Electronic Supplementary Material (ESI) for Metallomics.

	Uniprot Id	Entry name	Gene names	Protein names	Predicted Pattern	Number of iron ions	Iron role	EC number	Subcellular location	Membrane associated	Involvement in disease	Gene ontology (biological process)
1	P46952	3HAO_HUMAN	HAAO	3-hydroxyanthranilate 3,4-dioxygenase (EC 1.13.11.6) (3-hydroxyanthranilate oxygenase) (3-HAO) (3-hydroxyanthranilic acid dioxygenase) (HAD)	H47-E53-H91	1 Fe cation	Catalytic	1.13.11.6	Cytoplasm	No		NAD biosynthetic process [GO:0009435]; neuron cellular homeostasis [GO:0070050]; quinolinate biosynthetic process [GO:0019805]; response to cadmium ion [GO:0046686]; response to zinc ion [GO:0010043]; tryptophan catabolic process [GO:0006569]
2	O00767	ACOD_HUMAN	SCD	Acyl-CoA desaturase (EC 1.14.19.1) (Delta(9)-desaturase) (Delta-9 desaturase) (Fatty acid desaturase) (Stearoyl-CoA desaturase) (hSCD1)	H120-H125-H157-H161; H160-H269-H298-H302	2 Fe cations	Catalytic	1.14.19.1	Endoplasmic reticulum	Yes		long-chain fatty-acyl-CoA biosynthetic process [GO:0035338]; unsaturated fatty acid biosynthetic process [GO:0006636]
3	Q6ZNF0	ACP7_HUMAN	ACP7 PAPL PAPL1	Acid phosphatase type 7 (EC 3.1.3.2) (Purple acid phosphatase long form)	D141-D170-Y173-H335	1 Fe cation	Catalytic	3.1.3.2	Extracellular space	No		
4	Q96SZ5	AEDO_HUMAN	ADO C10orf22	2-aminoethanethiol dioxygenase (EC 1.13.11.19) (Cysteamine dioxygenase)	H112-H114-H193	1 Fe cation	Catalytic	1.13.11.19	Unknown	No		oxidation-reduction process [GO:0055114]; sulfur amino acid catabolic process [GO:0000098]
5	Q13686	ALKB1_HUMAN	ALKBH1 ABH ABH1 ALKBH	Nucleic acid dioxygenase ALKBH1 (EC 1.14.11.-) (Alkylated DNA repair protein alkB homolog 1) (Alpha-ketoglutarate-dependent dioxygenase ABH1) (DNA 6mA demethylase) (DNA N6-methyl adenine demethylase) (EC 1.14.11.-) (DNA lyase ABH1) (EC 4.2.99.18) (DNA oxidative demethylase ALKBH1) (EC 1.14.11.33) (tRNA N1-methyl adenine demethylase) (EC 1.14.11.-)	H231-D233-H287	1 Fe cation	Catalytic	1.14.11.-; 4.2.99.18; 1.14.11.33	Mitochondrion, Nucleus	No		developmental growth [GO:0048589]; DNA dealkylation involved in DNA repair [GO:0006307]; DNA demethylation [GO:0080111]; DNA repair [GO:0006281]; in utero embryonic development [GO:0001701]; negative regulation of neuron apoptotic process [GO:0043524]; neuron migration [GO:0001764]; neuron projection development [GO:0031175]; oxidative demethylation [GO:0070989]; oxidative single-stranded DNA demethylation [GO:0035552]; placenta development [GO:0001890]; regulation of mitochondrial translation [GO:0070129]; regulation of translational elongation [GO:0006448]; regulation of translational initiation [GO:0006446]; RNA repair [GO:0042245]; tRNA demethylation [GO:1990983]; tRNA wobble cytosine modification [GO:0002101]
6	Q6NS38	ALKB2_HUMAN	ALKBH2 ABH2	DNA oxidative demethylase ALKBH2 (EC 1.14.11.33) (Alkylated DNA repair protein alkB homolog 2) (Alpha-ketoglutarate-dependent dioxygenase alkB homolog 2) (Oxy DC1)	H171-D173-H236	1 Fe cation	Catalytic	1.14.11.33	Nucleus	No		DNA dealkylation involved in DNA repair [GO:0006307]; DNA demethylation [GO:0080111]; oxidative demethylation [GO:0070989]; oxidative DNA demethylation [GO:0035511]
7	Q96Q83	ALKB3_HUMAN	ALKBH3 ABH3 DEPC1	Alpha-ketoglutarate-dependent dioxygenase alkB homolog 3 (EC 1.14.11.54) (Alkylated DNA repair protein alkB homolog 3) (hABH3) (DEPC-1) (Prostate cancer antigen 1)	H191-D193-H257	1 Fe cation	Catalytic	1.14.11.54	Cytoplasm, Nucleus	No		cell proliferation [GO:0008283]; DNA dealkylation involved in DNA repair [GO:0006307]; DNA repair [GO:0006281]; oxidative single-stranded DNA demethylation [GO:0035552]; oxidative single-stranded RNA demethylation [GO:0035553]
8	Q9NXW9	ALKB4_HUMAN	ALKBH4 ABH4	Alpha-ketoglutarate-dependent dioxygenase alkB homolog 4 (EC 1.14.11.-) (Alkylated DNA repair protein alkB homolog 4)	H169-D171-H254	1 Fe cation	Catalytic	1.14.11.-	Cytoplasm, Nucleus	No		actomyosin structure organization [GO:0031032]; cleavage furrow ingression [GO:0036090]; protein demethylation [GO:0006482]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]
9	Q6P6C2	ALKB5_HUMAN	ALKBH5 ABH5 OFOXD1	RNA demethylase ALKBH5 (EC 1.14.11.-) (Alkylated DNA repair protein alkB homolog 5) (Alpha-ketoglutarate-dependent dioxygenase alkB homolog 5)	H204-D206-H266	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		cell differentiation [GO:0030154]; DNA dealkylation involved in DNA repair [GO:0006307]; mRNA export from nucleus [GO:0006406]; mRNA processing [GO:0006397]; oxidative single-stranded RNA demethylation [GO:0035553]; response to hypoxia [GO:0001666]; spermatogenesis [GO:0007283]
10	Q3KRA9	ALKB6_HUMAN	ALKBH6 ABH6	Alpha-ketoglutarate-dependent dioxygenase alkB homolog 6 (EC 1.14.11.-) (Alkylated DNA repair protein alkB homolog 6)	H114-D116-H182	1 Fe cation	Catalytic	1.14.11.-	Cytoplasm, Nucleus	No		

11	Q9BT30	ALKB7_HUMAN	ALKBH7 ABH7 SPATA11 UNQ6002/PRO34564	Alpha-ketoglutarate-dependent dioxygenase alkB homolog 7, mitochondrial (EC 1.14.11.-) (Alkylated DNA repair protein alkB homolog 7) (Spermatogenesis cell proliferation-related protein) (Spermatogenesis-associated protein 11)	H121-D123-H177	1 Fe cation	Catalytic	1.14.11.-	Mitochondrion	No		cellular response to DNA damage stimulus [GO:0006974]; fatty acid metabolic process [GO:0006631]; regulation of lipid storage [GO:0010883]; regulation of mitochondrial membrane permeability involved in programmed necrotic cell death [GO:1902445]
12	Q96BT7	ALKB8_HUMAN	ALKBH8 ABH8	Alkylated DNA repair protein alkB homolog 8 (EC 1.14.11.-) (Probable alpha-ketoglutarate-dependent dioxygenase ABH8) (S-adenosyl-L-methionine-dependent tRNA methyltransferase ABH8) (tRNA (carboxymethyluridine(34)-5-O)-methyltransferase ABH8) (EC 2.1.1.229)	H238-D240-H292	1 Fe cation	Catalytic	1.14.11.-; 2.1.1.229	Cytoplasm, Nucleus	No		cellular response to DNA damage stimulus [GO:0006974]; oxidation-reduction process [GO:0055114]; tRNA methylation [GO:0030488]; tRNA wobble uridine modification [GO:0002098]
13	Q6ZNB7	ALKMO_HUMAN	AGMO TMEM195	Alkylglycerol monooxygenase (EC 1.14.16.5) (Transmembrane protein 195)	H157-H161-H170-H173; H228-H250-H252-H253	2 Fe cations	Catalytic	1.14.16.5	Endoplasmic reticulum	Yes		ether lipid metabolic process [GO:0046485]; membrane lipid metabolic process [GO:0006643]; triglyceride biosynthetic process [GO:0019432]
14	Q12797	ASPH_HUMAN	ASPH BAH	Aspartyl/asparaginyl beta-hydroxylase (EC 1.14.11.16) (Aspartate beta-hydroxylase) (ASP beta-hydroxylase) (Peptide-aspartate beta-dioxygenase)	H679-H725	1 Fe cation	Catalytic	1.14.11.16	Endoplasmic reticulum	Yes	DISEASE: Facial dysmorphism, lens dislocation, anterior segment abnormalities, and spontaneous filtering blebs (FDLAB) [MIM:601552]: A syndrome characterized by dislocated crystalline lenses and anterior segment abnormalities in association with a distinctive facies involving flat cheeks and a beaked nose. Some affected individuals develop highly unusual non-traumatic conjunctival cysts (filtering blebs). {ECO:0000269|PubMed:24768550}. Note=The disease is caused by mutations affecting the gene represented in this entry.	activation of cysteine-type endopeptidase activity [GO:0097202]; activation of store-operated calcium channel activity [GO:0032237]; calcium ion transmembrane transport [GO:0070588]; cellular response to calcium ion [GO:0071277]; detection of calcium ion [GO:0005513]; face morphogenesis [GO:0060325]; ion transmembrane transport [GO:0034220]; limb morphogenesis [GO:0035108]; muscle contraction [GO:0006936]; negative regulation of cell proliferation [GO:0008285]; palate development [GO:0060021]; pattern specification process [GO:0007389]; peptidyl-aspartic acid hydroxylation [GO:0042264]; positive regulation of calcium ion transport into cytosol [GO:0010524]; positive regulation of intracellular protein transport [GO:0090316]; positive regulation of proteolysis [GO:0045862]; positive regulation of ryanodine-sensitive calcium-release channel activity [GO:0060316]; positive regulation of transcription, DNA-templated [GO:0045893]; regulation of cardiac conduction [GO:1903779]; regulation of cardiac muscle contraction by regulation of the release of sequestered calcium ion [GO:0010881]; regulation of cell communication by electrical coupling [GO:0010649]; regulation of inositol 1,4,5-trisphosphate-sensitive calcium-release channel activity [GO:0031585]; regulation of protein depolymerization [GO:1901879]; regulation of protein stability [GO:0031647]; regulation of release of sequestered calcium ion into cytosol by sarcoplasmic reticulum [GO:0010880]; regulation of ryanodine-sensitive calcium-release channel activity [GO:0060314]; response to ATP [GO:0033198]
15	Q6ICH7	ASPH2_HUMAN	ASPHD2	Aspartate beta-hydroxylase domain-containing protein 2 (EC 1.14.11.-)	H283-H328	1 Fe cation	Catalytic	1.14.11.-	Unknown	Yes		peptidyl-amino acid modification [GO:0018193]
16	Q9HAY6	BCDO1_HUMAN	BCO1 BCDO BCDO1 BCMO1	Beta,beta-carotene 15,15'-dioxygenase (EC 1.13.11.63) (Beta-carotene dioxygenase 1) (Beta-carotene oxygenase 1)	H172-H237-H308-H514	1 Fe cation	Catalytic	1.13.11.63	Unknown	No	DISEASE: Hypercarotenemia and vitamin A deficiency, autosomal dominant (ADHVAD) [MIM:115300]: A disorder characterized by increased serum beta-carotene, decreased conversion of beta-carotene to vitamin A and decreased serum vitamin A. {ECO:0000269|PubMed:17951468}. Note=The disease is caused by mutations affecting the gene represented in this entry.	beta-carotene metabolic process [GO:1901810]; retinal metabolic process [GO:0042574]; retinoid metabolic process [GO:0001523]; retinol metabolic process [GO:0042572]; vitamin A biosynthetic process [GO:0035238]
17	Q9BYV7	BCDO2_HUMAN	BCO2 BCDO2	Beta,beta-carotene 9',10'-oxygenase (EC 1.13.11.71) (B-diox-II) (Beta-carotene dioxygenase 2)	H226-H286-H357-H573	1 Fe cation	Catalytic	1.13.11.71	Mitochondrion	No		carotene catabolic process [GO:0016121]; carotene metabolic process [GO:0016119]; carotenoid metabolic process [GO:0016116]; oxidation-reduction process [GO:0055114]; regulation of mitochondrial membrane potential [GO:0051881]; regulation of reactive oxygen species metabolic process [GO:2000377]; retinal metabolic process [GO:0042574]; retinoic acid metabolic process [GO:0042573]; retinoid metabolic process [GO:0001523]; xanthophyll metabolic process [GO:0016122]

18	O75936	BODG_HUMAN	BBOX1 BBH BBOX	Gamma-butyrobetaine dioxygenase (EC 1.14.11.1) (Gamma-butyrobetaine hydroxylase) (Gamma-BBH) (Gamma-butyrobetaine,2-oxoglutarate dioxygenase)	H202-D204-H347	1 Fe cation	Catalytic	1.14.11.1	Cytoplasm	No		carnitine biosynthetic process [GO:0045329]
19	O95992	CH25H_HUMAN	CH25H	Cholesterol 25-hydroxylase (EC 1.14.99.38) (Cholesterol 25-monooxygenase) (h25OH)	H143-H147-H157-H161; H205-H238-H242-H243	2 Fe cations	Catalytic	1.14.99.38	Endoplasmic reticulum	Yes		B cell chemotaxis [GO:0035754]; bile acid biosynthetic process [GO:0006699]; cholesterol metabolic process [GO:0008203]; lipid metabolic process [GO:0006629]; sterol biosynthetic process [GO:0016126]
20	Q99807	COQ7_HUMAN	COQ7	5-demethoxyubiquinone hydroxylase, mitochondrial (DMQ hydroxylase) (EC 1.14.13.-) (Timing protein clk-1 homolog) (Ubiquinone biosynthesis monooxygenase COQ7)	E60-E90-H93-E178; E90-E142-E178-H181	2 Fe cations	Catalytic	1.14.13.-	Mitochondrion	Yes	DISEASE: Coenzyme Q10 deficiency, primary, 8 (COQ10D8) [MIM:616733]: An autosomal recessive disorder resulting from mitochondrial dysfunction and characterized by decreased levels of coenzyme Q10. Patients manifest neonatal lung hypoplasia, contractures, early infantile hypertension and cardiac hypertrophy, secondary to prenatal kidney dysplasia, with neonatal and infantile renal dysfunction. Clinical features also include progressive peripheral neuropathy, muscular hypotonia and atrophy, and mild psychomotor delay with hearing and visual impairment. {ECO:0000269|PubMed:26084283}. Note=The disease is caused by mutations affecting the gene represented in this entry.	negative regulation of transcription from RNA polymerase II promoter [GO:0000122]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; regulation of reactive oxygen species metabolic process [GO:2000377]; ubiquinone biosynthetic process [GO:0006744]
21	D3DRM8	D3DRM8_HUMAN	hCG_2040046	Galactose-1-phosphate uridylyltransferase	E154-H253-H271-H273	1 Fe cation	Catalytic	2.7.7.12	Unknown	No		
22	Q9BU89	DOHH_HUMAN	DOHH HLRC1	Deoxyhypusine hydroxylase (hDOHH) (EC 1.14.99.29) (Deoxyhypusine dioxygenase) (Deoxyhypusine monooxygenase) (HEAT-like repeat-containing protein 1)	H56-E57-H89-E90; H207-E208-H240-E241	2 Fe cations	Catalytic	1.14.99.29	Unknown	No		peptidyl-lysine modification to peptidyl-hypusine [GO:0008612]
23	Q9GZT9	EGLN1_HUMAN	EGLN1 C1orf12 PNAS-118 PNAS-137	Egl nine homolog 1 (EC 1.14.11.29) (Hypoxia-inducible factor prolyl hydroxylase 2) (HIF-PH2) (HIF-prolyl hydroxylase 2) (HPH-2) (Prolyl hydroxylase domain-containing protein 2) (PHD2) (SM-20)	H313-D315-H374	1 Fe cation	Catalytic	1.14.11.29	Cytoplasm, Nucleus	No	DISEASE: Erythrocytosis, familial, 3 (ECYT3) [MIM:609820]: An autosomal dominant disorder characterized by increased serum red blood cell mass, elevated serum hemoglobin and hematocrit, and normal serum erythropoietin levels. {ECO:0000269|PubMed:16407130, ECO:0000269|PubMed:17579185}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cardiac muscle tissue morphogenesis [GO:0055008]; cellular iron ion homeostasis [GO:0006879]; heart trabecula formation [GO:0060347]; labyrinthine layer development [GO:0060711]; negative regulation of cAMP catabolic process [GO:0030821]; negative regulation of cyclic-nucleotide phosphodiesterase activity [GO:0051344]; negative regulation of sequence-specific DNA binding transcription factor activity [GO:0043433]; oxygen homeostasis [GO:0032364]; peptidyl-proline hydroxylation to 4-hydroxy-L-proline [GO:0018401]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; regulation of angiogenesis [GO:0045765]; regulation of neuron death [GO:1901214]; regulation of transcription from RNA polymerase II promoter in response to hypoxia [GO:0061418]; response to hypoxia [GO:0001666]; response to nitric oxide [GO:0071731]; ventricular septum morphogenesis [GO:0060412]
24	Q96KS0	EGLN2_HUMAN	EGLN2 EIT6	Egl nine homolog 2 (EC 1.14.11.29) (Estrogen-induced tag 6) (HPH-3) (Hypoxia-inducible factor prolyl hydroxylase 1) (HIF-PH1) (HIF-prolyl hydroxylase 1) (HPH-1) (Prolyl hydroxylase domain-containing protein 1) (PHD1)	H297-D299-H358	1 Fe cation	Catalytic	1.14.11.29	Nucleus	No		cell redox homeostasis [GO:0045454]; intracellular estrogen receptor signaling pathway [GO:0030520]; peptidyl-proline hydroxylation to 4-hydroxy-L-proline [GO:0018401]; positive regulation of protein catabolic process [GO:0045732]; regulation of cell growth [GO:0001558]; regulation of neuron apoptotic process [GO:0043523]; regulation of transcription from RNA polymerase II promoter in response to hypoxia [GO:0061418]; response to hypoxia [GO:0001666]
25	Q9H6Z9	EGLN3_HUMAN	EGLN3	Egl nine homolog 3 (EC 1.14.11.29) (HPH-1) (Hypoxia-inducible factor prolyl hydroxylase 3) (HIF-PH3) (HIF-prolyl hydroxylase 3) (HPH-3) (Prolyl hydroxylase domain-containing protein 3) (PHD3)	H135-D137-H196	1 Fe cation	Catalytic	1.14.11.29	Cytoplasm, Nucleus	No		activation of cysteine-type endopeptidase activity involved in apoptotic process [GO:0006919]; apoptotic process [GO:0006915]; cellular response to DNA damage stimulus [GO:0006974]; peptidyl-proline hydroxylation to 4-hydroxy-L-proline [GO:0018401]; protein hydroxylation [GO:0018126]; regulation of cell proliferation [GO:0042127]; regulation of neuron apoptotic process [GO:0043523]; regulation of transcription from RNA polymerase II promoter in response to hypoxia [GO:0061418]; response to hypoxia [GO:0001666]

26	O95571	ETHE1_HUMAN	ETHE1 HSCO	Persulfide dioxygenase ETHE1, mitochondrial (EC 1.13.11.18) (Ethylmalonic encephalopathy protein 1) (Hepatoma subtracted clone one protein) (Sulfur dioxygenase ETHE1)	H79-H135-D154	1 Fe cation	Catalytic	1.13.11.18	Cytoplasm, Mitochondrion, Nucleus	No	DISEASE: Ethylmalonic encephalopathy (EE) [MIM:602473]: Autosomal recessive disorder characterized by neurodevelopmental delay and regression, recurrent petechiae, acrocyanosis, diarrhea, leading to death in the first decade of life. It is also associated with persistent lactic acidemia and ethylmalonic and methylsuccinic aciduria. {ECO:0000269|PubMed:14732903, ECO:0000269|PubMed:18593870, ECO:0000269|PubMed:23144459}. Note=The disease is caused by mutations affecting the gene represented in this entry.	glutathione metabolic process [GO:0006749]; hydrogen sulfide metabolic process [GO:0070813]; sulfide oxidation, using sulfide:quinone oxidoreductase [GO:0070221]
27	Q9UKA1	FBXL5_HUMAN	FBXL5 FBL4 FBL5 FLR1	F-box/LRR-repeat protein 5 (F-box and leucine-rich repeat protein 5) (F-box protein FBL4/FBL5) (p45SKP2-like protein)	H15-H57-E58-E61-E130; E61-H80-H126-E130	2 Fe cations	Substrate - regulation		Cytoplasm	No		iron ion homeostasis [GO:0055072]; positive regulation of cellular protein catabolic process [GO:1903364]; protein polyubiquitination [GO:0000209]; protein ubiquitination [GO:0016567]; SCF-dependent proteasomal ubiquitin-dependent protein catabolic process [GO:0031146]
28	Q9BXU8	FHL17_HUMAN	FTHL17	Ferritin heavy polypeptide-like 17 (Cancer/testis antigen 38) (CT38)	E28-D45-E50-E65-H66-E108-E135-Q142	Several Fe cations	Substrate - storage/transport		Unknown	No		intracellular sequestering of iron ion [GO:0006880]; iron ion transport [GO:0006826]
29	P0C7X4	FHL19_HUMAN	FTH1P19 FTHL19	Putative ferritin heavy polypeptide-like 19 (Ferritin heavy polypeptide 1 pseudogene 19)	D6-E13-E25-E28-E32-D95-D100	Several Fe cations	Substrate - storage/transport		Unknown	No		intracellular sequestering of iron ion [GO:0006880]; iron ion transport [GO:0006826]
30	Q16595	FRDA_HUMAN	FXN FRDA X25	Frataxin, mitochondrial (EC 1.16.3.1) (Friedreich ataxia protein) (Fxn) [Cleaved into: Frataxin intermediate form (i-FXN); Frataxin(56-210) (m56-FXN); Frataxin(78-210) (d-FXN) (m78-FXN); Frataxin mature form (Frataxin(81-210)) (m81-FXN)]	Unknown	1 Fe cation	Substrate - storage/transport	1.16.3.1	Cytoplasm, Mitochondrion	No	DISEASE: Friedreich ataxia (FRDA) [MIM:229300]: Autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region. {ECO:0000269|PubMed:10732799, ECO:0000269|PubMed:10874325, ECO:0000269|PubMed:19629184, ECO:0000269|PubMed:9150176, ECO:0000269|PubMed:9779809, ECO:0000269|PubMed:9989622, ECO:0000269|Ref.35, ECO:0000269|Ref.7, ECO:0000269|Ref.8}. Note=The disease is caused by mutations affecting the gene represented in this entry.	adult walking behavior [GO:0007628]; aerobic respiration [GO:0009060]; cellular iron ion homeostasis [GO:0006879]; cellular response to hydrogen peroxide [GO:0070301]; embryo development ending in birth or egg hatching [GO:0009792]; heme biosynthetic process [GO:0006783]; ion transport [GO:0006811]; iron incorporation into metallo-sulfur cluster [GO:0018283]; mitochondrion organization [GO:0007005]; negative regulation of apoptotic process [GO:0043066]; negative regulation of multicellular organism growth [GO:0040015]; negative regulation of organ growth [GO:0046621]; negative regulation of release of cytochrome c from mitochondria [GO:0090201]; oxidative phosphorylation [GO:0006119]; positive regulation of aconitate hydratase activity [GO:1904234]; positive regulation of catalytic activity [GO:0043085]; positive regulation of cell growth [GO:0030307]; positive regulation of cell proliferation [GO:0008284]; positive regulation of lyase activity [GO:0051349]; positive regulation of succinate dehydrogenase activity [GO:1904231]; proprioception [GO:0019230]; protein autoprocessing [GO:0016540]; regulation of ferrochelatase activity [GO:0010722]; response to iron ion [GO:0010039]; small molecule metabolic process [GO:0044281]
31	P02794	FRIH_HUMAN	FTH1 FTH FTHL6 OK/SW-cl.84 PIG15	Ferritin heavy chain (Ferritin H subunit) (EC 1.16.3.1) (Cell proliferation-inducing gene 15 protein) [Cleaved into: Ferritin heavy chain, N-terminally processed]	E28-D43-H58-Q59-E62-E63-E65-H66-E108-D132-Q142	Several Fe cations	Substrate - storage/transport	1.16.3.1	Unknown	No	DISEASE: Hemochromatosis 5 (HFE5) [MIM:615517]: A disorder of iron metabolism characterized by iron overload. Excess iron is deposited in a variety of organs leading to their failure, and resulting in serious illnesses including cirrhosis, hepatomas, diabetes, cardiomyopathy, arthritis, and hypogonadotropic hypogonadism. Severe effects of the disease usually do not appear until after decades of progressive iron loading. {ECO:0000269|PubMed:11389486}. Note=The disease is caused by mutations affecting the gene represented in this entry. In a Japanese family affected by HFE5, a single point mutation has been detected in the iron-responsive element (IRE) in the 5'-UTR of FTH1 mRNA. This mutation leads to an increased binding affinity for iron regulatory protein and thereby to the efficient suppression of mRNA translation.	cellular iron ion homeostasis [GO:0006879]; immune response [GO:0006955]; intracellular sequestering of iron ion [GO:0006880]; iron ion import [GO:0097286]; negative regulation of cell proliferation [GO:0008285]; negative regulation of fibroblast proliferation [GO:0048147]; neutrophil degranulation [GO:0043312]

32	P02792	FRIL_HUMAN	FTL	Ferritin light chain (Ferritin L subunit)	D39-D41-E46-E54-E57-E58-E61-E64-D128-E131	Several Fe cations	Substrate - storage/transport		Unknown	No	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. {ECO:0000269|PubMed:19176363}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Neurodegeneration with brain iron accumulation 3 (NBIA3) [MIM:606159]: A neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. It is characterized by a variety of neurological signs including parkinsonism, ataxia, corticospinal signs, mild non-progressive cognitive deficit and episodic psychosis. It is linked with decreased serum ferritin levels. {ECO:0000269|PubMed:16116125}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: L-ferritin deficiency (LFTD) [MIM:615604]: A condition characterized by low levels of ferritin in serum and tissues in the absence of other hematological symptoms. Seizures and mild neuropsychologic impairment may manifest in individuals with complete ferritin deficiency. {ECO:0000269|PubMed:23940258}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular iron ion homeostasis [GO:0006879]; intracellular sequestering of iron ion [GO:0006880]; iron ion homeostasis [GO:0055072]; iron ion transport [GO:0006826]; neutrophil degranulation [GO:0043312]
33	Q8N4E7	FTMT_HUMAN	FTMT	Ferritin, mitochondrial (EC 1.16.3.1)	E87-D104-H117-Q118-E121-E122-E124-H125-E167-D191-Q201-H233	Several Fe cations	Substrate - storage/transport	1.16.3.1	Mitochondrion	No		cellular iron ion homeostasis [GO:0006879]; intracellular sequestering of iron ion [GO:0006880]; iron ion transport [GO:0006826]; positive regulation of aconitate hydratase activity [GO:1904234]; positive regulation of cell proliferation [GO:0008284]; positive regulation of lyase activity [GO:0051349]; positive regulation of succinate dehydrogenase activity [GO:1904231]
34	Q9C0B1	FTO_HUMAN	FTO KIAA1752	Alpha-ketoglutarate-dependent dioxygenase FTO (EC 1.14.11.-) (Fat mass and obesity-associated protein)	H231-D233-H307	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No	DISEASE: Growth retardation, developmental delay, and facial dysmorphism (GDFD) [MIM:612938]: A severe polymalformation syndrome characterized by postnatal growth retardation, microcephaly, severe psychomotor delay, functional brain deficits and characteristic facial dysmorphism. In some patients, structural brain malformations, cardiac defects, genital anomalies, and cleft palate are observed. Early death occurs by the age of 3 years. {ECO:0000269|PubMed:19559399, ECO:0000269|PubMed:22002720, ECO:0000269|PubMed:26378117, ECO:0000269|PubMed:26697951}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. {ECO:0000269|PubMed:26287746}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A pathogenic intronic FTO variation (rs1421085) disrupts an evolutionarily conserved motif for ARID5B binding. Loss of ARID5B binding results in overexpression of two genes distal to FTO, IRX3 and IRX5. IRX3 and IRX5 overexpression shifts pre-adipocytes differentiation from brown to white fat cells, resulting in increased lipid storage and loss of mitochondrial thermogenesis. {ECO:0000269|PubMed:26287746}.	adipose tissue development [GO:0060612]; DNA dealkylation involved in DNA repair [GO:0006307]; DNA demethylation [GO:0080111]; oxidative demethylation [GO:0070989]; oxidative single-stranded DNA demethylation [GO:0035552]; oxidative single-stranded RNA demethylation [GO:0035553]; regulation of brown fat cell differentiation [GO:0090335]; regulation of lipid storage [GO:0010883]; regulation of multicellular organism growth [GO:0040014]; regulation of respiratory system process [GO:0044065]; regulation of white fat cell proliferation [GO:0070350]; RNA repair [GO:0042245]; temperature homeostasis [GO:0001659]

35	P07902	GALT_HUMAN	GALT	Galactose-1-phosphate uridylyltransferase (Gal-1-P uridylyltransferase) (EC 2.7.7.12) (UDP-glucose--hexose-1-phosphate uridylyltransferase)	H301-H319-H321	1 Fe cation	Catalytic	2.7.7.12	Unknown	No	DISEASE: Galactosemia (GALCT) [MIM:230400]: Inherited disorder of galactose metabolism that causes jaundice, cataracts, and mental retardation. {ECO:0000269|PubMed:10220154, ECO:0000269|PubMed:11754113, ECO:0000269|PubMed:11919338, ECO:0000269|PubMed:1373122, ECO:0000269|PubMed:1427861, ECO:0000269|PubMed:15841485, ECO:0000269|PubMed:1610789, ECO:0000269|PubMed:17041746, ECO:0000269|PubMed:17876724, ECO:0000269|PubMed:18956253, ECO:0000269|PubMed:1897530, ECO:0000269|PubMed:2011574, ECO:0000269|PubMed:22461411, ECO:0000269|PubMed:23022339, ECO:0000269|PubMed:25592817, ECO:0000269|PubMed:25614870, ECO:0000269|PubMed:7550229, ECO:0000269|PubMed:7887416, ECO:0000269|PubMed:7887417, ECO:0000269|PubMed:8112740, ECO:0000269|PubMed:8499924, ECO:0000269|PubMed:8598637, ECO:0000269|PubMed:8741038, ECO:0000269|PubMed:8869397,	galactose catabolic process [GO:0019388]; galactose metabolic process [GO:0006012]; UDP-glucose catabolic process [GO:0006258]
36	P09211	GSTP1_HUMAN	GSTP1 FAEES3 GST3	Glutathione S-transferase P (EC 2.5.1.18) (GST class-pi) (GSTP1-1)	Y8	1 Fe cation	Regulation - catalysis	2.5.1.18	Cytoplasm, Mitochondrion, Nucleus	No		animal organ regeneration [GO:0031100]; cellular response to cell-matrix adhesion [GO:0071460]; cellular response to epidermal growth factor stimulus [GO:0071364]; cellular response to glucocorticoid stimulus [GO:0071385]; cellular response to insulin stimulus [GO:0032869]; cellular response to lipopolysaccharide [GO:0071222]; central nervous system development [GO:0007417]; common myeloid progenitor cell proliferation [GO:0035726]; glutathione derivative biosynthetic process [GO:1901687]; glutathione metabolic process [GO:0006749]; linoleic acid metabolic process [GO:0043651]; negative regulation of acute inflammatory response [GO:0002674]; negative regulation of apoptotic process [GO:0043066]; negative regulation of biosynthetic process [GO:0009890]; negative regulation of ERK1 and ERK2 cascade [GO:0070373]; negative regulation of extrinsic apoptotic signaling pathway [GO:2001237]; negative regulation of fibroblast proliferation [GO:0048147]; negative regulation of I-kappaB kinase/NF-kappaB signaling [GO:0043124]; negative regulation of interleukin-1 beta production [GO:0032691]; negative regulation of JUN kinase activity [GO:0043508]; negative regulation of leukocyte proliferation [GO:0070664]; negative regulation of MAPK cascade [GO:0043409]; negative regulation of MAP kinase activity [GO:0043407]; negative regulation of monocyte chemotactic protein-1 production [GO:0071638]; negative regulation of nitric-oxide synthase biosynthetic process [GO:0051771]; negative regulation of protein kinase activity [GO:0006469]; negative regulation of smooth muscle cell chemotaxis [GO:0071672]; negative regulation of stress-activated MAPK cascade [GO:0032873]; negative regulation of tumor necrosis factor-mediated signaling pathway [GO:0010804]; negative regulation of tumor necrosis factor production [GO:0032720]; negative regulation of vascular smooth muscle cell proliferation [GO:1904706]; neutrophil degranulation [GO:0043312]; nitric

37	O43593	HAIR_HUMAN	HR	Lysine-specific demethylase hairless (EC 1.14.11.-)	C1007-E1009-H1125	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No	DISEASE: Alopecia universalis congenita (ALUNC) [MIM:203655]: A rare disorder characterized by loss of hair from the entire body. No hair are present in hair follicles on skin biopsy. {ECO:0000269|PubMed:12406339, ECO:0000269|PubMed:24334705, ECO:0000269|PubMed:9445480, ECO:0000269|PubMed:9736769}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Atrichia with papular lesions (APL) [MIM:209500]: An autosomal recessive disease characterized by papillary lesions over most of the body and almost complete absence of hair. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Hypotrichosis 4 (HYPT4) [MIM:146550]: An autosomal dominant condition characterized by reduced amount of hair, alopecia, little or no eyebrows, eyelashes or body hair, and coarse, wiry, twisted hair in early childhood. {ECO:0000269|PubMed:19122663, ECO:0000269|PubMed:24961381}. Note=The disease is caused by mutations affecting the gene represented in this entry.	histone H3-K9 demethylation [GO:0033169]; negative regulation of transcription, DNA-templated [GO:0045892]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]
38	Q96EW2	HBAP1_HUMAN	HSPBAP1 PASS1	HSPB1-associated protein 1 (27 kDa heat shock protein-associated protein 1) (Protein associated with small stress protein 1)	H175-D177-H257	1 Fe cation	Catalytic		Cytoplasm	No	DISEASE: Note=A chromosomal aberration involving HSPBAP1 has been found in a family with renal carcinoma (PubMed:12939738). Translocation t(2;3)(q35;q21) with the putative pseudogene DIRC3 (PubMed:12939738). Produces a hybrid mRNA encoding a truncated HSPBAP1 lacking the first 36 amino acids (PubMed:12939738). {ECO:0000269|PubMed:12939738}.	
39	P22830	HEMH_HUMAN	FECH	Ferrochelatase, mitochondrial (EC 4.99.1.1) (Heme synthase) (Protoheme ferro-lyase)	H263-E343	1 Fe cation	Substrate - biosynthesis	4.99.1.1	Mitochondrion	Yes	DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Erythropoietic protoporphyria is marked by excessive protoporphyrin in erythrocytes, plasma, liver and feces, and by widely varying photosensitive skin changes ranging from a burning or pruritic sensation to erythema, edema and wheals. {ECO:0000269|PubMed:10942404, ECO:0000269|PubMed:11375302, ECO:0000269|PubMed:12063482, ECO:0000269|PubMed:12601550, ECO:0000269|PubMed:1376018, ECO:0000269|PubMed:15286165, ECO:0000269|PubMed:17196862, ECO:0000269|PubMed:1755842, ECO:0000269|PubMed:7910885, ECO:0000269|PubMed:8757534, ECO:0000269|PubMed:9211198, ECO:0000269|PubMed:9585598, ECO:0000269|PubMed:9740232}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular response to dexamethasone stimulus [GO:0071549]; generation of precursor metabolites and energy [GO:0006091]; heme biosynthetic process [GO:0006783]; protoporphyrinogen IX metabolic process [GO:0046501]; response to arsenic-containing substance [GO:0046685]; response to drug [GO:0042493]; response to ethanol [GO:0045471]; response to insecticide [GO:0017085]; response to lead ion [GO:0010288]; response to light stimulus [GO:0009416]; response to methylmercury [GO:0051597]; response to platinum ion [GO:0070541]

40	P81172	HEPC_HUMAN	HAMP HEPC LEAP1 UNQ487/PRO1003	Hepcidin (Liver-expressed antimicrobial peptide 1) (LEAP-1) (Putative liver tumor regressor) (PLTR) [Cleaved into: Hepcidin-25 (Hepc25); Hepcidin-20 (Hepc20)]	Unknown	Unknown	Substrate - regulation		Extracellular space	No	DISEASE: Hemochromatosis 2B (HFE2B) [MIM:613313]: A juvenile form of hemochromatosis, a disorder of iron metabolism with excess deposition of iron in a variety of organs leading to their failure, bronze skin pigmentation, hepatic cirrhosis, arthropathy and diabetes. The most common symptoms of juvenile hemochromatosis at presentation are hypogonadism and cardiomyopathy. {ECO:0000269|PubMed:12915468, ECO:0000269|PubMed:14630809, ECO:0000269|PubMed:14633868, ECO:0000269|PubMed:14670915, ECO:0000269|PubMed:15099344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	acute-phase response [GO:0006953]; aging [GO:0007568]; antimicrobial humoral immune response mediated by antimicrobial peptide [GO:0061844]; cellular iron ion homeostasis [GO:0006879]; cellular response to bile acid [GO:1903413]; cellular response to interleukin-6 [GO:0071354]; cellular response to lipopolysaccharide [GO:0071222]; cellular response to tumor necrosis factor [GO:0071356]; cellular response to X-ray [GO:0071481]; defense response to bacterium [GO:0042742]; defense response to fungus [GO:0050832]; defense response to Gram-negative bacterium [GO:0050829]; defense response to Gram-positive bacterium [GO:0050830]; immune response [GO:0006955]; killing of cells of other organism [GO:0031640]; liver regeneration [GO:0097421]; multicellular organismal iron ion homeostasis [GO:0060586]; negative regulation of ferrous iron export [GO:1904039]; negative regulation of intestinal absorption [GO:1904479]; negative regulation of ion transmembrane transporter activity [GO:0032413]; negative regulation of iron channel activity [GO:1904255]; negative regulation of iron ion transmembrane transport [GO:0034760]; negative regulation of transcription by RNA polymerase II [GO:0000122]; positive regulation of cell growth involved in cardiac muscle cell development [GO:0061051]; positive regulation of protein polyubiquitination [GO:1902916]; positive regulation of receptor catabolic process [GO:2000646]; positive regulation of receptor internalization [GO:0002092]; response to erythropoietin [GO:0036017]; response to ethanol [GO:0045471]; response to iron ion [GO:0010039]; response to iron ion starvation [GO:1990641]; response to vitamin A [GO:0033189]; response to zinc ion [GO:0010043]
41	Q93099	HGD_HUMAN	HGD HGO	Homogentisate 1,2-dioxygenase (EC 1.13.11.5) (Homogentisate oxygenase) (Homogentisic acid oxidase) (Homogentisicase)	H335-E341-H371	1 Fe cation	Catalytic	1.13.11.5	Unknown	No	DISEASE: Alkaptonuria (AKU) [MIM:203500]: An autosomal recessive error of metabolism characterized by an increase in the level of homogentisic acid. The clinical manifestations are urine that turns dark on standing and alkalinization, black ochronotic pigmentation of cartilage and collagenous tissues, and spine arthritis. {ECO:0000269|PubMed:10205262, ECO:0000269|PubMed:10340975, ECO:0000269|PubMed:10482952, ECO:0000269|PubMed:10594001, ECO:0000269|PubMed:19862842, ECO:0000269|PubMed:21437689, ECO:0000269|PubMed:23353776, ECO:0000269|PubMed:23430897, ECO:0000269|PubMed:25681086, ECO:0000269|PubMed:8782815, ECO:0000269|PubMed:9154114, ECO:0000269|PubMed:9529363, ECO:0000269|PubMed:9630082}. Note=The disease is caused by mutations affecting the gene represented in this entry.	L-phenylalanine catabolic process [GO:0006559]; tyrosine catabolic process [GO:0006572]
42	Q9NWT6	HIF1N_HUMAN	HIF1AN FIH1	Hypoxia-inducible factor 1-alpha inhibitor (EC 1.14.11.30) (EC 1.14.11.n4) (Factor inhibiting HIF-1) (FIH-1) (Hypoxia-inducible factor asparagine hydroxylase)	H199-D201-H279	1 Fe cation	Catalytic	1.14.11.30; 1.14.11.n4	Cytoplasm, Nucleus	No		negative regulation of Notch signaling pathway [GO:0045746]; negative regulation of transcription from RNA polymerase II promoter in response to hypoxia [GO:0061428]; oxidation-reduction process [GO:0055114]; peptidyl-asparagine hydroxylation [GO:0042265]; peptidyl-aspartic acid hydroxylation [GO:0042264]; peptidyl-histidine hydroxylation [GO:0036138]; positive regulation of myoblast differentiation [GO:0045663]; positive regulation of vasculogenesis [GO:2001214]; regulation of transcription from RNA polymerase II promoter in response to hypoxia [GO:0061418]; transcription, DNA-templated [GO:0006351]
43	Q8IWW8	HOT_HUMAN	ADHFE1 HMFT2263	Hydroxyacid-oxoacid transhydrogenase, mitochondrial (HOT) (EC 1.1.99.24) (Alcohol dehydrogenase iron-containing protein 1) (ADHFe1) (Fe-containing alcohol dehydrogenase)	D242-H246-H330-H357	1 Fe cation	Catalytic	1.1.99.24	Mitochondrion	No		2-oxoglutarate metabolic process [GO:0006103]; molecular hydrogen transport [GO:0015993]
44	Q96IR7	HPDL_HUMAN	HPDL GLOXD1	4-hydroxyphenylpyruvate dioxygenase-like protein (EC 1.13.-.-) (Glyoxalase domain-containing protein 1)	H163-H258-E339	1 Fe cation	Catalytic	1.13.-.-	Unknown	No		aromatic amino acid family metabolic process [GO:0009072]

45	P32754	HPPD_HUMAN	HPD PPD	4-hydroxyphenylpyruvate dioxygenase (EC 1.13.11.27) (4-hydroxyphenylpyruvic acid oxidase) (4HPPD) (HPD) (HPPDase)	H183-H266-E349	1 Fe cation	Catalytic	1.13.11.27	Unknown	No	DISEASE: Tyrosinemia 3 (TYRSN3) [MIM:276710]: An inborn error of metabolism characterized by elevations of tyrosine in the blood and urine, seizures and mild mental retardation. {ECO:0000269|PubMed:10942115, ECO:0000269|PubMed:11073718}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Hawkinsinuria (HAWK) [MIM:140350]: An inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin, in the urine. {ECO:0000269|PubMed:11073718}. Note=The disease is caused by mutations affecting the gene represented in this entry.	L-phenylalanine catabolic process [GO:0006559]; tyrosine catabolic process [GO:0006572]
46	Q96NU7	HUTI_HUMAN	AMDHD1 HMFT1272	Probable imidazolonepropionase (EC 3.5.2.7) (Amidohydrolase domain-containing protein 1)	H87-H89-H260-D334	1 Fe or Zn cation	Catalytic - no redox	3.5.2.7	Unknown	No		histidine catabolic process [GO:0006548]; histidine catabolic process to glutamate and formamide [GO:0019556]; histidine catabolic process to glutamate and formate [GO:0019557]
47	Q15652	JHD2C_HUMAN	JMJD1C JHDM2C KIAA1380 TRIP8	Probable JmjC domain-containing histone demethylation protein 2C (EC 1.14.11.-) (Jumonji domain-containing protein 1C) (Thyroid receptor-interacting protein 8) (TR-interacting protein 8) (TRIP-8)	H2336-E2338-H2466	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		blood coagulation [GO:0007596]; histone H3-K9 demethylation [GO:0033169]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]
48	Q9H9V9	JMJD4_HUMAN	JMJD4	JmjC domain-containing protein 4 (Jumonji domain-containing protein 4)	H235-D237-H315	1 Fe cation	Catalytic		Unknown	No		
49	Q6NYC1	JMJD6_HUMAN	JMJD6 KIAA0585 PTDSR	Bifunctional arginine demethylase and lysyl-hydroxylase JMJD6 (EC 1.14.11.-) (Histone arginine demethylase JMJD6) (JmjC domain-containing protein 6) (Jumonji domain-containing protein 6) (Lysyl-hydroxylase JMJD6) (Peptide-lysine 5-dioxygenase JMJD6) (Phosphatidylserine receptor) (Protein PTDSR)	H187-D189-H273	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		cell surface receptor signaling pathway [GO:0007166]; erythrocyte development [GO:0048821]; heart development [GO:0007507]; histone H3-R2 demethylation [GO:0070078]; histone H4-R3 demethylation [GO:0070079]; kidney development [GO:0001822]; lung development [GO:0030324]; macrophage activation [GO:0042116]; mRNA processing [GO:0006397]; peptidyl-lysine hydroxylation to 5-hydroxy-L-lysine [GO:0018395]; recognition of apoptotic cell [GO:0043654]; regulation of mRNA splicing, via spliceosome [GO:0048024]; regulation of transcription, DNA-templated [GO:0006355]; retina development in camera-type eye [GO:0060041]; RNA splicing [GO:0008380]; sprouting angiogenesis [GO:0002040]; T cell differentiation in thymus [GO:0033077]; transcription, DNA-templated [GO:0006351]
50	P0C870	JMJD7_HUMAN	JMJD7	JmjC domain-containing protein 7 (Jumonji domain-containing protein 7)	H178-D180-H277	1 Fe cation	Catalytic		Unknown	No		
51	Q96S16	JMJD8_HUMAN	JMJD8 C16orf20 PP14397	JmjC domain-containing protein 8 (Jumonji domain-containing protein 8)	H249-H251-H318	1 Fe cation	Catalytic		Unknown	No		
52	Q9P272	K1456_HUMAN	KIAA1456 C8orf79	Probable tRNA methyltransferase 9-like protein (TRM9L) (EC 2.1.1.-)	H112	1 Fe cation	Catalytic	2.1.1.-	Unknown	No		tRNA modification [GO:0006400]; tRNA wobble uridine modification [GO:0002098]
53	Q9Y2K7	KDM2A_HUMAN	KDM2A CXXC8 FBL7 FBXL11 JHDM1A KIAA1004	Lysine-specific demethylase 2A (EC 1.14.11.27) (CXXC-type zinc finger protein 8) (F-box and leucine-rich repeat protein 11) (F-box protein FBL7) (F-box protein Lilina) (F-box/LRR-repeat protein 11) (JmjC domain-containing histone demethylation protein 1A) ([Histone-H3]-lysine-36 demethylase 1A)	H212-D214-Y222-H284	1 Fe cation	Catalytic	1.14.11.27	Nucleus	No		double-strand break repair via nonhomologous end joining [GO:0006303]; histone H3-K36 demethylation [GO:0070544]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]

54	Q8NHM5	KDM2B_HUMAN	KDM2B CXXC2 FBL10 FBXL10 JHDM1B PCCX2	Lysine-specific demethylase 2B (EC 1.14.11.27) (CXXC-type zinc finger protein 2) (F-box and leucine-rich repeat protein 10) (F-box protein FBL10) (F-box/LRR-repeat protein 10) (JmjC domain-containing histone demethylation protein 1B) (Jumonji domain-containing EMSY-interactor methyltransferase motif protein) (Protein JEMMA) (Protein-containing CXXC domain 2) ([Histone-H3]-lysine-36 demethylase 1B)	H242-D244-H314	1 Fe cation	Catalytic	1.14.11.27	Nucleus	No		embryonic camera-type eye morphogenesis [GO:0048596]; forebrain development [GO:0030900]; fourth ventricle development [GO:0021592]; hindbrain development [GO:0030902]; histone H2A monoubiquitination [GO:0035518]; initiation of neural tube closure [GO:0021993]; lateral ventricle development [GO:0021670]; midbrain development [GO:0030901]; midbrain-hindbrain boundary morphogenesis [GO:0021555]; negative regulation of neural precursor cell proliferation [GO:2000178]; negative regulation of neuron apoptotic process [GO:0043524]; negative regulation of transcription from RNA polymerase II promoter [GO:0000122]; positive regulation of cell growth [GO:0030307]; positive regulation of stem cell population maintenance [GO:1902459]; spermatogenesis [GO:0007283]; third ventricle development [GO:0021678]; transcription, DNA-templated [GO:0006351]
55	Q9Y4C1	KDM3A_HUMAN	KDM3A JHDM2A JMJD1 JMJD1A KIAA0742 TSGA	Lysine-specific demethylase 3A (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 2A) (Jumonji domain-containing protein 1A)	H1120-D1122-H1249	1 Fe cation	Catalytic	1.14.11.-	Cytoplasm, Nucleus	No		androgen receptor signaling pathway [GO:0030521]; formaldehyde biosynthetic process [GO:0046293]; histone H3-K9 demethylation [GO:0033169]; histone H3-K9 dimethylation [GO:0036123]; hormone-mediated signaling pathway [GO:0009755]; negative regulation of histone H3-K9 methylation [GO:0051573]; positive regulation of transcription, DNA-templated [GO:0045893]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; regulation of stem cell differentiation [GO:2000736]; regulation of stem cell population maintenance [GO:2000036]; spermatid nucleus elongation [GO:0007290]; transcription, DNA-templated [GO:0006351]
56	Q7LBC6	KDM3B_HUMAN	KDM3B C5orf7 JHDM2B JMJD1B KIAA1082	Lysine-specific demethylase 3B (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 2B) (Jumonji domain-containing protein 1B) (Nuclear protein 5qNCA)	H1604-H1689	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		histone H3-K9 demethylation [GO:0033169]; regulation of transcription, DNA-templated [GO:0006355]; response to cisplatin [GO:0072718]; transcription, DNA-templated [GO:0006351]
57	O75164	KDM4A_HUMAN	KDM4A JHDM3A JMJD2 JMJD2A KIAA0677	Lysine-specific demethylase 4A (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 3A) (Jumonji domain-containing protein 2A)	H188-E190-H276	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		cardiac muscle hypertrophy in response to stress [GO:0014898]; histone demethylation [GO:0016577]; negative regulation of astrocyte differentiation [GO:0048712]; negative regulation of autophagy [GO:0010507]; negative regulation of cell death [GO:0060548]; negative regulation of gene expression [GO:0010629]; negative regulation of histone H3-K9 trimethylation [GO:1900113]; negative regulation of transcription, DNA-templated [GO:0045892]; positive regulation of gene expression [GO:0010628]; positive regulation of neuron differentiation [GO:0045666]; response to nutrient levels [GO:0031667]; transcription, DNA-templated [GO:0006351]; viral process [GO:0016032]
58	O94953	KDM4B_HUMAN	KDM4B JHDM3B JMJD2B KIAA0876	Lysine-specific demethylase 4B (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 3B) (Jumonji domain-containing protein 2B)	H189-E191-H277	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]
59	Q9H3R0	KDM4C_HUMAN	KDM4C GASC1 JHDM3C JMJD2C KIAA0780	Lysine-specific demethylase 4C (EC 1.14.11.-) (Gene amplified in squamous cell carcinoma 1 protein) (GASC-1 protein) (JmjC domain-containing histone demethylation protein 3C) (Jumonji domain-containing protein 2C)	H190-E192-H278	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		blastocyst formation [GO:0001825]; histone H3-K9 demethylation [GO:0033169]; negative regulation of histone H3-K9 trimethylation [GO:1900113]; positive regulation of cell proliferation [GO:0008284]; positive regulation of gene expression [GO:0010628]; positive regulation of neuron differentiation [GO:0045666]; regulation of stem cell differentiation [GO:2000736]; regulation of stem cell population maintenance [GO:2000036]; regulation of transcription from RNA polymerase II promoter [GO:0006357]; transcription, DNA-templated [GO:0006351]
60	Q6B0I6	KDM4D_HUMAN	KDM4D JHDM3D JMJD2D	Lysine-specific demethylase 4D (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 3D) (Jumonji domain-containing protein 2D)	H192-E194-H280	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		cellular response to ionizing radiation [GO:0071479]; double-strand break repair via homologous recombination [GO:0000724]; histone H3-K9 demethylation [GO:0033169]; negative regulation of histone H3-K9 trimethylation [GO:1900113]; positive regulation of chromatin binding [GO:0035563]; positive regulation of double-strand break repair via nonhomologous end joining [GO:2001034]; regulation of protein phosphorylation [GO:0001932]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]

61	B2RXH2	KDM4E_HUMAN	KDM4E KDM4DL	Lysine-specific demethylase 4E (EC 1.14.11.-) (KDM4D-like protein) (Lysine-specific demethylase 4D-like)	H189-E191-H277	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		covalent chromatin modification [GO:0016569]; regulation of transcription, DNA-templated [GO:0006355]; transcription, DNA-templated [GO:0006351]
62	P29375	KDM5A_HUMAN	KDM5A JARID1A RBBP2 RBP2	Lysine-specific demethylase 5A (EC 1.14.11.-) (Histone demethylase JARID1A) (Jumonji/ARID domain-containing protein 1A) (Retinoblastoma-binding protein 2) (RBBP-2)	H483-E485-H571	1 Fe cation	Catalytic	1.14.11.-	Mitochondrion, Nucleus	No		circadian regulation of gene expression [GO:0032922]; histone H3-K4 demethylation [GO:0034720]; male gonad development [GO:0008584]; negative regulation of histone deacetylase activity [GO:1901726]; negative regulation of transcription from RNA polymerase II promoter [GO:0000122]; positive regulation of transcription, DNA-templated [GO:0045893]; regulation of sequence-specific DNA binding transcription factor activity [GO:0051090]; spermatogenesis [GO:0007283]; transcription from RNA polymerase II promoter [GO:0006366]
63	Q9UGL1	KDM5B_HUMAN	KDM5B JARID1B PLU1 RBBP2H1	Lysine-specific demethylase 5B (EC 1.14.11.-) (Cancer/testis antigen 31) (CT31) (Histone demethylase JARID1B) (Jumonji/ARID domain-containing protein 1B) (PLU-1) (Retinoblastoma-binding protein 2 homolog 1) (RBP2-H1)	H499-E501-H587	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		branching involved in mammary gland duct morphogenesis [GO:0060444]; cellular response to fibroblast growth factor stimulus [GO:0044344]; histone H3-K4 demethylation [GO:0034720]; lens fiber cell differentiation [GO:0070306]; mammary duct terminal end bud growth [GO:0060763]; negative regulation of transcription, DNA-templated [GO:0045892]; positive regulation of gene expression [GO:0010628]; positive regulation of mammary gland epithelial cell proliferation [GO:0033601]; post-embryonic development [GO:0009791]; regulation of estradiol secretion [GO:2000864]; regulation of transcription from RNA polymerase II promoter [GO:0006357]; response to fungicide [GO:0060992]; rhythmic process [GO:0048511]; single fertilization [GO:0007338]; transcription, DNA-templated [GO:0006351]; uterus morphogenesis [GO:0061038]
64	P41229	KDM5C_HUMAN	KDM5C DXS1272E JARID1C SMCX XE169	Lysine-specific demethylase 5C (EC 1.14.11.-) (Histone demethylase JARID1C) (Jumonji/ARID domain-containing protein 1C) (Protein SmcX) (Protein Xe169)	H514-E516-H602	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No	DISEASE: Mental retardation, X-linked, syndromic, Claes-Jensen type (MRXSCJ) [MIM:300534]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXSCJ patients manifest mental retardation associated with variable features such as slowly progressive spastic paraplegia, seizures, facial dysmorphism. {ECO:0000269|PubMed:15586325, ECO:0000269|PubMed:16538222, ECO:0000269|PubMed:16541399, ECO:0000269|PubMed:17320160, ECO:0000269|PubMed:17468742, ECO:0000269|PubMed:23356856, ECO:0000269|PubMed:25666439}. Note=The disease is caused by mutations affecting the gene represented in this entry.	histone H3-K4 demethylation [GO:0034720]; negative regulation of transcription, DNA-templated [GO:0045892]; response to toxic substance [GO:0009636]; rhythmic process [GO:0048511]; transcription, DNA-templated [GO:0006351]
65	Q9BY66	KDM5D_HUMAN	KDM5D HY HYA JARID1D KIAA0234 SMCY	Lysine-specific demethylase 5D (EC 1.14.11.-) (Histocompatibility Y antigen) (H-Y) (Histone demethylase JARID1D) (Jumonji/ARID domain-containing protein 1D) (Protein SmcY)	H504-E506-H592	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		histone H3-K4 demethylation [GO:0034720]; regulation of androgen receptor signaling pathway [GO:0060765]; regulation of transcription, DNA-templated [GO:0006355]; T cell antigen processing and presentation [GO:0002457]; transcription, DNA-templated [GO:0006351]
66	O15550	KDM6A_HUMAN	KDM6A UTX	Lysine-specific demethylase 6A (EC 1.14.11.-) (Histone demethylase UTX) (Ubiquitously-transcribed TPR protein on the X chromosome) (Ubiquitously-transcribed X chromosome tetratricopeptide repeat protein)	H1146-E1148-H1226	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No	DISEASE: Kabuki syndrome 2 (KABUK2) [MIM:300867]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269|PubMed:22197486}. Note=The disease is caused by mutations affecting the gene represented in this entry.	canonical Wnt signaling pathway [GO:0060070]; cardiovascular system development [GO:0072358]; heart morphogenesis [GO:0003007]; histone H3-K4 methylation [GO:0051568]; in utero embryonic development [GO:0001701]; mesodermal cell differentiation [GO:0048333]; multicellular organism growth [GO:0035264]; neural tube closure [GO:0001843]; notochord morphogenesis [GO:0048570]; positive regulation of gene expression [GO:0010628]; respiratory system process [GO:0003016]; somite rostral/caudal axis specification [GO:0032525]

67	O15054	KDM6B_HUMAN	KDM6B JMJD3 KIAA0346	Lysine-specific demethylase 6B (EC 1.14.11.-) (JmjC domain-containing protein 3) (Jumonji domain-containing protein 3) (Lysine demethylase 6B)	H1390-E1392-H1470	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		cardiac muscle cell differentiation [GO:0055007]; cell fate commitment [GO:0045165]; cellular response to hydrogen peroxide [GO:0070301]; endothelial cell differentiation [GO:0045446]; hippocampus development [GO:0021766]; inflammatory response to antigenic stimulus [GO:0002437]; mesodermal cell differentiation [GO:0048333]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; response to activity [GO:0014823]; response to fungicide [GO:0060992]
68	Q6ZMT4	KDM7A_HUMAN	KDM7A JHDM1D KDM7 KIAA1718	Lysine-specific demethylase 7A (EC 1.14.11.-) (JmjC domain-containing histone demethylation protein 1D) (Lysine-specific demethylase 7)	H282-D284-Y292-H354	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		histone H3-K27 demethylation [GO:0071557]; histone H3-K36 demethylation [GO:0070544]; histone H3-K9 demethylation [GO:0033169]; histone H4-K20 demethylation [GO:0035574]; midbrain development [GO:0030901]; positive regulation of transcription, DNA-templated [GO:0045893]; transcription, DNA-templated [GO:0006351]
69	Q8N371	KDM8_HUMAN	KDM8 JMJD5	Lysine-specific demethylase 8 (EC 1.14.11.27) (JmjC domain-containing protein 5) (Jumonji domain-containing protein 5)	H321-D323-H400	1 Fe cation	Catalytic	1.14.11.27	Nucleus	No		G2/M transition of mitotic cell cycle [GO:0000086]; histone H3-K36 demethylation [GO:0070544]; positive regulation of transcription, DNA-templated [GO:0045893]; transcription, DNA-templated [GO:0006351]
70	P18054	LOX12_HUMAN	ALOX12 12LO LOG12	Arachidonate 12-lipoxygenase, 12S-type (12S-LOX) (12S-lipoxygenase) (EC 1.13.11.31) (Lipoxin synthase 12-LO) (EC 3.3.2.-) (Platelet-type lipoxygenase 12)	H360-H365-H540	1 Fe cation	Catalytic	1.13.11.31; 3.3.2.-	Cytoplasm	Yes	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:17460548}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Gln at position 261 may confer interindividual susceptibility to esophageal cancer (PubMed:17460548). {ECO:0000269|PubMed:17460548}.; DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:17151091}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Gln at position 261 may confer interindividual susceptibility to colorectal cancer (PubMed:17460548). {ECO:0000269|PubMed:17460548}.	aging [GO:0007568]; arachidonic acid metabolic process [GO:0019369]; cellular response to lipid [GO:0071396]; establishment of skin barrier [GO:0061436]; fatty acid oxidation [GO:0019395]; hepoxilin biosynthetic process [GO:0051122]; hepoxilin metabolic process [GO:0051121]; leukotriene A4 metabolic process [GO:1901751]; linoleic acid metabolic process [GO:0043651]; lipoxin A4 biosynthetic process [GO:2001303]; lipoxin B4 biosynthetic process [GO:2001306]; lipoxin metabolic process [GO:2001300]; lipoxygenase pathway [GO:0019372]; movement of cell or subcellular component [GO:0006928]; negative regulation of apoptotic process [GO:0043066]; negative regulation of muscle cell apoptotic process [GO:0010656]; negative regulation of platelet aggregation [GO:0090331]; positive regulation of angiogenesis [GO:0045766]; positive regulation of blood vessel diameter [GO:0097755]; positive regulation of cell adhesion [GO:0045785]; positive regulation of cell growth [GO:0030307]; positive regulation of cell migration [GO:0030335]; positive regulation of cell proliferation [GO:0008284]; positive regulation of cysteine-type endopeptidase activity involved in apoptotic process [GO:0043280]; positive regulation of endothelial cell differentiation [GO:0045603]; positive regulation of endothelial cell migration [GO:0010595]; positive regulation of gene expression [GO:0010628]; positive regulation of mitochondrial depolarization [GO:0051901]; positive regulation of smooth muscle cell proliferation [GO:0048661]; reactive oxygen species metabolic process [GO:0072593]; superoxide anion generation [GO:0042554]
71	P16050	LOX15_HUMAN	ALOX15 LOG15	Arachidonate 15-lipoxygenase (15-LOX) (15-LOX-1) (EC 1.13.11.33) (12/15-lipoxygenase) (Arachidonate 12-lipoxygenase, leukocyte-type) (12-LOX) (EC 1.13.11.31) (Arachidonate omega-6 lipoxygenase)	H360-H365-H540	1 Fe cation	Catalytic	1.13.11.33; 1.13.11.31	Cytoplasm, Cell membrane	Yes	DISEASE: Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Met at position 560 may confer interindividual susceptibility to coronary artery disease (CAD) (PubMed:17959182). {ECO:0000269|PubMed:17959182}.	apoptotic cell clearance [GO:0043277]; arachidonic acid metabolic process [GO:0019369]; bone mineralization [GO:0030282]; cellular response to calcium ion [GO:0071277]; cellular response to interleukin-13 [GO:0035963]; hepoxilin biosynthetic process [GO:0051122]; inflammatory response [GO:0006954]; leukotriene metabolic process [GO:0006691]; lipoxin A4 biosynthetic process [GO:2001303]; lipoxygenase pathway [GO:0019372]; negative regulation of adaptive immune response [GO:0002820]; ossification [GO:0001503]; phosphatidylethanolamine biosynthetic process [GO:0006646]; positive regulation of actin filament polymerization [GO:0030838]; positive regulation of cell-substrate adhesion [GO:0010811]; positive regulation of ERK1 and ERK2 cascade [GO:0070374]; positive regulation of heterotypic cell-cell adhesion [GO:0034116]; regulation of engulfment of apoptotic cell [GO:1901074]; regulation of peroxisome proliferator activated receptor signaling pathway [GO:0035358]; response to endoplasmic reticulum stress [GO:0034976]; wound healing [GO:0042060]
72	P09917	LOX5_HUMAN	ALOX5 LOG5	Arachidonate 5-lipoxygenase (5-LO) (5-lipoxygenase) (EC 1.13.11.34)	H368-H373-H551	1 Fe cation	Catalytic	1.13.11.34	Cytoplasm, Nucleus	Yes		leukotriene biosynthetic process [GO:0019370]; leukotriene metabolic process [GO:0006691]; leukotriene production involved in inflammatory response [GO:0002540]; lipoxin metabolic process [GO:2001300]; lipoxygenase pathway [GO:0019372]; neutrophil degranulation [GO:0043312]

73	Q9BYJ1	LOXE3_HUMAN	ALOXE3	Hydroperoxide isomerase ALOXE3 (EC 5.4.4.7) (Epidermis-type lipoxygenase 3) (Epidermal LOX-3) (e-LOX-3) (eLOX-3) (Hydroperoxy icosatetraenoate dehydratase) (EC 4.2.1.152)	H408-H413-H588	1 Fe cation	Catalytic	5.4.4.7; 4.2.1.152	Cytoplasm	No	DISEASE: Ichthyosis, congenital, autosomal recessive 3 (ARCI3) [MIM:606545]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:11773004, ECO:0000269|PubMed:15629692, ECO:0000269|PubMed:16116617, ECO:0000269|PubMed:19131948, ECO:0000269|PubMed:19890349}. Note=The disease is caused by mutations affecting the gene represented in this entry.	arachidonic acid metabolic process [GO:0019369]; ceramide biosynthetic process [GO:0046513]; establishment of skin barrier [GO:0061436]; fat cell differentiation [GO:0045444]; hepoxilin biosynthetic process [GO:0051122]; linoleic acid metabolic process [GO:0043651]; lipoxygenase pathway [GO:0019372]; peroxisome proliferator activated receptor signaling pathway [GO:0035357]; sensory perception of pain [GO:0019233]; sphingolipid metabolic process [GO:0006665]
74	O75342	LX12B_HUMAN	ALOX12B	Arachidonate 12-lipoxygenase, 12R-type (12R-LOX) (12R-lipoxygenase) (EC 1.13.11.-) (Epidermis-type lipoxygenase 12)	H398-H403-H578	1 Fe cation	Catalytic	1.13.11.-	Cytoplasm	No	DISEASE: Ichthyosis, congenital, autosomal recessive 2 (ARCI2) [MIM:242100]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:11773004, ECO:0000269|PubMed:15629692, ECO:0000269|PubMed:16116617, ECO:0000269|PubMed:19131948, ECO:0000269|PubMed:19890349}. Note=The disease is caused by mutations affecting the gene represented in this entry.	arachidonic acid metabolic process [GO:0019369]; ceramide biosynthetic process [GO:0046513]; establishment of skin barrier [GO:0061436]; hepoxilin biosynthetic process [GO:0051122]; linoleic acid metabolic process [GO:0043651]; lipoxygenase pathway [GO:0019372]; oxidation-reduction process [GO:0055114]; positive regulation of gene expression [GO:0010628]; positive regulation of MAPK cascade [GO:0043410]; positive regulation of mucus secretion [GO:0070257]; protein lipidation [GO:0006497]; sphingolipid metabolic process [GO:0006665]
75	O15296	LX15B_HUMAN	ALOX15B	Arachidonate 15-lipoxygenase B (15-LOX-B) (EC 1.13.11.33) (15-lipoxygenase 2) (15-LOX-2) (Arachidonate 15-lipoxygenase type II) (Linoleate 13-lipoxygenase 15-LOb) (EC 1.13.11.-)	H373-H378-H553	1 Fe cation	Catalytic	1.13.11.33; 1.13.11.-	Nucleus	No		apoptotic process [GO:0006915]; arachidonic acid metabolic process [GO:0019369]; hepoxilin biosynthetic process [GO:0051122]; linoleic acid metabolic process [GO:0043651]; lipid metabolic process [GO:0006629]; lipoxygenase pathway [GO:0019372]; negative regulation of cell cycle [GO:0045786]; negative regulation of cell migration [GO:0030336]; negative regulation of cell proliferation [GO:0008285]; negative regulation of growth [GO:0045926]; positive regulation of chemokine secretion [GO:0090197]; positive regulation of keratinocyte differentiation [GO:0045618]; positive regulation of macrophage derived foam cell differentiation [GO:0010744]; positive regulation of peroxisome proliferator activated receptor signaling pathway [GO:0035360]; prostate gland development [GO:0030850]; regulation of epithelial cell differentiation [GO:0030856]
76	P53582	MAP11_HUMAN	METAP1 KIAA0094	Methionine aminopeptidase 1 (MAP 1) (MetAP 1) (EC 3.4.11.18) (Peptidase M 1)	D220-D231-H294-E327-E358	1 Divalent cation	Catalytic	3.4.11.18	Cytoplasm	No		N-terminal protein amino acid modification [GO:0031365]; peptidyl-methionine modification [GO:0018206]; platelet aggregation [GO:0070527]; regulation of rhodopsin mediated signaling pathway [GO:0022400]; regulation of translation [GO:0006417]

77	Q6UB28	MAP12_HUMAN	METAP1D MAP1D	Methionine aminopeptidase 1D, mitochondrial (MAP 1D) (MetAP 1D) (EC 3.4.11.18) (Methionyl aminopeptidase type 1D, mitochondrial) (Peptidase M 1D)	D178-D189-H252-E284-E315	1 Divalent cation	Catalytic	3.4.11.18	Mitochondrion	No		N-terminal protein amino acid modification [GO:0031365]; peptidyl-methionine modification [GO:0018206]
78	P50579	MAP2_HUMAN	METAP2 MNPEP P67EIF2	Methionine aminopeptidase 2 (MAP 2) (MetAP 2) (EC 3.4.11.18) (Initiation factor 2-associated 67 kDa glycoprotein) (p67) (p67eIF2) (Peptidase M)	D251-D262-H331-E364-E459	1 Divalent cation	Catalytic	3.4.11.18	Cytoplasm	No		N-terminal protein amino acid modification [GO:0031365]; peptidyl-methionine modification [GO:0018206]; protein processing [GO:0016485]; regulation of rhodopsin mediated signaling pathway [GO:0022400]
79	Q9NYZ2	MFRN1_HUMAN	SLC25A37 MFRN MSCP HT015	Mitoferrin-1 (Mitochondrial iron transporter 1) (Mitochondrial solute carrier protein) (Solute carrier family 25 member 37)	Unknown	Unknown	Substrate - transport		Mitochondrion	Yes		iron ion homeostasis [GO:0055072]; mitochondrial iron ion transport [GO:0048250]
80	Q96A46	MFRN2_HUMAN	SLC25A28 MFRN2 NPD016	Mitoferrin-2 (Mitochondrial RNA-splicing protein 3/4 homolog) (MRS3/4) (hMRS3/4) (Mitochondrial iron transporter 2) (Solute carrier family 25 member 28)	Unknown	Unknown	Substrate - transport		Mitochondrion	Yes		iron ion homeostasis [GO:0055072]; mitochondrial iron ion transport [GO:0048250]
81	Q9UGB7	MIOX_HUMAN	MIOX ALDRL6 KSP32 RSOR	Inositol oxygenase (EC 1.13.99.1) (Aldehyde reductase-like 6) (Kidney-specific protein 32) (Myo-inositol oxygenase) (MI oxygenase) (Renal-specific oxidoreductase)	H98-H123-D124-D253; D124-H194-H220	2 Fe cations	Catalytic	1.13.99.1	Cytoplasm	No		inositol catabolic process [GO:0019310]
82	O15442	MPPD1_HUMAN	MPPED1 C22orf1 FAM1A	Metallophosphoesterase domain-containing protein 1 (EC 3.1.-.-) (Adult brain protein 239) (239AB)	D97-H99-D118-H286; H245-H284-N149	2 Divalent cations	Catalytic	3.1.-.-	Unknown	No		
83	P49959	MRE11_HUMAN	MRE11 HNGS1 MRE11A	Double-strand break repair protein MRE11 (Double-strand break repair protein MRE11A) (Meiotic recombination 11 homolog 1) (MRE11 homolog 1) (Meiotic recombination 11 homolog A) (MRE11 homolog A)	D20-H22-D60	1 Fe cation	Catalytic		Nucleus	No	DISEASE: Ataxia-telangiectasia-like disorder 1 (ATLD1) [MIM:604391]: A rare disorder characterized by progressive cerebellar ataxia, dysarthria, abnormal eye movements, and absence of telangiectasia. ATLD patients show normal levels of total IgG, IgA and IgM, although there may be reduced levels of specific functional antibodies. At the cellular level, ATLD exhibits hypersensitivity to ionizing radiation and radioresistant DNA synthesis. {ECO:0000269|PubMed:10612394}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Note=Defects in MRE11 can be a cause of nephronophthisis-related ciliopathies (NPHP-RC), a group of recessive diseases that affect kidney, retina and brain. A homozygous truncating mutation MRE11 has been found in patients with cerebellar vermis hypoplasia, ataxia and dysarthria. {ECO:0000269|PubMed:22863007}.	cell proliferation [GO:0008283]; cellular response to DNA damage stimulus [GO:0006974]; DNA double-strand break processing [GO:0000729]; DNA duplex unwinding [GO:0032508]; DNA recombination [GO:0006310]; DNA repair [GO:0006281]; DNA replication [GO:0006260]; DNA synthesis involved in DNA repair [GO:0000731]; double-strand break repair [GO:0006302]; double-strand break repair via homologous recombination [GO:0000724]; double-strand break repair via nonhomologous end joining [GO:0006303]; intra-S DNA damage checkpoint [GO:0031573]; mitotic G2 DNA damage checkpoint [GO:0007095]; negative regulation of apoptotic process [GO:0043066]; negative regulation of DNA endoreduplication [GO:0032876]; positive regulation of kinase activity [GO:0033674]; positive regulation of protein autophosphorylation [GO:0031954]; positive regulation of telomere maintenance [GO:0032206]; positive regulation of type I interferon production [GO:0032481]; reciprocal meiotic recombination [GO:0007131]; regulation of mitotic recombination [GO:0000019]; regulation of signal transduction by p53 class mediator [GO:1901796]; sister chromatid cohesion [GO:0007062]; strand displacement [GO:0000732]; synapsis [GO:0007129]; telomere maintenance via telomerase [GO:0007004]; telomeric 3' overhang formation [GO:0031860]; viral process [GO:0016032]
84	Q15800	MSMO1_HUMAN	MSMO1 DESP4 ERG25 SC4MOL	Methylsterol monooxygenase 1 (EC 1.14.13.72) (C-4 methylsterol oxidase)	Unknown	1 Fe cation	Catalytic	1.14.13.72	Endoplasmic reticulum	Yes	DISEASE: Microcephaly, congenital cataract, and psoriasiform dermatitis (MCCPD) [MIM:616834]: An autosomal recessive inborn error of cholesterol metabolism characterized by accumulation of a large amount of methylsterols, particularly dimethylsterols, in affected individuals. Patients manifest psoriasiform dermatitis, arthralgias, congenital cataracts, microcephaly, and developmental delay. {ECO:0000269|PubMed:21285510, ECO:0000269|PubMed:24144731}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cholesterol biosynthetic process [GO:0006695]; fatty acid metabolic process [GO:0006631]; steroid metabolic process [GO:0008202]; sterol biosynthetic process [GO:0016126]

85	Q9BV57	MTND_HUMAN	ADI1 MTCBP1 HMFT1638	1,2-dihydroxy-3-keto-5-methylthiopentene dioxygenase (EC 1.13.11.54) (Acireductone dioxygenase (Fe(2+)-requiring)) (ARD) (Fe-ARD) (Membrane-type 1 matrix metalloproteinase cytoplasmic tail-binding protein 1) (MTCBP-1) (Submergence-induced protein-like factor) (Sip-L)	H88-H90-E94-H133	1 Fe cation	Catalytic	1.13.11.54	Cytoplasm, Cell membrane, Nucleus	Yes		L-methionine salvage from methylthioadenosine [GO:0019509]
86	P80188	NGAL_HUMAN	LCN2 HNL NGAL	Neutrophil gelatinase-associated lipocalin (NGAL) (25 kDa alpha-2-microglobulin-related subunit of MMP-9) (Lipocalin-2) (Oncogene 24p3) (Siderocalin LCN2) (p25)	Y126-K145-K154	Binds ferric siderophore	Substrate - transport		Extracellular space	No		antimicrobial humoral response [GO:0019730]; cellular iron ion homeostasis [GO:0006879]; cellular response to hydrogen peroxide [GO:0070301]; cellular response to interleukin-1 [GO:0071347]; cellular response to lipopolysaccharide [GO:0071222]; cellular response to nutrient levels [GO:0031669]; cellular response to tumor necrosis factor [GO:0071356]; extrinsic apoptotic signaling pathway in absence of ligand [GO:0097192]; innate immune response [GO:0045087]; ion transport [GO:0006811]; neutrophil degranulation [GO:0043312]; positive regulation of cell projection organization [GO:0031346]; positive regulation of gene expression [GO:0010628]; protein homotrimerization [GO:0070207]; response to drug [GO:0042493]; response to herbicide [GO:0009635]; response to mycotoxin [GO:0010046]; response to virus [GO:0009615]; siderophore transport [GO:0015891]
87	Q9GZT8	NIF3L_HUMAN	NIF3L1 ALS2CR1 MDS015 My018	NIF3-like protein 1 (Amyotrophic lateral sclerosis 2 chromosomal region candidate gene 1 protein)	H93-H339-E343	1 Fe cation	Catalytic		Cytoplasm, Nucleus	No		negative regulation of nucleic acid-templated transcription [GO:1903507]; neuron differentiation [GO:0030182]; positive regulation of transcription, DNA-templated [GO:0045893]
88	P49279	NRAM1_HUMAN	SLC11A1 LSH NRAMP NRAMP1	Natural resistance-associated macrophage protein 1 (NRAMP 1) (Solute carrier family 11 member 1)	Unknown	Unknown	Substrate - transport		Unknown	Yes		activation of protein kinase activity [GO:0032147]; antigen processing and presentation of peptide antigen [GO:0048002]; antimicrobial humoral response [GO:0019730]; cadmium ion transmembrane transport [GO:0070574]; cell redox homeostasis [GO:0045454]; cellular cadmium ion homeostasis [GO:0006876]; cellular iron ion homeostasis [GO:0006879]; defense response to bacterium [GO:0042742]; defense response to Gram-negative bacterium [GO:0050829]; defense response to protozoan [GO:0042832]; divalent metal ion export [GO:0070839]; inflammatory response [GO:0006954]; interleukin-2 production [GO:0032623]; interleukin-3 production [GO:0032632]; iron ion homeostasis [GO:0055072]; iron ion transport [GO:0006826]; L-arginine import [GO:0043091]; macrophage activation [GO:0042116]; manganese ion transport [GO:0006828]; MHC class II biosynthetic process [GO:0045342]; mRNA stabilization [GO:0048255]; multicellular organismal iron ion homeostasis [GO:0060586]; negative regulation of cytokine production [GO:0001818]; neutrophil degranulation [GO:0043312]; nitrite transport [GO:0015707]; phagocytosis [GO:0006909]; positive regulation of cytokine production [GO:0001819]; positive regulation of dendritic cell antigen processing and presentation [GO:0002606]; positive regulation of gene expression [GO:0010628]; positive regulation of interferon-gamma production [GO:0032729]; positive regulation of phagocytosis [GO:0050766]; positive regulation of T-helper 1 type immune response [GO:0002827]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; respiratory burst [GO:0045730]; response to bacterium [GO:0009617]; response to interferon-gamma [GO:0034341]; response to lipopolysaccharide [GO:0032496]; T cell cytokine production [GO:0002369]; T cell proliferation involved in immune response [GO:0002309]; vacuolar acidification [GO:0007035]; wound healing [GO:0042060]

89	P49281	NRAM2_HUMAN	SLC11A2 DCT1 DMT1 NRAMP2 OK/SW-cl.20	Natural resistance-associated macrophage protein 2 (NRAMP 2) (Divalent cation transporter 1) (Divalent metal transporter 1) (DMT-1) (Solute carrier family 11 member 2)	Unknown	Unknown	Substrate - transport		Cell membrane, Endosome	Yes	DISEASE: Anemia, hypochromic microcytic, with iron overload 1 (AHMIO1) [MIM:206100]: A hematologic disease characterized by abnormal hemoglobin content in the erythrocytes which are reduced in size. The disorder is due to an error of iron metabolism that results in high serum iron, massive hepatic iron deposition, and absence of sideroblasts and stainable bone marrow iron store. Despite adequate transferrin-iron complex, delivery of iron to the erythroid bone marrow is apparently insufficient for the demands of hemoglobin synthesis. {ECO:0000269|PubMed:15459009, ECO:0000269|PubMed:16160008, ECO:0000269|PubMed:16439678}. Note=The disease is caused by mutations affecting the gene represented in this entry.	activation of cysteine-type endopeptidase activity involved in apoptotic process [GO:0006919]; cadmium ion transmembrane transport [GO:0070574]; cellular iron ion homeostasis [GO:0006879]; cellular response to oxidative stress [GO:0034599]; cobalt ion transport [GO:0006824]; copper ion transport [GO:0006825]; dendrite morphogenesis [GO:0048813]; detection of oxygen [GO:0003032]; erythrocyte development [GO:0048821]; ferrous iron import [GO:0070627]; ferrous iron transport [GO:0015684]; heme biosynthetic process [GO:0006783]; lead ion transport [GO:0015692]; learning or memory [GO:0007611]; manganese ion transport [GO:0006828]; multicellular organismal iron ion homeostasis [GO:0060586]; nickel cation transport [GO:0015675]; response to hypoxia [GO:0001666]; response to iron ion [GO:0010039]; vanadium ion transport [GO:0015676]
90	Q8N543	OGFD1_HUMAN	OGFOD1 KIAA1612 TPA1	Prolyl 3-hydroxylase OGFOD1 (EC 1.14.11.-) (2-oxoglutarate and iron-dependent oxygenase domain-containing protein 1) (Termination and polyadenylation 1 homolog)	H155-D157-H218	1 Fe cation	Catalytic	1.14.11.-	Cytoplasm, Nucleus	No		cell proliferation [GO:0008283]; peptidyl-proline hydroxylation [GO:0019511]; protein hydroxylation [GO:0018126]; regulation of translational termination [GO:0006449]; stress granule assembly [GO:0034063]
91	Q6N063	OGFD2_HUMAN	OGFOD2	2-oxoglutarate and iron-dependent oxygenase domain-containing protein 2 (EC 1.14.11.-)	H235-D237-H290	1 Fe cation	Catalytic	1.14.11.-	Unknown	No		
92	Q6PK18	OGFD3_HUMAN	OGFOD3 C17orf101	2-oxoglutarate and iron-dependent oxygenase domain-containing protein 3 (EC 1.14.11.-)	H230-D232-H288	1 Fe cation	Catalytic	1.14.11.-	Unknown	Yes		
93	Q9NPF4	OSGEP_HUMAN	OSGEP GCPL1	Probable tRNA N6-adenosine threonylcarbamoyltransferase (EC 2.3.1.234) (N6-L-threonylcarbamoyladenine synthase) (t(6)A synthase) (O-sialoglycoprotein endopeptidase) (hOSGEP) (t(6)A37 threonylcarbamoyladenosine biosynthesis protein OSGEP) (tRNA threonylcarbamoyladenosine biosynthesis protein OSGEP)	H109-H113-Y130-D294	1 Divalent cation	Catalytic	2.3.1.234	Cytoplasm, Nucleus	No		tRNA threonylcarbamoyladenosine modification [GO:0002949]
94	Q32P28	P3H1_HUMAN	P3H1 GROS1 LEPRE1 PSEC0109	Prolyl 3-hydroxylase 1 (EC 1.14.11.7) (Growth suppressor 1) (Leucine- and proline-enriched proteoglycan 1) (Leprecan-1)	H587-D589-H659	1 Fe cation	Catalytic	1.14.11.7	Endoplasmic reticulum	No	DISEASE: Osteogenesis imperfecta 8 (OI8) [MIM:610915]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI8 is characterized by disproportionate short stature, severe osteoporosis, shortening of the long bones, white sclerae, a round face and a short barrel-shaped chest. {ECO:0000269|PubMed:17277775, ECO:0000269|PubMed:19088120}. Note=The disease is caused by mutations affecting the gene represented in this entry. A splice site mutation leading to the absence of isoform 1 has been reported in 2 OI8 patients. Isoform 1 is the only form predicted to be located in the endoplasmic reticulum, which the appropriate location for the catalysis of collagen hydroxylation. These patients show indeed severely reduced COL1A1 hydroxylation (PubMed:19088120). {ECO:0000269|PubMed:19088120}.	bone development [GO:0060348]; chaperone-mediated protein folding [GO:0061077]; collagen metabolic process [GO:0032963]; negative regulation of cell proliferation [GO:0008285]; negative regulation of post-translational protein modification [GO:1901874]; protein folding [GO:0006457]; protein hydroxylation [GO:0018126]; protein stabilization [GO:0050821]; regulation of protein secretion [GO:0050708]

95	Q8IVL5	P3H2_HUMAN	P3H2 LEPREL1 MLAT4	Prolyl 3-hydroxylase 2 (EC 1.14.11.7) (Leprecan-like protein 1) (Myxoid liposarcoma-associated protein 4)	H580-D582-H652	1 Fe cation	Catalytic	1.14.11.7	Endoplasmic reticulum, Golgi apparatus	No	DISEASE: Myopia, high, with cataract and vitreoretinal degeneration (MCVD) [MIM:614292]: A disorder characterized by severe myopia with variable expressivity of cataract and vitreoretinal degeneration. Some patients manifest lens subluxation, lens instability and retinal detachment. {ECO:0000269|PubMed:21885030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	collagen metabolic process [GO:0032963]; negative regulation of cell proliferation [GO:0008285]; peptidyl-proline hydroxylation [GO:0019511]
96	Q8IVL6	P3H3_HUMAN	P3H3 LEPREL2	Prolyl 3-hydroxylase 3 (EC 1.14.11.7) (Leprecan-like protein 2) (Protein B)	H584-D586-H656	1 Fe cation	Catalytic	1.14.11.7	Endoplasmic reticulum	No		collagen metabolic process [GO:0032963]; negative regulation of cell proliferation [GO:0008285]
97	P13674	P4HA1_HUMAN	P4HA1 P4HA	Prolyl 4-hydroxylase subunit alpha-1 (4-PH alpha-1) (EC 1.14.11.2) (Procollagen-proline,2-oxoglutarate-4-dioxygenase subunit alpha-1)	H429-D431-H500	1 Fe cation	Catalytic	1.14.11.2	Endoplasmic reticulum	No		collagen fibril organization [GO:0030199]; peptidyl-proline hydroxylation to 4-hydroxy-L-proline [GO:0018401]
98	O15460	P4HA2_HUMAN	P4HA2 UNQ290/PRO330	Prolyl 4-hydroxylase subunit alpha-2 (4-PH alpha-2) (EC 1.14.11.2) (Procollagen-proline,2-oxoglutarate-4-dioxygenase subunit alpha-2)	H430-D432-H501	1 Fe cation	Catalytic	1.14.11.2	Endoplasmic reticulum	No	DISEASE: Myopia 25, autosomal dominant (MYP25) [MIM:617238]: A refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269|PubMed:25741866}. Note=The disease is caused by mutations affecting the gene represented in this entry.	
99	Q7Z4N8	P4HA3_HUMAN	P4HA3 UNQ711/PRO1374	Prolyl 4-hydroxylase subunit alpha-3 (4-PH alpha-3) (EC 1.14.11.2) (Procollagen-proline,2-oxoglutarate-4-dioxygenase subunit alpha-3)	H440-D442-H510	1 Fe cation	Catalytic	1.14.11.2	Endoplasmic reticulum	No		
100	Q9NXG6	P4HTM_HUMAN	P4HTM PH4	Transmembrane prolyl 4-hydroxylase (P4H-TM) (EC 1.14.11.-) (Hypoxia-inducible factor prolyl hydroxylase 4) (HIF-PH4) (HIF-prolyl hydroxylase 4) (HPH-4)	H328-D330-H441	1 Fe cation	Catalytic	1.14.11.-	Endoplasmic reticulum	Yes		regulation of erythrocyte differentiation [GO:0045646]
101	O14832	PAHX_HUMAN	PHYH PAHX	Phytanoyl-CoA dioxygenase, peroxisomal (EC 1.14.11.18) (Phytanic acid oxidase) (Phytanoyl-CoA alpha-hydroxylase) (PhyH)	H175-D177-H264	1 Fe cation	Catalytic	1.14.11.18	Peroxisome	No	DISEASE: Refsum disease (RD) [MIM:266500]: A rare autosomal recessive peroxisomal disorder characterized by the accumulation of the branched-chain fatty acid, phytanic acid, in blood and tissues. Cardinal clinical features are retinitis pigmentosa, peripheral neuropathy, cerebellar ataxia, and elevated protein levels in the cerebrospinal fluid (CSF). Half of all patients exhibit generalized, mild to moderate ichthyosis resembling ichthyosis vulgaris. Less constant features are nerve deafness, anosmia, skeletal abnormalities, cataracts and cardiac impairment. {ECO:0000269|PubMed:10709665, ECO:0000269|PubMed:10767344, ECO:0000269|PubMed:14974078, ECO:0000269|PubMed:9326939, ECO:0000269|PubMed:9326940}. Note=The disease is caused by mutations affecting the gene represented in this entry.	2-oxoglutarate metabolic process [GO:0006103]; fatty acid alpha-oxidation [GO:0001561]; isoprenoid metabolic process [GO:0006720]; methyl-branched fatty acid metabolic process [GO:0097089]

102	P00439	PH4H_HUMAN	PAH	Phenylalanine-4-hydroxylase (PAH) (EC 1.14.16.1) (Phe-4-monooxygenase)	H285-H290-E330	1 Fe cation	Catalytic	1.14.16.1	Unknown	No	DISEASE: Phenylketonuria (PKU) [MIM:261600]: Autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. {ECO:0000269|PubMed:10200057, ECO:0000269|PubMed:10679941, ECO:0000269|PubMed:11180595, ECO:0000269|PubMed:11326337, ECO:0000269|PubMed:11385716, ECO:0000269|PubMed:11461196, ECO:0000269|PubMed:12501224, ECO:0000269|PubMed:1355066, ECO:0000269|PubMed:1363837, ECO:0000269|PubMed:1363838, ECO:0000269|PubMed:1671810, ECO:0000269|PubMed:1672290, ECO:0000269|PubMed:1672294, ECO:0000269|PubMed:1679030, ECO:0000269|PubMed:1709636, ECO:0000269|PubMed:18538294, ECO:0000269|PubMed:1975559, ECO:0000269|PubMed:2014802,	catecholamine biosynthetic process [GO:0042423]; cellular amino acid biosynthetic process [GO:0008652]; L-phenylalanine catabolic process [GO:0006559]; neurotransmitter biosynthetic process [GO:0042136]
103	O75151	PHF2_HUMAN	PHF2 CENP-35 KIAA0662	Lysine-specific demethylase PHF2 (EC 1.14.11.-) (GRC5) (PHD finger protein 2)	H249-D251-Y321	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		liver development [GO:0001889]; negative regulation of chromatin silencing at rDNA [GO:0061188]; protein demethylation [GO:0006482]; transcription, DNA-templated [GO:0006351]
104	Q9UPP1	PHF8_HUMAN	PHF8 KIAA1111 ZNF422	Histone lysine demethylase PHF8 (EC 1.14.11.27) (PHD finger protein 8)	H283-D285-Y293-H355	1 Fe cation	Catalytic	1.14.11.27	Nucleus	No	DISEASE: Mental retardation, X-linked, syndromic, Siderius type (MRXSSD) [MIM:300263]: A syndrome characterized by mild to borderline mental retardation with or without cleft lip/cleft palate. {ECO:0000269|PubMed:16199551, ECO:0000269|PubMed:17661819, ECO:0000269|PubMed:20101266, ECO:0000269|PubMed:20208542, ECO:0000269|PubMed:20346720, ECO:0000269|PubMed:20421419, ECO:0000269|PubMed:20548336, ECO:0000269|PubMed:20622853, ECO:0000269|PubMed:20622854}. Note=The disease is caused by mutations affecting the gene represented in this entry.	brain development [GO:0007420]; G1/S transition of mitotic cell cycle [GO:0000082]; histone H3-K27 demethylation [GO:0071557]; histone H3-K36 demethylation [GO:0070544]; histone H3-K9 demethylation [GO:0033169]; histone H4-K20 demethylation [GO:0035574]; negative regulation of chromatin silencing at rDNA [GO:0061188]; positive regulation of transcription, DNA-templated [GO:0045893]; positive regulation of transcription from RNA polymerase I promoter [GO:0045943]; transcription, DNA-templated [GO:0006351]
105	Q5SRE7	PHYD1_HUMAN	PHYHD1	Phytanoyl-CoA dioxygenase domain-containing protein 1 (EC 1.-.-.-)	H156-D158-H246	1 Fe cation	Catalytic	1.-.-.-	Unknown	No		
106	O00625	PIR_HUMAN	PIR	Pirin (EC 1.13.11.24) (Probable quercetin 2,3-dioxygenase PIR) (Probable quercetinase)	H56-H58-H101-E103	1 Fe cation	Catalytic	1.13.11.24	Cytoplasm, Nucleus	No		monocyte differentiation [GO:0030224]; regulation of transcription, DNA-templated [GO:0006355]; transcription from RNA polymerase II promoter [GO:0006366]
107	Q02809	PLOD1_HUMAN	PLOD1 LLH PLOD	Procollagen-lysine,2-oxoglutarate 5-dioxygenase 1 (EC 1.14.11.4) (Lysyl hydroxylase 1) (LH1)	H656-D658-H708	1 Fe cation	Catalytic	1.14.11.4	Endoplasmic reticulum	Yes	DISEASE: Ehlers-Danlos syndrome 6 (EDS6) [MIM:225400]: A connective tissue disorder characterized by generalized joint hypermobility, hyperextensible skin, atrophic cutaneous scars due to tissue fragility, progressive kyphoscoliosis already present at birth, ocular manifestations, arterial rupture, easy bruising, severe neonatal muscle hypotonia and delayed motor development. {ECO:0000269|PubMed:10686424, ECO:0000269|PubMed:15666309, ECO:0000269|PubMed:15854030, ECO:0000269|PubMed:15979919, ECO:0000269|PubMed:8163671, ECO:0000269|PubMed:9617436}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular protein modification process [GO:0006464]; epidermis development [GO:0008544]; hydroxylysine biosynthetic process [GO:0046947]; oxidation-reduction process [GO:0055114]; peptidyl-lysine hydroxylation [GO:0017185]; response to hypoxia [GO:0001666]

108	O00469	PLOD2_HUMAN	PLOD2	Procollagen-lysine,2-oxoglutarate 5-dioxygenase 2 (EC 1.14.11.4) (Lysyl hydroxylase 2) (LH2)	H666-D668-H718	1 Fe cation	Catalytic	1.14.11.4	Endoplasmic reticulum	Yes	DISEASE: Bruck syndrome 2 (BRKS2) [MIM:609220]: An autosomal recessive disease characterized by generalized osteopenia, congenital joint contractures, fragile bones with onset of fractures in infancy or early childhood, short stature, severe limb deformity, progressive scoliosis, and pterygia. It is distinguished from osteogenesis imperfecta by the absence of hearing loss and dentinogenesis imperfecta, and by the presence of clubfoot and congenital joint limitations. {ECO:0000269|PubMed:12881513, ECO:0000269|PubMed:15523624}. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect leading to Bruck syndrome is an aberrant cross-linking of bone collagen, due to underhydroxylation of lysine residues within the telopeptides of type I collagen, whereas the lysine residues in the triple helix are normal.; DISEASE: Note=PLOD2 mutations give rise to a broad variety of phenotypes with variable degrees of severity of bone fragility and joint contractures. Disease-associated mutations have been found in patients with autosomal recessive osteogenesis imperfecta (AR-OI) (PubMed:22689593). {ECO:0000269|PubMed:22689593}.	cellular protein modification process [GO:0006464]; hydroxylysine biosynthetic process [GO:0046947]; peptidyl-lysine hydroxylation [GO:0017185]; response to hypoxia [GO:0001666]
109	O60568	PLOD3_HUMAN	PLOD3	Procollagen-lysine,2-oxoglutarate 5-dioxygenase 3 (EC 1.14.11.4) (Lysyl hydroxylase 3) (LH3)	H667-D669-H719	1 Fe cation	Catalytic	1.14.11.4	Endoplasmic reticulum	Yes	DISEASE: Lysyl hydroxylase 3 deficiency (LH3 deficiency) [MIM:612394]: Connective tissue disorder. The syndrome is characterized by congenital malformations severely affecting many tissues and organs and revealing features of several collagen disorders, most of them involving COL2A1 (type II collagen). The findings suggest that the failure of lysyl hydroxylation and hydroxylysyl carbohydrate addition, which affects many collagens, is the molecular basis of this syndrome. {ECO:0000269|PubMed:18834968}. Note=The disease is caused by mutations affecting the gene represented in this entry.	basement membrane assembly [GO:0070831]; cellular response to hormone stimulus [GO:0032870]; collagen fibril organization [GO:0030199]; endothelial cell morphogenesis [GO:0001886]; epidermis morphogenesis [GO:0048730]; hydroxylysine biosynthetic process [GO:0046947]; in utero embryonic development [GO:0001701]; lung morphogenesis [GO:0060425]; neural tube development [GO:0021915]; peptidyl-lysine hydroxylation [GO:0017185]; protein localization [GO:0008104]; protein O-linked glycosylation [GO:0006493]; vasodilation [GO:0042311]
110	P62136	PP1A_HUMAN	PPP1CA PPP1A	Serine/threonine-protein phosphatase PP1-alpha catalytic subunit (PP-1A) (EC 3.1.3.16)	D64-H66-D92	1 Fe cation	Catalytic	3.1.3.16	Cytoplasm, Nucleus	No		beta-catenin destruction complex disassembly [GO:1904886]; branching morphogenesis of an epithelial tube [GO:0048754]; cell cycle [GO:0007049]; cell division [GO:0051301]; circadian regulation of gene expression [GO:0032922]; dephosphorylation [GO:0016311]; entrainment of circadian clock by photoperiod [GO:0043153]; glycogen metabolic process [GO:0005977]; lung development [GO:0030324]; negative regulation of protein binding [GO:0032091]; positive regulation of extrinsic apoptotic signaling pathway in absence of ligand [GO:2001241]; protein dephosphorylation [GO:0006470]; regulation of canonical Wnt signaling pathway [GO:0060828]; regulation of circadian rhythm [GO:0042752]; regulation of glycogen biosynthetic process [GO:0005979]; regulation of glycogen catabolic process [GO:0005981]; regulation of translational initiation by eIF2 alpha dephosphorylation [GO:0036496]

111	Q08209	PP2BA_HUMAN	PPP3CA CALNA CNA	Serine/threonine-protein phosphatase 2B catalytic subunit alpha isoform (EC 3.1.3.16) (CAM-PRP catalytic subunit) (Calmodulin-dependent calcineurin A subunit alpha isoform)	D90-H92-D118	1 Fe cation	Catalytic	3.1.3.16	Cell membrane, Nucleus	Yes		calcineurin-NFAT signaling cascade [GO:0033173]; calcium ion transport [GO:0006816]; cardiac muscle hypertrophy in response to stress [GO:0014898]; cellular response to drug [GO:0035690]; cellular response to glucose stimulus [GO:0071333]; dephosphorylation [GO:0016311]; excitatory postsynaptic potential [GO:0060079]; Fc-epsilon receptor signaling pathway [GO:0038095]; G1/S transition of mitotic cell cycle [GO:0000082]; modulation of synaptic transmission [GO:0050804]; multicellular organismal response to stress [GO:0033555]; negative regulation of chromatin binding [GO:0035562]; negative regulation of dendrite morphogenesis [GO:0050774]; negative regulation of insulin secretion [GO:0046676]; negative regulation of production of miRNAs involved in gene silencing by miRNA [GO:1903799]; positive regulation of cardiac muscle hypertrophy in response to stress [GO:1903244]; positive regulation of connective tissue replacement [GO:1905205]; positive regulation of NFAT protein import into nucleus [GO:0051533]; positive regulation of sequence-specific DNA binding transcription factor activity [GO:0051091]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; protein dephosphorylation [GO:0006470]; protein import into nucleus [GO:0006606]; response to amphetamine [GO:0001975]; response to calcium ion [GO:0051592]; skeletal muscle fiber development [GO:0048741]; T cell activation [GO:0042110]; transition between fast and slow fiber [GO:0014883]; Wnt signaling pathway, calcium modulating pathway [GO:0007223]
112	P16298	PP2BB_HUMAN	PPP3CB CALNA2 CALNB CNA2	Serine/threonine-protein phosphatase 2B catalytic subunit beta isoform (EC 3.1.3.16) (CAM-PRP catalytic subunit) (Calmodulin-dependent calcineurin A subunit beta isoform)	D99-H101-D127	1 Fe cation	Catalytic	3.1.3.16	Unknown	No		axon extension [GO:0048675]; calcineurin-NFAT signaling cascade [GO:0033173]; calcium ion regulated exocytosis [GO:0017156]; cellular response to drug [GO:0035690]; dephosphorylation [GO:0016311]; Fc-epsilon receptor signaling pathway [GO:0038095]; heart development [GO:0007507]; learning [GO:0007612]; locomotion involved in locomotory behavior [GO:0031987]; lymphangiogenesis [GO:0001946]; memory [GO:0007613]; negative regulation of T cell mediated cytotoxicity [GO:0001915]; positive regulation of insulin secretion involved in cellular response to glucose stimulus [GO:0035774]; positive regulation of NFAT protein import into nucleus [GO:0051533]; positive regulation of transcription, DNA-templated [GO:0045893]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; protein dephosphorylation [GO:0006470]; protein phosphorylation [GO:0006468]; regulation of insulin secretion [GO:0050796]; regulation of synaptic plasticity [GO:0048167]; response to cytokine [GO:0034097]; signal transduction [GO:0007165]; social behavior [GO:0035176]; T cell activation [GO:0042110]; T cell differentiation [GO:0030217]; T cell homeostasis [GO:0043029]; T cell proliferation [GO:0042098]; Wnt signaling pathway, calcium modulating pathway [GO:0007223]
113	P48454	PP2BC_HUMAN	PPP3CC CALNA3 CNA3	Serine/threonine-protein phosphatase 2B catalytic subunit gamma isoform (EC 3.1.3.16) (CAM-PRP catalytic subunit) (Calcineurin, testis-specific catalytic subunit) (Calmodulin-dependent calcineurin A subunit gamma isoform)	D86-H88-D114	1 Fe cation	Catalytic	3.1.3.16	Unknown	No		brain development [GO:0007420]; positive regulation of protein insertion into mitochondrial membrane involved in apoptotic signaling pathway [GO:1900740]
114	P13686	PPA5_HUMAN	ACP5	Tartrate-resistant acid phosphatase type 5 (TR-AP) (EC 3.1.3.2) (Tartrate-resistant acid ATPase) (TrATPase) (Type 5 acid phosphatase)	D33-D71-Y74-H242; D71-N110-H205-H240	2 Fe cations	Catalytic	3.1.3.2	Unknown	No	DISEASE: Spondyloenchondrodysplasia with immune dysregulation (SPENCDI) [MIM:607944]: A disease characterized by vertebral and metaphyseal dysplasia, spasticity with cerebral calcifications, and strong predisposition to autoimmune diseases. The skeletal dysplasia is characterized by radiolucent and irregular spondylar and metaphyseal lesions that represent islands of chondroid tissue within bone. {ECO:0000269|PubMed:21217752, ECO:0000269|PubMed:21217755}. Note=The disease is caused by mutations affecting the gene represented in this entry. ACP5 inactivating mutations result in a functional excess of phosphorylated osteopontin causing deregulation of osteopontin signaling and consequential autoimmune disease.	riboflavin metabolic process [GO:0006771]

115	Q7KZA3	Q7KZA3_HUMAN	DKFZp686P18130	Ferrochelatase	Unknown	1 Fe cation	Substrate - biosinthesis	4.99.1.1	Unknown	No		ferrochelatase activity
116	Q9H6W3	RIOX1_HUMAN	RIOX1 C14orf169 MAPJD NO66	Ribosomal oxygenase 1 (60S ribosomal protein L8 histidine hydroxylase) (Bifunctional lysine-specific demethylase and histidyl-hydroxylase NO66) (EC 1.14.11.-) (EC 1.14.11.27) (Histone lysine demethylase NO66) (Myc-associated protein with JmjC domain) (Nucleolar protein 66) (hsNO66) (Ribosomal oxygenase NO66) (ROX)	H340-D342-H405	1 Fe cation	Catalytic	1.14.11.-; 1.14.11.27	Nucleus	No		chromatin remodeling [GO:0006338]; histone H3-K36 demethylation [GO:0070544]; histone H3-K4 demethylation [GO:0034720]; negative regulation of osteoblast differentiation [GO:0045668]; negative regulation of transcription, DNA-templated [GO:0045892]; peptidyl-arginine hydroxylation [GO:0030961]; transcription, DNA-templated [GO:0006351]
117	Q8IUF8	RIOX2_HUMAN	RIOX2 MDIG MINA MINA53 NO52	Ribosomal oxygenase 2 (60S ribosomal protein L27a histidine hydroxylase) (Bifunctional lysine-specific demethylase and histidyl-hydroxylase MINA) (EC 1.14.11.-) (Histone lysine demethylase MINA) (MYC-induced nuclear antigen) (Mineral dust-induced gene protein) (Nucleolar protein 52) (Ribosomal oxygenase MINA) (ROX)	H179-D181-H240	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		chromatin remodeling [GO:0006338]; negative regulation of transcription, DNA-templated [GO:0045892]; peptidyl-arginine hydroxylation [GO:0030961]; ribosome biogenesis [GO:0042254]; transcription, DNA-templated [GO:0006351]
118	P31350	RIR2_HUMAN	RRM2 RR2	Ribonucleoside-diphosphate reductase subunit M2 (EC 1.17.4.1) (Ribonucleotide reductase small chain) (Ribonucleotide reductase small subunit)	D138-E169-H172; E169-E232-E266-H269	2 Fe cations	Catalytic	1.17.4.1	Cytoplasm	No		deoxyribonucleotide biosynthetic process [GO:0009263]; DNA replication [GO:0006260]; G1/S transition of mitotic cell cycle [GO:0000082]; nucleobase-containing small molecule interconversion [GO:0015949]; protein heterotetramerization [GO:0051290]; regulation of transcription involved in G1/S transition of mitotic cell cycle [GO:0000083]
119	Q7LG56	RIR2B_HUMAN	RRM2B P53R2	Ribonucleoside-diphosphate reductase subunit M2 B (EC 1.17.4.1) (TP53-inducible ribonucleotide reductase M2 B) (p53-inducible ribonucleotide reductase small subunit 2-like protein) (p53R2)	D100-E131-H134; E131-E194-E228-H231	2 Fe cations	Catalytic	1.17.4.1	Cytoplasm, Nucleus	No	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Mitochondrial DNA depletion syndrome 8B (MTDPS8B) [MIM:612075]: A disease due to mitochondrial dysfunction and characterized by ophthalmoplegia, ptosis, gastrointestinal dysmotility, cachexia, peripheral neuropathy. {ECO:0000269|PubMed:19667227}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 5 (PEOA5) [MIM:613077]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and	deoxyribonucleoside triphosphate metabolic process [GO:0009200]; deoxyribonucleotide biosynthetic process [GO:0009263]; DNA repair [GO:0006281]; kidney development [GO:0001822]; mitochondrial DNA replication [GO:0006264]; negative regulation of intrinsic apoptotic signaling pathway by p53 class mediator [GO:1902254]; nucleobase-containing small molecule interconversion [GO:0015949]; renal system process [GO:0003014]; response to amine [GO:0014075]; response to oxidative stress [GO:0006979]
120	Q96AT9	RPE_HUMAN	RPE HUSSY-17	Ribulose-phosphate 3-epimerase (EC 5.1.3.1) (Ribulose-5-phosphate-3-epimerase)	H35-D37-H70-D175	1 Divalent cation	Catalytic - no redox	5.1.3.1	Unknown	No		carbohydrate metabolic process [GO:0005975]; cellular carbohydrate metabolic process [GO:0044262]; pentose catabolic process [GO:0019323]; pentose-phosphate shunt [GO:0006098]; pentose-phosphate shunt, non-oxidative branch [GO:0009052]

121	Q16518	RPE65_HUMAN	RPE65	Retinoid isomerohydrolase (EC 3.1.1.64) (All-trans-retinyl-palmitate hydrolase) (Retinal pigment epithelium-specific 65 kDa protein) (Retinol isomerase)	H180-H241-H313-H527	1 Fe cation	Catalytic	3.1.1.64	Cytoplasm, Cell membrane	Yes	DISEASE: Leber congenital amaurosis 2 (LCA2) [MIM:204100]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:10090910, ECO:0000269|PubMed:10766140, ECO:0000269|PubMed:11462243, ECO:0000269|PubMed:14611946, ECO:0000269|PubMed:14962443, ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:16205573, ECO:0000269|PubMed:17297704, ECO:0000269|PubMed:17724218, ECO:0000269|PubMed:18682808, ECO:0000269|PubMed:9326941, ECO:0000269|PubMed:9801879}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Retinitis pigmentosa 20 (RP20) [MIM:613794]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well.	cellular response to electrical stimulus [GO:0071257]; circadian rhythm [GO:0007623]; detection of light stimulus involved in visual perception [GO:0050908]; insulin receptor signaling pathway [GO:0008286]; neural retina development [GO:0003407]; regulation of rhodopsin gene expression [GO:0007468]; retina homeostasis [GO:0001895]; retinal metabolic process [GO:0042574]; retina morphogenesis in camera-type eye [GO:0060042]; retinoid metabolic process [GO:0001523]; visual perception [GO:0007601]; vitamin A metabolic process [GO:0006776]
122	Q2QD12	RPEL1_HUMAN	RPEL1	Ribulose-phosphate 3-epimerase-like protein 1 (EC 5.1.3.1) (Ribulose-5-phosphate-3-epimerase-like protein 1)	H35-D37-H70-D175	1 Divalent cation	Catalytic - no redox	5.1.3.1	Unknown	No		cellular carbohydrate metabolic process [GO:0044262]; pentose catabolic process [GO:0019323]; pentose-phosphate shunt, non-oxidative branch [GO:0009052]
123	Q9NP59	S40A1_HUMAN	SLC40A1 FPN1 IREG1 SLC11A3 MSTP079	Solute carrier family 40 member 1 (Ferroportin-1) (Iron-regulated transporter 1)	Unknown	Unknown	Substrate - transport		Cell membrane	Yes	DISEASE: Hemochromatosis 4 (HFE4) [MIM:606069]: A disorder of iron metabolism characterized by iron overload. Excess iron is deposited in a variety of organs leading to their failure, and resulting in serious illnesses including cirrhosis, hepatomas, diabetes, cardiomyopathy, arthritis, and hypogonadotropic hypogonadism. Severe effects of the disease usually do not appear until after decades of progressive iron loading. {ECO:0000269|PubMed:10747949, ECO:0000269|PubMed:11431687, ECO:0000269|PubMed:11518736, ECO:0000269|PubMed:12091366, ECO:0000269|PubMed:12091367, ECO:0000269|PubMed:12123233, ECO:0000269|PubMed:12406098, ECO:0000269|PubMed:12730114, ECO:0000269|PubMed:12857562, ECO:0000269|PubMed:12865285, ECO:0000269|PubMed:15338274, ECO:0000269|PubMed:15466004, ECO:0000269|PubMed:16351644}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular iron ion homeostasis [GO:0006879]; endothelium development [GO:0003158]; ferrous iron export across plasma membrane [GO:1903988]; iron ion transmembrane transport [GO:0034755]; lymphocyte homeostasis [GO:0002260]; multicellular organismal iron ion homeostasis [GO:0060586]; negative regulation of apoptotic process [GO:0043066]; positive regulation of transcription by RNA polymerase II [GO:0045944]; regulation of transcription from RNA polymerase II promoter in response to iron [GO:0034395]; spleen trabecula formation [GO:0060345]
124	O75845	SC5D_HUMAN	SC5D SC5DL	Lathosterol oxidase (EC 1.14.19.20) (C-5 sterol desaturase) (Delta(7)-sterol 5-desaturase) (Delta(7)-sterol C5(6)-desaturase) (Lathosterol 5-desaturase) (Sterol-C5-desaturase)	H138-H142-H151-H155; H209-H228-H232-H233	2 Fe cations	Catalytic	1.14.19.20	Endoplasmic reticulum	Yes	DISEASE: Lathosterolosis (LATHST) [MIM:607330]: Autosomal recessive disorder characterized by a complex phenotype, including multiple congenital anomalies, mental retardation, and liver disease. {ECO:0000269|PubMed:12189593, ECO:0000269|PubMed:12812989}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cholesterol biosynthetic process via desmosterol [GO:0033489]; cholesterol biosynthetic process via lathosterol [GO:0033490]; lipid metabolic process [GO:0006629]
125	Q86SK9	SCD5_HUMAN	SCD5 ACOD4 SCD2 SCD4	Stearoyl-CoA desaturase 5 (EC 1.14.19.1) (Acyl-CoA-desaturase 4) (HSCD5) (Stearoyl-CoA 9-desaturase) (Stearoyl-CoA desaturase 2)	H94-H99-H131-H134; H135-H243-H272-H276	2 Fe cations	Catalytic	1.14.19.1	Endoplasmic reticulum	Yes		long-chain fatty-acyl-CoA biosynthetic process [GO:0035338]; unsaturated fatty acid biosynthetic process [GO:0006636]

126	Q8NFU7	TET1_HUMAN	TET1 CXXC6 KIAA1676 LCX	Methylcytosine dioxygenase TET1 (EC 1.14.11.n2) (CXXC-type zinc finger protein 6) (Leukemia-associated protein with a CXXC domain) (Ten-eleven translocation 1 gene protein)	H1672-D1674-H2028	1 Fe cation	Catalytic	1.14.11.n2	Nucleus	No	DISEASE: Note=A chromosomal aberration involving TET1 may be a cause of acute leukemias (PubMed:12646957). Translocation t(10;11)(q22;q23) with KMT2A/MLL1. This is a rare chromosomal translocation 5' KMT2A/MLL1-TET1 3' (PubMed:12124344, PubMed:12646957). {ECO:0000269|PubMed:12124344, ECO:0000269|PubMed:12646957}.	covalent chromatin modification [GO:0016569]; DNA demethylation [GO:0080111]; inner cell mass cell differentiation [GO:0001826]; negative regulation of methylation-dependent chromatin silencing [GO:0090310]; positive regulation of cell proliferation [GO:0008284]; positive regulation of histone methylation [GO:0031062]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; protein O-linked glycosylation [GO:0006493]; stem cell population maintenance [GO:0019827]; transcription, DNA-templated [GO:0006351]
127	Q6N021	TET2_HUMAN	TET2 KIAA1546 Nbla00191	Methylcytosine dioxygenase TET2 (EC 1.14.11.n2)	H1382-D1384-H1881	1 Fe cation	Catalytic	1.14.11.n2	Unknown	No	DISEASE: Note=TET2 is frequently mutated in myeloproliferative disorders (MPD). These constitute a heterogeneous group of disorders, also known as myeloproliferative diseases or myeloproliferative neoplasms (MPN), characterized by cellular proliferation of one or more hematologic cell lines in the peripheral blood, distinct from acute leukemia. Included diseases are: essential thrombocythemia, polycythemia vera, primary myelofibrosis (chronic idiopathic myelofibrosis). Bone marrow samples from patients display uniformly low levels of hmC in genomic DNA compared to bone marrow samples from healthy controls as well as hypomethylation relative to controls at the majority of differentially methylated CpG sites.; DISEASE: Polycythemia vera (PV) [MIM:263300]: A myeloproliferative disorder characterized by abnormal proliferation of all hematopoietic bone marrow elements, erythroid hyperplasia, an absolute increase in total blood volume, but also by myeloid leukocytosis, thrombocytosis and splenomegaly. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Note=TET2 is frequently mutated in systemic mastocytosis; also known as systemic mast cell disease. A condition with features in common with myeloproliferative diseases. It is a clonal disorder of the mast cell and its precursor cells. The clinical symptoms and signs of systemic mastocytosis are due to accumulation of clonally derived mast cells in different tissues, including bone marrow, skin, the gastrointestinal tract, the liver, and the spleen.; DISEASE: Myelodysplastic syndrome (MDS) [MIM:614286]: A	5-methylcytosine catabolic process [GO:0006211]; cell cycle [GO:0007049]; cytosine metabolic process [GO:0019858]; DNA demethylation [GO:0080111]; hematopoietic stem cell homeostasis [GO:0061484]; hemoglobin metabolic process [GO:0020027]; histone H3-K4 trimethylation [GO:0080182]; kidney development [GO:0001822]; liver morphogenesis [GO:0072576]; myeloid cell differentiation [GO:0030099]; myeloid progenitor cell differentiation [GO:0002318]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; post-embryonic development [GO:0009791]; protein O-linked glycosylation [GO:0006493]; response to organic cyclic compound [GO:0014070]; spleen development [GO:0048536]
128	O43151	TET3_HUMAN	TET3 KIAA0401	Methylcytosine dioxygenase TET3 (EC 1.14.11.n2)	H942-D944-H1538	1 Fe cation	Catalytic	1.14.11.n2	Cytoplasm, Nucleus	No		DNA demethylation [GO:0080111]; DNA demethylation of male pronucleus [GO:0044727]; histone H3-K4 trimethylation [GO:0080182]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; protein O-linked glycosylation [GO:0006493]
129	Q9NVH6	TMLH_HUMAN	TMLHE TMLH	Trimethyllysine dioxygenase, mitochondrial (EC 1.14.11.8) (Epsilon-trimethyllysine 2-oxoglutarate dioxygenase) (Epsilon-trimethyllysine hydroxylase) (TML hydroxylase) (TML-alpha-ketoglutarate dioxygenase) (TML dioxygenase) (TMLD)	H242-D244-H389	1 Fe cation	Catalytic	1.14.11.8	Mitochondrion	No	DISEASE: Autism, X-linked 6 (AUTSX6) [MIM:300872]: A form of autism, a complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. AUTSX6 patients may respond favorably to carnitine supplementation. {ECO:0000269|PubMed:21865298, ECO:0000269|PubMed:23092983}. Note=The disease is caused by mutations affecting the gene represented in this entry.	carnitine biosynthetic process [GO:0045329]; negative regulation of oxidoreductase activity [GO:0051354]
130	Q0P6H9	TMM62_HUMAN	TMEM62	Transmembrane protein 62	D63-H65-D99	1 Fe cation	Catalytic		Unknown	Yes		
131	Q6ZT21	TMPPE_HUMAN	TMPPE	Transmembrane protein with metallophosphoesterase domain (EC 3.1.-.-)	D214-H216-D246-H393; N277-H369-H391	2 Divalent cations	Catalytic	3.1.-.-	Unknown	Yes		
132	P17752	TPH1_HUMAN	TPH1 TPH TPRH TRPH	Tryptophan 5-hydroxylase 1 (EC 1.14.16.4) (Tryptophan 5-monooxygenase 1)	H272-H277-E317	1 Fe cation	Catalytic	1.14.16.4	Unknown	No		aromatic amino acid family metabolic process [GO:0009072]; bone remodeling [GO:0046849]; circadian rhythm [GO:0007623]; indolalkylamine biosynthetic process [GO:0046219]; mammary gland alveolus development [GO:0060749]; negative regulation of ossification [GO:0030279]; positive regulation of fat cell differentiation [GO:0045600]; response to immobilization stress [GO:0035902]; serotonin biosynthetic process [GO:0042427]

133	Q8IWU9	TPH2_HUMAN	TPH2 NTPH	Tryptophan 5-hydroxylase 2 (EC 1.14.16.4) (Neuronal tryptophan hydroxylase) (Tryptophan 5-monooxygenase 2)	H318-H323-E363	1 Fe cation	Catalytic	1.14.16.4	Unknown	No	DISEASE: Major depressive disorder (MDD) [MIM:608516]: A common psychiatric disorder. It is a complex trait characterized by one or more major depressive episodes without a history of manic, mixed, or hypomanic episodes. A major depressive episode is characterized by at least 2 weeks during which there is a new onset or clear worsening of either depressed mood or loss of interest or pleasure in nearly all activities. Four additional symptoms must also be present including changes in appetite, weight, sleep, and psychomotor activity; decreased energy; feelings of worthlessness or guilt; difficulty thinking, concentrating, or making decisions; or recurrent thoughts of death or suicidal ideation, plans, or attempts. The episode must be accompanied by distress or impairment in social, occupational, or other important areas of functioning. {ECO:0000269|PubMed:15629698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.; DISEASE: Attention deficit-hyperactivity disorder 7 (ADHD7) [MIM:613003]: A neurobehavioral developmental disorder primarily characterized by the coexistence of attentional problems and hyperactivity, with each behavior occurring infrequently alone. {ECO:0000269|PubMed:18347598}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Naturally occurring variants of TPH2 with impaired enzyme activity could cause deficiency of serotonin production and result in an increased risk of developing behavioral disorders.	aromatic amino acid family metabolic process [GO:0009072]; cellular response to lithium ion [GO:0071285]; circadian rhythm [GO:0007623]; indolalkylamine biosynthetic process [GO:0046219]; response to activity [GO:0014823]; response to calcium ion [GO:0051592]; response to estrogen [GO:0043627]; response to glucocorticoid [GO:0051384]; response to nutrient levels [GO:0031667]; serotonin biosynthetic process [GO:0042427]
134	P02787	TRFE_HUMAN	TF PRO1400	Serotransferrin (Transferrin) (Beta-1 metal-binding globulin) (Siderophilin)	D82-Y114-Y207-H268; D411-D445-Y536-H604	2 Fe cations	Substrate - transport		Extracellular space	No	DISEASE: Atransferrinemia (ATRAF) [MIM:209300]: A rare autosomal recessive disorder characterized by abnormal synthesis of transferrin leading to iron overload and microcytic hypochromic anemia. {ECO:0000269|PubMed:11110675, ECO:0000269|PubMed:15466165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular iron ion homeostasis [GO:0006879]; cellular response to iron ion [GO:0071281]; ferrous iron import across plasma membrane [GO:0098707]; iron ion homeostasis [GO:0055072]; membrane organization [GO:0061024]; platelet degranulation [GO:0002576]; positive regulation of receptor-mediated endocytosis [GO:0048260]; regulation of protein stability [GO:0031647]; retina homeostasis [GO:0001895]; transferrin transport [GO:0033572]
135	P02788	TRFL_HUMAN	LTF GIG12 LF	Lactotransferrin (Lactoferrin) (EC 3.4.21.-) (Growth-inhibiting protein 12) (Talalactoferrin) [Cleaved into: Lactoferricin-H (Lfcin-H); Kaliocin-1; Lactoferroxin-A; Lactoferroxin-B; Lactoferroxin-C]	D79-Y111-Y211-H272; D414-Y454-Y547-H616	2 Fe cations	Substrate - transport	3.4.21.-	Cytoplasm, Extracellular space	No		antibacterial humoral response [GO:0019731]; antifungal humoral response [GO:0019732]; antimicrobial humoral response [GO:0019730]; bone morphogenesis [GO:0060349]; cellular protein metabolic process [GO:0044267]; humoral immune response [GO:0006959]; innate immune response in mucosa [GO:0002227]; ion transport [GO:0006811]; iron assimilation by chelation and transport [GO:0033214]; negative regulation by host of viral process [GO:0044793]; negative regulation of apoptotic process [GO:0043066]; negative regulation of ATPase activity [GO:0032780]; negative regulation of cysteine-type endopeptidase activity [GO:2000117]; negative regulation of lipopolysaccharide-mediated signaling pathway [GO:0031665]; negative regulation of osteoclast development [GO:2001205]; negative regulation of single-species biofilm formation in or on host organism [GO:1900229]; negative regulation of tumor necrosis factor (ligand) superfamily member 11 production [GO:2000308]; negative regulation of viral genome replication [GO:0045071]; negative regulation of viral process [GO:0048525]; neutrophil degranulation [GO:0043312]; ossification [GO:0001503]; positive regulation of bone mineralization involved in bone maturation [GO:1900159]; positive regulation of chondrocyte proliferation [GO:1902732]; positive regulation of I-kappaB kinase/NF-kappaB signaling [GO:0043123]; positive regulation of NF-kappaB transcription factor activity [GO:0051092]; positive regulation of osteoblast differentiation [GO:0045669]; positive regulation of osteoblast proliferation [GO:0033690]; positive regulation of protein serine/threonine kinase activity [GO:0071902]; positive regulation of toll-like receptor 4 signaling pathway [GO:0034145]; regulation of cytokine production [GO:0001817]; regulation of tumor necrosis factor production [GO:0032680]; retina homeostasis [GO:0001895]; transcription, DNA-templated [GO:0006351]
136	P08582	TRFM_HUMAN	MELTF MAP97 MFI2	Melanotransferrin (Melanoma-associated antigen p97) (CD antigen CD228)	D78-Y107-Y210-H279; Y451-Y556-H625	2 Fe cations	Substrate - transport		Cell membrane	Yes		C-terminal protein lipidation [GO:0006501]; iron ion homeostasis [GO:0055072]; iron ion import [GO:0097286]; negative regulation of substrate adhesion-dependent cell spreading [GO:1900025]; positive regulation of extracellular matrix disassembly [GO:0090091]; positive regulation of plasminogen activation [GO:0010756]

137	P07101	TY3H_HUMAN	TH TYH	Tyrosine 3-monooxygenase (EC 1.14.16.2) (Tyrosine 3-hydroxylase) (TH)	H361-H366-E406	1 Fe cation	Catalytic	1.14.16.2	Unknown	No	DISEASE: Segawa syndrome autosomal recessive (ARSEGS) [MIM:605407]: A form of DOPA-responsive dystonia presenting in infancy or early childhood. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Some cases present with parkinsonian symptoms in infancy. Unlike all other forms of dystonia, it is an eminently treatable condition, due to a favorable response to L-DOPA. {ECO:0000269|PubMed:10585338, ECO:0000269|PubMed:11196107, ECO:0000269|PubMed:11246459, ECO:0000269|PubMed:15505183, ECO:0000269|PubMed:15747353, ECO:0000269|PubMed:16049992, ECO:0000269|PubMed:17696123, ECO:0000269|PubMed:18058633, ECO:0000269|PubMed:18554280, ECO:0000269|PubMed:19491146, ECO:0000269|PubMed:20056467, ECO:0000269|PubMed:20430833, ECO:0000269|PubMed:21940685, ECO:0000269|PubMed:22264700, ECO:0000269|PubMed:22815559, ECO:0000269|PubMed:23762320, ECO:0000269|PubMed:23939262, ECO:0000269|PubMed:24753243, ECO:0000269|PubMed:7814018,	aminergic neurotransmitter loading into synaptic vesicle [GO:0015842]; anatomical structure morphogenesis [GO:0009653]; animal organ morphogenesis [GO:0009887]; catecholamine biosynthetic process [GO:0042423]; cellular response to drug [GO:0035690]; cellular response to glucose stimulus [GO:0071333]; cellular response to growth factor stimulus [GO:0071363]; cellular response to manganese ion [GO:0071287]; cellular response to nicotine [GO:0071316]; cerebral cortex development [GO:0021987]; circadian sleep/wake cycle [GO:0042745]; dopamine biosynthetic process [GO:0042416]; dopamine biosynthetic process from tyrosine [GO:0006585]; eating behavior [GO:0042755]; embryonic camera-type eye morphogenesis [GO:0048596]; epinephrine biosynthetic process [GO:0042418]; eye photoreceptor cell development [GO:0042462]; fatty acid metabolic process [GO:0006631]; glycoside metabolic process [GO:0016137]; heart development [GO:0007507]; heart morphogenesis [GO:0003007]; isoquinoline alkaloid metabolic process [GO:0033076]; learning [GO:0007612]; locomotory behavior [GO:0007626]; mating behavior [GO:0007617]; memory [GO:0007613]; multicellular organism aging [GO:0010259]; neurotransmitter biosynthetic process [GO:0042136]; norepinephrine biosynthetic process [GO:0042421]; phthalate metabolic process [GO:0018963]; phytoalexin metabolic process [GO:0052314]; pigmentation [GO:0043473]; regulation of heart contraction [GO:0008016]; response to activity [GO:0014823]; response to amphetamine [GO:0001975]; response to corticosterone [GO:0051412]; response to electrical stimulus [GO:0051602]; response to estradiol [GO:0032355]; response to ethanol [GO:0045471]; response to ether [GO:0045472]; response to herbicide [GO:0009635]; response to hypoxia [GO:0001666]; response to immobilization stress [GO:0035902]; response to isolation stress [GO:0035900]; response to light stimulus [GO:0009416];
138	A2RUC4	TYW5_HUMAN	TYW5 C2orf60	tRNA wybutosine-synthesizing protein 5 (hTYW5) (EC 1.14.11.42) (tRNA(Phe) (7-(3-amino-3-carboxypropyl)wyosine(37)-C(2))-hydroxylase)	H160-D162-H235	1 Fe cation	Catalytic	1.14.11.42	Unknown	No		tRNA modification [GO:0006400]; wybutosine biosynthetic process [GO:0031591]
139	O14607	UTY_HUMAN	UTY KDM6C	Histone demethylase UTY (EC 1.14.11.-) (Ubiquitously-transcribed TPR protein on the Y chromosome) (Ubiquitously-transcribed Y chromosome tetratricopeptide repeat protein)	H1093-E1095-H1173	1 Fe cation	Catalytic	1.14.11.-	Nucleus	No		regulation of gene expression [GO:0010468]