	Uniprot Id	Entry name	Gene names	Protein names	Predicted Pattern	Number of cofactors	Iron-cofactor role	EC number	Subcellular location	Membrane associated	Involvement in disease	Gene ontology (biological process)
1	O75027	ABCB7_HUMAN	ABCB7 ABC7	ATP-binding cassette sub-family B member 7, mitochondrial (ATP-binding cassette transporter 7) (ABC transporter 7 protein)	Unknown	Fe_2S_2	Substrate - transport		Mitochondrion	Yes	DISEASE: Anemia, sideroblastic, spinocerebellar ataxia (ASAT) [MIM:301310]: A X-linked recessive disorder characterized by an infantile to early childhood onset of non-progressive cerebellar ataxia and mild anemia, with hypochromia and microcytosis. {ECO:0000269|PubMed:10196363, ECO:0000269|PubMed:11050011, ECO:0000269|PubMed:11843825, ECO:0000269|PubMed:22398176). Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular iron ion homeostasis [GO:0006879]; transmembrane transport [GO:0055085]; transport [GO:0006810]
2	P61221	ABCE1_HUMAN	ABCE1 RLI RNASEL1 RNASELI RNS4I OK/SW-cl.40	ATP-binding cassette sub-family E member 1 (2'-5'-oligoadenylate-binding protein) (HuHP68) (RNase L inhibitor) (Ribonuclease 4 inhibitor) (RNS4I)	C16-C21-C25-C29-C55-C58-C61-C65	2 × Fe_4S_4	Unknown		Cytoplasm, Mitochondrion, Cell membrane	Yes		negative regulation of endoribonuclease activity [GO:0060702]; regulation of type I interferon-mediated signaling pathway [GO:0060338]; ribosomal subunit export from nucleus [GO:0000054]; translational initiation [GO:0006413]; translational termination [GO:0006415]; viral process [GO:0016032]
3	P21399	ACOC_HUMAN	ACO1 IREB1	Cytoplasmic aconitate hydratase (Aconitase) (EC 4.2.1.3) (Citrate hydro-lyase) (Ferritin repressor protein) (Iron regulatory protein 1) (IRP1) (Iron-responsive element-binding protein 1) (IRE-BP 1)	C437-C503-C506	Fe_4S_4	Substrate - sensor	4.2.1.3	Cytoplasm	No		cellular iron ion homeostasis [GO:0006879]; citrate metabolic process [GO:0006101]; intestinal absorption [GO:0050892]; post-embryonic development [GO:0009791]; regulation of translation [GO:0006417]; response to iron(II) ion [GO:0010040]; tricarboxylic acid cycle [GO:0006099]
4	Q99798	ACON_HUMAN	ACO2	Aconitate hydratase, mitochondrial (Aconitase) (EC 4.2.1.3) (Citrate hydro-lyase)	C385-C448-C451	Fe_4S_4	Unknown	4.2.1.3	Mitochondrion	No	DISEASE: Infantile cerebellar-retinal degeneration (ICRD) [MIM:614559]: A severe autosomal recessive neurodegenerative disorder characterized by onset between ages 2 and 6 months of truncal hypotonia, athetosis, seizures, and ophthalmologic abnormalities, particularly optic atrophy and retinal degeneration. Affected individuals show profound psychomotor retardation, with only some achieving rolling, sitting, or recognition of family. Brain MRI shows progressive cerebral and cerebellar degeneration. {ECO:0000269|PubMed:22405087, ECO:0000269|PubMed:25351951). Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Optic atrophy 9 (OPA9) [MIM:616289]: A condition that features progressive visual loss in association with optic atrophy. Atrophy of the optic disk indicates a deficiency in the number of nerve fibers which arise in the retina and converge to form the optic disk, optic nerve, optic chiasm and optic tracts. {ECO:0000269|PubMed:25351951). Note=The disease is caused by mutations affecting the gene represented in this entry.	citrate metabolic process [GO:0006101]; generation of precursor metabolites and energy [GO:0006091]; isocitrate metabolic process [GO:0006102]; liver development [GO:0001889]; response to isolation stress [GO:0035900]; tricarboxylic acid cycle [GO:0006099]
5	P10109	ADX_HUMAN	FDX1 ADX	Adrenodoxin, mitochondrial (Adrenal ferredoxin) (Ferredoxin-1) (Hepatoredoxin)	C106-C112-C115-C152	Fe_2S_2	Electron transfer		Mitochondrion	No		C21-steroid hormone biosynthetic process [GO:0006700]; cellular response to cAMP [GO:0071320]; cellular response to forskolin [GO:1904322]; cholesterol metabolic process [GO:0008203]; hormone biosynthetic process [GO:0042446]; small molecule metabolic process [GO:0044281]; sterol metabolic process [GO:0016125]
6	Q96NN9	AIFM3_HUMAN	AIFM3 AIFL	Apoptosis-inducing factor 3 (EC 1.-.-.-) (Apoptosis-inducing factor-like protein)	C109-H111-C128-H131	Fe_2S_2 (predicted)	Unknown	1.-.-.-	Mitochondrion, Nucleus	No		execution phase of apoptosis [GO:0097194]
7	Q06278	AOXA_HUMAN	AOX1 AO	Aldehyde oxidase (EC 1.2.3.1) (Aldehyde oxidase 1) (Azaheterocycle hydroxylase) (EC 1.17.3.-)	C44-C49-C52-C74; C114-C117-C149-C151	2 × Fe_2S_2	Electron transfer	1.2.3.1; 1.17.3.-	Cytoplasm	No		drug metabolic process [GO:0017144]; oxidation-reduction process [GO:0055114]; vitamin B6 metabolic process [GO:0042816]; xanthine catabolic process [GO:0009115]
8	Q9Y3E2	BOLA1_HUMAN	BOLA1 CGI-143	BolA-like protein 1 (hBolA)	Unknown	Fe_2S_2 shared with GLRX	Substrate - biosinthesis		Mitochondrion	No		
9	Q9H3K6	BOLA2_HUMAN	BOLA2 BOLA2A My016; BOLA2B	BolA-like protein 2	Unknown	Fe_2S_2 shared with GLRX	Substrate - biosinthesis		Cytoplasm, Nucleus	No		[2Fe-2S] cluster assembly [GO:0044571]; interleukin-12-mediated signaling pathway [GO:0035722]; protein maturation by iron-sulfur cluster transfer [GO:0097428]
10	Q53S33	BOLA3_HUMAN	BOLA3	BolA-like protein 3	Unknown	Fe_2S_2 shared with GLRX	Substrate - biosinthesis		Mitochondrion	No	DISEASE: Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia (MMDS2) [MIM:614299]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. Some patients show failure to thrive, pulmonary hypertension, hypotonia and irritability. Biochemical features include severe combined deficiency of the 2-oxoacid dehydrogenases, defective lipoic acid synthesis and reduction in activity of mitochondrial respiratory chain complexes. {ECO:0000269|PubMed:21944046, ECO:0000269|PubMed:22562699, ECO:0000269|PubMed:24334290, ECO:0000269|PubMed:26741492). Note=The disease is caused by mutations affecting the gene represented in this entry.	
11	Q5VV42	CDKAL_HUMAN	CDKAL1	Threonylcarbamoyladenosine tRNA methylthiotransferase (EC 2.8.4.5) (CDK5 regulatory subunit-associated protein 1-like 1) (tRNA-t(6)A37 methylthiotransferase)	C73-C109-C138; C214-C218-C221	2 × Fe_4S_4	Catalytic	2.8.4.5	Endoplasmic reticulum	Yes	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:17460697, ECO:0000269|PubMed:17463246). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	maintenance of translational fidelity [GO:1990145]; tRNA modification [GO:0006400]
12	Q9NZ45	CISD1_HUMAN	CISD1 C10orf70 ZCD1	CDGSH iron-sulfur domain-containing protein 1 (MitoNEET)	C72-C74-C83-H87	Fe_2S_2	Substrate - biogenesis		Mitochondrion	Yes		regulation of cellular respiration [GO:0043457]
13	Q8N5K1	CISD2_HUMAN	CISD2 CDGSH2 ERIS ZCD2	CDGSH iron-sulfur domain-containing protein 2 (Endoplasmic reticulum intermembrane small protein) (MitoNEET-related 1 protein) (Miner1) (Nutrient-deprivation autophagy factor-1) (NAF-1)	C99-C101-C110-H114	Fe_2S_2	Unknown		Endoplasmic reticulum, Mitochondrion	Yes	DISEASE: Wolfram syndrome 2 (WFS2) [MIM:604928]: A rare disorder characterized by juvenile-onset insulin-dependent diabetes mellitus with optic atrophy. Other manifestations include diabetes insipidus, sensorineural deafness, dementia, psychiatric illnesses. WFS2 patients additionally show a strong bleeding tendency and gastrointestinal ulceration. Diabetes insipidus may be absent. {ECO:0000269|PubMed:17846994). Note=The disease is caused by mutations affecting the gene represented in this entry.	autophagy of mitochondrion [GO:0000422]; multicellular organism aging [GO:0010259]; regulation of autophagy [GO:0010506]
14	P0C7P0	CISD3_HUMAN	CISD3	CDGSH iron-sulfur domain-containing protein 3, mitochondrial (MitoNEET-related protein 2) (Miner2)	C60-C62-C71-H75; C98-C100-C109-H113	2 × Fe_2S_2	Unknown		Mitochondrion	No		
15	Q96SZ6	CKSP1_HUMAN	CDK5RAP1 C20orf34 CGI-05 HSPC167	CDK5 regulatory subunit-associated protein 1 (CDK5 activator-binding protein C42)	C109-C145-C183; C258-C262-C265	2 × Fe_4S_4	Catalytic		Unknown	No		brain development [GO:0007420]; mitochondrial tRNA modification [GO:0070900]; negative regulation of cyclin-dependent protein serine/threonine kinase activity [GO:0045736]; positive regulation of mitochondrial translation [GO:0070131]; positive regulation of translational fidelity [GO:0045903]; regulation of neuron differentiation [GO:0045664]
16	Q9Y471	CMAH_HUMAN	CMAHP CMAH	Inactive cytidine monophosphate-N-acetylneuraminic acid hydroxylase (CMP-NeuAc hydroxylase-like protein) (Cytidine monophosphate-N-acetylneuraminic acid hydroxylase pseudogene)	Unknown	Fe_2S_2	Unknown		Cytoplasm, Nucleus	Yes		regulation of Wnt signaling pathway [GO:0030111]
17	Q6FI81	CPIN1_HUMAN	CIAPIN1 CUA001 PRO0915	Anamorsin (Cytokine-induced apoptosis inhibitor 1) (Fe-S cluster assembly protein DRE2 homolog)	C237-C246-C249-C251	2 × Fe_2S_2	Substrate - biogenesis		Cytoplasm, Mitochondrion, Nucleus	Yes		apoptotic process [GO:0006915]; hemopoiesis [GO:0030097]; iron-sulfur cluster assembly [GO:0016226]; negative regulation of apoptotic process [GO:0043066]

18	Q96FC9	DDX11_HUMAN	DDX11 CHL1 CHLR1 KRG2	ATP-dependent DNA helicase DDX11 (EC 3.6.4.12) (CHL1-related protein 1) (hCHLR1) (DEAD/H-box protein 11) (Keratinocyte growth factor-regulated gene 2 protein) (KRG-2)	C267-C285-C315-C350	Fe_4S_4	Structural - Regulatory	3.6.4.12	Cytoplasm, Nucleus	No	DISEASE: Warsaw breakage syndrome (WBRS) [MIM:613398]: A syndrome characterized by severe microcephaly, pre- and postnatal growth retardation, facial dysmorphism and abnormal skin pigmentation. Additional features include high arched palate, coloboma of the right optic disk, deafness, ventricular septal defect, toes and fingers abnormalities. At cellular level, drug-induced chromosomal breakage, a feature of Fanconi anemia, and sister chromatid cohesion defects, a feature of Roberts syndrome, coexist. {ECO:0000269|PubMed:20137776, ECO:0000269|PubMed:23033317, ECO:0000269|PubMed:26089203}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular response to bleomycin [GO:1904976]; cellular response to cisplatin [GO:0072719]; cellular response to DNA damage stimulus [GO:0006974]; cellular response to hydroxyurea [GO:0072711]; DNA duplex unwinding [GO:0032508]; DNA repair [GO:0006281]; G-quadruplex DNA unwinding [GO:0044806]; IRE1-mediated unfolded protein response [GO:0036498]; multicellular organism development [GO:0007275]; negative regulation of protein binding [GO:0032091]; nucleolar chromatin organization [GO:1990700]; positive regulation of chromatin binding [GO:0035563]; positive regulation of double-strand break repair [GO:2000781]; positive regulation of endodeoxyribonuclease activity [GO:0032079]; positive regulation of sister chromatid cohesion [GO:0045876]; positive regulation of transcription of nuclear large rRNA transcript from RNA polymerase I promoter [GO:1901838]; replication fork processing [GO:0031297]; sister chromatid cohesion [GO:0007062]; transcription, DNA-templated [GO:0006351]; viral process [GO:0016032]
19	Q92771	DDX12_HUMAN	DDX12P CHLR2 DDX12	Putative ATP-dependent RNA helicase DDX12 (EC 3.6.4.13) (CHL1-related protein 2) (hCHLR2) (DEAD/H box protein 12)	C286-C304-C334-C369	Fe_4S_4	Structural - Regulatory	3.6.4.13	Nucleus	No		cell cycle [GO:0007049]; nucleobase-containing compound metabolic process [GO:0006139]
20	P51530	DNA2_HUMAN	DNA2 DNA2L KIAA0083	DNA replication ATP-dependent helicase/nuclease DNA2 (hDNA2) (DNA replication ATP-dependent helicase-like homolog) [Includes: DNA replication nuclease DNA2 (EC 3.1.-.-); DNA replication ATP-dependent helicase DNA2 (EC 3.6.4.12)]	C136-C393-C396-C402	Fe_4S_4	Structural - Regulatory	3.1.-.-; 3.6.4.12	Mitochondrion, Nucleus	No	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 (PEOA6) [MIM:615156]: A disorder characterized by muscle weakness, mainly affecting the lower limbs, external ophthalmoplegia, exercise intolerance, and mitochondrial DNA deletions on muscle biopsy. Symptoms may appear in childhood or adulthood and show slow progression. {ECO:0000269|PubMed:23352259}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Seckel syndrome 8 (SCKL8) [MIM:615807]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:24389050}. Note=The disease is caused by mutations affecting the gene represented in this entry.	base-excision repair [GO:0006284]; DNA double-strand break processing [GO:0000729]; DNA replication [GO:0006260]; DNA replication, Okazaki fragment processing [GO:0033567]; DNA replication, removal of RNA primer [GO:0043137]; DNA replication checkpoint [GO:0000076]; DNA synthesis involved in DNA repair [GO:0000731]; G-quadruplex DNA unwinding [GO:0044806]; mitochondrial DNA repair [GO:0043504]; mitochondrial DNA replication [GO:0006264]; mitotic telomere maintenance via semi-conservative replication [GO:1902990]; nucleic acid phosphodiester bond hydrolysis [GO:0090305]; positive regulation of DNA replication [GO:0045740]; regulation of signal transduction by p53 class mediator [GO:1901796]; strand displacement [GO:0000732]; t-circle formation [GO:0090656]; telomere maintenance [GO:0000723]; telomere maintenance via semi-conservative replication [GO:0032201]
21	Q9BZG8	DPH1_HUMAN	DPH1 DPH2L DPH2L1 OVCA1	2-(3-amino-3-carboxypropyl)histidine synthase subunit 1 (EC 2.5.1.108) (Diphthamide biosynthesis protein 1) (Diphtheria toxin resistance protein 1) (Ovarian cancer-associated gene 1 protein) (S-adenosyl-L-methionine:L-histidine 3-amino-3-carboxypropyltransferase 1)	C115-C219-C347	Fe_4S_4	Catalytic	2.5.1.108	Cytoplasm, Nucleus	No	DISEASE: Developmental delay with short stature, dysmorphic features, and sparse hair (DEDSSH) [MIM:616901]: An autosomal recessive syndrome characterized by intellectual disability, short stature, and craniofacial and ectodermal anomalies including scaphocephaly with or without craniosynostosis, prominent forehead, sparse eyebrows and hair, hypoplastic toenails and, in some cases, dental anomalies. {ECO:0000269|PubMed:25558065, ECO:0000269|PubMed:26220823}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cell proliferation [GO:0008283]; peptidyl-diphthamide biosynthetic process from peptidyl-histidine [GO:0017183]
22	Q9BQC3	DPH2_HUMAN	DPH2 DPH2L2	2-(3-amino-3-carboxypropyl)histidine synthase subunit 2 (EC 2.5.1.108) (Diphthamide biosynthesis protein 2) (Diphtheria toxin resistance protein 2) (S-adenosyl-L-methionine:L-histidine 3-amino-3-carboxypropyltransferase 2)	C88-C341	Fe_4S_4	Catalytic	2.5.1.108	Unknown	No		peptidyl-diphthamide biosynthetic process from peptidyl-histidine [GO:0017183]
23	P28340	DPOD1_HUMAN	POLD1 POLD	DNA polymerase delta catalytic subunit (EC 2.7.7.7) (EC 3.1.11.-) (DNA polymerase subunit delta p125)	C1058-C1061-C1071-C1076	Fe_4S_4	Structural - Regulatory	2.7.7.7; 3.1.11.-	Nucleus	No	DISEASE: Colorectal cancer 10 (CRCS10) [MIM:612591]: 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:23263490, ECO:0000269|PubMed:24501277}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.; DISEASE: Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL) [MIM:615381]: An autosomal dominant systemic disorder characterized by prominent loss of subcutaneous fat, metabolic abnormalities including insulin resistance and diabetes mellitus, sclerodermatous skin, and a facial appearance characterized by mandibular hypoplasia. Sensorineural deafness occurs late in the first or second decades of life. {ECO:0000269|PubMed:23770608}. Note=The disease is caused by mutations affecting the gene represented in this entry.	base-excision repair, gap-filling [GO:0006287]; cellular response to UV [GO:0034644]; DNA damage response, detection of DNA damage [GO:0042769]; DNA ligation [GO:0006266]; DNA repair [GO:0006281]; DNA replication [GO:0006260]; DNA replication proofreading [GO:0045004]; DNA synthesis involved in DNA repair [GO:0000731]; fatty acid homeostasis [GO:0055089]; mismatch repair [GO:0006298]; nucleotide-excision repair, DNA gap filling [GO:0006297]; nucleotide-excision repair, DNA incision [GO:0033683]; nucleotide-excision repair, DNA incision, 5'-to lesion [GO:0006296]; response to UV [GO:0009411]; telomere maintenance [GO:0000723]; telomere maintenance via semi-conservative replication [GO:0032201]; transcription-coupled nucleotide-excision repair [GO:0006283]; translesion synthesis [GO:0019985]
24	Q07864	DPOE1_HUMAN	POLE POLE1	DNA polymerase epsilon catalytic subunit A (EC 2.7.7.7) (DNA polymerase II subunit A)	C2221-C2224-C2236-C2238	Fe_4S_4	Structural - Regulatory	2.7.7.7	Nucleus	No	DISEASE: Colorectal cancer 12 (CRCS12) [MIM:615083]: 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. CRCS12 is characterized by a high-penetrance predisposition to the development of colorectal adenomas and carcinomas, with a variable tendency to develop multiple and large tumors. Onset is usually before age 40 years. The histologic features of the tumors are unremarkable. {ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24501277, ECO:0000269|PubMed:25860647, ECO:0000269|PubMed:27573199}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.; DISEASE: Facial dysmorphism, immunodeficiency, livedo, and short stature (FILS) [MIM:615139]: A syndrome characterized by mild facial dysmorphism, mainly malar hypoplasia, livedo on the skin since birth, and immunodeficiency resulting in recurrent infections. Growth impairment is observed during early childhood and results in variable short stature in adulthood. {ECO:0000269|PubMed:23230001}. Note=The disease is caused by mutations affecting the gene represented in this entry.	base-excision repair, gap-filling [GO:0006287]; DNA replication [GO:0006260]; DNA replication initiation [GO:0006270]; DNA replication proofreading [GO:0045004]; DNA synthesis involved in DNA repair [GO:0000731]; embryonic organ development [GO:0048568]; G1/S transition of mitotic cell cycle [GO:0000082]; leading strand elongation [GO:0006272]; nucleotide-excision repair, DNA gap filling [GO:0006297]; telomere maintenance via semi-conservative replication [GO:0032201]

25	P09884	DPOLA_HUMAN	POLA1 POLA	DNA polymerase alpha catalytic subunit (EC 2.7.7.7) (DNA polymerase alpha catalytic subunit p180)	C1348-C1353-C1371-C1374	Fe_4S_4	Structural - Regulatory	2.7.7.7	Cytoplasm, Nucleus	No	DISEASE: Pigmentary disorder, reticulate, with systemic manifestations, X-linked (PDR) [MIM:301220]: A X-linked recessive disorder characterized by recurrent infections and sterile inflammation in various organs. Diffuse skin hyperpigmentation with a distinctive reticulate pattern is universally evident by early childhood. This is later followed in many patients by hypohidrosis, corneal inflammation and scarring, enterocolitis that resembles inflammatory bowel disease, and recurrent urethral strictures. Melanin and amyloid deposition is present in the dermis. Affected males also have a characteristic facies with frontally upswept hair and flared eyebrows. Female carriers have only restricted pigmentary changes along Blaschko's lines. {ECO:0000269|PubMed:27019227}. Note=The disease is caused by mutations affecting the gene represented in this entry. XLPDR is caused by a recurrent intronic mutation that results in missplicing and reduced POLA1 expression. This leads to a decrease in cytosolic RNA:DNA hybrids and constitutive activation of type I interferon responses, but has no effect on cell replication. {ECO:0000269|PubMed:27019227}.	cell proliferation [GO:0008283]; DNA replication [GO:0006260]; DNA replication, synthesis of RNA primer [GO:0006269]; DNA replication initiation [GO:0006270]; DNA strand elongation involved in DNA replication [GO:0006271]; double-strand break repair via nonhomologous end joining [GO:0006303]; G1/S transition of mitotic cell cycle [GO:0000082]; lagging strand elongation [GO:0006273]; leading strand elongation [GO:0006272]; regulation of transcription involved in G1/S transition of mitotic cell cycle [GO:0000083]; telomere maintenance via semi-conservative replication [GO:0032201]; viral process [GO:0016032]
26	Q12882	DPYD_HUMAN	DPYD	Dihydropyrimidine dehydrogenase [NADP(+)] (DHPDHase) (DPD) (EC 1.3.1.2) (Dihydrothymine dehydrogenase) (Dihydrouracil dehydrogenase)	C79-C82-C87-C91; C130-C136-C140-C156; C953-C956-C959-C963; C986-C989-C992-C996	4 × Fe_4S_4	Unknown	1.3.1.2	Cytoplasm	No	DISEASE: Dihydropyrimidine dehydrogenase deficiency (DPYDD) [MIM:274270]: A metabolic disorder with large phenotypic variability, ranging from no symptoms to a convulsive disorder with motor and mental retardation. It is characterized by persistent urinary excretion of excessive amounts of uracil, thymine and 5-hydroxymethyluracil. Patients suffering from this disease show a severe reaction to the anticancer drug 5-fluorouracil. {ECO:0000269|PubMed:14702039, ECO:0000269|PubMed:16710414, ECO:0000269|PubMed:9266349, ECO:0000269|PubMed:9439663}. Note=The disease is caused by mutations affecting the gene represented in this entry.	beta-alanine biosynthetic process [GO:0019483]; purine nucleobase catabolic process [GO:0006145]; pyrimidine nucleobase catabolic process [GO:0006208]; pyrimidine nucleoside catabolic process [GO:0046135]; thymidine catabolic process [GO:0006214]; thymine catabolic process [GO:0006210]; uracil catabolic process [GO:0006212]
27	Q9H9T3	ELP3_HUMAN	ELP3	Elongator complex protein 3 (hELP3) (EC 2.3.1.48)	C99-C109-C112	Fe_4S_4	Catalytic	2.3.1.48	Cytoplasm	No	DISEASE: Note=ELP3 genetic variations may be associated with an increased risk for neurodegeneration and motor neuron diseases. {ECO:0000303|PubMed:18996918}.	central nervous system development [GO:0007417]; histone H3 acetylation [GO:0043966]; histone H4 acetylation [GO:0043967]; neuron migration [GO:0001764]; positive regulation of cell migration [GO:0030335]; regulation of transcription from RNA polymerase II promoter [GO:0006357]; transcription elongation from RNA polymerase II promoter [GO:0006368]
28	P18074	ERCC2_HUMAN	ERCC2 XPD XPDC	TFIIH basal transcription factor complex helicase XPD subunit (EC 3.6.4.12) (Basic transcription factor 2 80 kDa subunit) (BTF2 p80) (CXPD) (DNA excision repair protein ERCC-2) (DNA repair protein complementing XP-D cells) (TFIIH basal transcription factor complex 80 kDa subunit) (TFIIH 80 kDa subunit) (TFIIH p80) (Xeroderma pigmentosum group D-complementing protein)	C116-C134-C155-C190	Fe_4S_4	Structural - Regulatory	3.6.4.12	Cytoplasm, Nucleus	No	DISEASE: Xeroderma pigmentosum complementation group D (XP-D) [MIM:278730]: An autosomal recessive pigmentary skin disorder characterized by solar hypersensitivity of the skin, high predisposition for developing cancers on areas exposed to sunlight and, in some cases, neurological abnormalities. The skin develops marked freckling and other pigmentation abnormalities. Some XP-D patients present features of Cockayne syndrome, including cachectic dwarfism, pigmentary retinopathy, ataxia, decreased nerve conduction velocities. The phenotype combining xeroderma pigmentosum and Cockayne syndrome traits is referred to as XP-CS complex. {ECO:0000269|PubMed:10447254, ECO:0000269|PubMed:11709541, ECO:0000269|PubMed:15494306, ECO:0000269|PubMed:7585650, ECO:0000269|PubMed:7825573, ECO:0000269|PubMed:7849702, ECO:0000269|PubMed:9101292}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Trichothiodystrophy 1, photosensitive (TTD1) [MIM:601675]: A form of trichothiodystrophy, an autosomal recessive disease characterized by sulfur-deficient brittle hair and multisystem variable abnormalities. The spectrum of clinical features varies from mild disease with only hair involvement to severe disease with cutaneous, neurologic and profound developmental defects. Ichthyosis, intellectual and developmental disabilities, decreased fertility, abnormal characteristics at birth, ocular abnormalities, short stature, and infections are common manifestations. There are both photosensitive and non-photosensitive forms of the disorder. TTD1 patients manifest cutaneous photosensitivity. {ECO:0000269|PubMed:11242112, ECO:0000269|PubMed:7920640, ECO:0000269|PubMed:8571952, ECO:0000269|PubMed:9195225, ECO:0000269|PubMed:9238033, ECO:0000269|PubMed:9758621}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Cerebro-oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic	7-methylguanosine mRNA capping [GO:0006370]; aging [GO:0007568]; apoptotic process [GO:0006915]; bone mineralization [GO:0030282]; cell proliferation [GO:0008283]; central nervous system myelin formation [GO:0032289]; chromosome segregation [GO:0007059]; embryonic cleavage [GO:0040016]; embryonic organ development [GO:0048568]; erythrocyte maturation [GO:0043249]; extracellular matrix organization [GO:0030198]; global genome nucleotide-excision repair [GO:0070911]; hair cell differentiation [GO:0035315]; hair follicle maturation [GO:0048820]; hematopoietic stem cell differentiation [GO:0060218]; in utero embryonic development [GO:0001701]; multicellular organism growth [GO:0035264]; nucleotide-excision repair [GO:0006289]; nucleotide-excision repair, DNA duplex unwinding [GO:0000717]; nucleotide-excision repair, DNA incision [GO:0033683]; nucleotide-excision repair, DNA incision, 3'-to lesion [GO:0006295]; nucleotide-excision repair, DNA incision, 5'-to lesion [GO:0006296]; nucleotide-excision repair, preincision complex assembly [GO:0006294]; nucleotide-excision repair, preincision complex stabilization [GO:0006293]; positive regulation of DNA binding [GO:0043388]; positive regulation of transcription, DNA-templated [GO:0045893]; positive regulation of transcription from RNA polymerase II promoter [GO:0045944]; post-embryonic development [GO:0009791]; protein phosphorylation [GO:0006468]; regulation of mitotic cell cycle phase transition [GO:1901990]; response to hypoxia [GO:0001666]; response to oxidative stress [GO:0006979]; spinal cord development [GO:0021510]; termination of RNA polymerase I transcription [GO:0006363]; transcription-coupled nucleotide-excision repair [GO:0006283]; transcription elongation from RNA polymerase II promoter [GO:0006368]; transcription elongation from RNA polymerase I promoter [GO:0006362]; transcription from RNA polymerase II promoter [GO:0006366];
29	Q16134	ETFD_HUMAN	ETFDH	Electron transfer flavoprotein-ubiquinone oxidoreductase, mitochondrial (ETF-QO) (ETF-ubiquinone oxidoreductase) (EC 1.5.5.1) (Electron-transferring-flavoprotein dehydrogenase) (ETF dehydrogenase)	C561-C586-C589-C592	Fe_4S_4	Electron transfer	1.5.5.1	Mitochondrion	Yes	DISEASE: Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:12359134, ECO:0000269|PubMed:12815589, ECO:0000269|PubMed:16527485, ECO:0000269|PubMed:17412732, ECO:0000269|PubMed:19249206, ECO:0000269|PubMed:20370797}. Note=The disease is caused by mutations affecting the gene represented in this entry.	electron transport chain [GO:0022900]; fatty acid beta-oxidation using acyl-CoA dehydrogenase [GO:0033539]; respiratory electron transport chain [GO:0022904]; response to oxidative stress [GO:0006979]
30	Q9BX63	FANCJ_HUMAN	BRIP1 BACH1 FANCJ	Fanconi anemia group J protein (Protein FACJ) (EC 3.6.4.13) (ATP-dependent RNA helicase BRIP1) (BRCA1-associated C-terminal helicase 1) (BRCA1-interacting protein C-terminal helicase 1) (BRCA1-interacting protein 1)	C283-C298-C310-C350	Fe_4S_4	Structural - Regulatory	3.6.4.13	Nucleus	No	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:11301010, ECO:0000269|PubMed:14983014}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.; DISEASE: Fanconi anemia complementation group J (FANCJ) [MIM:609054]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:16116423, ECO:0000269|PubMed:16116424, ECO:0000269|PubMed:20639400}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular response to angiotensin [GO:1904385]; cellular response to hypoxia [GO:0071456]; cellular response to vitamin [GO:0071295]; chiasma assembly [GO:0051026]; DNA damage checkpoint [GO:0000077]; DNA replication [GO:0006260]; DNA synthesis involved in DNA repair [GO:0000731]; double-strand break repair [GO:0006302]; double-strand break repair involved in meiotic recombination [GO:1990918]; meiotic DNA double-strand break processing involved in reciprocal meiotic recombination [GO:0010705]; negative regulation of cell proliferation [GO:0008285]; negative regulation of gene expression [GO:0010629]; regulation of signal transduction by p53 class mediator [GO:1901796]; regulation of transcription from RNA polymerase II promoter [GO:0006357]; response to toxic substance [GO:0009636]; seminiferous tubule development [GO:0072520]; spermatid development [GO:0007286]; spermatogonial cell division [GO:0007284]; strand displacement [GO:0000732]
31	Q6P4F2	FDX2_HUMAN	FDX2 FDX1L	Ferredoxin-2, mitochondrial (Adrenodoxin-like protein) (Ferredoxin-1-like protein)	C105-C111-C114-C151	Fe_2S_2	Substrate - biogenesis		Mitochondrion	No		C21-steroid hormone biosynthetic process [GO:0006700]; small molecule metabolic process [GO:0044281]; sterol metabolic process [GO:0016125]

32	P80404	GABT_HUMAN	ABAT GABAT	4-aminobutyrate aminotransferase, mitochondrial (EC 2.6.1.19) ((S)-3-amino-2-methylpropionate transaminase) (EC 2.6.1.22) (GABA aminotransferase) (GABA-AT) (Gamma-amino-N-butyrate transaminase) (GABA transaminase) (GABA-T) (L-AIBAT)	C163-C166	Fe_2S_2 per homodimer	Unknown	2.6.1.19; 2.6.1.22	Mitochondrion	No	DISEASE: GABA transaminase deficiency (GABATD) [MIM:613163]: An enzymatic deficiency resulting in psychomotor retardation, hypotonia, hyperreflexia, lethargy, refractory seizures, and EEG abnormalities. {ECO:0000269|PubMed:10407778}. Note=The disease is caused by mutations affecting the gene represented in this entry.	aging [GO:0007568]; behavioral response to cocaine [GO:0048148]; cerebellum development [GO:0021549]; copulation [GO:0007620]; exploration behavior [GO:0035640]; gamma-aminobutyric acid biosynthetic process [GO:0009449]; gamma-aminobutyric acid catabolic process [GO:0009450]; locomotory behavior [GO:0007626]; negative regulation of blood pressure [GO:0045776]; negative regulation of dopamine secretion [GO:0033602]; negative regulation of gamma-aminobutyric acid secretion [GO:0014053]; negative regulation of platelet aggregation [GO:0090331]; neurotransmitter catabolic process [GO:0042135]; positive regulation of aspartate secretion [GO:1904450]; positive regulation of dopamine metabolic process [GO:0045964]; positive regulation of heat generation [GO:0031652]; positive regulation of inhibitory postsynaptic potential [GO:0097151]; positive regulation of insulin secretion [GO:0032024]; positive regulation of prolactin secretion [GO:1902722]; positive regulation of uterine smooth muscle contraction [GO:0070474]; response to drug [GO:0042493]; response to ethanol [GO:0045471]; response to hypoxia [GO:0001666]; response to iron ion [GO:0010039]; response to nicotine [GO:0035094]
33	Q9NS18	GLRX2_HUMAN	GLRX2 GRX2 CGI-133	Glutaredoxin-2, mitochondrial	C77	Fe_2S_2 per homodimer	Substrate - biosinthesis		Mitochondrion	No		aging [GO:0007568]; apoptotic process [GO:0006915]; cell differentiation [GO:0030154]; cell redox homeostasis [GO:0045454]; cellular response to superoxide [GO:0071451]; DNA protection [GO:0042262]; glutathione metabolic process [GO:0006749]; regulation of signal transduction [GO:0009966]; regulation of transcription, DNA-templated [GO:0006355]; response to hydrogen peroxide [GO:0042542]; response to organic substance [GO:0010033]; response to redox state [GO:0051775]; response to temperature stimulus [GO:0009266]
34	O76003	GLRX3_HUMAN	GLRX3 PICOT TXNL2 HUSSY-22	Glutaredoxin-3 (PKC-interacting cousin of thioredoxin) (PICOT) (PKC-theta-interacting protein) (PKCq-interacting protein) (Thioredoxin-like protein 2)	C159-C261	Fe_2S_2 shared with partner	Substrate - biosinthesis		Cytoplasm, Cell membrane	Yes		[2Fe-2S] cluster assembly [GO:0044571]; cell redox homeostasis [GO:0045454]; negative regulation of cardiac muscle hypertrophy [GO:0010614]; protein maturation by iron-sulfur cluster transfer [GO:0097428]; regulation of the force of heart contraction [GO:0002026]
35	Q86SX6	GLRX5_HUMAN	GLRX5 C14orf87	Glutaredoxin-related protein 5, mitochondrial (Monothiol glutaredoxin-5)	C67	Fe_2S_2	Substrate - biogenesis		Mitochondrion	No	DISEASE: Anemia, sideroblastic, 3, pyridoxine-refractory (SIDBA3) [MIM:616860]: A form of sideroblastic anemia, a bone marrow disorder defined by the presence of pathologic iron deposits in erythroblast mitochondria. Sideroblastic anemia is characterized by anemia of varying severity, hypochromic peripheral erythrocytes, systemic iron overload secondary to chronic ineffective erythropoiesis, and the presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-loaded mitochondria clustered around the nucleus. SIDBA3 is refractory to treatment with vitamin B6, while iron chelation therapy may result in clinical improvement. SIDBA3 inheritance is autosomal recessive. {ECO:0000269|PubMed:17485548, ECO:0000269|PubMed:20364084, ECO:0000269|PubMed:25342667, ECO:0000269|PubMed:26100117}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Spasticity, childhood-onset, with hyperglycinemia (SPAHGC) [MIM:616859]: An autosomal recessive disorder characterized by childhood-onset of spasticity, spinal lesions, leukodystrophy, optic atrophy in some patients, non-ketotic hyperglycinemia, and defective enzymatic glycine cleavage. Glycine levels in the cerebrospinal fluid are mildly increased in some but not all patients. The increase is less pronounced than in patients with classic non-ketotic hyperglycinemia. {ECO:0000269|PubMed:24334290}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cell redox homeostasis [GO:0045454]; hemopoiesis [GO:0030097]; protein lipoylation [GO:0009249]; small molecule metabolic process [GO:0044281]
36	A8MXD5	GRCR1_HUMAN	GRXCR1 DFNB25	Glutaredoxin domain-containing cysteine-rich protein 1	C156	Fe_2S_2 (predicted)	Unknown		Unknown	No	DISEASE: Deafness, autosomal recessive, 25 (DFNB25) [MIM:613285]: A form of non-syndromic sensorineural deafness characterized by moderate to severe or profound hearing loss which is progressive in some individuals but not in others. Speech development is impaired in some but not all affected individuals, and vestibular dysfunction is observed in some affected individuals. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:20137774, ECO:0000269|PubMed:20137778}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cell redox homeostasis [GO:0045454]; inner ear receptor cell development [GO:0060119]; inner ear receptor stereocilium organization [GO:0060122]; negative regulation of phosphatase activity [GO:0010923]; sensory perception of sound [GO:0007605]; vestibular receptor cell development [GO:0060118]
37	P22830	HEMH_HUMAN	FECH	Ferrochelatase, mitochondrial (EC 4.99.1.1) (Heme synthase) (Protoheme ferro-lyase)	C196-C403-C406-C411	Fe_2S_2	Regulatory	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]
38	P48200	IREB2_HUMAN	IREB2	Iron-responsive element-binding protein 2 (IRE-BP 2) (Iron regulatory protein 2) (IRP2)	C512-C578-C581	Fe_4S_4	Substrate - sensor		Cytoplasm	No		cellular iron ion homeostasis [GO:0006879]; iron ion transport [GO:0006826]; metabolic process [GO:0008152]
39	Q9BUE6	ISCA1_HUMAN	ISCA1 HBLD2 GK004	Iron-sulfur cluster assembly 1 homolog, mitochondrial (HESB-like domain-containing protein 2) (Iron-sulfur assembly protein IscA) (hIscA)	C57-C121-C123	Fe_2S_2/Fe_4S_4	Substrate - biogenesis		Mitochondrion	No	DISEASE: Multiple mitochondrial dysfunctions syndrome 5 (MMDS5) [MIM:617613]: An autosomal recessive, severe disorder characterized by early onset neurological deterioration, seizures, cerebral and cerebellar leukodystrophy, dysmyelination, cortical migrational abnormalities, lactic acidosis and early demise. {ECO:0000269|PubMed:28356563}. Note=The disease is caused by mutations affecting the gene represented in this entry.	iron-sulfur cluster assembly [GO:0016226]; protein maturation by iron-sulfur cluster transfer [GO:0097428]; small molecule metabolic process [GO:0044281]

40	Q86U28	ISCA2_HUMAN	ISCA2 HBLD1	Iron-sulfur cluster assembly 2 homolog, mitochondrial (HESB-like domain-containing protein 1)	C79-C144-C146	Fe_2S_2/Fe_4S_4	Substrate - biogenesis		Mitochondrion	No	DISEASE: Multiple mitochondrial dysfunctions syndrome 4 (MMDS4) [MIM:616370]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. {ECO:0000269|PubMed:25539947}. Note=The disease is caused by mutations affecting the gene represented in this entry.	iron-sulfur cluster assembly [GO:0016226]; protein maturation [GO:0051604]; protein maturation by iron-sulfur cluster transfer [GO:0097428]; small molecule metabolic process [GO:0044281]
41	Q9H1K1	ISCU_HUMAN	ISCU NIFUN	Iron-sulfur cluster assembly enzyme ISCU, mitochondrial (NifU-like N-terminal domain-containing protein) (NifU-like protein)	C69-C95-H137-C138	Fe_2S_2	Substrate - biogenesis		Mitochondrion	No	DISEASE: Myopathy with exercise intolerance Swedish type (MEIS) [MIM:255125]: Autosomal recessive metabolic disease characterized by lifelong severe exercise intolerance, in which minor exertion causes fatigue of active muscles, shortness of breath, and cardiac palpitations in association with lactic acidosis. The biochemical phenotype is characterized by a deficiency in mitochondrial iron-sulfur proteins and impaired muscle oxidative metabolism. {ECO:0000269|PubMed:18304497}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular iron ion homeostasis [GO:0006879]; iron-sulfur cluster assembly [GO:0016226]; protein maturation by iron-sulfur cluster transfer [GO:0097428]; small molecule metabolic process [GO:0044281]
42	O43766	LIAS_HUMAN	LIAS LAS HUSSY-01	Lipoyl synthase, mitochondrial (EC 2.8.1.8) (Lipoate synthase) (LS) (Lip-syn) (Lipoic acid synthase)	C106-C111-C117; C137-C141-C144	2 × Fe_4S_4	Electron transfer, Catalytic	2.8.1.8	Mitochondrion	No	DISEASE: Hyperglycinemia, lactic acidosis, and seizures (HGCLAS) [MIM:614462]: An enzymatic defect resulting in an autosomal recessive disorder of mitochondrial metabolism. It is characterized by early-onset lactic acidosis, severe encephalomyopathy, and a pyruvate oxidation defect. Affected individuals have neonatal-onset epilepsy, poor growth, psychomotor retardation, muscular hypotonia, lactic acidosis, and elevated glycine concentration in plasma and urine. {ECO:0000269|PubMed:22152680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	cellular nitrogen compound metabolic process [GO:0034641]; inflammatory response [GO:0006954]; lipoate biosynthetic process [GO:0009107]; neural tube closure [GO:0001843]; protein lipoylation [GO:0009249]; response to lipopolysaccharide [GO:0032496]; response to oxidative stress [GO:0006979]
43	Q9NZB8	MOCS1_HUMAN	MOCS1 MIG11	Molybdenum cofactor biosynthesis protein 1 (Cell migration-inducing gene 11 protein) (Molybdenum cofactor synthesis-step 1 protein A-B) [Includes: GTP 3',8-cyclase (EC 4.1.99.22) (Molybdenum cofactor biosynthesis protein A); Cyclic pyranopterin monophosphate synthase (EC 4.6.1.17) (Molybdenum cofactor biosynthesis protein C)]	C80-C84-C87; C312-C315-C329	2 × Fe_4S_4	Catalytic, Structural	4.1.99.22; 4.6.1.17	Unknown	No	DISEASE: Molybdenum cofactor deficiency, complementation group A (MOCODA) [MIM:252150]: An autosomal recessive metabolic disorder leading to the pleiotropic loss of molybdoenzyme activities. It is clinically characterized by onset in infancy of poor feeding, intractable seizures, severe psychomotor retardation, and death in early childhood in most patients. {ECO:0000269|PubMed:12754701, ECO:0000269|PubMed:16021469, ECO:0000269|PubMed:9731530, ECO:0000269|PubMed:9921896}. Note=The disease is caused by mutations affecting the gene represented in this entry.	molybdopterin cofactor biosynthetic process [GO:0032324]; Mo-molybdopterin cofactor biosynthetic process [GO:0006777]
44	Q9UIF7	MUTYH_HUMAN	MUTYH MYH	Adenine DNA glycosylase (EC 3.2.2.-) (MutY homolog) (hMYH)	C287-C294-C297-C303	Fe_4S_4	Structural - Regulatory	3.2.2.-	Mitochondrion, Nucleus	No	DISEASE: Familial adenomatous polyposis 2 (FAP2) [MIM:608456]: A condition characterized by the development of multiple colorectal adenomatous polyps, benign neoplasms derived from glandular epithelium. Some affected individuals may develop colorectal carcinoma. {ECO:0000269|PubMed:11818965, ECO:0000269|PubMed:12606733, ECO:0000269|PubMed:12853198, ECO:0000269|PubMed:15366000, ECO:0000269|PubMed:16134147, ECO:0000269|PubMed:16287072, ECO:0000269|PubMed:16557584, ECO:0000269|PubMed:16941501, ECO:0000269|PubMed:18091433, ECO:0000269|PubMed:18515411, ECO:0000269|PubMed:19953527, ECO:0000269|PubMed:20418187, ECO:0000269|PubMed:20848659, ECO:0000269|PubMed:25820570, ECO:0000269|PubMed:26694661}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Gastric cancer (GASC) [MIM:613659]: A malignant disease which starts in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. The term gastric cancer or gastric carcinoma refers to adenocarcinoma of the stomach that accounts for most of all gastric malignant tumors. Two main histologic types are recognized, diffuse type and intestinal type carcinomas. Diffuse tumors are poorly differentiated infiltrating lesions, resulting in thickening of the stomach. In contrast, intestinal tumors are usually exophytic, often ulcerating, and associated with intestinal metaplasia of the stomach, most often observed in sporadic disease. {ECO:0000269|PubMed:15273732, ECO:0000269|PubMed:25820570}. Note=The gene represented in this entry may be involved in disease pathogenesis. Somatic mutations contribute to the development of a sub-set of sporadic gastric cancers in carriers of Helicobacter pylori (PubMed:15273732). {ECO:0000269|PubMed:15273732}.	depurination [GO:0045007]; DNA repair [GO:0006281]; mismatch repair [GO:0006298]
45	Q9UHQ1	NARF_HUMAN	NARF	Nuclear prelamin A recognition factor (Iron-only hydrogenase-like protein 2) (IOP2)	C172-C228-C374-C378	2 × Fe_4S_4	Unknown		Nucleus	No		
46	Q9H6Q4	NARFL_HUMAN	NARFL PRN	Cytosolic Fe-S cluster assembly factor NARFL (Iron-only hydrogenase-like protein 1) (IOP1) (Nuclear prelamin A recognition factor-like protein) (Protein related to Narf)	C24-C71-C74-C77; C190-C246-C395-C399	2 × Fe_4S_4	Substrate - biogenesis		Unknown	No		hematopoietic progenitor cell differentiation [GO:0002244]; iron-sulfur cluster assembly [GO:0016226]; oxygen homeostasis [GO:0032364]; regulation of gene expression [GO:0010468]; response to hypoxia [GO:0001666]
47	P28331	NDUS1_HUMAN	NDUFS1	NADH-ubiquinone oxidoreductase 75 kDa subunit, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (Complex I-75kD) (CI-75kD)	C64-C75-C78-C92; H124-C128-C131-C137; C176-C179-C182-C226	2 × Fe_4S_4, Fe_2S_2	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	Yes	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:11349233}. Note=The disease is caused by mutations affecting the gene represented in this entry.	apoptotic mitochondrial changes [GO:0008637]; ATP metabolic process [GO:0046034]; cellular respiration [GO:0045333]; mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]; reactive oxygen species metabolic process [GO:0072593]; regulation of mitochondrial membrane potential [GO:0051881]
48	O75306	NDUS2_HUMAN	NDUFS2	NADH dehydrogenase [ubiquinone] iron-sulfur protein 2, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (Complex I-49kD) (CI-49kD) (NADH-ubiquinone oxidoreductase 49 kDa subunit)	C326-C332-C347	Fe_4S_4	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	Yes	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:11220739}. Note=The disease is caused by mutations affecting the gene represented in this entry.	mitochondrial ATP synthesis coupled electron transport [GO:0042775]; mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]; response to oxidative stress [GO:0006979]

49	O75251	NDUS7_HUMAN	NDUFS7	NADH dehydrogenase [ubiquinone] iron-sulfur protein 7, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (Complex I-20kD) (CI-20kD) (NADH-ubiquinone oxidoreductase 20 kDa subunit) (PSST subunit)	C88-C89-C153-C183	Fe_4S_4	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	No	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:10360771}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:10330338}. Note=The disease is caused by mutations affecting the gene represented in this entry.	mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]
50	O00217	NDUS8_HUMAN	NDUFS8	NADH dehydrogenase [ubiquinone] iron-sulfur protein 8, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (Complex I-23kD) (CI-23kD) (NADH-ubiquinone oxidoreductase 23 kDa subunit) (TYKY subunit)	C111-C114-C117-C160; C121-C150-C153-C156	2 × Fe_4S_4	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	No	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:9837812}. Note=The disease is caused by mutations affecting the gene represented in this entry.	mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]; response to oxidative stress [GO:0006979]
51	P49821	NDUV1_HUMAN	NDUFV1 UQOR1	NADH dehydrogenase [ubiquinone] flavoprotein 1, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (Complex I-51kD) (CI-51kD) (NADH dehydrogenase flavoprotein 1) (NADH-ubiquinone oxidoreductase 51 kDa subunit)	C379-C382-C385-C425	Fe_4S_4	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	Yes	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:10080174}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:10080174, ECO:0000269|PubMed:11349233}. Note=The disease is caused by mutations affecting the gene represented in this entry.	mitochondrial ATP synthesis coupled electron transport [GO:0042775]; mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]
52	P19404	NDUV2_HUMAN	NDUFV2	NADH dehydrogenase [ubiquinone] flavoprotein 2, mitochondrial (EC 1.6.5.3) (EC 1.6.99.3) (NADH-ubiquinone oxidoreductase 24 kDa subunit)	C135-C140-C176-C180	Fe_2S_2	Electron transfer	1.6.5.3; 1.6.99.3	Mitochondrion	Yes		cardiac muscle tissue development [GO:0048738]; mitochondrial electron transport, NADH to ubiquinone [GO:0006120]; mitochondrial respiratory chain complex I assembly [GO:0032981]; nervous system development [GO:0007399]
53	Q9Y697	NFS1_HUMAN	NFS1 NIFS HUSSY-08	Cysteine desulfurase, mitochondrial (EC 2.8.1.7)	C381	Fe_2S_2	Substrate - biogenesis	2.8.1.7	Mitochondrion	No		[2Fe-2S] cluster assembly [GO:0044571]; iron incorporation into metallo-sulfur cluster [GO:0018283]; molybdopterin cofactor biosynthetic process [GO:0032324]; Mo-molybdopterin cofactor biosynthetic process [GO:0006777]; protein complex assembly [GO:0006461]; small molecule metabolic process [GO:0044281]; sulfur amino acid metabolic process [GO:0000096]
54	Q9UMS0	NFU1_HUMAN	NFU1 HIRIP5 CGI-33	NFU1 iron-sulfur cluster scaffold homolog, mitochondrial (HIRA-interacting protein 5)	C210-C213	Fe_4S_4	Substrate - biogenesis		Cytoplasm, Mitochondrion	No	DISEASE: Multiple mitochondrial dysfunctions syndrome 1 (MMDS1) [MIM:605711]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. Some patients show failure to thrive, pulmonary hypertension, hypotonia and irritability. Biochemical features include severe combined deficiency of the 2-oxoacid dehydrogenases, defective lipoic acid synthesis and reduction in activity of mitochondrial respiratory chain complexes. {ECO:0000269|PubMed:21944046, ECO:0000269|PubMed:22077971, ECO:0000269|PubMed:25918518, ECO:0000269|PubMed:28161430, ECO:0000269|PubMed:28906594}. Note=The disease is caused by mutations affecting the gene represented in this entry.	iron-sulfur cluster assembly [GO:0016226]
55	P78549	NTH_HUMAN	NTHL1 NTH1 OCTS3	Endonuclease III-like protein 1 (hNTH1) (EC 3.2.2.-) (EC 4.2.99.18) (Bifunctional DNA N-glycosylase/DNA-(apurinic or apyrimidinic site) lyase) (DNA glycosylase/AP lyase)	C290-C297-C300-C306	Fe_4S_4	Structural - Regulatory	3.2.2.-; 4.2.99.18	Mitochondrion, Nucleus	No	DISEASE: Familial adenomatous polyposis 3 (FAP3) [MIM:616415]: A form of familial adenomatous polyposis, a condition characterized by the development of multiple colorectal adenomatous polyps, benign neoplasms derived from glandular epithelium. Some affected individuals may develop colorectal carcinoma. {ECO:0000269|PubMed:25938944}. Note=The disease is caused by mutations affecting the gene represented in this entry.	base-excision repair, AP site formation [GO:0006285]; depyrimidination [GO:0045008]; nucleotide-excision repair, DNA incision, 5'-to lesion [GO:0006296]
56	P53384	NUBP1_HUMAN	NUBP1 NBP NBP1	Cytosolic Fe-S cluster assembly factor NUBP1 (Nucleotide-binding protein 1) (NBP 1)	C8-C22-C25-C31; C235-C238	Fe_4S_4, Fe_4S_4 shared with NUBP2	Substrate - biogenesis		Cytoplasm, Nucleus	No		cell growth [GO:0016049]; cell projection organization [GO:0030030]; cellular iron ion homeostasis [GO:0006879]; centrosome localization [GO:0051642]; iron-sulfur cluster assembly [GO:0016226]; negative regulation of centrosome duplication [GO:0010826]; protein localization to cell cortex [GO:0072697]
57	Q9Y5Y2	NUBP2_HUMAN	NUBP2	Cytosolic Fe-S cluster assembly factor NUBP2 (Nucleotide-binding protein 2) (NBP 2)	C196-C199	Fe_4S_4 shared with NUBP1	Substrate - biogenesis		Cytoplasm, Nucleus	No		cell projection organization [GO:0030030]; iron-sulfur cluster assembly [GO:0016226]
58	Q8TB37	NUBPL_HUMAN	NUBPL C14orf127	Iron-sulfur protein NUBPL (IND1 homolog) (Nucleotide-binding protein-like) (huInd1)	C244-C247	Fe_2S_2/Fe_4S_4	Substrate - biogenesis		Mitochondrion	No	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:20818383, ECO:0000269|PubMed:23553477}. Note=The disease is caused by mutations affecting the gene represented in this entry.	mitochondrial respiratory chain complex I assembly [GO:0032981]; mitochondrion morphogenesis [GO:0070584]
59	P49643	PRI2_HUMAN	PRIM2 PRIM2A	DNA primase large subunit (EC 2.7.7.-) (DNA primase 58 kDa subunit) (p58)	C287-C367-C384-C424	Fe_4S_4	Structural - Regulatory	2.7.7.-	Unknown	No		DNA replication, synthesis of RNA primer [GO:0006269]; DNA replication initiation [GO:0006270]; G1/S transition of mitotic cell cycle [GO:0000082]; telomere maintenance via semi-conservative replication [GO:0032201]

60	Q06203	PUR1_HUMAN	PPAT GPAT	Amidophosphoribosyltransferase (ATase) (EC 2.4.2.14) (Glutamine phosphoribosylpyrophosphate amidotransferase) (GPAT)	C280-C426-C503-C506	Fe_4S_4	Unknown	2.4.2.14	Unknown	No		'de novo' IMP biosynthetic process [GO:0006189]; animal organ regeneration [GO:0031100]; cellular response to drug [GO:0035690]; cellular response to insulin stimulus [GO:0032869]; G1/S transition of mitotic cell cycle [GO:0000082]; glutamine catabolic process [GO:0006543]; kidney development [GO:0001822]; lactation [GO:0007595]; maternal process involved in female pregnancy [GO:0060135]; nucleoside metabolic process [GO:0009116]; protein homotetramerization [GO:0051289]; purine nucleobase biosynthetic process [GO:0009113]; purine nucleotide biosynthetic process [GO:0006164]; purine ribonucleoside monophosphate biosynthetic process [GO:0009168]
61	O60673	REV3L_HUMAN	REV3L POLZ REV3	DNA polymerase zeta catalytic subunit (EC 2.7.7.7) (Protein reversionless 3-like) (REV3-like) (hREV3)	C3086-C3089-C3099-C3104	Fe_4S_4	Structural - Regulatory	2.7.7.7	Nucleus	No		DNA-dependent DNA replication [GO:0006261]; error-prone translesion synthesis [GO:0042276]
62	Q8TAC1	RFESD_HUMAN	RFESD	Rieske domain-containing protein	C57-H59-C80-H83	Fe_2S_2 (predicted)	Unknown		Unknown	No		
63	Q9HA92	RSAD1_HUMAN	RSAD1	Radical S-adenosyl methionine domain-containing protein 1, mitochondrial (EC 1.3.99.-) (Oxygen-independent coproporphyrinogen-III oxidase-like protein RSAD1)	C49-C53-C56	Fe_4S_4	Catalytic	1.3.99.-	Mitochondrion	No		porphyrin-containing compound biosynthetic process [GO:0006779]
64	Q8WXG1	RSAD2_HUMAN	RSAD2 CIG5	Radical S-adenosyl methionine domain-containing protein 2 (Cytomegalovirus-induced gene 5 protein) (Viperin) (Virus inhibitory protein, endoplasmic reticulum-associated, interferon-inducible)	C83-C87-C90	Fe_4S_4	Unknown		Cytoplasm, Endoplasmic reticulum, Golgi apparatus, Mitochondrion	Yes		CD4-positive, alpha-beta T cell activation [GO:0035710]; CD4-positive, alpha-beta T cell differentiation [GO:0043367]; defense response to virus [GO:0051607]; negative regulation of protein secretion [GO:0050709]; negative regulation of viral genome replication [GO:0045071]; positive regulation of T-helper 2 cell cytokine production [GO:2000553]; positive regulation of toll-like receptor 7 signaling pathway [GO:0034157]; positive regulation of toll-like receptor 9 signaling pathway [GO:0034165]; response to virus [GO:0009615]; type I interferon signaling pathway [GO:0060337]; viral process [GO:0016032]
65	Q9NZ71	RTEL1_HUMAN	RTEL1 C20orf41 KIAA1088 NHL	Regulator of telomere elongation helicase 1 (EC 3.6.4.12) (Novel helicase-like)	C145-C163-C172-C207	Fe_4S_4	Structural - Regulatory	3.6.4.12	Nucleus	No	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453664, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad {PubMed:23329068}. {ECO:0000269|PubMed:23329068}.; DISEASE: Dyskeratosis congenita, autosomal dominant, 4 (DKCA4) [MIM:615190]: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269|PubMed:23329068}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Pulmonary fibrosis, and/or bone marrow failure, telomere-related, 3 (PFBMFT3) [MIM:616373]: A	DNA duplex unwinding [GO:0032508]; DNA repair [GO:0006281]; mitotic telomere maintenance via semi-conservative replication [GO:1902990]; negative regulation of DNA recombination [GO:0045910]; negative regulation of t-circle formation [GO:1904430]; negative regulation of telomere maintenance in response to DNA damage [GO:1904506]; positive regulation of telomere capping [GO:1904355]; positive regulation of telomere maintenance [GO:0032206]; positive regulation of telomere maintenance via telomere lengthening [GO:1904358]; positive regulation of telomeric loop disassembly [GO:1904535]; regulation of double-strand break repair via homologous recombination [GO:0010569]; replication fork processing [GO:0031297]; strand displacement [GO:0000732]; telomere maintenance [GO:0000723]; telomere maintenance in response to DNA damage [GO:0043247]; telomeric loop disassembly [GO:0090657]
66	P21912	SDHB_HUMAN	SDHB SDH SDH1	Succinate dehydrogenase [ubiquinone] iron-sulfur subunit, mitochondrial (EC 1.3.5.1) (Iron-sulfur subunit of complex II) (Ip)	C93-C98-C101-C113; C186-C189-C192-C253; C196-C243-C249	Fe_2S_2, Fe_3S_4, Fe_4S_4	Electron transfer	1.3.5.1	Mitochondrion	Yes	DISEASE: Pheochromocytoma (PCC) [MIM:171300]: A catecholamine-producing tumor of chromaffin tissue of the adrenal medulla or sympathetic paraganglia. The cardinal symptom, reflecting the increased secretion of epinephrine and norepinephrine, is hypertension, which may be persistent or intermittent. {ECO:0000269|PubMed:11404820, ECO:0000269|PubMed:12000816, ECO:0000269|PubMed:12618761, ECO:0000269|PubMed:14500403, ECO:0000269|PubMed:14974914, ECO:0000269|PubMed:15328326, ECO:0000269|PubMed:17634472}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.; DISEASE: Paragangliomas 4 (PGL4) [MIM:115310]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11404820, ECO:0000269|PubMed:11897817, ECO:0000269|PubMed:14715873, ECO:0000269|PubMed:14974914, ECO:0000269|PubMed:15328326}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Paraganglioma and gastric stromal sarcoma (PGGSS) [MIM:606864]: Gastrointestinal stromal tumors may be sporadic or inherited in an autosomal dominant manner, alone or as a component of a syndrome associated with other tumors, such as in the context of neurofibromatosis type 1 (NF1). Patients have both gastrointestinal stromal tumors and paragangliomas. Susceptibility to the tumors was inherited in an apparently autosomal dominant manner, with incomplete penetrance. {ECO:0000269|PubMed:17804857}. Note=The disease is caused by mutations affecting the gene represented in this entry.; DISEASE: Cowden syndrome 2 (CWS2) [MIM:612359]: A form of Cowden syndrome, a hamartomatous polyposis syndrome with age-related penetrance. Cowden syndrome is characterized by hamartomatous	aerobic respiration [GO:0009060]; respiratory electron transport chain [GO:0022904]; succinate metabolic process [GO:0006105]; tricarboxylic acid cycle [GO:0006099]
67	Q6NUM6	TYW1B_HUMAN	TYW1B RSAFD2	S-adenosyl-L-methionine-dependent tRNA 4-demethylwyosine synthase (EC 4.1.3.44) (Radical S-adenosyl methionine and flavodoxin domain-containing protein 2) (tRNA wybutosine-synthesizing protein 1 homolog B)	C352-C356-C359	Fe_4S_4	Catalytic	4.1.3.44	Unknown	No		oxidation-reduction process [GO:0055114]; tRNA processing [GO:0008033]
68	P47985	UCRI_HUMAN	UQCRFS1	Cytochrome b-c1 complex subunit Rieske, mitochondrial (EC 1.10.2.2) (Complex III subunit 5) (Cytochrome b-c1 complex subunit 5) (Rieske iron-sulfur protein) (RISP) (Rieske protein UQCRFS1) (Ubiquinol-cytochrome c reductase iron-sulfur subunit) [Cleaved into: Cytochrome b-c1 complex subunit 9 (Su9) (Subunit 9) (8 kDa subunit 9) (Complex III subunit IX) (Cytochrome b-c1 complex subunit 11) (Ubiquinol-cytochrome c reductase 8 kDa protein)]	C217-H219-C236-H239	Fe_2S_2	Electron transfer	1.10.2.2	Mitochondrion	Yes		mitochondrial electron transport, ubiquinol to cytochrome c [GO:0006122]; response to antibiotic [GO:0046677]; response to drug [GO:0042493]; response to hormone [GO:0009725]

69	P0C7P4	UCRIL_HUMAN	UQCRFS1P1 UQCRFSL1	Putative cytochrome b-c1 complex subunit Rieske-like protein 1 (Ubiquinol-cytochrome c reductase Rieske iron-sulfur subunit pseudogene 1)	C226-H228-C231-C245-H248	Fe_2S_2 (predicted)	Unknown		Unknown	No		
70	P47989	XDH_HUMAN	XDH XDHA	Xanthine dehydrogenase/oxidase [Includes: Xanthine dehydrogenase (XD) (EC 1.17.1.4); Xanthine oxidase (XO) (EC 1.17.3.2) (Xanthine oxidoreductase) (XOR)]	C43-C48-C51-C73; C113-C116-C148-C150	2 × Fe_2S_2	Electron transfer	1.17.1.4; 1.17.3.2	Cytoplasm, Extracellular space, Peroxisome	No	DISEASE: Xanthinuria 1 [XAN1] [MIM:278300]: A disorder characterized by excretion of very large amounts of xanthine in the urine and a tendency to form xanthine stones. Uric acid is strikingly diminished in serum and urine. XAN1 is due to isolated xanthine dehydrogenase deficiency. Patients can metabolize allopurinol. [ECO:0000269|PubMed:10844591, ECO:0000269|PubMed:11379872, ECO:0000269|PubMed:14551354, ECO:0000269|PubMed:9153281]. 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]; lactation [GO:0007595]; negative regulation of endothelial cell differentiation [GO:0045602]; negative regulation of endothelial cell proliferation [GO:0001937]; negative regulation of gene expression [GO:0010629]; negative regulation of protein kinase B signaling [GO:0051898]; negative regulation of protein phosphorylation [GO:0001933]; negative regulation of vascular endothelial growth factor signaling pathway [GO:1900747]; negative regulation of vasculogenesis [GO:2001213]; positive regulation of p38MAPK cascade [GO:1900745]; positive regulation of reactive oxygen species metabolic process [GO:2000379]; purine nucleotide catabolic process [GO:0006195]; xanthine catabolic process [GO:0009115]