteasome-mediated ubiquitin-dependent protein catabolic process, positive regulation of MAPK cascade, positive regulation of neuron apoptotic process, canonical Wnt signaling pathway involved in positive regulation of epithelial to mesenchymal transition, canonical Wnt signaling pathway involved in negative regulation of apoptotic process, canonical Wnt signaling pathway involved in mesenchymal stem cell differentiation, canonical Wnt signaling pathway involved in osteoblast differentiation, bone resorption, positive regulation of endothelial cell differentiation, positive regulation of osteoblast differentiation, positive regulation of osteoclast differentiation, positive regulation of fibroblast growth factor receptor signaling pathway, regulation of angiogenesis, negative regulation of transcription, DNA-templated, positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, negative regulation of mitotic cell cycle, embryonic, regulation of fibroblast proliferation, cell maturation, synaptic vesicle transport, thymus development, oocyte development, embryonic foregut morphogenesis, positive regulation of skeletal muscle tissue development, regulation of smooth muscle cell proliferation, negative regulation of oligodendrocyte differentiation, regulation of neurogenesis, synapse organization, positive regulation of sequence-specific DNA binding transcription factor activity, smooth muscle cell differentiation, positive regulation of muscle cell differentiation, positive regulation of muscle cell differentiation, positive regulation of histone H3-K4 methylation, regulation of timing of anagen, positive regulation of telomerase activity, oviduct development, canonical Wnt signaling pathway, regulation of transcription initiation from RNA polymerase II promoter, trachea formation, epithelial tube branching involved in lung morphogenesis, lung cell differentiation, lung-associated mesenchyme development, lung induction, epithelial cell differentiation involved in prostate gland development, positive regulation of epithelial cell proliferation involved in prostate gland development, hair follicle placode formation, regulation of canonical Wnt signaling pathway, mesenchymal cell proliferation involved in lung development, positive regulation of branching involved in lung morphogenesis, endothelial tube morphogenesis, fungiform papilla formation, canonical Wnt signaling pathway involved in positive regulation of cardiac outflow tract cell proliferation, sympathetic ganglion development, cranial ganglion development, regulation of centromeric sister chromatid cohesion, cellular response to growth factor stimulus, cellular response to indole-3-methanol, renal vesicle formation, renal inner medulla development, renal outer medulla development, nephron tubule formation, regulation of nephron tubule epithelial cell differentiation, mesenchymal stem cell differentiation, negative regulation of canonical Wnt signaling pathway, regulation of calcium ion import, synaptic vesicle clustering, cell-cell adhesion, negative regulation of oxidative stress-induced neuron death, positive regulation of core promoter binding, beta-catenin-TCF complex assembly, beta-catenin destruction complex disassembly, cranial skeletal system development, midbrain dopaminergic neuron differentiation, canonical Wnt signaling pathway involved in midbrain dopaminergic neuron differentiation, neuron projection extension, embryonic brain development, dorsal root ganglion development, regulation of protein localization to cell surface, positive regulation of determination of dorsal identity, negative regulation of apoptotic signaling pathway

GOTERM_CC_DIRECT

euchromatin, spindle pole, nucleus, nucleoplasm, transcription factor complex, cytoplasm, centrosome, cytosol, plasma membrane, cell-cell junction, adherens junction, fascia adherens, bicellular tight junction, focal adhesion, cell cortex, membrane, basolateral plasma membrane, apicolateral plasma membrane, lateral plasma membrane, catenin complex, flotillin complex, Z disc, amellipodium, cell junction, beta-catenin destruction complex, microvillus membrane, macromolecular complex, protein-DNA complex, Scrib-APC-beta-catenin complex, presynaptic membrane, cell projection, apical part of cell, synapse, postsynaptic membrane, perinuclear region of cytoplasm, extracellular exosome, beta-catenin-TCF7L2 complex, cell periphery, Schaffer collateral - CA1 synapse, presynaptic active zone cytoplasmic component, postsynaptic density, intracellular component, Beta-catenin-ICAT complex, beta-catenin-TCF complex, Wnt signalosome,

GOTERM_MF_DIRECT

RNA polymerase II transcription factor binding, RNA polymerase II activating transcription factor binding, transcription cofactor binding, chromatin binding, transcription coactivator activity, protein binding, beta-catenin binding, protein C-terminus binding, transcription factor binding, ligand-dependent nuclear receptor binding, enzyme binding, kinase binding, protein kinase binding, protein phosphatase binding, estrogen receptor binding, nuclear hormone receptor binding, ion channel binding, alpha-catenin binding, cadherin binding, SMAD binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, I-SMAD binding, repressing transcription factor binding, disordered domain specific binding, histone methyltransferase binding,

INTERPRO **KEGG PATHWAY** Armadillo, Armadillo-like helical, Beta-catenin, Armadillo-type fold,

Rap1 signaling pathway, Wnt signaling pathway, Hippo signaling pathway, Focal adhesion, Adherens junction, Signaling pathways regulating pluripotency of stem cells, Leukocyte transendothelial migration, Melanogenesis, Thyroid hormone signaling pathway, Cushing syndrome, Alcoholic liver disease, Alzheimer disease, Pathways of neurodegeneration - multiple diseases, Bacterial invasion of epithelial cells, Salmonella infection, Hepatitis C, Human cytomegalovirus infection, Human papillomavirus infection, Kaposi sarcoma-associated herpesvirus infection, Pathways in cancer, Proteoglycans in cancer, <u>Colorectal cancer</u>, <u>Endometrial cancer</u>, <u>Prostate cancer</u>, Thyroid cancer, <u>Basal cell carcinoma</u>, <u>Breast cancer</u>, <u>Hepatocellular carcinoma</u>, <u>Gastric cancer</u>, <u>Arrhythmogenic right ventricular</u> cardiomyopathy, Fluid shear stress and atherosclerosis

OMIM_DISEASE

Colorectal cancer, somatic, Hepatocellular carcinoma, somatic, Pilomatricoma, somatic, Medulloblastoma, somatic, Ovarian cancer, somatic, Neurodevelopmental disorder with spastic diplegia and visual defects, Exudative vitreoretinopathy 7,

SMART

UP_KW_DISEASE

UP_SEQ_FEATURE

UP_KW_BIOLOGICAL_PROCESS

Cell adhesion, Neurogenesis, Transcription, Transcription regulation, Wnt signaling pathway, Host-virus interaction,

UP_KW_CELLULAR_COMPONENT Cytoskeleton, Membrane, Nucleus, Synapse, Cytoplasm, Cell junction, Cell projection, Cell membrane, Disease variant, Mental retardation

UP_KW_DOMAIN

Repeat, UP_KW_MOLECULAR_FUNCTION Activator,

UP KW PTM

Acetylation, Glycoprotein, Phosphoprotein, S-nitrosylation, Ubl conjugation,

CARBOHYD:O-linked (GlcNAc) serine; alternate, COMPBIAS:Polar residues, MUTAGEN:F->A: Abolishes CTNNBIP1 binding; when CARBOHYD:O-linked (GICNAC) serine; alternate, COMPBIAS:Polar residues, MUTAGEN:F->A: Abolishes CINNBIP1 binding; when associated with A-661., MUTAGEN:F->A: Abolishes or strongly reduces AXIN2 binding, and strongly reduces or abolishes LEF1 binding., MUTAGEN:H->A: Abolishes or strongly reduces AXIN1 and AXIN2 binding. Strongly reduces phosphorylation and degradation; when associated with A-386 and A-383., MUTAGEN:K->A: Abolishes APC binding., MUTAGEN:K->A: Abolishes TCF7L2 and LEF1 binding., MUTAGEN:K->A: Abolishes or strongly reduces AXIN1 and AXIN2 binding., MUTAGEN:K->A: Strongly reduces or abolishes LEF1 binding., MUTAGEN:K->E: Abolishes TCF7L2 binding., MUTAGEN:L->A: Abolishes interaction with BCL9 but no effect on interaction with CDH3; when associated with A-159., MUTAGEN:L->A: No effect on interaction with BCL9 but no effect on interaction with BCL9 and CDH3. MUTAGEN:L->A: No effect on interaction with BCL9 and CDH3. with BCL9 and CDH3., MUTAGEN:L->A: No effect on interaction with BCL9 and CDH3. Abolishes interaction with BCL9 but no effect on interaction with CDH3; when associated with A-156., MUTAGEN:N->A: Abolishes TCF7L2 and LEF1 binding., MUTAGEN:R->A: Abolishes CTNNBIP1 binding; when associated with A-660., MUTAGEN:R->A: Abolishes TCF7L2 binding, and strongly reduces or abolishes LEF1 binding., MUTAGEN:R->A: Strongly reduces APC binding. Strongly reduces phosphorylation and degradation; when associated with A-260 and A-383., MUTAGEN:S->F: No effect., MUTAGEN:W->A: Abolishes APC binding. Strongly reduces phosphorylation and and A-363, MOTAGEN:5-7F: No effect, MOTAGEN:W-7A: Abolishes APC billioning, Strongly reduces principling price and BCL9L., MUTAGEN:Y->F: Abolishes phosphorylation by PTK6., REGION:Interaction with BCL9 and BCL9L., MUTAGEN:Y->F: Abolishes phosphorylation by PTK6., REGION:Interaction with BCL9, REGION:Interaction with SCRIB, REGION:Interaction with VCL, REPEAT:ARM, REPEAT:ARM 1, REPEAT:ARM 10, REPEAT:ARM 11, REPEAT:ARM 12, REPEAT:ARM 2, REPEAT:ARM 3, REPEAT:ARM 4, REPEAT:ARM 5, REPEAT:ARM 6, REPEAT:ARM 7, REPEAT:ARM 8, REPEAT:ARM 9,

COL1A1

collagen type I alpha 1 chain(COL1A1)

Related Genes

Homo sapiens

GOTERM_BP_DIRECT

skeletal system development, ossification, blood vessel development, osteoblast differentiation, intramembranous ossification, endochondral ossification, cell adhesion, blood coagulation, visual perception, sensory perception of sound, response to xenobiotic stimulus, response to mechanical stimulus, positive regulation of epithelial to mesenchymal transition, negative regulation of cell-substrate adhesion, protein transport, platelet activation, extracellular matrix organization, collagen fibril organization, positive regulation of cell migration, response to corticosteroid, response to estradiol, collagen biosynthetic process, protein localization to nucleus, tooth mineralization, collagen-activated tyrosine kinase receptor signaling pathway, wound healing, response to drug, response to hydrogen peroxide, response to peptide hormone, skin development, skin morphogenesis, cellular response to fibroblast growth factor stimulus, tooth eruption, positive regulation of transcription, DNA-templated, embryonic skeletal system development, regulation of immune response, leukocyte migration, response to cAMP, response to hyperoxia, face morphogenesis, bone trabecula formation, cartilage development involved in endochondral bone morphogenesis, cellular response to amino acid stimulus, cellular response to mechanical stimulus, cellular response to retinoic acid, cellular response to vitamin E, cellular response to tumor necrosis factor, cellular response to epidermal growth factor stimulus, cellular response to transforming growth factor beta stimulus, positive regulation of canonical Wnt signaling pathway, cellular response to fluoride,

GOTERM_CC_DIRECT

extracellular region, collagen trimer, collagen type I trimer, extracellular space, cytoplasm, endoplasmic reticulum lumen, Golgi

apparatus, integral component of membrane, secretory granule, extracellular matrix

GOTERM_MF_DIRECT protease binding, extracellular matrix structural constituent, protein binding, extracellular matrix structural constituent conferring

tensile strength, identical protein binding, metal ion binding, platelet-derived growth factor binding,

Fibrillar collagen, C-terminal, von Willebrand factor, type C, Collagen triple helix repeat,

KEGG PATHWAY PI3K-Akt signaling pathway, Focal adhesion, ECM-receptor interaction, Platelet activation, Relaxin signaling pathway, AGE-RAGE

signaling pathway in diabetic complications, Protein digestion and absorption, <u>Amoebiasis</u>, Human papillomavirus infection,

Proteoglycans in cancer, Diabetic cardiomyopathy,

OMIM_DISEASE Caffey disease, Ehlers-Danlos syndrome, arthrochalasia type, 1, Osteogenesis imperfecta, type I, Osteogenesis imperfecta, type II,

Osteogenesis imperfecta, type IV, Bone mineral density variation QTL, osteoporosis, Osteogenesis imperfecta, type III, Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1,

COLFI, VWC,

UP_KW_CELLULAR_COMPONENT E racellular matrix, Membrane, Secreted,

UP KW DISEASE ase variant, <u>Dwarfism</u>, <u>Ehlers-Danlos syndrome</u>, <u>Osteogenesis imperfecta</u>,

UP KW DOMAIN Collagen, Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_LIGAND Calcium, Metal-binding

UP_KW_PTM Glycoprotein, Hydroxylation, Phosphoprotein, Pyrrolidone carboxylic acid, Disulfide bond,

UP_SEQ_FEATURE

CARBOHYD:N-linked (GlcNAc...) asparagine, CARBOHYD:O-linked (Gal...) hydroxylysine; alternate, COMPBIAS:Basic and acidic residues, COMPBIAS:Pro residues, DISULFID:Interchain, DOMAIN:Fibrillar collagen NC1, DOMAIN:VWFC, METAL:Calcium, METAL:Calcium; via carbonyl oxygen, MOTIF:Cell attachment site, PROPEP:C-terminal propeptide, PROPEP:N-terminal propeptide, REGION:Disordered, REGION:Nonhelical region (C-terminal), REGION:Nonhelical region, (N-terminal), REGION:Triple-helical region, SITE:Cleavage; by collagenase, SITE:Cleavage; by procollagen C-endopeptidase, SITE:Cleavage; by procollagen N-endopeptidase, TRANSMEM:Helical,

CDK12 cyclin dependent kinase 12(CDK12) Related Genes

GOTERM BP DIRECT

transcription elongation from RNA polymerase II promoter, mRNA processing, protein phosphorylation, RNA splicing, positive regulation of transcription elongation from RNA polymerase II promoter, regulation of MAP kinase activity, regulation of RNA splicing, protein autophosphorylation, regulation of cell cycle, phosphorylation of RNA polymerase II C-terminal domain, negative regulation of

stem cell differentiation.

GOTERM CC DIRECT cyclin-dependent protein kinase holoenzyme complex, cyclin K-CDK12 complex, nucleoplasm, cyclin/CDK positive

transcription elongation factor complex, nuclear speck, nuclear cyclin-dependent protein kinase holoenzyme complex,

protein kinase activity, cyclin-dependent protein serine/threonine kinase activity, protein serine/threonine/tyrosine kinase activity, protein binding, ATP binding, RNA polymerase II carboxy-terminal domain kinase activity, kinase activity, protein kinase binding, cyclin

INTERPRO Protein kinase, catalytic domain, Serine/threonine-protein kinase, active site, Protein kinase-like domain, Protein kinase, ATP binding

site,

SMART S_TKc,

UP_KW_BIOLOGICAL_PROCESS mRNA processing, mRNA splicing,

UP_KW_CELLULAR_COMPONENT Nucleus

GOTERM_MF_DIRECT

UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Kinase, Serine/threonine-protein kinase, Transferase UP KW PTN Phosphoprotein, Ubl conjugation, Isopeptide bond,

UP_SEQ_FEATURE ACT_SITE:Proton acceptor, BINDING:ATP, COMPBIAS:Basic and acidic residues, COMPBIAS:Basic residues, COMPBIAS:Polar residues,

COMPBIAS: Pro residues, CROSSLNK: Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), DOMAIN: Protein kinase, MUTAGEN: D->N: Abolishes kinase activity., NP_BIND: ATP, REGION: Disordered,

CDKN2A cyclin dependent kinase inhibitor 2A(CDKN2A)

BBID $1. RB phosphoE2F,\ 26. cyclin-CDK_complexes,\ 94. E2F_transcriptional_activity_cell_cycle, \\$

BIOCARTA Tumor Suppressor Arf Inhibits Ribosomal Biogenesis, Cyclins and Cell Cycle Regulation, CTCF, Cell Cycle,

GOTERM_BP_DIRECT G1/S transition of mitotic cell cycle, protein polyubiquitination, mitophagy, negative regulation of cell-matrix adhesion, rRNA

processing, negative regulation of protein kinase activity, apoptotic process, activation of cysteine-type endopeptidase activity involved in apoptotic process, cell cycle, cell cycle arrest, Ras protein signal transduction, negative regulation of cell proliferation, apoptotic mitochondrial changes, regulation of G2/M transition of mitotic cell cycle, positive regulation of gene expression, negative regulation of cell growth, negative regulation of B cell proliferation, regulation of protein stability, protein destabilization, negative regulation of NF-kappaB transcription factor activity, negative regulation of immature T cell proliferation in thymus, positive regulation of protein sumoylation, positive regulation of smooth muscle cell apoptotic process, senescence-associated heterochromatin focus assembly, negative regulation of phosphorylation, positive regulation of apoptotic process, positive regulation of DNA damage response, signal transduction by p53 class mediator, negative regulation of cyclin-dependent protein serine/threonine kinase activity, negative regulation of transcription, DNA-templated, positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, regulation of protein export from nucleus, somatic stem cell division, protein stabilization, negative regulation of ubiquitin-protein transferase activity, regulation of cell cycle, mitochondrial depolarization, regulation of transcription initiation from RNA polymerase II promoter, protein K63-linked ubiquitination, positive regulation of cell cycle arrest, cellular senescence, replicative senescence, intrinsic apoptotic signaling pathway, positive regulation of protein localization to nucleus, regulation of signal transduction by p53 class mediator, regulation of signal transduction by p53 class mediator, regulation of apoptotic DNA fragmentation, negative regulation of proteolysis involved in cellular protein catabolic process, regulation of protein targeting to mitochondrion, negative regulation of ubiquitin protein ligase activity, amyloid fibril formation, regulation of G1/S transition of mitotic cell cycle, negative regulation of protein ubiquitination involved in ubiquitin-dependent protein catabolic process, positive regulation of macrophage apoptotic process, negative regulation of G1/S transition of mitotic cell cycle, negative regulation of protein neddylation, positive regulation of cellular senescence,

GOTERM CC DIRECT nucleus, nucleoplasm, nucleolus, cytoplasm, mitochondrion, mitochondrial matrix, cytosol, macromolecular complex, senescence-

associated heterochromatin focus.

GOTERM_MF_DIRECT p53 binding, DNA binding, RNA binding, cyclin-dependent protein serine/threonine kinase inhibitor activity, protein binding,

transcription factor binding, kinase activity, SUMO transferase activity, protein kinase binding, NF-kappaB binding, ubiquitin-protein transferase inhibitor activity, MDM2/MDM4 family protein binding, disordered domain specific binding, ubiquitin ligase inhibitor activity,

INTERPRO Cyclin-dependent kinase inhibitor 2A, Ankyrin repeat-containing domain.

KEGG_PATHWAY Endocrine resistance, Platinum drug resistance, Cell cycle, p53 signaling pathway, Cellular senescence, Cushing syndrome, Human

cytomegalovirus infection, Human T-cell leukemia virus 1 infection, Pathways in cancer, Viral carcinogenesis, MicroRNAs in cancer Pancreatic cancer, Glioma, Melanoma, Bladder cancer, Chronic myeloid leukemia, Non-small cell lung cancer, Hepatocellular

carcinoma,

OMIM_DISEASE Melanoma, cutaneous malignant, 2, Melanoma and neural system tumor syndrome, Melanoma-pancreatic cancer syndrome,

UP_KW_BIOLOGICAL_PROCESS Apoptosis, Cell cycle, rRNA processing, Transcription, Transcription regulation, Ubl conjugation pathway,

UP_KW_CELLULAR_COMPONENT Mitochondrion, Nucleus, Cytoplasm, UP KW DISEASE Tumor suppressor, Disease variant, Li-Fraumeni syndrome, Li-Fraumeni syndrome,

UP KW DOMAIN ANK repeat, Repeat

UP_KW_MOLECULAR_FUNCTION Cyclin, DNA-binding, Kinase, Transferase, UP_KW_PTM Acetylation, Phosphoprotein, Ubl conjugation,

UP_SEQ_FEATURE REGION: Disordered, REGION: Interaction with CDK5RAP3 and MDM2, REPEAT: ANK 1, REPEAT: ANK 2, REPEAT: ANK 3, REPEAT: ANK 4,

EGFR epidermal growth factor receptor(EGFR) BIOCARTA Agrin in Postsynaptic Differentiation, Angiotensin II mediated activation of JNK Pathway via Pyk2 dependent signaling, Role of EGF

Receptor Transactivation by GPCRs in Cardiac Hypertrophy, CBL mediated ligand-induced downregulation of EGF receptors, The role of FYVE-finger proteins in vesicle transport, EGF Signaling Pathway, Map Kinase Inactivation of SMRT Corepressor, Neuroregulin receptor degredation protein-1 Controls ErbB3 receptor recycling, Erk1/Erk2 Mapk Signaling pathway, Role of ERBB2 in Signal Transduction and Oncology, Keratinocyte Differentiation, mCalpain and friends in Cell motility, Sprouty regulation of tyrosine kinase signals, Telomeres,

Telomerase, Cellular Aging, and Immortality, Trefoil Factors Initiate Mucosal Healing,

GOTERM BP DIRECT MAPK cascade, cell morphogenesis, ossification, embryonic placenta development, positive regulation of protein phosphorylation, hair follicle development, regulation of transcription from RNA polymerase II promoter, translation, receptor-mediated endocytosis,

response to osmotic stress, signal transduction, cell surface receptor signaling pathway, transmembrane receptor protein tyrosi kinase signaling pathway, epidermal growth factor receptor signaling pathway, activation of phospholipase C activity, multicellular organism development, salivary gland morphogenesis, midgut development, learning or memory, circadian rhythm, positive regulation organism development, salval y gland moliphogenesis, indigate development, learning of mentiory, circalar hydrin, positive regulation of netric oxide mediated signal transduction, magnesium ion homeostasis, regulation of phosphatidylinositol 3-kinase signaling, diterpenoid metabolic process, peptidyl-tyrosine phosphorylation, viral life cycle, cerebral cortex cell migration, cell differentiation, positive regulation of cell growth, lung development, positive regulation of cell migration, positive regulation of superoxide anion generation, positive regulation of peptidyl-serine phosphorylation, response to cobalamin, response to hydroxylsoflavone, positive regulation of kinase activity, cellular response to reactive oxygen species, cellular response to drug, peptidyl-tyrosine autophosphorylation, ERBB2 signaling pathway, negative regulation of epidermal growth factor receptor signaling pathway, wound healing, negative regulation of protein catabolic process, positive regulation of phosphorylation, ovulation cycle, hydrogen peroxide metabolic process, activation of phospholipase A2 activity by calcium-mediated signaling, negative regulation of apoptotic process, positive regulation of MAP kinase activity, tongue development, positive regulation of cyclin-dependent protein serine/threonine kinase activity, positive regulation of DNA repair, positive regulation of DNA replication, negative regulation of Notch signaling pathway, positive regulation of bone resorption, positive regulation of transcription, DNA-templated, positive regulation of vasoconstriction, negative regulation of mitotic cell cycle, positive regulation of transcription from RNA polymerase II promoter, regulation of JNK cascade, viral entry into host cell, protein autophosphorylation, astrocyte activation, positive regulation of fibroblast proliferation, digestive tract morphogenesis, positive regulation of smooth muscle cell proliferation, neuron projection morphogenesis, positive regulation of epithelial cell proliferation, positive regulation of inflammatory response, regulation of peptidyl-tyrosine phosphorylation, regulation of nitric-oxide synthase activity, protein insertion into membrane, response to calcium ion, positive regulation of protein kinase B signaling, positive regulation of synaptic transmission, glutamatergic, positive regulation of glial cell proliferation, morphogenesis of an epithelial fold, membrane organization, eyelid development in camera-type eye, response to UV-A, positive regulation of mucus secretion, regulation of ERK1 and ERK2 cascade, positive regulation of ERK1 and ERK2 cascade, cellular response to amino acid stimulus, cellular response to mechanical stimulus, cellular response to cadmium ion, cellular response to epidermal growth factor stimulus, cellular response to estradiol stimulus, cellular response to dexamethasone stimulus, positive regulation of canonical Wnt signaling pathway, liver regeneration, cell-cell adhesion, positive regulation of protein kinase C activity, positive regulation of G1/S transition of mitotic cell cycle, negative regulation of ERBB signaling pathway, positive regulation of NIK/NF-kappaB signaling, positive regulation of prolactin secretion, positive regulation of protein localization to plasma membrane, positive regulation of production of miRNAs involved in gene silencing by miRNA, negative regulation of cardiocyte differentiation,

GOTERM CC DIRECT

Golgi membrane, extracellular space, nucleus, cytoplasm, endosome, endoplasmic reticulum membrane, plasma membrane, integral component of plasma membrane, focal adhesion, basal plasma membrane, cell surface, endosome membrane, membrane, integral component of membrane, basolateral plasma membrane, apical plasma membrane, cell junction, clathrin-coated endocytic vesicle membrane, early endosome membrane, nuclear membrane, ruffle membrane, macromolecular complex, receptor complex, membrane raft, synapse, perinuclear region of cytoplasm, Shc-EGFR complex, multivesicular body, internal vesicle lumen, intracellular vesicle,

GOTERM_MF_DIRECT virus receptor activity, chromatin binding, double-stranded DNA binding, protein kinase activity, MAP kinase kinase kinase activity,

protein serine/threonine/tyrosine kinase activity, protein tyrosine kinase activity, transmembrane receptor protein tyrosine kinase activity, transmembrane receptor protein tyrosine kinase activity, transmembrane signaling receptor activity, epidermal growth factor-activated receptor activity, integrin binding, protein binding, calmodulin binding, ATP binding, kinase activity, enzyme binding, kinase binding, protein kinase binding, protein phosphatase binding, nitric-oxide synthase regulator activity, ubiquitin protein ligase binding, identical protein binding, cadherin binding, epidermal growth factor binding, actin filament binding, ATPase binding,

EGF receptor, L domain, Protein kinase, catalytic domain, Serine-threonine/tyrosine-protein kinase catalytic domain, Furin-like cysteine-rich domain, Furin-like repeat, Tyrosine-protein kinase, active site, Insulin-like growth factor binding protein, N-terminal, Protein kinase-like domain, Tyrosine protein kinase, EGF/ERB/XmrK receptor, Protein kinase, ATP binding site, Tyrosine-protein kinase, INTERPRO

KEGG PATHWAY

EGFR tyrosine kinase inhibitor resistance, Endocrine resistance, MAPK signaling pathway, ErbB signaling pathway, Ras signaling pathway, Rap1 signaling pathway, Calcium signaling pathway, HIF-1 signaling pathway, FoxO signaling pathway, Phospholipase D signaling pathway, Endocytosis, PI3K-Akt signaling pathway, Foxal adhesion, Adherens junction, Gap junction, JAK-STAT signaling pathway, Regulation of actin cytoskeleton, GnRH signaling pathway, Estrogen signaling pathway, Oxytocin signaling pathway, Relaxin signaling pathway, Parathyroid hormone synthesis, secretion and action, Cushing syndrome, Epithelial cell signaling in Helicobacter pylori infection, Shigellosis, Hepatitis C, Human cytomegalovirus infection, Human papillomavirus infection, <u>Coronavirus disease -</u>
<u>COVID-19</u>, Pathways in cancer, Proteoglycans in cancer, <u>MicroRNAs in cancer</u>, Chemical carcinogenesis - receptor activation, <u>Chemical</u>

carcinogenesis - reactive oxygen species, Colorectal cancer, Pancreatic cancer, Endometrial cancer, Glioma, Prostate cancer, Melanoma, Bladder cancer, Non-small cell lung cancer, Breast cancer, Hepatocellular carcinoma, Gastric cancer, Central carbon metabolism in cancer, Choline metabolism in cancer, PD-L1 expression and PD-1 checkpoint pathway in cancer,

OMIM_DISEASE Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, Nonsmall cell lung cancer, susceptibility to, Inflammatory skin and bowel disease, neonatal, 2

PIR SUPERFAMILY tyrosine-protein kinase, EGF receptor type,

TyrKc, FU,

UP_KW_BIOLOGICAL_PROCESS

UP_KW_CELLULAR_COMPONENT Endoplasmic reticulum, Golgi apparatus, Membrane, Nucleus, Secreted, Endosome, Cell membrane,

UP KW DISEASE Disease variant, Proto-oncogene

UP_KW_DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix

UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Developmental protein, Kinase, Receptor, Transferase, Tyrosine-protein kinase, Host cell receptor for virus entry, Developmental

UP KW PTM Glycoprotein, Lipoprotein, Methylation, Palmitate, Phosphoprotein, Ubl conjugation, Disulfide bond, Isopeptide bond,

UP_SEQ_FEATURE ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GlcNAc...) (complex) asparagine; atypical; partial, CARBOHYD:N-

linked (GlcNAc...) (high mannose) asparagine, CARBOHYD:N-linked (GlcNAc...) asparagine; atypical, CARBOHYD:N-linked (GlcNAc...) asparagine; atypical, CARBOHYD:N-linked (GlcNAc...) asparagine; partial, COMPBIAS:Polar residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in ubiquitin), DOMAIN:Furin-like, DOMAIN:GF_recep_IV, DOMAIN:PK_Tyr_Ser-Thr,

DOMAIN: Protein kinase, DOMAIN: Recep_L_domain, LIPID: S-palmitoyl cysteine, MUTAGEN: C->A: Decreased palmitoylation., MUTAGEN: D->A: Increased EGF binding; when associated with A-590 and A-609., MUTAGEN: D->A: Strongly reduced phosphorylation., MUTAGEN: DGPH->AGPA: Decreases intramolecular interactions and facilitates EGF binding, MUTAGEN: E->A: Reduced phosphorylation., MUTAGEN: ED->RK: Constitutively activated kinase., MUTAGEN: H->A: Increased EGF binding; when associated with A-587; A-590 and A-609., MUTAGEN: H->A: Abolishes phosphorylation., MUTAGEN: H->A: Increased EGF binding; when associated with A-587; A-590 and A-609., MUTAGEN: H->A: Abolishes phosphorylation., MUTAGEN: MutaGEN

EIF4G1

eukarvotic translation initiation factor 4 gamma 1(EIF4G1)

Related Genes

Homo sapiens

BIOCARTA
GOTERM_BP_DIRECT

Regulation of eIF4e and p70 S6 Kinase, Eukaryotic protein translation, Internal Ribosome entry pathway, mTOR Signaling Pathway,

nuclear-transcribed mRNA catabolic process, nonsense-mediated decay, behavioral fear response, cap-dependent translational initiation, translational initiation, regulation of translational initiation, negative regulation of autophagy, negative regulation of peptidyl-threonine phosphorylation, positive regulation of cell death, viral process, positive regulation of cell growth, cellular response to nutrient levels, positive regulation of cellular protein metabolic process, developmental process, positive regulation of peptidyl-serine phosphorylation, cellular macromolecule biosynthetic process, positive regulation of translation in response to endoplasmic reticulum stress, regulation of mRNA stability, positive regulation of neuron differentiation, regulation of gene silencing by miRNA, regulation of cellular response to stress, energy homeostasis, positive regulation of G1/S transition of mitotic cell cycle, negative regulation of neuron death, positive regulation of eukaryotic translation initiation factor 4F complex assembly, regulation of presynapse assembly, positive regulation of mRNA cap binding, positive regulation of miRNA mediated inhibition of translation, regulation of polysome binding,

GOTERM_CC_DIRECT GOTERM_MF_DIRECT nucleus, cytoplasm, cytosol, polysome, cytoplasmic stress granule, membrane, eukaryotic translation initiation factor 4F complex, RNA binding, mRNA binding, translation initiation factor activity, protein binding, ATP binding, translation factor activity, RNA binding, eukaryotic initiation factor 4E binding, translation initiation factor binding, identical protein binding, binding, binding,

INTERPRO W2 domain, MIF4G-like, type 3, Initiation factor eIF-4

ERPRO W2 domain, MIF4G-like, type 3, Initiation factor eIF-4 gamma, MA3, MIF4-like, type 1/2/3, Armadillo-type fold,

KEGG_PATHWAY

OMIM_DISEASE

SMART

Viral myocarditis,
Parkinson disease 18.

eIF5C, MIF4G, MA3,

UP_KW_BIOLOGICAL_PROCESS Protein biosynthesis, Translation regulation, Host-virus interaction,

UP_KW_CELLULAR_COMPONENT Cytoplasm,

UP_KW_DISEASE <u>Disease variant</u>, <u>Neurodegeneration</u>, <u>Parkinson disease</u>, <u>Parkinsonism</u>,

UP_KW_MOLECULAR_FUNCTION Initiation factor, RNA-binding, Translational shunt,

 UP_KW_PTM
 Acetylation, Methylation, Phosphoprotein,

 UP_SEQ_FEATURE
 COMPBIAS:Acidic residues, COMPBIAS:Ba

COMPBIAS:Acidic residues, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:MI, DOMAIN:MIF4G, DOMAIN:W2, MUTAGEN:DPNQ->AAAA: Loss of PABPC1 binding; when associated with 174-AAAA-178., MUTAGEN:F->A: Abolishes binding to EIF4A; when associated with A-973., MUTAGEN:FE->AK: Abolishes binding to EIF4A; when associated with A-841 and A-842., MUTAGEN:G->A,V,W,R,E: Reduced cleavage by protease 2A from human rhinovirus 2., MUTAGEN:I->A: Abolishes binding to EIF4A; when associated with A-895 and A-95., MUTAGEN:I->A: Loss of PABPC1 binding, MUTAGEN:I->A: Abolishes binding to EIF4A; when associated with 184-AAAA-187., MUTAGEN:L->A: Abolishes binding to EIF4A; when associated with A-767 and A-775., MUTAGEN:L->A: Abolishes binding to EIF4A; when associated with A-895 and A-92., MUTAGEN:L->A: Abolishes binding to EIF4A; when associated with A-895 and A-92., MUTAGEN:L->A: Abolishes binding to EIF4A; when associated with A-895 and K-851., MUTAGEN:L->A: Abolishes binding to EIF4A; when associated with A-989., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-896., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-896., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when associated with A-8976., MUTAGEN:LL->A: Abolishes binding to EIF4A; when as

FN1
GOTERM_BP_DIRECT

fibronectin 1(FN1) Related Genes Homo sapiens angiogenesis, regulation of protein phosphorylation, platelet degranulation, acute-phase response, cell-substrate junction assembly,

cell adhesion, cell-matrix adhesion, calcium-independent cell-matrix adhesion, integrin-mediated signaling pathway, nervous system development, heart development, positive regulation of cell proliferation, regulation of cell shape, response to wounding, positive regulation of gene expression, positive regulation of peptidase activity, positive regulation of phosphatidylinositol 3-kinase signaling, peptide cross-linking, cytokine-mediated signaling pathway, extracellular matrix organization, integrin activation, substrate adhesion-dependent cell spreading, endodermal cell differentiation, post-translational protein modification, cellular protein metabolic process, positive regulation of axon extension, positive regulation of fibroblast proliferation, leukocyte migration, interaction with symbiont, regulation of ERK1 and ERK2 cascade, platelet aggregation, negative regulation of transforming growth factor beta production, blood coagulation, fibrin clot formation, neural crest cell migration involved in autonomic nervous system development, positive regulation of substrate-dependent cell migration, cell attachment to substrate,

GOTERM_CC_DIRECT

extracellular region, fibrinogen complex, <u>basement membrane</u>, <u>extracellular space</u>, <u>endoplasmic reticulum lumen</u>, <u>endoplasmic reticulum-Golgi intermediate compartment</u>, <u>plasma membrane</u>, <u>apical plasma membrane</u>, <u>extracellular matrix</u>, <u>platelet alpha granule lumen</u>, <u>extracellular exosome</u>, blood microparticle,

GOTERM_MF_DIRECT

protease binding, receptor binding, integrin binding, extracellular matrix structural constituent, protein binding, collagen binding, protein C-terminus binding, heparin binding, peptidase activator activity, enzyme binding, identical protein binding, proteoglycan binding, chaperone binding, disordered domain specific binding,

INTERPRO

OMIM_DISEASE

Fibronectin, type I, Fibronectin, type II, collagen-binding, Fibronectin, type III, Immunoglobulin-like fold, Kringle-like fold,

KEGG_PATHWAY

PI3K-Akt signaling pathway, Focal adhesion, ECM-receptor interaction, Regulation of actin cytoskeleton, AGE-RAGE signaling pathway in diabetic complications, Bacterial invasion of epithelial cells, Yersinia infection, Amoebiasis, Human papillomavirus infection, Pathways

in cancer, Proteoglycans in cancer, Small cell lung cancer,
Spondylometaphyseal dysplasia, corner fracture type, Glomerulopathy with fibronectin deposits 2,

SMART FN1, FN2, FN3,

UP_KW_BIOLOGICAL_PROCESS Acute phase, Angiogenesis, Cell adhesion, Cell shape,

UP_KW_CELLULAR_COMPONENT Extracellular matrix, Secreted,

UP KW DISEASE Disease variant, Dwarfism,

UP_KW_DOMAIN Repeat, Signal, UP_KW_MOLECULAR_FUNCTION Heparin-binding,

UP KW PTM UP_SEQ_FEATURE Glycoprotein, Oxidation, Phosphoprotein, Sulfation, Pyrrolidone carboxylic acid, Disulfide bond, Isopeptide bond

CARBOHYD:N-linked (GlcNAc...) (complex) asparagine, CARBOHYD:N-linked (GlcNAc...) asparagine, CARBOHYD:O-linked (GalNAc...) threonine, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, CROSSLNK:Isoglutamyl lysine isopeptide (Gln-Lys) (interchain with K-?), DISULFID:Interchain (with C-2458), DISULFID:Interchain (with C-2462), DOMAIN:Fibronectin type-I, DOMAIN:Fibronectin type-I 1, DOMAIN:Fibronectin type-I 10, DOMAIN:Fibronectin type-I 11, DOMAIN:Fibronectin type-I 12, DOMAIN:Fibronectin type-I 2, DOMAIN:Fibronectin type-I 3, DOMAIN:Fibronectin type-I 4, DOMAIN:Fibronectin type-I 5, DOMAIN:Fibronectin type-I 6, DOMAIN:Fibronectin type-I 7, DOMAIN:Fibronectin type-I 8, DOMAIN:Fibronectin type-I 9, DOMAIN:Fibronectin type-II, DOMAIN:Fibronectin type-II 1, DOMAIN:Fibronectin type-II 2, DOMAIN:Fibronectin type-III, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 11, DOMAIN:Fibronectin type-III 12, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 12, DOMAIN:Fibronectin type-III 11, DOMAIN:Fibronectin type-III 12, DOMAIN:Fibronectin type-III 13; extra domain A, DOMAIN:Fibronectin type-III 14, DOMAIN:Fibronectin type-III 15, DOMAIN:Fibronectin type-III 16, DOMAIN:Fibronectin type-III 2, DOMAIN:Fibronectin type-III 3, DOMAIN:Fibronectin type-III 4, DOMAIN:Fibronectin type-III 5, DOMAIN:Fibronectin type-III 6, DOMAIN:Fibronectin type-III 7, DOMAIN:Fibronectin type-III 8; extra domain B, DOMAIN:Fibronectin type-III 9, MOTIF:Cell attachment site, MUTAGEN:DOMAIN:Fibronectin type-III 9, MOTIF:DOMAIN:Fibronectin type-III 17, DOMAIN:Fibronectin type-III 17, DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 17, DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 17, DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 17, DOMAIN:Fibronectin type-III 18, MOTIF:DOMAIN:Fibronectin type-III 18, MOT enhanced ability to form fibronectin aggregates; when associated with A-691 and A-694., MUTAGEN:E->A: Slightly enhanced ability to form fibronectin aggregates; when associated with A-694 and A-696., MUTAGEN:F->A: Loss of ability to form fibronectin aggregates; when associated with A-695., MUTAGEN:I->A: Little effect on ability to form fibronectin aggregates; when associated with A-642; A-682 and A-692., MUTAGEN:I->A: Little effect on ability to form fibronectin aggregates; when associated with A-642; A-684 and A-692., MUTAGEN:I->A: Little effect on ability to form fibronectin aggregates; when associated with A-642; A-684 and A-692., MUTAGEN:I->A: Little effect on secondary structure nor on fibronectin binding nor on activation of p38 K but abolity: When associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-642 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683. MUTAGEN:I->A: Soverely compromised ability to form fibronectin aggregates; when associated with A-642 and A-683. MUTAGEN:L->A: Severely compromised ability to form fibronectin aggregates; when associated with A-641 and A-683., MUTAGEN:R->A: Slightly enhanced ability to form fibronectin aggregates; when associated with A-691 and A-696., MUTAGEN:S->A: Severely compromised ability to form fibronectin aggregates; when associated with A-641 and A-681., MUTAGEN:V->A: Little effect on ability to form fibronectin aggregates; when associated with A-642; A-682 and A-684., MUTAGEN:Y->A: No effect on secondary structure nor on fibronectin binding nor on activation of p38 kinase but abolishes polymerization activity; when associated with A-663., MUTAGEN:Y->A: Severely compromised ability to form fibronectin aggregates; when associated with A-681 and A-683., REGION:Binds to FBLN1, REGION:Cell-attachment, REGION:Collagen-binding, REGION:Critical for collagen binding, REGION:Disordered, REGION:Fibrin- and heparin-binding 1, REGION:Fibrin-binding 2, REGION:Heparin-binding 2, REGION:V region (type III connecting segment, IIICS),

SITE:Important for superfibronectin formation, KEAP1 kelch like ECH associated protein 1(KEAP1) Related Genes Homo sapiens

BIOCARTA

Oxidative Stress Induced Gene Expression Via Nrf2,

GOTERM BP DIRECT

in utero embryonic development, regulation of transcription, DNA-templated, ubiquitin-dependent protein catabolic process, regulation of autophagy, viral process, protein ubiquitination, protein deubiquitination, positive regulation of proteasomal ubiquitin-dependent protein catabolic process, cellular response to oxidative stress, cytoplasmic sequestering of transcription factor, negative regulation of sequence-specific DNA binding transcription factor activity, post-translational protein modification, regulation of epidermal cell differentiation, cellular response to interleukin-4,

GOTERM_CC_DIRECT

nucleoplasm, cytoplasm, endoplasmic reticulum, cytosol, actin filament, inclusion body, midbody, Cul3-RING ubiquitin ligase complex, centriolar satellite,

GOTERM_MF_DIRECT

protein binding, transcription factor binding, identical protein binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, disordered domain specific binding,

INTERPRO KEGG_PATHWAY BTB/POZ-like, Kelch repeat type 1, BTB/POZ fold, BTB/Kelch-associated, Kelch-type beta propeller, Kelch-like protein, gigaxonin,

Ubiquitin mediated proteolysis, Parkinson disease, Pathways in cancer, Chemical carcinogenesis - reactive oxygen species,

Hepatocellular carcinoma, Fluid shear stress and atherosclerosis kelch-like protein, gigaxonin type,

PIR_SUPERFAMILY SMART

BTB, Kelch, SM00875.

UP_KW_BIOLOGICAL_PROCESS

Ubl conjugation pathway, Host-virus interaction,

UP_KW_CELLULAR_COMPONENT Nucleus, Cytoplasm, UP_KW_DOMAIN

UP KW PTN

Repeat, Kelch repeat,

UP_SEQ_FEATURE

S-nitrosylation, Ubl conjugation, COMPBIAS: Basic and acidic residues, COMPBIAS: Pro residues, CROSSLNK: N5-[4-(S-L-cysteinyl)-5-methyl-1H-imidazol-2-yl]-L-

ornithine (Arg-Cys) (interchain with C-151 in KEAP1), CROSSLNK:N5-[4-(S-L-cysteinyl)-5-methyl-1H-imidazol-2-yl]-L-ornithine (Cys-Arg) (interchain with R-135 in KEAP1), DOMAIN:BACK, DOMAIN:BTB, MUTAGEN:C->S,N,D,L: Substitution with a small side chain that prevents covalent modification by an electrophile; promotes constitutive ubiquitination of NFE2L2/NRF2 and subsequent repression of phase 2 detoxifying enzymes. Resistance of ubiquitination of PGAM5 to inhibition by oxidative stress and sulforaphane. Impaired interaction with CUL3. Reduced formation of a high-molecular mass KEAP1 molecule when methylglyoxal accumulates., MUTAGEN:C->S: Abolishes repression of NFE2L2/NRF2-dependent gene expression. Slows down degradation of NFE2L2/NRF2., MUTAGEN:C->W,Y: Substitution with a bulky side chain that mimicks covalent modification by an electrophile; prevents ubiquitination and degradation of NFE2L2/NRF2, leading to constitutive activation of NFE2L2/NRF2 and subsequent expression of phase 2 detoxifying enzymes., MUTAGEN:F->A: Abolishes repression of NFE2L2/NRF2-dependent gene expression., MUTAGEN:H->A: Loss of interaction with NFE2L2/NRF2. Abolishes repression of NFE2L2/NRF2-dependent gene expression. Does not affect interaction with SQSTM1/p62., MUTAGEN:IEG->AAA: Increases ubiquitination and proteolytic degradation., MUTAGEN:L->A: Loss of export from nucleus; when associated with A-308., MUTAGEN:H->A: Abolished interaction with NFE2L2/NRF2; when associated with 123-A--A-127., MUTAGEN:N->A: Loss of interaction with NFE2L2/NRF2. Strongly reduces repression of NFE2L2/NRF2-dependent gene expression. Impaired interaction with NFE2L2/NRF2. Abolishes repression of NFE2L2/NRF2. SQSTM1/p62., MUTAGEN:R->A: Loss of interaction with NFE2L2/NRF2. Abolishes repression of NFE2L2/NRF2-dependent gene expression. Loss of interaction with PGAMS. Does not affect interaction with SQSTM1/p62., MUTAGEN:R->A: Reduced formation of a high-molecular mass KEAP1 molecule when methylglyoxal accumulates., MUTAGEN:VSIEG->ASAEA: Abolished interaction with NFE2L2/NRF2; when associated with 161-A-A-162., MUTAGEN:Y->A: Loss of interaction with NFE2L2/NRF2. Strongly reduces repression of NFE2L2/NRF2-dependent gene expression. Abolishes interaction with SQSTM1/p62., MUTAGEN:Y->A: Loss of interaction with NFE2L2/NRF2. Strongly reduces repression of NFE2L2/NRF2-dependent gene expression. Abolishes interaction with SQSTM1/p62., MUTAGEN:Y->A: Loss of interaction with NFE2L2/NRF2-dependent gene expression. repression of NFE2L2/NRF2-dependent gene expression. Abolishes interaction with SQSTM1/po2., MOTAGEN:Y->A: Loss of interaction with NFE2L2/NRF2. Strongly reduces repression of NFE2L2/NRF2-dependent gene expression. Loss of interaction with PGAM5., MUTAGEN:Y->A: Loss of interaction with NFE2L2/NRF2. Strongly reduces repression of NFE2L2/NRF2-dependent gene expression. Loss of interaction with PGAM5. Abolishes interaction with SQSTM1/p62., MUTAGEN:YQI->AAA: Increases ubiquitination and proteolytic degradation., REGION:Disordered, REPEAT:Kelch 1, REPEAT:Kelch 2, REPEAT:Kelch 3, REPEAT:Kelch 4, REPEAT:Kelch 5, REPEAT:Kelch 6, SITE:Sensor for electrophilic agents,

KDR

kinase insert domain receptor(KDR)

Related Genes

BIOCARTA GOTERM BP DIRECT Actions of Nitric Oxide in the Heart, VEGF, Hypoxia, and Angiogenesis,

angiogenesis, ovarian follicle development, branching involved in blood vessel morphogenesis, vasculogenesis, positive regulation of protein phosphorylation, positive regulation of endothelial cell proliferation, lymph vessel development, cell migration involved in sprouting angiogenesis, positive regulation of mesenchymal cell proliferation, hematopoietic progenitor cell differentiation, endocardium development, endothelium development, transmembrane receptor protein tyrosine kinase signaling pathway, multicellular organism development, positive regulation of cell proliferation, regulation of cell shape, positive regulation of endothelial cell migration, negative regulation of gene expression, positive regulation of phosphatidylinositol 3-kinase signaling, viral process, positive regulation of macroautophagy, cell migration, peptidyl-tyrosine phosphorylation, extracellular matrix organization, positive regulation of cell migration, positive regulation of BMP signaling pathway, positive regulation of kinase activity, embryonic hemopoiesis, calcium-mediated signaling using intracellular calcium source, cellular response to vascular endothelial growth factor stimulus, vascular endothelial growth factor receptor-2 signaling pathway, positive regulation of endothelial cell chemotaxis by VEGF- activated vascular endothelial growth factor receptor signaling pathway, peptidyl-tyrosine autophosphorylation, vascular endothelial growth factor signaling pathway, negative regulation of apoptotic process, surfactant homeostasis, positive regulation of MAPK cascade, protein kinase B signaling, positive regulation of blood vessel endothelial cell migration, cell fate commitment, endothelial cell differentiation, positive regulation of angiogenesis, protein autophosphorylation, vascular endothelial growth factor receptor signaling pathway, lung alveolus development, cell maturation, post-embryonic camera-type eye morphogenesis, positive regulation of positive chemotaxis, positive regulation of nitric-oxide synthase biosynthetic process, positive regulation of focal adhesion assembly, positive regulation of mitochondrial depolarization, calcium ion homeostasis, vascular wound healing, ERK1 and ERK2 cascade, positive regulation of ERK1 and ERK2 cascade, positive regulation of cell migration involved in sprouting angiogenesis, positive regulation of mitochondrial fission, regulation of hematopoietic progenitor cell differentiation, regulation of bone development, cellular response to hydrogen sulfide, negative regulation of endothelial cell apoptotic process, positive regulation of vasculogenesis,

GOTERM_CC_DIRECT

extracellular region, nucleus, endosome, early endosome, endoplasmic reticulum, Golgi apparatus, plasma membrane, integral component of plasma membrane, external side of plasma membrane, cell junction, receptor complex, membrane raft, sorting

endosome,

GOTERM_MF_DIRECT

protein serine/threonine/tyrosine kinase activity, protein tyrosine kinase activity, transmembrane receptor protein tyrosine kinase activity, vascular endothelial growth factor-activated receptor activity, integrin binding, protein binding, ATP binding, transferase activity, growth factor binding, vascular endothelial growth factor binding, dentical protein binding, cadherin binding, Hsp90 protein binding,

INTERPRO

Protein kinase, catalytic domain, Serine-threonine/tyrosine-protein kinase catalytic domain, Tyrosine-protein kinase, receptor class III, conserved site, Immunoglobulin subtype 2, Immunoglobulin subtype, Immunoglobulin-like domain, Tyrosine-protein kinase, active site, Tyrosine-protein kinase, vascular endothelial growth factor receptor 2 (VEGFR2), Protein kinase-like domain, Immunoglobulin Iset, Immunoglobulin, Immunoglobulin-like fold, Protein kinase, ATP binding site, Tyrosine-protein kinase, catalytic domain,

KEGG_PATHWAY

EGFR tyrosine kinase inhibitor resistance, MAPK signaling pathway, Ras signaling pathway, Rap1 signaling pathway, Calcium signaling pathway, PI3K-Akt signaling pathway, VEGF signaling pathway, Focal adhesion, Proteoglycans in cancer, Fluid shear stress and

TvrKc, IGc2, IG

OMIM_DISEASE

Hemangioma, capillary infantile, somatic, Hemangioma, capillary infantile, susceptibility to,

SMART

Angiogenesis, Differentiation, Host-virus interaction,

ATP-binding, Nucleotide-binding,

UP_KW_BIOLOGICAL_PROCESS

UP_KW_CELLULAR_COMPONENT Endoplasmic reticulum, Membrane, Nucleus, Cytoplasm, Secreted, Cell junction, Endosome, Cytoplasmic vesicle, Cell membrane,

UP KW DOMAIN

Immunoglobulin domain, Repeat, Signal, Transmembrane, Transmembrane helix,

UP KW LIGAND UP_KW_MOLECULAR_FUNCTION

<u>Developmental protein</u>, <u>Kinase</u>, <u>Receptor</u>, <u>Transferase</u>, <u>Tyrosine-protein kinase</u>, <u>Developmental protein</u>, Glycoprotein, Phosphoprotein, Ubl conjugation, Disulfide bond

UP_KW_PTM UP_SEQ_FEATURE

ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GIcNAc...) asparagine, COMPBIAS:Polar residues, DISULFID:Redox-

ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GlcNAc...) asparagine, COMPBIAS:Polar residues, DISULFID:Redox-active, DOMAIN:Ig-like C2-type 1, DOMAIN:Ig-like C2-type 2, DOMAIN:Ig-like C2-type 3, DOMAIN:Ig-like C2-type 4, DOMAIN:Ig-like C2-type 5, DOMAIN:Ig-like C2-type 6, DOMAIN:Ig-like C2-type 7, DOMAIN:PK_Tyr_Ser-Thr, DOMAIN:Protein kinase, MUTAGEN:C->A: Significantly higher kinase activity., MUTAGEN:D->A: Strongly reduced autophosphorylation and activation of MAP kinases., MUTAGEN:K->M: Loss of enzyme activity., MUTAGEN:R->A: Strongly reduced autophosphorylation and activation of MAP kinases., MUTAGEN:Y->F: Abolishes phosphorylation of PLCG1 and MAP kinases in response to VEGFA., MUTAGEN:Y->F: Abolishes stimulation of nitric oxide synthesis., MUTAGEN:Y->F: Loss of phosphorylation site. Abolishes reorganization of the actin cytoskeleton in response to VEGFA., MUTAGEN:Y->F: Strongly reduced autophosphorylation. Abolishes phosphorylation of downstream signaling proteins; when associated with F-1054, MUTAGEN:Y->F: Strongly reduced autophosphorylation. Abolishes phosphorylation of downstream signaling proteins; when associated with F-1059., MUTAGEN:Y->F: Strongly reduced autophosphorylation. Abolishes phosphorylation. Reduces phosphorylation of PLCG1., NP_BIND:ATP, REGION:Disordered, SITE:Interaction with SHB, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

KMT2A GOTERM_BP_DIRECT

chromatin organization, apoptotic process, positive regulation of transporter activity, circadian regulation of gene expression, embryonic hemopoiesis, histone H4-K16 acetylation, histone H3-K4 dimethylation, regulation of megakaryocyte differentiation positive regulation of transcription, DNA-templated, positive regulation of transcription from RNA polymerase II promoter, histone H3-K4 methylation, positive regulation of histone H3-K4 methylation, macromolecular complex assembly, regulation of histone H3-K14 acetylation, histone H3-K4 trimethylation, histone H3-K4 monomethylation, regulation of hematopoietic stem cell differentiation, negative regulation of DNA methylation, regulation of histone H3-K9 acetylation,

GOTERM_CC_DIRECT

nucleus, nucleoplasm, cytosol, histone methyltransferase complex, MLL1 complex,

lysine methyltransferase 2A(KMT2A)

GOTERM_MF_DIRECT

DNA binding, AT DNA binding, protein binding, zinc ion binding, histone methyltransferase activity (H3-K4 specific), identical protein

binding, protein homodimerization activity, unmethylated CpG binding, lysine-acetylated histone binding,

INTERPRO

SMART

SET domain, Bromodomain, Zinc finger, PHD-type, Zinc finger, CXXC-type, Post-SET domain, FY-rich, N-terminal, FY-rich, C-terminal, Zinc finger, FYVE/PHD-type, Zinc finger, RING/FYVE/PHD-type, Methyltransferase, trithorax, Zinc finger, PHD-finger,

KEGG PATHWAY

Lysine degradation, Metabolic pathways, Cushing syndrome, Transcriptional misregulation in cancer,

OMIM_DISEASE PIR_SUPERFAMILY

protein methyltransferase, trithorax protein type, PHD, BROMO, SET, PostSET, FYRN, FYRC,

Metal-binding, Zinc, S-adenosyl-L-methionine,

Wiedemann-Steiner syndrome

UP_KW_BIOLOGICAL_PROCESS

Apoptosis, Biological rhythms, Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus,

Proto-oncogene,

UP KW DISEASE UP_KW_DOMAIN Bromodomain, Coiled coil, Repeat, Zinc-finger,

UP_KW_MOLECULAR_FUNCTION

Chromatin regulator, DNA-binding, Methyltransferase, Transferase,

UP KW PTM

UP_SEQ_FEATURE

UP KW LIGAND

Acetylation, Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

BINDING:S-adenosyl-L-methionine, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), DNA_BIND:A.T hook 1, DNA_BIND:A.T hook 2, DNA_BIND:A.T hook 3, DOMAIN:Bromo; divergent, DOMAIN:CXXC-type, DOMAIN:FYR C-terminal, DOMAIN:FYR N-terminal, DOMAIN:Post-SET, DOMAIN:SET, METAL:Zinc, METAL:Zinc 1, METAL:Zinc 2, MOTIF:9aaTAD, MOTIF:Integrase domain-binding motif 1 (IBM1), MOTIF:Integrase domain-binding motif 2 (IBM2), MOTIF:Menin-binding motif (MBM), MOTIF:WDR5 interaction motif (WIN), MUTAGEN:A->R: Reduced interaction with MEN1., MUTAGEN:C->A,S: Abolished auto-methylation., MUTAGEN:C->A: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:C->A: Impairs zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:C->A: No effect on stability or DNA-binding., MUTAGEN:C->D: Abolishes DNA-binding and increases CpG methylation of the HOXA9 promoter region. Does not lead to the development of leukemia when overexpressed in mice as gene fusion with MLLT3., MUTAGEN:D->A: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:D->A: Abolishes zinc-binding and stability of the enzyme alone or in the control of the con leukemia when overexpressed in mice as gene fusion with MLLT3, MUTAGEN:D->A: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:D->A: Does not affect methyltransferase activity of the enzyme alone or in complex; when associated with A-3872., MUTAGEN:D->A: Impairs DNA-binding., MUTAGEN:DG->AA: Reduces cleavage without abolishing it. Abolishes cleavage by TASP1; when associated with 2718-A--A-2720., MUTAGEN:DGV->AAA: Abolishes cleavage by TASP1; when associated with 2666-A-A-2667., MUTAGEN:E->A: Does not affect methyltransferase activity of the enzyme alone or in complex; when associated with A-3869., MUTAGEN:E->Q: Loss of interaction with PSIP1; when associated with Q-144 and A-148., MUTAGEN:F->A: Loss of interaction with PSIP1; when associated with Q-146 and A-148., MUTAGEN:F->A: Reduced interaction with PSIP1. Loss of interaction with PSIP1; when associated with A-149 or Q-144 and Q-146., MUTAGEN:F->A: Reduced interaction with PSIP1; when associated with A-129 or A-132., MUTAGEN:F->A: Weakly affects interaction with PSIP1 whereas

significantly decreases interaction of KMT2A-MEN1 complex with PSIP1. Reduced interaction with PSIP1; when associated with A-133., MUTAGEN:F->H,Y: Reduced interaction with MEN1., MUTAGEN:H->A; Slight decrease in interaction with WDR5., MUTAGEN:H->Y: Increased interaction with WDR5., MUTAGEN:K->A: Affects methyltransferase activity of the enzyme alone, while it does not affect methyltransferase activity in complex; when associated with A-3874., MUTAGEN:K->A: Impairs DNA-binding., MUTAGEN:KFGG->AAAA: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:L->A: Loss of >AAAA: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:L->A: Loss of interaction with PSIP1; when associated with A-148., MUTAGEN:N->A: Mildly decreases DNA-binding., MUTAGEN:M->A: No effect on DNA-binding., MUTAGEN:N->A: Impairs DNA-binding., MUTAGEN:N->A: Loss of the histone H3 methyltransferase activity. Abolishes interaction with S-adenosyl-L-methionine., MUTAGEN:N->A: No effect on stability or DNA-binding., MUTAGEN:N->T: Leads to stable interaction with ASH2L and RBBP5 in the absence of WDR5; when associated with V-3867., MUTAGEN:N->T: Leads to stable interaction with ASH2L and RBBP5 in the absence of WDR5; when associated with V-3867., MUTAGEN:P->A: Reduced interaction with MEN1., MUTAGEN:Q->A: Decreases affinity for histone H3K4me3., MUTAGEN:Q->A: Impairs DNA-binding., MUTAGEN:Q->A: No effect on stability or DNA-binding., MUTAGEN:Q->A: Slightly affects methyltransferase activity of the enzyme alone, while it impairs methyltransferase activity in complex; when associated with A-3871., MUTAGEN:Q->V: Leads to stable interaction with ASH2L and RBBP5 in the absence of WDR5; when associated with I-3861., MUTAGEN:Q->V: Leads to stable interaction with ASH2L and RBBP5 in the absence of WDR5; when associated with I-3861., MUTAGEN:Q->V: Leads to stable interaction with ASH2L and RBBP5 in the absence of WDR5; when associated with I-3861., MUTAGEN:R->A: Abolishes zinc-binding and stability of the CXXC-type zinc finger and causes loss of DNA-binding., MUTAGEN:R->A: Disrupts interaction with MSH2L and RBBP5 in the absence of WDR5; when associated with I-3861., MUTAGEN:R->A: Loss of interaction with the WRAD complex and WDR5., MUTAGEN:R->A: No effect on stability or DNA-binding., MUTAGEN:R->A: Reduced interaction with MEN1., MUTAGEN:R->E: Reduced interaction with MEN1; when associated with E-25., MUTAGEN:R->E: Reduced interaction with MEN1; when associated with E-25. Phosphomimetic mutant. MEN1; when associated with E-25., MUTAGEN:S->A: Increased interaction with WDR5., MUTAGEN:S->D: Phosphomimetic mutant. Significant increase in interaction with PSIP1; when associated with D-136., MUTAGEN:S->D: Phosphomimetic mutant. Significant increase in interaction with PSIP1; when associated with D-142., MUTAGEN:V->A: Decreases binding affinity for PPIE., MUTAGEN:V->A: Reduced interaction with PSIP1; when associated with A-133., MUTAGEN:W->A: Abolishes interaction with histone H3K4me3., MUTAGEN:W->A: Reduced interaction with MEN1., MUTAGEN:W->E: Decreases affinity for histone H3K4me3., MUTAGEN:Y->A,F: Impairs methyltransferase activity toward unmodified or monomethylated H3K4me., MUTAGEN:Y->A: Affects methyltransferase MUTAGEN:Y->A: MotaGEN:Y->A: Arects metryltansferase activity toward unmodified or monomethylated H3K4me3., MUTAGEN:Y->A: Impairs methyltransferase activity toward unmodified or monomethylated H3K4me., MUTAGEN:Y->A: May perturb protein folding and thereby decrease binding affinity for PPIE., MUTAGEN:Y->F: Shifts from a specific monomethyltransferase to a di- and trimethyltransferase activity, MUTAGEN:Y->F: Slightly affects methyltransferase activity toward unmodified or monomethylated H3K4me., REGION:Disordered, REGION:Disordered, REGION:Disordered, REGION:Disordered, REGION:Disordered, REGION:Disordered of the protein of the pro methyltransferase activity toward unmodified or monomethylated H3K4mes, ReGION:Disordered, ReGION:Interaction with histone H3K4me3, REGION:S-adenosyl-L-methionine binding, SITE:Breakpoint for translocation to form KMT2A-AF3P21 and KMT2A-KNL1 oncogenes, SITE:Breakpoint for translocation to form KMT2A-FF4 fusion protein, SITE:Breakpoint for translocation to form KMT2A-CENPK oncogene, SITE:Breakpoint for translocation to form KMT2A-FRYL fusion protein, SITE:Breakpoint for translocation to form KMT2A-GAS7 oncogene, SITE:Breakpoint for translocation to form KMT2A-LPP, SITE:Breakpoint for translocation to form KMT2A-ZFYVE19 oncogene, SITE:Cleavage; by TASP1, site 1, SITE:Cleavage; by TASP1, site 2, SITE:Important for WDR5-recognition and binding, ZN_FING:C2HC pre-PHD-type, ZN_FING:CXXC-type, ZN_FING:PHD-type 1, ZN_FING:PHD-type 2, ZN_FING:PHD-type 3, ZN_FING:PHD-type 4,

KMT2C Ivsine methyltransferase 2C(KMT2C)

COG_ONTOLOGY General function prediction only,

GOTERM_BP_DIRECT chromatin organization, regulation of transcription, DNA-templated, regulation of megakaryocyte differentiation, positive regulation of transcription from RNA polymerase II promoter, histone H3-K4 monomethylation,

GOTERM_CC_DIRECT nucleus, nucleoplasm, histone methyltransferase complex, MLL3/4 complex,

GOTERM_MF_DIRECT DNA binding, transcription coactivator activity, RNA binding, protein binding, transferase activity, transferring acyl groups, histone

methyltransferase activity, histone binding, histone methyltransferase activity (H3-K4 specific), metal ion binding,

HMG-I/HMG-Y, DNA-binding, conserved site, <u>SET domain</u>, Zinc finger, RING-type, Zinc finger, PHD-type, <u>Post-SET domain</u>, FY-rich, <u>N-terminal</u>, FY-rich, C-terminal, High mobility group (HMG) box domain, Zinc finger, FYVE/PHD-type, Zinc finger, RING/FYVE/PHD-type, INTERPRO

Zinc finger, PHD-finger,

KEGG_PATHWAY Lysine degradation, Metabolic pathways,

OMIM_DISEASE Kleefstra syndrome 2,

RING, PHD, SET, HMG, PostSET, FYRN, FYRC,

UP KW BIOLOGICAL PROCESS Transcription, Transcription regulation,

UP_KW_CELLULAR_COMPONENT Nucleus

UP_KW_DISEASE Disease variant, Mental retardation, UP KW DOMAIN Coiled coil, Repeat, Zinc-finger,

UP_KW_LIGAND Metal-binding, Zinc, S-adenosyl-L-methionine,

UP_KW_MOLECULAR_FUNCTION Activator, Acyltransferase, Chromatin regulator, DNA-binding, Methyltransferase, Transferase,

UP KW PTM Acetylation, Lipoprotein, Methylation, Palmitate, Phosphoprotein,

BINDING:S-adenosyl-L-methionine, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DNA_BIND:A.T hook, DOMAIN:DHHC, DOMAIN:PYR C-terminal, DOMAIN:FYR N-terminal, DOMAIN:PHD-type, DOMAIN:POst-SET, UP_SEQ_FEATURE

DOMAIN:FIX Hook, DOMAIN:BIT, C-terminal, DOMAIN:FIX N-terminal, DOMAIN:FIX N-terminal, DOMAIN:FIX D H3K4me1 into H3K4me2., REGION:Disordered, REGION:S-adenosyl-L-methionine binding, ZN_FING:C2HC pre-PHD-type 1; degenerate, ZN_FING:C2HC pre-PHD-type 2, ZN_FING:PHD-type 1, ZN_FING:PHD-type 2, ZN_FING:PHD-type 3, ZN_FING:PHD-type 4, ZN_FING:PHD-ty

4, ZN_FING:PHD-type 5, ZN_FING:PHD-type 6, ZN_FING:PHD-type 7, ZN_FING:PHD-type 8, ZN_FING:RING-type,

MMP16 matrix metallopeptidase 16(MMP16)

GOTERM_BP_DIRECT

skeletal system development, endochondral ossification, proteolysis, protein processing, extracellular matrix disassembly, extracellular matrix organization, collagen catabolic process, chondrocyte proliferation, embryonic cranial skeleton morphogenesis, regulation of

catalytic activity, craniofacial suture morphogenesis

GOTERM_CC_DIRECT extracellular region, Golgi lumen, plasma membrane, integral component of plasma membrane, cell surface, extracellular matrix,

GOTERM MF DIRECT metalloendopeptidase activity, enzyme activator activity, zinc ion binding, metalloaminopeptidase activity,

INTERPRO Hemopexin-like domain, Peptidase M10, metallopeptidase, Peptidoglycan binding-like, Peptidase, metallopeptidase, Hemopexin,

conserved site, Hemopexin-like repeats, Peptidase M10A, cysteine switch, zinc binding site, Peptidase M10A, Peptidase M10A, matrix metallopeptidase, C-terminal, Metallopeptidase, catalytic domain,

KEGG_PATHWAY Parathyroid hormone synthesis, secretion and action, MicroRNAs in cancer,

PIR_SUPERFAMILY matrix metalloproteinase, stromelysin type,

SMART HX, ZnMc

UP KW BIOLOGICAL PROCESS Collagen degradation,

UP_KW_CELLULAR_COMPONENT Extracellular matrix, Membrane, Secreted, Cell membrane, UP_KW_DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_LIGAND Calcium, Metal-binding, Zinc, UP_KW_MOLECULAR_FUNCTION Hydrolase, Metalloprotease, Protease,

UP KW PTM Cleavage on pair of basic residues, Glycoprotein, Zymogen, Disulfide bond,

CARBOHYD:N-linked (GlcNac...) asparagine, COMPBIAS:Pro residues, DOMAIN:PG_binding_1, METAL:Calcium 1, METAL:Calcium 1; via carbonyl oxygen, METAL:Calcium 2, METAL:Calcium 2; via carbonyl oxygen, METAL:Zinc 1, METAL:Zinc 1; in inhibited form, METAL:Zinc 2; catalytic, MOTIF:Cysteine switch, REGION:Disordered, REPEAT:Hemopexin 1, REPEAT:Hemopexin 2, REPEAT:Hemopexin 2, REPEAT:Hemopexin 3, REPEAT:Hemopexin UP_SEQ_FEATURE

Related Genes

3, REPEAT: Hemopexin 4, TOPO_DOM: Cytoplasmic, TOPO_DOM: Extracellular, TRANSMEM: Helical,

BIOCARTA Inhibition of Matrix Metalloproteinases,

GOTERM_BP_DIRECT

matrix metallopeptidase 2(MMP2)

angiogenesis, ovarian follicle development, ovulation from ovarian follicle, luteinization, response to hypoxia, blood vessel maturation, intramembranous ossification, proteolysis, negative regulation of cell adhesion, heart development, embryo implantation, parturition, aging, response to xenobiotic stimulus, response to mechanical stimulus, peripheral nervous system axon regeneration, response to activity, cell migration, cytokine-mediated signaling pathway, extracellular matrix disassembly, extracellular matrix organization, positive regulation of cell migration, collagen catabolic process, response to retinoic acid, cellular response to reactive oxygen species, response to nicotine, endodermal cell differentiation, response to hydrogen peroxide, positive regulation of apoptotic process, response to estrogen, cellular protein metabolic process, negative regulation of vasoconstriction, ephrin receptor signaling pathway, positive regulation of smooth muscle cell proliferation, tissue remodeling, response to electrical stimulus, response to hyperoxia, face morphogenesis, bone trabecula formation, prostate gland epithelium morphogenesis, cellular response to amino acid stimulus, cellular response to interleukin-1, cellular response to estradiol stimulus, cellular response to UV-A, cellular response to fluid shear stress, response to beta-amyloid, positive regulation of vascular smooth muscle cell proliferation,

GOTERM_CC_DIRECT extracellular region, extracellular space, nucleus, mitochondrion, plasma membrane, sarcomere, extracellular matrix

fibronectin binding, endopeptidase activity, metalloendopeptidase activity, serine-type endopeptidase activity, protein binding, metallopeptidase activity, zinc ion binding, GOTERM_MF_DIRECT

INTERPRO Fibronectin, type II, collagen-binding, Hemopexin-like domain, Peptidase M10, metallopeptidase, Peptidoglycan binding-like, Peptidase,

metallopeptidase, Kringle-like fold, Hemopexin, conserved site, Hemopexin-like repeats, Peptidase M10A, cysteine switch, zinc binding site, Peptidase M10A, Metallopeptidase, catalytic domain,

Endocrine resistance, Leukocyte transendothelial migration, GnRH signaling pathway, Estrogen signaling pathway, Relaxin signaling pathway, AGE-RAGE signaling pathway in diabetic complications, Pathways in cancer, Proteoglycans in cancer, Bladder cancer, Diab KEGG_PATHWAY

cardiomyopathy, Fluid shear stress and atherosclerosis, Multicentric osteolysis, nodulosis, and arthropathy,

PIR_SUPERFAMILY matrix metalloproteinase, stromelysin type,

SMART FN2, HX, ZnMc

OMIM_DISEASE

UP KW BIOLOGICAL PROCESS Angiogenesis, Collagen degradation,

UP_KW_CELLULAR_COMPONENT Extracellular matrix, Membrane, Mitochondrion, Nucleus, Cytoplasm, Secreted,

UP KW DISEASE Disease variant UP_KW_DOMAIN Repeat, Signal,

UP KW LIGAND Calcium, Metal-binding, Zinc UP_KW_MOLECULAR_FUNCTION Hydrolase, Metalloprotease, Protease,

UP_KW_PTM Autocatalytic cleavage, Glycoprotein, Phosphoprotein, Zymogen, Disulfide bond,

UP_SEQ_FEATURE CARBOHYD:N-linked (GlcNAc...) asparagine, DOMAIN:Fibronectin type-II, DOMAIN:Fibronectin type-II 1, DOMAIN:Fibronectin type-II

2, DOMAIN:Fibronectin type-II 3, DOMAIN:Peptidase_M10, DOMAIN:ZnMc, METAL:Calcium 1, METAL:Calcium 1; via carbonyl oxygen, METAL:Calcium 2, METAL:Calcium 2; via carbonyl oxygen, METAL:Calcium 3, METAL:Calcium 3; via carbonyl oxygen, METAL:Calcium 4; via carbonyl oxygen, METAL:Zinc 1; catalytic, METAL:Zinc 1; in inhibited form, METAL:Zinc 2, MOTIF:Cysteine switch,

PROPEP:Activation peptide, REGION:Collagen-binding, REGION:Collagenase-like 1, REGION:Collagenase-like 2, REGION:Required for inhibitor TIMP2 binding, REPEAT:Hemopexin, REPEAT:Hemopexin 1, REPEAT:Hemopexin 2, REPEAT:Hemopexin 3, REPEAT:Hemopexin 3

MAP3K4 mitogen-activated protein kinase kinase kinase 4(MAP3K4) Related Genes Homo sapiens

BBID 100.MAPK signaling cascades. **BIOCARTA** MAPKinase Signaling Pathway,

MAPK cascade, activation of MAPKK activity, placenta development, protein phosphorylation, response to UV-C, regulation of gene GOTERM_BP_DIRECT

expression, male germ-line sex determination, positive regulation of telomere maintenance via telomerase, intracellular signal transduction, positive regulation of JUN kinase activity, positive regulation of telomerase activity, chorionic trophoblast cell differentiation, positive regulation of p38MAPK cascade, positive regulation of telomere capping,

GOTERM CC DIRECT cytoplasm, perinuclear region of cytoplasm,

protein kinase activity, MAP kinase kinase kinase activity, protein serine/threonine/tyrosine kinase activity, protein binding, ATP binding, metal ion binding, GOTERM_MF_DIRECT

INTERPRO Protein kinase, catalytic domain, Serine/threonine-protein kinase, active site, Protein kinase-like domain, Protein kinase, ATP binding

KEGG_PATHWAY MAPK signaling pathway, GnRH signaling pathway,

S_TKc UP_KW_CELLULAR_COMPONENT Cytoplasm, UP KW DOMAIN Coiled coil

UP KW LIGAND ATP-binding, Magnesium, Metal-binding, Nucleotide-binding, UP_KW_MOLECULAR_FUNCTION Kinase, Serine/threonine-protein kinase, Transferase

UP_KW_PTM Phosphoprotein,

UP_SEQ_FEATURE ACT_SITE:Proton acceptor, BINDING:ATP, COMPBIAS:Acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues,

DOMAIN: Protein kinase, MUTAGEN: K->R: Loss of activity., NP_BIND: ATP, REGION: Disordered,

neural cell adhesion molecule 1(NCAM1) Related Genes NCAM1 Homo sapiens

BIOCARTA Synaptic Proteins at the Synaptic Junction,

GOTERM_BP_DIRECT MAPK cascade, cell adhesion, axon guidance, viral entry into host cell, interferon-gamma-mediated signaling pathway, commissural

neuron axon guidance, regulation of semaphorin-plexin signaling pathway,

GOTERM_CC_DIRECT Golgi membrane, extracellular region, cytosol, plasma membrane, external side of plasma membrane, cell surface, membrane, integral component of membrane, anchored component of membrane, neuron projection,

GOTERM_MF_DIRECT virus receptor activity, protein binding, identical protein binding INTERPRO

Immunoglobulin subtype 2, Immunoglobulin subtype, Fibronectin, type III, Immunoglobulin-like domain, Neural cell adhesion,

Immunoglobulin I-set, Immunoglobulin-like fold,

KEGG_PATHWAY Cell adhesion molecules, Prion disease, SMART

UP KW BIOLOGICAL PROCESS

Cell adhesion, Host-virus interaction, UP_KW_CELLULAR_COMPONENT Membrane, Secreted, Cell membrane,

UP KW DOMAIN Immunoglobulin domain, Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_MOLECULAR_FUNCTION Receptor, Host cell receptor for virus entry,

UP_KW_PTM Glycoprotein, GPI-anchor, Lipoprotein, Phosphoprotein, Disulfide bond,

UP_SEQ_FEATURE

CARBOHYD:N-linked (GlcNAc...) asparagine, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, DOMAIN:Fibronectin type-III, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 2, DOMAIN:Ig-like, DOMAIN:Ig-like C2-type 1, DOMAIN:Ig-like C2-type 2, DOMAIN:Ig-like C2-type 3, DOMAIN:Ig-like C2-type 4, DOMAIN:Ig-like C2-type 5, LIPID:GPI-anchor amidated asparagine, REGION:Disordered, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

NF1 neurofibromin 1(NF1) Related Genes Homo sapiens

BIOCARTA Chromatin Remodeling by hSWI/SNF ATP-dependent Complexes,

GOTERM_BP_DIRECT MAPK cascade, osteoblast differentiation, metanephros development, response to hypoxia, liver development, negative regulation of endothelial cell proliferation, positive regulation of endothelial cell proliferation, regulation of cell-matrix adhesion, negative regulation

of cell-matrix adhesion, negative regulation of protein kinase activity, cell communication, signal transduction, Ras protein signal transduction, negative regulation of neuroblast proliferation, brain development, peripheral nervous system development, heart development, skeletal muscle tissue development, negative regulation of cell proliferation, visual learning, extrinsic apoptotic signaling pathway via death domain receptors, regulation of gene expression, Schwann cell development, phosphatidylinositol 3-kinase signaling, negative regulation of angiogenesis, spinal cord development, amygdala development, forebrain astrocyte development, neural tube development, cerebral cortex development, myelination in peripheral nervous system, actin cytoskeleton organization, extracellular matrix organization, collagen fibril organization, adrenal gland development, negative regulation of cell migration, negative regulation of vascular endothelial growth factor receptor signaling pathway, regulation of synaptic transmission, GABAergic, cellular response to heat, negative regulation of Rac protein signal transduction, reelin-mediated signaling pathway, wound healing, cellular response to heat, negative regulation of Rac protein signal transduction, reelin-mediated signaling, pathway, wound healing, negative regulation of protein import into nucleus, positive regulation of apoptotic process, regulation of GTPase activity, negative regulation of MAPK cascade, pigmentation, positive regulation of neuron apoptotic process, regulation of blood vessel endothelial cell migration, positive regulation of GTPase activity, regulation of bone resorption, negative regulation of osteoclast differentiation, regulation of glial cell differentiation, positive regulation of adenylate cyclase activity, regulation of aniogenesis, negative regulation of neurotransmitter secretion, negative regulation of fibroblast proliferation, regulation of long-term neuronal synaptic plasticity, sympathetic nervous system development, camera-type eye morphogenesis, negative regulation of astrocyte differentiation, negative regulation of oligodendrocyte differentiation, smooth muscle tissue development, hair follicle maturation, artery morphogenesis, forebrain morphogenesis, cognition, gamma-aminobutyric acid secretion, neurotransmission, observational learning, regulation of long-term synaptic potentiation, positive regulation of glutamate secretion, neurotransmission, observational learning, regulation of long-term synaptic potentiation, positive regulation of

extrinsic apoptotic signaling pathway in absence of ligand, <u>cleus, nucleolus, cytoplasm, cytosol, membrane, integral component of membrane, axon, dendrite, presynapse, </u>

GOTERM_MF_DIRECT GTPase activator activity, protein binding, phosphatidylethanolamine binding, phosphatidylcholine binding,

INTERPRO CRAL-TRIO domain, Ras GTPase-activating protein, Rho GTPase activation protein, Pleckstrin homology-like domain, Armadillo-type

fold, Ras GTPase-activating protein, conserved site,

KEGG_PATHWAY EGFR tyrosine kinase inhibitor resistance, MAPK signaling pathway, Ras signaling pathway,

OMIM DISEASE Neurofibromatosis, type 1, Neurofibromatosis, familial spinal, Watson syndrome, Neurofibromatosis-Noonan syndrome, Leukemia,

juvenile myelomonocytic

SMART RasGAP, SEC14, UP_KW_CELLULAR_COMPONENT Membrane, Nucleus,

UP_KW_DISEASE Tumor suppressor, Disease variant,

UP KW DOMAIN Coiled coil, Transmembrane, Transmembrane helix

UP KW LIGAND Lipid-binding, UP_KW_MOLECULAR_FUNCTION GTPase activation. UP KW PTM

Acetylation, Phosphoprotein,

UP_SEQ_FEATURE COMPBIAS: Polar residues, COMPBIAS: Pro residues, DOMAIN: CRAL-TRIO, DOMAIN: Ras-GAP, MOTIF: Bipartite nuclear localization

signal, MUTAGEN:K->A: Reduces phospholipid binding; when associated with A-1691; A-169 and A-1769., MUTAGEN:K->A: Reduces phospholipid binding; when associated with A-1691; A-169 and A-1771., MUTAGEN:Missing: Reduces protein stability., MUTAGEN:R->A: Reduces phospholipid binding; when associated with A-1691; A-1695 and A-1771., MUTAGEN:R->A: Reduces phospholipid binding; when associated with A-1691; A-1769 and A-1771., REGION:Disordered, REGION:Lipid binding, TRANSMEM:Helical,

NTRK2 neurotrophic receptor tyrosine kinase 2(NTRK2)

GOTERM_BP_DIRECT

OMIM DISEASE

GOTERM_CC_DIRECT

vasculogenesis, neuron migration, positive regulation of protein phosphorylation, transmembrane receptor protein tyrosine kinase signaling pathway, activation of phospholipase C activity, multicellular organism development, nervous system development, learning, circadian rhythm, feeding behavior, positive regulation of cell proliferation, positive regulation of gene expression, positive regulation of neuron projection development, glutamate secretion, positive regulation of phosphatidylinositol 3-kinase signaling, peptidyl-tyrosine phosphorylation, neuronal action potential propagation, central nervous system neuron development, cerebral cortex development myelination in peripheral nervous system, cell differentiation, neuron differentiation, brain-derived neurotrophic factor receptor signaling pathway, positive regulation of peptidyl-serine phosphorylation, positive regulation of kinase activity, neurotrophin signaling pathway, mechanoreceptor differentiation, regulation of GTPase activity, positive regulation of MAPK cascade, negative regulation of neuron apoptotic process, retinal rod cell development, protein autophosphorylation, neurotrophin TRK receptor signaling pathway, oligodendrocyte differentiation, peripheral nervous system neuron development, positive regulation of axonogenesis, regulation of protein kinase B signaling, positive regulation of synapse assembly, long-term synaptic potentiation, cellular response to amino acid stimulus, trans-synaptic signaling by neuropeptide, modulating synaptic transmission, negative regulation of beta-amyloid formation, positive regulation of non-membrane spanning protein tyrosine kinase activity, cellular response to nerve growth factor stimulus, cellular response to brain-derived neurotrophic factor stimulus, negative regulation of anoikis,

GOTERM CC DIRECT early endosome, cytosol, plasma membrane, integral component of plasma membrane, postsynaptic density, integral component of

membrane, axon, dendrite, early endosome membrane, terminal bouton, dendritic spine, receptor complex, axon perinuclear region of cytoplasm,

GOTERM_MF_DIRECT protease binding, protein serine/threonine/tyrosine kinase activity, transmembrane receptor protein tyrosine kinase activity,

neurotrophin receptor activity, protein binding, ATP binding, kinase activity, protein homodimerization activity, neurotrophin binding, brain-derived neurotrophic factor binding, brain-derived neurotrophic factor-activity,

INTERPRO Leucine-rich repeat-containing N-terminal, Cysteine-rich flanking region, C-terminal, Protein kinase, catalytic domain, Serine-

International Communication of the Internation of the International Communication of t

KEGG_PATHWAY MAPK signaling pathway, Ras signaling pathway, Calcium signaling pathway, PI3K-Akt signaling pathway, Neurotrophin signaling

Obesity, hyperphagia, and developmental delay, Developmental and epileptic encephalopathy 58,

LRRNT, LRRCT, TyrKc, IGc2, IG, UP_KW_BIOLOGICAL_PROCESS Differentiation, Neurogenesis,

UP_KW_CELLULAR_COMPONENT Membrane, Synapse, Cytoplasm, Cell junction, Cell projection, Endosome, Cell membrane,

UP KW DISEASE Disease variant, Obesity, Epilepsy,

UP KW DOMAIN Immunoglobulin domain, Leucine-rich repeat, Repeat, Signal, Transmembrane, Transmembrane helix,

UP KW LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Developmental protein, Kinase, Receptor, Transferase, Tyrosine-protein kinase, Developmental protein,

UP_KW_PTM Glycoprotein, Phosphoprotein, Ubl conjugation, Disulfide bond

UP SEQ FEATURE ACT_SITE:Proton acceptor, BINDING:ATP, CARBOHYD:N-linked (GlcNAc...) asparagine, COMPBIAS:Polar residues, DOMAIN:Iq-like,

Homo sapiens

DOMAIN:Ig-like C2-type 1, DOMAIN:Ig-like C2-type 2, DOMAIN:LRRCT, DOMAIN:LRRNT, DOMAIN:Protein kinase, NP_BIND:ATP, REGION:Disordered, REGION:Interaction with MAPK8IP3/JIP3, REPEAT:LRR 1, REPEAT:LRR 2, SITE:Interaction with PLC-gamma-1, SITE:Interaction with PLCG1, SITE:Interaction with SH2D1A, SITE:Interaction with SHC1, TOPO_DOM:Cytoplasmic, TOPO_DOM: Extracellular, TRANSMEM: Helical,

NUP98 nucleoporin 98 and 96 precursor(NUP98)

GOTERM_BP_DIRECT

regulation of glycolytic process, mRNA export from nucleus, tRNA export from nucleus, proteolysis, protein import into nucleus, nucleocytoplasmic transport, nuclear pore organization, mitotic spindle organization, mitotic nuclear envelope reassembly, protein transport, viral process, protein sumoylation, viral life cycle, viral transcription, positive regulation of transcription, DNA-templated positive regulation of mRNA splicing, via spliceosome, mRNA transport, nuclear pore complex assembly, regulation of gene silencing by miRNA, intracellular transport of virus, regulation of cellular response to heat,

GOTERM_CC_DIRECT

kinetochore, nucleus, nuclear envelope, nuclear pore, nucleoplasm, chromosome, cytosol, nuclear body, nuclear pore outer ring, nuclear membrane, nuclear periphery, nuclear inclusion body, intracellular membrane-bounded organelle, host cell, nuclear pore nuclear basket, ribonucleoprotein complex,

GOTERM_MF_DIRECT

transcription coactivator activity, mRNA binding, transporter activity, protein binding, nuclear localization sequence binding, serine-type peptidase activity, structural constituent of nuclear pore, histone-lysine N-methyltransferase activity, metal ion binding, promoter-specific chromatin binding,

INTERPRO

PWWP, SET domain, Zinc finger, PHD-type, Post-SET domain, AWS, Peptidase S59, nucleoporin, Zinc finger, FYVE/PHD-type, Zinc finger, RING/FYVE/PHD-type, Zinc finger, PHD-type, conserved site, Zinc finger, PHD-finger, Nuclear protein 96, Nucleoporin FG repeat,

KEGG PATHWAY

Nucleocytoplasmic transport, Amyotrophic lateral sclerosis, Influenza A,

SMART

UP_KW_BIOLOGICAL_PROCESS

mRNA transport, Protein transport, Translocation, Transport, Host-virus interaction,

UP KW MOLECULAR FUNCTION

UP_KW_CELLULAR_COMPONENT Chromosome, Membrane, Nucleus, Nuclear pore complex,

UP KW DOMAIN

Coiled coil, Repeat,

PHD, PWWP, SET, PostSET, AWS,

UP KW LIGAND

Metal-binding, Zinc, S-adenosyl-L-methionine, Chromatin regulator, Hydrolase, Methyltransferase, Protease, Serine protease, Transferase,

UP_KW_PTM

Acetylation, Autocatalytic cleavage, Phosphoprotein, Ubl conjugation, Isopeptide bond,

UP_SEQ_FEATURE

ACT_SITE:Nucleophile, COMPBIAS:Basic and acidic residues, COMPBIAS:Basic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2), CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO2); alternate, DOMAIN:AWS, DOMAIN:Nup96, DOMAIN:PHD-type, DOMAIN:PWWP, DOMAIN:Peptidase S59, DOMAIN:Post-SET, DOMAIN:SET, MUTAGEN:FSKY->SSKR: Loss of processing. Loss of nuclear membrane DOMAIN: Peptidase S59, DOMAIN: Post-SET, DOMAIN: SET, MUTAGEN: FSKY-SSKR: Loss of processing. Loss of nuclear membrane localization., MUTAGEN: H->A, Q: Moderate reduction in autoprocessing., MUTAGEN: K->A: No effect in autoprocessing. MUTAGEN: K->A: No effect on autoprocessing. Severe loss of autoprocessing; when associated with A-879., MUTAGEN: N->A: Slight reduction in autoprocessing, MUTAGEN: S->A: Loss of autoprocessing. Loss of nuclear membrane localization., REGION: Disordered, REGION: FG repeats 1, REGION: FG repeats 2, REGION: GLEBS; interaction with RAE1, SITE: Breakpoint for translocation to form NUP98-CCDC28A, SITE: Breakpoint for translocation to form NUP98-PHF23 oncogene, SITE: Breakpoint for translocation to form the NUP98-RAP1GDS1 fusion protein. SITE: Breakpoint for translocation to form the NUP98-RAP1GDS1 fusion protein, SITE: Breakpoint for translocation to form the NUP98-RAP1GDS1 fusion protein. SITE: SITE: Breakpoint for translocation to form the NUP98-RAP1GDS1 fusion protein. SITE: Clearly autophysics. translocation to form the NUP98-RAP1GDS1 fusion protein, SITE: Cleavage; by autolysis,

PIK3CA

phosphatidylinositol-4.5-bisphosphate 3-kinase catalytic subunit alpha(PIK3CA)

Related Genes

BIOCARTA

Role of nicotinic acetylcholine receptors in the regulation of apoptosis, AKT Signaling Pathway, Tumor Suppressor Arf Inhibits
Ribosomal Biogenesis, Regulation of BAD phosphorylation, B Cell Survival Pathway, Role of P13K subunit p85 in regulation of Actin
Organization and Cell Migration, Transcription factor CREB and its extracellular signals, CTCF, The Co-Stimulatory Signal During T-cell
Activation, CXCR4 Signaling Pathway, Erk and PI-3 Kinase Are Necessary for Collagen Binding in Corneal Epithelia, Phospholipids as signalling intermediaries, EGF Signaling Pathway, Regulation of eIF4e and p70 S6 Kinase, Role of Erk5 in Neuronal Receptor I Signaling in Mast Cells, Corticosteroids and cardioprotection, Growth Hormone Signaling Pathway, Inhibition of Cellular Proliferation by Gleevec, Inactivation of Gsk3 by AKT causes accumulation of b-catenin in Alveolar Macrophages, Human Cytomegalovirus and Map Kinase Pathways, Control of skeletal myogenesis by HDAC & calcium/calmodulin-dependent kinase (CaMK), Role of ERBB2 in Signal Transduction and Oncology, Skeletal muscle hypertrophy is regulated via AKT/mTOR pathway, IGF-1 Signaling Pathway, Multiple antiapoptotic pathways from IGF-1R signaling lead to BAD phosphorylation, IL-2 Receptor Beta Chain in T cell Activation, IL-7 Signal Transduction, Insulin Signaling lead to BAD phosphorylation, IL-2 Receptor Beta Chain in 1 Ceil Activation, IL-7 Signal Transduction, Insulin Signaling Pathway, The IGF-1 Receptor and Longevity, Signaling of Hepatocyte Growth Factor Receptor, mTOR Signaling Pathway, NFAT and Hypertrophy of the heart (Transcription in the broken heart), Nerve growth factor pathway (NGF), Ras-Independent pathway in NK cell-mediated cytotoxicity, Thrombin signaling and protease-activated receptors, PDGF Signaling Pathway, Phospholipase C Signaling Pathway, Mechanism of Gene Regulation by Peroxisome Proliferators via PPARa(alpha), PTEN dependent cell cycle arrest and apoptosis, Rac 1 cell motility signaling pathway, Influence of Ras and Rho proteins on G1 to S Transition, Ras Signaling Pathway, T Cell Receptor Signaling Pathway, Trefoil Factors Initiate Mucosal Healing, TPO Signaling Pathway, Transition, Transition, Transition, Pathway, VEGF, Hypoxia, and Angiogenesis,

GOTERM_BP_DIRECT

MAPK cascade, angiogenesis, liver development, vasculature development, glucose metabolic process, protein phosphorylation phosphatidylinositol biosynthetic process, phagocytosis, epidermal growth factor receptor signaling pathway, G-protein coupled receptor signaling pathway, axon guidance, regulation of gene expression, positive regulation of lamellipodium assembly, negative regulation of gene expression, phosphatidylinositol 3-kinase signaling, positive regulation of phosphatidylinositol 3-kinase signaling, response to activity, response to muscle inactivity, negative regulation of macroautophagy, phosphorylation, cell migration, cytokine-mediated signaling pathway, actin cytoskeleton organization, platelet activation, negative regulation of actin filament depolymerization, T cell costimulation, positive regulation of TOR signaling, activation of protein kinase activity, positive regulation of peptidyl-serine phosphorylation, response to muscle stretch, phosphatidylinositol-3-phosphate biosynthetic process, insulin receptor signaling pathway via phosphatidylinositol 3-kinase, vascular endothelial growth factor signaling pathway, Fc-epsilon receptor signaling pathway, Fc-gamma receptor signaling pathway involved in phagocytosis, ERBB2 signaling pathway, regulation of multicellular organism growth, response to leucine, anoikis, regulation of cellular respiration, protein kinase B signaling, negative regulation of neuron apoptotic process, endothelial cell migration, hypomethylation of CpG island, phosphatidylinositol phosphorylation, vascular endothelial growth factor receptor signaling pathway, phosphatidylinositol-mediated signaling, positive regulation of smooth muscle cell proliferation, T cell receptor signaling pathway, leukocyte migration, positive regulation of protein kinase B signaling, relaxation of cardiac muscle, cardiac muscle contraction, adipose tissue development, cellular response to glucose stimulus, cellular response to hydrostatic pressure, response to dexamethasone, cardiac muscle cell contraction, energy homeostasis, response to butyrate, negative regulation of fibroblast apoptotic process, regulation of genetic imprinting, negative regulation of anoikis,

GOTERM CC DIRECT

cytoplasm, cytosol, plasma membrane, phosphatidylinositol 3-kinase complex, phosphatidylinositol 3-kinase complex, class IA, phosphatidylinositol 3-kinase complex, class IB, intercalated disc, membrane, lamellipodium, perinuclear region of cytoplasm,

GOTERM_MF_DIRECT

protein serine/threonine kinase activity, protein serine/threonine/tyrosine kinase activity, protein binding, ATP binding, kinase activity, 1-phosphatidylinositol-3-kinase activity, protein kinase activator activity, phosphatidylinositol 3-kinase activity, 1-phosphatidylinositol-4-phosphate 3-kinase activity, insulin receptor substrate binding, phosphatidylinositol-4,5-bisphosphate 3-kinase activity, phosphatidylinositol-3,4-bisphosphate 5-kinase activity,

INTERPRO

Phosphatidylinositol 3-kinase Ras-binding (PI3K RBD) domain, Phosphatidylinositol 3-/4-kinase, catalytic domain, Phosphoinositide 3-kinase, accessory (PIK) domain, Phosphatidylinositol 3-kinase C2 (PI3K C2) domain, Phosphatidylinositol 3-kinase adaptor-binding (PI3K ABD) domain, Protein kinase-like domain, Phosphatidylinositol Kinase, Armadillo-type fold, Phosphatidylinositol 3/4-kinase, conserved site,

KEGG_PATHWAY

Inositol phosphate metabolism, Metabolic pathways, EGFR tyrosine kinase inhibitor resistance, Endocrine resistance, Platinum drug resistance, ErbB signaling pathway, Ras signaling pathway, Rap1 signaling pathway, cAMP signaling pathway, Chemokine signaling pathway, HIF-1 signaling pathway, FoxO signaling pathway, Phosphatidylinositol signaling system, Sphingolipid signaling pathway, Phospholipase D signaling pathway, Autophagy - animal, mTOR signaling pathway, PI3K-Akt signaling pathway, AMPK signaling pathway, Apoptosis, Longevity regulating pathway, Longevity regulating pathway - multiple species, Cellular senescence, Axon guidance, VEGF signaling pathway, Osteoclast differentiation, Focal adhesion, Signaling pathways regulating pluripotency of stem cells, Platelet activation, Neutrophil extracellular trap formation, Toll-like receptor signaling pathway, C-type lectin receptor signaling pathway, JAK-STAT signaling pathway, Natural killer cell mediated cytotoxicity, T cell receptor signaling pathway, Bc cell receptor signaling pathway, Fc epsilon RI signaling pathway, Fc epamma R-mediated phagocytosis, TNF signaling pathway, Leukocyte transendothelial migration, Neurotrophin signaling pathway, Cholinergic synapse, Inflammatory mediator regulation of TRP channels, Regulation of actin cytoskeleton, Insulin signaling pathway, Projecterone-mediated oocyte maturation, Estrogen signaling pathway, GnRH secretion, Type II diabetes mellitus, Insulin resistance, Non-alcoholic fatty liver disease, AGE-RAGE signaling pathway in diabetic complications, Growth hormone synthesis, secretion and action, Aldosterone-regulated sodium reabsorption, Carbohydrate digestion and absorption, Alzheimer disease, Spinocerebellar ataxia, Prion disease, Bacterial invasion of epithelial cells, Shigellosis, Salmonella infection, Versinia infection, Chaqas disease, Amoebiasis, Hepatitis C, Hepatitis B, Measles, Human cytomegalovirus infection, Influenza A, Human appillomavirus infection, Human T-cell leukemia virus 1 infection, Kaposi sarcoma-associated herpesvirus infection, Herpes simplex virus 1 infection, Epstein-Barr virus infection, Human immunodeficiency virus 1 infection, Coronavirus disease - COVID-19, Pathways in cancer, Viral carcinogenesis, Proteoglycans in cancer, MicroRNAs in cancer, Chemical carcinogenesis - receptor activation, Chemical carcinogenesis - receptor activation, Chemical carcinogenesis - receptor activation, Gastric cancer, Renal cell carcinoma, Pancreatic cancer, Endometrial cancer, Glioma, Prostate cancer, Hepatocellular carcinoma, Gastric cancer, Central carbon metabolism in cancer, Choline metabolism in cancer, PD-1 expression and PD-1 checkpoint pathway in cancer, Diabetic cardiomyopathy, Li

OMIM_DISEASE

Breast cancer, somatic, Colorectal cancer, somatic, Hepatocellular carcinoma, somatic, Macrodactyly, somatic, Nevus, epidermal, somatic, Ovarian cancer, somatic, Keratosis, seborrheic, somatic, Nonsmall cell lung cancer, somatic, Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, CLOVE syndrome, somatic, CLAPO syndrome, somatic, Gastric cancer, somatic, Coundary of the Cound

Cowden syndrome 5

SMART
PI3K C2, PI3K p85B, PI3K rbd, PI3Ka, PI3Kc,
UP_KW_BIOLOGICAL_PROCESS
UP_KW_DISEASE
Disease variant, Proto-oncogene,

UP_KW_DOMAIN Coiled coil,

UP_KW_LIGAND ATP-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION

PRKCB

Kinase, Serine/threonine-protein kinase, Transferase,

protein kinase C beta(PRKCB)

UP_SEQ_FEATURE

DOMAIN:C2 PI3K-type, DOMAIN:PI3K-ABD, DOMAIN:PI3K-RBD, DOMAIN:PI3K/PI4K, DOMAIN:PIK helical, SITE:Implicated in the recognition of ATP as well as PIP2. Also crucial for autophosphorylation of the p85alpha subunit,

BIOCARTA

Attenuation of GPCR Signaling, Oxidative Stress Induced Gene Expression Via Nrf2, Angiotensin II mediated activation of JNK Pathway via Pyk2 dependent signaling, BCR Signaling Pathway, Bioactive Peptide Induced Signaling Pathway, Effects of calcineurin in Keratinocyte Differentiation, Role of EGF Receptor Transactivation by GPCRs in Cardiac Hypertrophy, CBL mediated ligand-induced downregulation of EGF receptors, CCR3 signaling in Eosinophils, Pertussis toxin-insensitive CCR5 Signaling in Macrophage, Cadmium induces DNA synthesis and proliferation in macrophages, Apoptotic Signaling in Response to DNA Damage, Transcription factor CREB and its extracellular signals, CXCR4 Signaling Pathway, Phospholipids as signalling intermediaries, EGF Signaling Pathway, Regulation of eIF4e and p70 S6 Kinase, g-Secretase mediated Erb84 Signaling Pathway, Fc Epsilon Receptor I Signaling in Mast Cells, Growth Hormone Signaling Pathway, Signaling Pathway from G-Protein Families, Ion Channel and Phorbal Esters Signaling Pathway, Keratinocyte Differentiation, Role of MEF2D in T-cell Apoptosis, PKC-catalyzed phosphorylation of inhibitory phosphoprotein of myosin phosphatase, Nitric Oxide Signaling Pathway, Thrombin signaling and protease-activated receptors, PDGF Signaling Pathway, Activation of PKC through G protein coupled receptor, Phospholipase C d1 in phospholipid associated cell signaling, Phospholipase C Signaling Pathway, Mechanism of Gene Regulation by Peroxisome Proliferators via PPARa(alpha), Links between Pyk2 and Map Kinases, Aspirin Blocks Signaling Pathway, Involved in Platelet Activation, Activation of Src by Protein-tyrosine phosphatase alpha, T Cell Receptor Signaling Pathway, TPO Signaling Pathway, Trka Receptor Signaling Pathway, VEGF, Hypoxia, and Angiogenesis,

GOTERM_BP_DIRECT

adaptive immune response, chromatin organization, regulation of transcription from RNA polymerase II promoter, protein phosphorylation, calcium ion transport, cellular calcium ion homeostasis, apoptotic process, mitotic nuclear envelope disassembly, signal transduction, regulation of glucose transport, negative regulation of glucose transport, peptidyl-serine phosphorylation, platelet activation, positive regulation of vascular endothelial growth factor receptor signaling pathway, histone H3-T6 phosphorylation, intracellular signal transduction, B cell activation, lipoprotein transport, positive regulation of I-kappaB kinase/NF-kappaB signaling, regulation of myeloid cell differentiation, positive regulation of angiogenesis, positive regulation of transcription, DNA-templated, negative regulation of insulin receptor signaling pathway, B cell receptor signaling pathway, positive regulation of B cell receptor signaling pathway, positive regulation of NF-kappaB transcription factor activity, cellular response to carbohydrate stimulus, regulation of synaptic vesicle exocytosis,

GOTERM_CC_DIRECT
GOTERM MF DIRECT

nucleus, nucleoplasm, cytoplasm, cytosol, plasma membrane, spectrin, calyx of Held, extracellular exosome, presynaptic cytosol, chromatin binding, protein kinase activity, protein serine/threonine kinase activity, protein kinase C activity, calcium-dependent protein kinase C activity, protein serine/threonine/tyrosine kinase activity, protein kinase C binding, calcium channel regulator activity, protein binding, ATP binding, zinc ion binding, ligand-dependent nuclear receptor transcription coactivator activity, histone kinase activity (H3-T6 specific), histone binding, androgen receptor binding,

INTERPRO

C2 calcium-dependent membrane targeting, Protein kinase, catalytic domain, AGC-kinase, C-terminal, Protein kinase C-like, phorbol ester/diacylglycerol binding, Serine/threonine-protein kinase, active site, Protein kinase-like domain, Protein kinase C, alpha/beta/gamma types, Protein kinase C mu-related, Protein kinase, ATP binding site, Protein kinase, C-terminal, Diacylglycerol/phorbol-ester binding,

KEGG PATHWAY

EGFR tyrosine kinase inhibitor resistance, MAPK signaling pathway, ErbB signaling pathway, Ras signaling pathway, Rap1 signaling pathway, Calcium signaling pathway, Chemokine signaling pathway, NF-kappa B signaling pathway, HIF-1 signaling pathway, Phosphatidylinositol signaling system, Sphingolipid signaling pathway, mTOR signaling pathway, Yascular smooth muscle contraction, Wnt signaling pathway, VEGF signaling pathway, Focal adhesion, Gap junction, Neutrophil extracellular trap formation, Natural killer cell mediated cytotoxicity, B cell receptor signaling pathway, Fc gamma R-mediated phagocytosis, Leukocyte transendothelial migration, Circadian entrainment, Long-term potentiation, Retrograde endocannabinoid signaling, Glutamatergic synapse, Cholinergic synapse, Serotonergic synapse, GABAergic synapse, Dopaminergic synapse, Long-term depression, Inflammatory mediator regulation of TRP channels, Insulin secretion, GnRH signaling pathway, Melanogenesis, Thyroid hormone synthesis, Thyroid hormone signaling pathway, Oxytocin signaling pathway, Aldosterone synthesis and secretion, Parathyroid hormone synthesis, secretion and action, GnRH secretion, Insulin resistance, AGE-RAGE signaling pathway in diabetic complications, Growth hormone synthesis, secretion and action, Aldosterone-regulated sodium reabsorption, Endocrine and other factor-regulated calcium reabsorption, Salivary secretion, Pancreatic secretion, Carbohydrate digestion and absorption, Spinocerebellar ataxia, Pathways of neurodegeneration - multiple diseases, Amphetamine addiction, Morphine addiction, Leishmaniasis, African trypanosomiasis, Amoebiasis, Hepatitis B, Human cytomegalovirus infection, Influenza A, Human immunodeficiency virus 1 infection, Coronavirus disease - COVID-19, Pathways in cancer, Proteoglycans in cancer, MicroRNAs in cancer, Chemical carcinogenesis - receptor activation, Glioma, Non-small cell lung cancer, Hepatocellular carcinoma, Choline metabolism in cancer, Diabetic cardiomyopathy,

PIR_SUPERFAMILY <u>protein kinase C, alpha/beta/gamma types</u>

SMART <u>C1</u>, <u>S TK X</u>, <u>S TKc</u>, <u>C2</u>

UP_KW_BIOLOGICAL_PROCESS Apoptosis, Immunity, Transcription, Transcription regulation, Adaptive immunity,

UP_KW_CELLULAR_COMPONENT Membrane, Nucleus, Cytoplasm,

UP_KW_DOMAIN Repeat, Zinc-finger,

 UP_KW_LIGAND
 ATP-binding, Calcium, Metal-binding, Nucleotide-binding, Zinc,

UP_KW_MOLECULAR_FUNCTION Chromatin regulator, Kinase, Serine/threonine-protein kinase, Transferase,

UP_KW_PTM Acetylation, Phosphoprotein,

UP_SEQ_FEATURE

ACT_SITE:Proton acceptor, BINDING:ATP, COMPBIAS:Basic and acidic residues, COMPBIAS:Polar residues, DOMAIN:AGC-kinase C-terminal, DOMAIN:C2, DOMAIN:Protein kinase, METAL:Calcium 1, METAL:Calcium 1; via carbonyl oxygen, METAL:Calcium 2, METAL:Calcium 2; via carbonyl oxygen, METAL:Calcium 3, METAL:Calcium 3; via carbonyl oxygen, NP_BIND:ATP, REGION:Disordered,

ZN_FING:Phorbol-ester/DAG-type 1, ZN_FING:Phorbol-ester/DAG-type 2,

PTPRD protein tyrosine phosphatase receptor type D(PTPRD) Related Genes Homo sapiens

GOTERM_BP_DIRECT protein dephosphorylation, phosphate-containing compound metabolic process, heterophilic cell-cell adhesion via plasma membrane cell adhesion molecules, transmembrane receptor protein tyrosine phosphatase signaling pathway, neuron differentiation, peptidyltyrosine dephosphorylation, negative regulation of JAK-STAT cascade, positive regulation of dendrite morphogenesis, regulation of

immune response, modulation of synaptic transmission, positive regulation of synapse assembly, presynaptic membrane assembly, trans-synaptic signaling by trans-synaptic complex, synaptic membrane adhesion, regulation of presynapse assembly,

GOTERM_CC_DIRECT plasma membrane, integral component of plasma membrane, integral component of membrane, extracellular exosome, Schaffer collateral - CA1 synapse, hippocampal mossy fiber to CA3 synapse, glutamatergic synapse, integral component of presynaptic

GOTERM MF DIRECT protein tyrosine phosphatase activity, transmembrane receptor protein tyrosine phosphatase activity, receptor binding, protein

binding, structural constituent of muscle, cell adhesion molecule binding,

INTERPRO Protein-tyrosine phosphatase, receptor/non-receptor type, Protein-tyrosine/Dual specificity phosphatase, Protein-tyrosine phosphatase, catalytic, Immunoglobulin subtype 2, Immunoglobulin subtype, Fibronectin, type III, Immunoglobulin-like domain, Immunoglobulin I-set, Immunoglobulin-like fold, Protein-tyrosine phosphatase, active site, Obscurin-myosin light chain kinase,

SMART FN3, PTPc, PTPc motif, IGc2, IG

UP_KW_CELLULAR_COMPONENT Membrane

UP_KW_DOMAIN Immunoglobulin domain, Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_MOLECULAR_FUNCTION Hydrolase, Receptor, Protein phosphatase,

UP KW PTM Glycoprotein, Disulfide bond

UP_SEQ_FEATURE

ACT_SITE:Phosphocysteine intermediate, BINDING:Substrate, CARBOHYD:N-linked (GlcNac...) asparagine, COMPBIAS:Polar residues, DOMAIN:Fibronectin type-III, DOMAIN:Fibronectin type-III 1, DOMAIN:Fibronectin type-III 2, DOMAIN:Fibronectin type-III 3, DOMAIN:Fibronectin type-III 4, DOMAIN:Fibronectin type-III 5, DOMAIN:Fibronectin type-III 6, DOMAIN:Fibronectin type-III 7, DOMAIN:Fibronectin type-III 8, DOMAIN:Ig-like, DOMAIN:Ig-like C2-type 1, DOMAIN:Ig-like C2-type 2, DOMAIN:Ig-like C2-type 3, DOMAIN:Tyrosine-protein phosphatase, DOMAIN:Tyrosine-protein phosphatase, DOMAIN:Tyrosine-protein phosphatase 2, MUTAGEN:R->A: 2.5-fold reduction in cleavage. 10-fold reduction in cleavage; when associated with A-1178., REGION:Disordered, REGION:Interaction with IL1RAPL1, REGION:Mini-exon peptide A9; sufficient for interaction with IL1RAPL1, REGION:Mini-exon peptide B; required for interaction with SLITRK2 and in the function in pre-synaptic differentiation; Acts as an adjustable linker to control relative positions and orientations of the PTPRD second and third immunoglobilin domains for their simultaneous interactions with the first immunoglobilin domain of IL1RAPL1 and IL1RAP; Modulates affinity for IL1RAPL1 and IL1RAP, REGION:Substrate binding, SITE:Cleavage, SITE:Required for interaction with IL1RAP, TOPO_DOM:Cytoplasmic, TOPO_DOM:Extracellular, TRANSMEM:Helical,

Related Genes RYR2 rvanodine receptor 2(RYR2)

BIOCARTA Actions of Nitric Oxide in the Heart,

GOTERM_BP_DIRECT response to hypoxia, regulation of heart rate, embryonic heart tube morphogenesis, left ventricular cardiac muscle tissue

morphogenesis, cardiac muscle hypertrophy, detection of calcium ion, calcium ion transport, cellular calcium ion homeostasis, positive regulation of heart rate, regulation of cardiac muscle contraction by regulation of the release of sequestered calcium ion, regulation of cardiac muscle contraction by calcium ion signaling, release of sequestered calcium ion into cytosol by sarcoplasmic reticulum, response to muscle activity, calcium-mediated signaling, response to caffeine, ion transmembrane transport, calcium-mediated signaling using intracellular calcium source, response to muscle stretch, release of sequestered calcium ion into cytosol, positive regulation of sequestering of calcium ion, regulation of cytosolic calcium ion concentration, response to redox state, regulation of cardiac muscle contraction, cardiac muscle contraction, calcium ion transport into cytosol, sarcoplasmic reticulum calcium ion transport, cellular response to caffeine, cellular response to epinephrine stimulus, establishment of protein localization to endoplasmic reticulum, ventricular cardiac muscle cell action potential, Purkinje myocyte to ventricular cardiac muscle cell signaling, cell communication by electrical coupling involved in cardiac conduction, type B pancreatic cell apoptotic process, positive regulation of the force of heart contraction, regulation of AV node cell action potential, regulation of SA node cell action potential, regulation of atrial

cardiac muscle cell action potential, regulation of ventricular cardiac muscle cell action potential, positive regulation of calcium-transporting ATPase activity, regulation of cardiac conduction,

GOTERM CC DIRECT smooth endoplasmic reticulum, plasma membrane, junctional sarcoplasmic reticulum membrane, membrane, integral component of

membrane, sarcoplasmic reticulum, Z disc, cytoplasmic vesicle membrane, macromolecular complex, sarcoplasmic reticulum membrane, calcium channel complex, sarcolemma,

GOTERM_MF_DIRECT ryanodine-sensitive calcium-release channel activity, calcium channel activity, calcium ion binding, protein binding, calmodulin binding, calcium-release channel activity, enzyme binding, protein kinase A catalytic subunit binding, protein kinase A regulatory subunit binding, identical protein binding, protein self-association, suramin binding, ion channel binding, calcium-induced calcium release

activity

INTERPRO Intracellular calcium-release channel, B30.2/SPRY domain, EF-hand domain, Ryanodine receptor Ryr, SPla/RYanodine receptor SPRY, Ion transport domain, Ryanodine Receptor TM 4-6, EF-hand-like domain, Concanavalin A-like lectin/glucanase, subgroup, Ryanodine

receptor, RyR/IP3R Homology associated domain, Inositol 1,4,5-trisphosphate/ryanodine receptor, MIR motif

KEGG PATHWAY

Calcium signaling pathway, cAMP signaling pathway, Cardiac muscle contraction, Adrenergic signaling in cardiomyocytes, Apelin signaling pathway, Circadian entrainment, Insulin secretion, Oxytocin signaling pathway, Pancreatic secretion, Prion disease, Pathways of neurodegeneration - multiple diseases, Hypertrophic cardiomyopathy, Arrhythmogenic right ventricular cardiomyopathy, Dilated

cardiomyopathy, Diabetic cardiomyopathy,

OMIM_DISEASE Ventricular arrythmias due to cardiac ryanodine receptor calcium release deficiency syndrome, Arrhythmogenic right ventricular

dysplasia 2, Ventricular tachycardia, catecholaminergic polymorphic, 1,

SMART SPRY, MIR

UP_KW_BIOLOGICAL_PROCESS Calcium transport, Ion transport, Transport, UP_KW_CELLULAR_COMPONENT Membrane, Sarcoplasmic reticulum, UP_KW_DISEASE Cardiomyopathy, Disease variant,

UP_KW_DOMAIN Coiled coil, Repeat, Transmembrane, Transmembrane helix,

UP KW LIGAND Calcium,

UP_KW_MOLECULAR_FUNCTION Calcium channel, Calmodulin-binding, Developmental protein, Ion channel, Receptor, Ligand-gated ion channel, Developmental

protein,

UP KW PTM

UP_SEQ_FEATURE COMPBIAS:Basic and acidic residues, DOMAIN:B30.2/SPRY, DOMAIN:B30.2/SPRY 1, DOMAIN:B30.2/SPRY 2, DOMAIN:B30.2/SPRY 3, DOMAIN:EF-hand, DOMAIN:MIR, DOMAIN:MIR 1, DOMAIN:MIR 2, DOMAIN:MIR 3, DOMAIN:MIR 4, DOMAIN:MIR 5, INTRAMEM:Pore-

forming, MUTAGEN:G->A: Changed ryanodine-sensitive calcium-release channel activity characterized by increased sensitivity to cytosolic calcium activation., MUTAGEN:H->N: Changed ryanodine-sensitive calcium-release channel activity characterized by increased sensitivity to cytosolic calcium activation., MUTAGEN:H->N: Changed ryanodine-sensitive calcium-release channel activity characterized by increased sensitivity to cytosolic calcium activation., MUTAGEN:H->Q: Changed ryanodine-sensitive calcium-release channel activity characterized by increased sensitivity to cytosolic calcium activation., MUTAGEN:M->I: Changed ryanodine-sensitive calcium-release channel activity characterized by increased sensitivity to cytosolic calcium activation., MUTAGEN:S->A: Abolishes phosphorylation by PKA., REGION:4 X approximate repeats, REGION:Disordered, REGION:Interaction with CALM, REPEAT:1, REPEAT:2, REPEAT:3, REPEAT:4, TOPO_DOM:Cytoplasmic, TRANSMEM:Helical,

STK11 serine/threonine kinase 11(STK11) Related Genes Homo sapiens

GOTERM_BP_DIRECT regulation of cell growth, tissue homeostasis, vasculature development, protein phosphorylation, protein dephosphorylation,

autophagy, cellular response to DNA damage stimulus, cell cycle arrest, spermatogenesis, axonogenesis, negative regulation of cell proliferation, response to ionizing radiation, positive regulation of autophagy, response to activity, peptidyl-threonine phosphorylation,

establishment of cell polarity, regulation of Wnt signaling pathway, negative regulation of cell growth, positive regulation of transforming growth factor beta receptor signaling pathway, activation of protein kinase activity, response to glucagon, response to lipid, glucose homeostasis, anoikis, positive thymic T cell selection, positive regulation of gluconeogenesis, protein autophosphorylation, regulation of dendrite morphogenesis, positive regulation of axonogenesis, T cell receptor signaling pathway, Golgi localization, regulation of cell cycle, regulation of protein kinase B signaling, canonical Wnt signaling pathway, negative regulation of epithelial cell proliferation involved in prostate gland development, G1 to G0 transition, cellular response to UV-B intrinsic apoptotic signaling pathway by p53 class mediator, negative regulation of canonical Wnt signaling pathway, response to thyroid hormone, dendrite extension, positive regulation of protein localization to nucleus, positive regulation of vesicle transport along microtubule, regulation of signal transduction by p53 class mediator, negative regulation of TORC1 signaling,

GOTERM_CC_DIRECT nucleus, nucleoplasm, cytoplasm, mitochondrion, cytosol, membrane, Z disc, extracellular exosome, serine/threonine protein kinase

magnesium ion binding, p53 binding, protein kinase activity, protein serine/threonine kinase activity, protein serine/threonine/tyrosine kinase activity, protein binding, ATP binding, kinase activity, LRR domain binding, protein kinase activator activity, macromolecular complex binding, metal ion binding, GOTERM_MF_DIRECT

INTERPRO Protein kinase, catalytic domain, Serine/threonine-protein kinase, active site, Protein kinase-like domain, Protein kinase, ATP binding

FoxO signaling pathway, Autophagy - animal, mTOR signaling pathway, PI3K-Akt signaling pathway, AMPK signaling pathway, Longevity regulating pathway, Tight junction, Adipocytokine signaling pathway, KEGG_PATHWAY

OMIM DISEASE Melanoma, malignant, somatic, Peutz-Jeghers syndrome, Pancreatic cancer, somatic, Testicular tumor, somatic,

SMAR1

UP_KW_BIOLOGICAL_PROCESS Apoptosis, Autophagy, Cell cycle, Differentiation, DNA damage, Spermatogenesis,

UP_KW_CELLULAR_COMPONENT Membrane, Mitochondrion, Nucleus, Cytoplasm,

UP_KW_DISEASE Tumor suppressor, Disease variant,

UP KW LIGAND ATP-binding, Magnesium, Manganese, Metal-binding, Nucleotide-binding,

UP_KW_MOLECULAR_FUNCTION Kinase, Serine/threonine-protein kinase, Transferase

UP KW PTM Acetylation, Lipoprotein, Methylation, Palmitate, Phosphoprotein, Prenylation,

UP_SEQ_FEATURE ACT SITE:Proton acceptor, BINDING:ATP, COMPBIAS:Basic and acidic residues, DOMAIN:Protein kinase, LIPID:S-farnesyl cysteine,

LIPID:S-palmitoyl cysteine, MUTAGEN:D->A: Loss of kinase activity, MUTAGEN:F->A: No effect. Impaired formation of a heterotrimeric complex with STRADA and CAB39; when associated with A-74., MUTAGEN:K->I: Loss of kinase activity, leading to greatly reduced autophosphorylation., MUTAGEN:K->M: Loss of kinase activity, leading to reduced autophosphorylation and acting as a dominant-negative mutant., MUTAGEN:K->Q: No effect on basal nucleocytoplasmic localization, but fails to translocate to the cytoplasm when coexpressed with SIRT1., MUTAGEN:K->R: Enhanced phosphorylation at Thr-336 and Ser-428, enhanced cytoplasmic localization and increased kinase activity., MUTAGEN:K->R: No effect on kinase activity., MUTAGEN:R->A: Impaired formation of a heterotrimeric complex with STRADA and CAB39; when associated with A-204., MUTAGEN:5->A,E: No effect on kinase activity., MUTAGEN:5->A,E: No effect on kinase activity., MUTAGEN:5->A: Reduced phosphorylation., NP_BIND:ATP, PROPEP:Removed in mature form, REGION:Disordered, REGION:Sufficient

for interaction with SIRT1,

SORCS3 sortilin related VPS10 domain containing receptor 3(SORCS3) Related Genes

GOTERM BP DIRECT neuropeptide signaling pathway, learning, memory, neurotrophin TRK receptor signaling pathway, regulation of long term synaptic

GOTERM CC DIRECT plasma membrane, membrane, integral component of membrane, glutamatergic synapse, integral component of postsynaptic density

GOTERM_MF_DIRECT protein binding, neuropeptide receptor activity,

PKD domain, VPS10, Immunoglobulin-like fold, WD40/YVTN repeat-like-containing domain, INTERPRO

VPS10, SMART UP_KW_CELLULAR_COMPONENT Membrane

UP_KW_DOMAIN Repeat, Signal, Transmembrane, Transmembrane helix,

UP_KW_MOLECULAR_FUNCTION Receptor, UP_KW_PTM Glycoprotein,

UP SEQ FEATURE CARBOHYD:N-linked (GlcNac...) asparagine, COMPBIAS:Basic and acidic residues, DOMAIN:PKD, DOMAIN:Sortilin-Vps10, REGION:Disordered, REPEAT:BNR 1, REPEAT:BNR 2, REPEAT:BNR 3, REPEAT:BNR 4, REPEAT:BNR 5, REPEAT:BNR 6,

TOPO_DOM:Cytoplasmic, TOPO_DOM:Lumenal, TRANSMEM:Helical, SVFP1

n Willebrand factor type A, EGF and pentraxin domain containing 1(SVEP1) GOTERM_BP_DIRECT lymph circulation, cell adhesion, epidermis development, gene expression, lymph vessel morphogenesis, Tie signaling pathway,

GOTERM CC DIRECT extracellular region, extracellular space, nucleus, cytoplasm, membrane

GOTERM_MF_DIRECT chromatin binding, calcium ion binding, carbohydrate binding,

INTERPRO EGF-type aspartate/asparagine hydroxylation site, Sushi/SCR/CCP, Epidermal growth factor-like domain, Pentaxin, EGF-like calcium-

binding, von Willebrand factor, type A, Hyalin, Insulin-like growth factor binding protein, N-terminal, Tyrosine-protein kinase ephrin type A/B receptor-like, EGF-like, conserved site, EGF, extracellular, Concanavalin A-like lectin/glucanase, subgroup, EGF-like calcium-

binding, conserved site,

SMART CCP, PTX, EGF_CA, EGF, VWA,

UP_KW_BIOLOGICAL_PROCESS Cell adhesion,

UP_KW_CELLULAR_COMPONENT Membrane, Cytoplasm, Secreted UP_KW_DOMAIN EGF-like domain, Repeat, Signal, Sushi,

UP KW LIGAND

BBID

UP KW PTM Glycoprotein, Disulfide bond

UP_SEQ_FEATURE

CARBOHYD:N-linked (GlcNac...) asparagine, COMPBIAS:Polar residues, DOMAIN:EGF-like, DOMAIN:EGF-like 1, DOMAIN:EGF-like 2; calcium-binding, DOMAIN:EGF-like 3; calcium-binding, DOMAIN:EGF-like 4; calcium-binding, DOMAIN:EGF-like 5; calcium-binding, DOMAIN:EGF-like 6; calcium-binding, DOMAIN:EGF-like 7; calcium-binding, DOMAIN:EGF-like 8, DOMAIN:EGF-like 9, DOMAIN:HYR, DOMAIN:HYR 1, DOMAIN:HYR 2, DOMAIN:Pentraxin (PTX), DOMAIN:Sushi, DOMAIN:Sushi 1, DOMAIN:Sushi 10, DOMAIN:Sushi 11, DOMAIN:Sushi 12, DOMAIN:Sushi 13, DOMAIN:Sushi 14, DOMAIN:Sushi 15, DOMAIN:Sushi 16, DOMAIN:Sushi 17, DOMAIN:Sushi 18, DOMAIN:Sushi 12, DOMAIN:Sushi 3, DOMAIN:Sushi 14, DOMAIN:Sushi 12, DOMAIN:Sushi 12, DOMAIN:Sushi 13, DOMAIN:Sushi 14, DOMAIN:Sushi 14, DOMAIN:Sushi 29, DOMAIN:Sushi 30, DOMAIN:Sushi 30, DOMAIN:Sushi 31, DOMAIN:Sushi 32, DOMAIN:Sushi 33, DOMAIN:Sushi 34, DOMAIN:Sushi 4, DOMAIN:Sushi 5, DOMAIN:Sushi 6, DOMAIN:Sushi 7, DOMAIN:Sushi 8, DOMAIN:Sushi 9, DOMAIN:Sushi 9, MOTIF:Cell attachment site, REGION:Disordered, REGION:O-

alvcosvlated at one site.

TP53 tumor protein p53(TP53) Related Genes Homo sapiens

1.RBphosphoE2F, 152.altered synaptic signalling-neurodegenerative disorders, 157.14-3-3 Cell Cycle,

38.Cell_cycle_arrest_and_apoptosis_ceramide, 94.E2F_transcriptional_activity_cell_cycle, 99.NF-kB_activation,

BIOCARTA Tumor Suppressor Arf Inhibits Ribosomal Biogenesis, ATM Signaling Pathway, Role of BRCA1, BRCA2 and ATR in Cancer Susceptibility, BTG family proteins and cell cycle regulation, Apoptotic Signaling in Response to DNA Damage, CTCF, Estrogen-responsive protein Efp controls cell cycle and breast tumors growth, Cell Cycle, Cell Cycle, Hypoxia and p53 in the Cardiovascular system, p53 Signaling Pathway, Regulation of cell cycle progression by Plk3, Regulation of transcriptional activity by PML, RB Tumor Suppressor/Checkpoint Signaling in response to DNA damage, Double Stranded RNA Induced Gene Expression, Telomeres, Telomerase, Cellular Aging, and Immortality, Overview of telomerase protein component gene hTert Transcriptional Regulation, Chaperones modulate interferor

GOTERM BP DIRECT

negative regulation of transcription from RNA polymerase II promoter, DNA strand renaturation, in utero embryonic development, somitogenesis, release of cytochrome c from mitochondria, hematopoietic progenitor cell differentiation, T cell proliferation involved in immune response, B cell lineage commitment, T cell lineage commitment, response to ischemia, nucleotide-excision repair, doublestrand break repair, regulation of transcription, DNA-templated, regulation of transcription from RNA polymerase II promoter, protein import into nucleus, autophagy, apoptotic process, cellular response to DNA damage stimulus, DNA damage response, signal transduction by p53 class mediator resulting in cell cycle arrest, DNA damage response, signal transduction by p53 class mediator resulting in transcription of p21 class mediator, response to oxidative stress, ER overload response, cell cycle, cell cycle arrest, transforming growth factor beta receptor signaling pathway, Ras protein signal transduction, gastrulation, negative regulation of neuroblast proliferation, cell aging, protein localization, negative regulation of DNA replication, negative regulation of cell proliferation, determination of adult lifespan, mRNA transcription, rRNA transcription, response to salt stress, response to X-ray, response to gamma radiation, positive regulation of gene expression, positive regulation of cardiac muscle cell apoptotic process, viral process, protein deubiquitination, cytokine-mediated signaling pathway, cerebellum development, negative regulation of cell growth, DNA damage response, signal transduction by p53 class mediator, negative regulation of transforming growth factor beta receptor signaling pathway, positive regulation of histone deacetylation, chromatin assembly, mitotic G1 DNA damage checkpoint, <u>T cell differentiation in thymus</u>, tumor necrosis factor-mediated signaling pathway, regulation of tissue remodeling, cellular protein localization, cellular response to UV, multicellular organism growth, cellular response to drug, positive regulation of mitochondrial membrane permeability, positive regulation of transcription from RNA polymerase II promoter in response to stress, cellular response to glucose starvation, response to drug, intrinsic apoptotic signaling pathway in response to DNA damage by p53 class mediator, regulation of apoptotic process, positive regulation of apoptotic process, negative regulation of apoptotic process, entrainment of circadian clock by photoperiod, mitochondrial DNA repair, regulation of DNA damage response, signal transduction by p53 class mediator, positive regulation of neuron apoptotic process, negative regulation of proteolysis, negative regulation of transcription, DNA-templated, positive regulation of proteolysis, negative regulation of proteolysis, negative regulation of positive regulation of proteolysis, negative re regulation of transcription, DNA-templated, positive regulation of RNA polymerase II transcriptional preinitiation complex assembly, positive regulation of transcription from RNA polymerase II promoter, response to antibiotic, positive regulation of protein export from nucleus, negative regulation of fibroblast proliferation, rhythmic process, circadian behavior, bone marrow development, embryonic organ development, positive regulation of peptidyl-tyrosine phosphorylation, protein stabilization, negative regulation of helicase activity, protein tetramerization, chromosome organization, neuron apoptotic process, regulation of cell cycle, negative regulation of telomerase activity, hematopoietic stem cell differentiation, regulation of transcription initiation from RNA polymerase II promoter, interferon-gamma-mediated signaling pathway, cardiac septum morphogenesis, positive regulation of transcription from RNA polymerase II promoter in response to hypoxia, macromolecular complex assembly, intrinsic apoptotic signaling pathway in response to endoplasmic reticulum stress, positive regulation of thymocyte apoptotic process, necroptotic process, positive regulation of cell cycle arrest, cellular response to hypoxia, cellular response to xenobiotic stimulus, cellular response to ionizing radiation, cellular response to gamma radiation, cellular response to UV-C, mitotic cell cycle arrest, signal transduction by p53 class mediator, intrinsic apoptotic signaling pathway by p53 class mediator, cellular response to actinomycin D, positive regulation of release of cytochrome c from mitochondria, positive regulation of cell aging, replicative senescence, oxidative stress-induced premature senescence, intrinsic apoptotic signaling pathway, oligodendrocyte apoptotic process, positive regulation of execution phase of apoptosis, positive regulation of protein insertion into mitochondrial membrane involved in apoptotic signaling pathway, negative regulation of macromitophagy, regulation of signal transduction by p53 class mediator, regulation of mitochondrial membrane permeability involved in apoptotic process, regulation of intrinsic apoptotic signaling pathway by p53 class mediator, regulation of cell cycle G2/M phase transition, positive regulation of pri-miRNA transcription from RNA polymerase II promoter, negative regulation of G1 to G0 transition, negative regulation of production of miRNAs involved in gene silencing by miRNA, positive regulation of production of miRNAs involved in gene silencing by miRNA, positive regulation of production of miRNAs involved in gene silencing by miRNA, negative regulation of glucose catabolic process to lactate via pyruvate, negative regulation of pentose-phosphate shunt, intrinsic apoptotic signaling pathway in response to hypoxia, regulation of transcription from RNA polymerase II promoter in response to DNA damage, positive regulation of transcription from RNA polymerase II promoter in response to endoplasmic reticulum stress, regulation of fibroblast apoptotic process, negative regulation of reactive oxygen species metabolic process, positive regulation of reactive oxygen species metabolic process, regulation of cellular senescence, positive regulation of intrinsic apoptotic signaling pathway

GOTERM_CC_DIRECT

chromatin, nucleus, nucleoplasm, replication fork, transcription factor complex, nucleolus, cytoplasm, mitochondrion, mitochondrial matrix, endoplasmic reticulum, centrosome, microtubule organizing center, cytosol, integral component of membrane, nuclear matrix nuclear body, PML body, transcriptional repressor complex, macromolecular complex, site of double-strand break,

GOTERM_MF_DIRECT

transcription regulatory region sequence-specific DNA binding, RNA polymerase II core promoter proximal region sequence-specific DNA binding, RNA polymerase II transcription factor activity, sequence-specific DNA binding, core promoter proximal region sequence-specific DNA binding, core promoter sequence-specific DNA binding, RNA polymerase II transcription factor binding, TFIID-class transcription factor binding, bacterial-type RNA polymerase transcriptional activator activity, sequence-specific DNA binding, transcriptional repressor activity, RNA polymerase II transcription regulatory region sequence-specific binding, transcriptional activator activity, RNA polymerase II transcription regulatory region sequence-specific binding, protease binding, p53 binding, DNA binding, chromatin binding, transcription factor activity, sequence-specific DNA binding, mRNA 3'-UTR binding, copper ion binding, protein binding, transcription factor binding, zinc ion binding, enzyme binding, protein kinase binding, protein phosphatase binding, receptor tyrosine kinase binding, ubiquitin protein ligase binding, histone deacetylase regulator activity, histone acetyltransferase binding, protein binding, annealing helicase activity, identical protein binding, histone deacetylase binding, protein self-association, metal ion binding, protein heterodimerization activity, protein N-terminus binding, chaperone binding, protein phosphatase 2A binding, RNA polymerase II sequence-specific DNA binding transcription factor binding, MDM2/MDM4 family protein binding, disordered domain specific binding, promoter-specific chromatin binding,

INTERPRO

p53 tumour suppressor family, p53-like transcription factor, DNA-binding, p53, tetramerisation domain, p53, DNA-binding domain, p53/RUNT-type transcription factor, DNA-binding domain, p53 transactivation domain, PLAA family ubiquitin binding, PFU,

KEGG PATHWAY

Endocrine resistance, Platinum drug resistance, MAPK signaling pathway, Sphingolipid signaling pathway, Cell cycle, p53 signaling pathway, Mitophagy - animal, PI3K-Akt signaling pathway, Apoptosis, Longevity regulating pathway, Ferroptosis, Cellular senescence, Wnt signaling pathway, Neurotrophin signaling pathway, Thyroid hormone signaling pathway, Parkinson disease, Amyotrophic lateral sclerosis, Huntington disease, Shigellosis, Hepatitis C, Hepatitis B, Measles, Human cytomegalovirus infection, Human papillomavirus infection, Human T-cell leukemia virus 1 infection, Kaposi sarcoma-associated herpesvirus infection, Herpes simplex virus 1 infection, Epstein-Barr virus infection, Pathways in cancer, Transcriptional misregulation in cancer, Viral carcinogenesis, Proteoglycans in cancer, MicroRNAs in cancer, Colorectal cancer, Pancreatic cancer, Endometria cancer, Giloma, Prostate cancer, Thyroid cancer, Basal cell carcinoma, Melanoma, Bladder cancer, Chronic myeloid leukemia, Small cell lung cancer, Non-small cell lung cancer, Breast cancer, Hepatocellular carcinoma, Gastric cancer, Central carbon metabolism in cancer, Lipid and atherosclerosis, Fluid shear stress and

OMIM_DISEASE

Breast cancer, somatic, Colorectal cancer, Hepatocellular carcinoma, somatic, Glioma susceptibility 1, Li-Fraumeni syndrome, Adrenocortical carcinoma, pediatric, Osteosarcoma, Choroid plexus papilloma, Nasopharyngeal carcinoma, somatic, Basal cell carcinoma 7, Bone marrow failure syndrome 5, Pancreatic cancer, somatic 260350,

UP_KW_BIOLOGICAL_PROCESS

Apoptosis, Biological rhythms, Cell cycle, Transcription, Transcription regulation, Host-virus interaction, Necrosis,

UP KW DISEASE

UP_KW_CELLULAR_COMPONENT Cytoskeleton, Endoplasmic reticulum, Membrane, Mitochondrion, Nucleus, Cytoplasmic Tumor suppressor, Disease variant, Li-Fraumeni syndrome, Li-Fraumeni syndrome,

UP KW DOMAIN Transmembrane, Transmembrane helix

UP_KW_LIGAND Metal-binding, Zinc

UP_KW_MOLECULAR_FUNCTION Activator, DNA-binding, Repressor,

UP_KW_PTM UP SEQ FEATURE Acetylation, Glycoprotein, Methylation, Phosphoprotein, Ubl conjugation, Isopeptide bond,

COMPBIAS:Basic and acidic residues, COMPBIAS:Basic residues, COMPBIAS:Polar residues, COMPBIAS:Pro residues, CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in SUMO), CROSSLNK:Glycyl lysine isopeptide (Lys-Gly) (interchain with G-Cter in lysine isopeptide (Lys-Giy) (interchain with G-Cter in Sumo), CROSSLINK:GlyCri Iysine isopeptide (Lys-Giy) (interchain with G-Cter in ubiquitin), DOMAIN:P53, DOMAIN:P53_tetramer, DOMAIN:PFU, DOMAIN:TAD2, METAL:Zinc, MOTIF:Bipartite nuclear localization signal, MOTIF:Nuclear export signal, MOTIF:TADII, MOTIF:[KR]-[STA]-K motif, MUTAGEN:E->A: Abolishes SUMO1 conjugation., MUTAGEN:F->A: Reduced SUMO1 conjugation., MUTAGEN:G->E: Abolishes binding to USP7., MUTAGEN:K->A: Abolishes SUMO1 conjugation, in vitro and in vivo., MUTAGEN:K->A: Abolishes acetylation by CREBBP., MUTAGEN:K-

>A: Loss of nuclear localization; when associated with A-319 and A-320, MUTAGEN:K->A: Loss of nuclear localization; when associated with A-319 and A-321, MUTAGEN:K->A: Loss of nuclear localization; when associated with A-320 and A-321, MUTAGEN:K->R: Abolishes dimethylation by EHMT1 and EHMT2., MUTAGEN:K->R: Abolishes monomethylation by KMT5A., MUTAGEN:K->R: Abolishes ubiquitination by MUL1., MUTAGEN:K->R: Induces a decrease in methylation by SMYD2., MUTAGEN:K->A: Induces a decrease in protein stabilization., MUTAGEN:KK->RR: Abolishes polyubiquitination by MKRN1., MUTAGEN:L->A: Abolishes S-315 phosphorylation by CDK2/cyclin A., MUTAGEN:LW->QS: Loss of interaction with MDM2, leading to constitutively increased TP53 protein levels., MUTAGEN:Missing: Alters interaction with WWOX., MUTAGEN:P->D: Abolishes binding to USP7., MUTAGEN:R->S: Does not induce SNA11 degradation., MUTAGEN:RGRER->KGKEK: Reduced methylation by PRMT5. Reduced nuclear localization. Decreased binding to promoters of target genes. Reduced transcriptional activity. Decrease in cell cycle arrest., MUTAGEN:S->A: Abolishes binding to USP7., MUTAGEN:S->A: Abolishes phosphorylation by DYRK2 and HIPK2 and acetylation of K-382 by CREBBP, MUTAGEN:S->A: Abolishes phosphorylation site. Abolishes strongly phosphorylation., MUTAGEN:S->A: Abolishes phosphorylation., MUTAGEN:S->A: Abolishes phosphorylation by MAFKAPK5., MUTAGEN:S->A: Loss of interaction with PPP2R5C, PPP2CA AND PPP2R1A., MUTAGEN:S->D: Abolishes phosphorylation by MAFKAPK5., MUTAGEN:S->D: Constitutively increased TP53 protein levels., MUTAGEN:S->E: Inhibits slightly its transcriptional activity., MUTAGEN:S->E: Inhibits strongly its transcriptional activity., MUTAGEN:T->A: No effect SUM01 conjugation., MUTAGEN:T->A: No effect on interaction with MDM2 and increase in protein levels after DNA damage., MUTAGEN:T->E: Inhibits strongly its transcriptional activity., REGION:Interaction with AUTAGEN:T->A: No effect SUM01 conjugation., MUTAGEN:T->A: No effect on interaction with CARM1, REGION:Interaction