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DISEASE: Warsaw breakage syndrome (WBRS) [MIM:613398]: A syndrome characterized by bone marrow failures, pancytopenia, and purpura; premature osteoporosis, facial dysmorphism, and malformations of the limbs; learning disabilities, and severe behavior problems; mental retardation and intellectual disability, short stature, and craniofacial and ectodermal anomalies including cleft palate, coloboma of the right optic disk, deafness, ventricular septal defect, toes and fingers anomalies, and renal anomalies; and death at early ages due to bone fractures and infections. The disease is caused by mutations affecting the gene represented in this entry.

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DISEASE: Friedenreich Syndrome (FRDS) [MIM:614541]: An autosomal recessive disorder characterized by severe microcephaly, pre- and postnatal growth retardation, facial dysmorphism, including scaphocephaly with or without craniosynostosis, prominent forehead, sparse eyebrows and hair, hypoplastic toenails and, in some cases, dental anomalies. The disease is caused by mutations affecting the gene represented in this entry.

DISEASE: Ooglio (OOG) [MIM:607861]: A rare autosomal recessive disorder characterized by proportionate dwarfism of unknown etiology. The disease is caused by mutations affecting the gene represented in this entry.

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DISEASE: Dublin-Oakley syndrome (DOS) [MIM:614767]: An autosomal recessive disorder characterized by severe microcephaly, pre- and postnatal growth retardation, facial dysmorphism, including scaphocephaly with or without craniosynostosis, prominent forehead, sparse eyebrows and hair, hypoplastic toenails and, in some cases, dental anomalies. The disease is caused by mutations affecting the gene represented in this entry.

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DISEASE: Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL) [MIM:615381]: An autosomal dominant systemic disorder characterized by severe microcephaly, pre- and postnatal growth retardation, facial dysmorphism, including scaphocephaly with or without craniosynostosis, prominent forehead, sparse eyebrows and hair, hypoplastic toenails and, in some cases, dental anomalies. The disease is caused by mutations 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 severe microcephaly, pre- and postnatal growth retardation, facial dysmorphism, including scaphocephaly with or without craniosynostosis, prominent forehead, sparse eyebrows and hair, hypoplastic toenails and, in some cases, dental anomalies. The disease is caused by mutations affecting the gene represented in this entry.

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**DNA polymerase alpha catalytic subunit (EC 2.7.7.7)** (DNA polymerase alpha)

- **DISEASE:** Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by features resembling Cockayne syndrome traits is referred to as XP-CS complex.

- **DISEASE:** Breast cancer (BC) [MIM:114480]: A common malignancy originating from C21-steroid hormone biosynthetic process [GO:0006700]; small molecule Structural - Regulatory 3.6.4.13 Nucleus  No

**Dihydropyrimidine dehydrogenase [NADP(+)] (DHPDHase) (DPD) (EC 1.3.1.2)** 29 Q16134 ETFD_HUMAN ETFDH

- **DISEASE:** Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by features resembling Cockayne syndrome traits is referred to as XP-CS complex.

**helicase BRIP1) (BRCA1-associated C-terminal helicase 1) (BRCA1-interacting QO) (ETF-ubiquinone oxidoreductase) (EC 1.5.5.1) (Electron-transferring-transcription factor complex 80 kDa subunit) (TFIIH 80 kDa subunit) (TFIIH p80) protein ERCC-2) (DNA repair protein complementing XP-D cells) (TFIIH basal transcription factor 2 80 kDa subunit) (BTF2 p80) (CXPD) (DNA excision repair C105-C111-C114-C151 Fe

**Electron transfer (respiratory chain-flavoprotein oxidase, cytochrome b-c1 complex 0) (EC 1.1.1.1)**

- **DISEASE:** Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by features resembling Cockayne syndrome traits is referred to as XP-CS complex.

**Fatty acid beta-oxidation using acyl-CoA dehydrogenase (3-ketoacyl-CoA thioesterase) (EC 1.1.99.2)**

- **DISEASE:** Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by features resembling Cockayne syndrome traits is referred to as XP-CS complex.

**Histone H3 acetylation**

- **DISEASE:** Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic oculo-facio-skeletal syndrome 2 (COFS2) [MIM:610756]: A disorder of prenatal onset characterized by features resembling Cockayne syndrome traits is referred to as XP-CS complex.
4-aminobutyrate aminotransferase, mitochondrial (EC 2.6.1.19) (S)-3-amino-2-
\[2Fe-2S\] cluster assembly [GO:0044571]; cell redox homeostasis
DISEASE: Deafness, autosomal recessive, 25 (DFNB25) [MIM:613285]: A form of non-
DISEASE: Multiple mitochondrial dysfunctions syndrome 5 (MMDS5) [MIM:617613]: An
Substrate - biogenesis  Mitochondrion  No
DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria.
2
4
DISEASE: Sideroblastic anemia, juvenile (SJJ) or sideroblastic anemia, juvenile type 2
\[2Fe-2S\] cluster assembly 1 homolog, mitochondrial (HESB-like domain-
Glutaredoxin domain-containing cysteine-rich protein 1
Glutaredoxin-3 (PKC-interacting cousin of thioredoxin) (PICOT) (PKC-theta-
\[2Fe-2S\] partner
Substrate - biosynthesis  Cytoplasm, Cell
membrane
the gene represented in this entry.
{ECO:0000269|PubMed:28356563}. Note=The disease is caused by mutations affecting
migrational abnormalities, lactic acidosis and early demise.
deterioration, seizures, cerebral and cerebellar leukodystrophy, dysmyelination, cortical
autosomal recessive, severe disorder characterized by early onset neurological
deficiency resulting in psychomotor retardation, hypotonia, hyperreflexia, lethargy,
deficiency resulting in psychomotor retardation, hypotonia, hyperreflexia, lethargy,
inheritance is autosomal recessive. {ECO:0000269|PubMed:17485548,
presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-
pathologic iron deposits in erythroblast mitochondria. Sideroblastic anemia is
form of sideroblastic anemia, a bone marrow disorder defined by the presence of
supplementation, 25(1)OH-D3 is required for absorption and utilization of dietary iron.
NOTE: For more information about sideroblastic anemia and iron metabolism see
Oligonucleotide treatment has been shown to improve learning, memory and other cognitive
functions in mice with kainate-induced neurotoxicity. Oligonucleotide treatment had no
effects on memory in controls. {ECO:0000269|PubMed:14266250}. Note=The disease is caused by
Spinocerebellar ataxia type 13 (SCA13) [MIM:614178]: An autosomal recessive disorder
for refraction. Clinical findings include nystagmus, cerebellar ataxia, dysarthria, hyperreflexia,
early demise.
EDS-VI or Hidinger-Latermark syndrome, familial (HLSFS) [MIM:129100]: An autosomal
regulatory 4.99.1.1  Mitochondrion  Yes
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  Yes
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  No
Regulatory 4.99.1.1  Mitochondrion  No
Iron-sulfur cluster assembly 2 homolog, mitochondrial (HESB-like domain)

Mitochondrion, Nucleus

DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the nuclear prelamin A recognition factor (Iron-only hydrogenase-like protein 2)

Electron transfer 1.6.5.3; 1.6.99.3  Mitochondrion  Yes

NADH dehydrogenase [ubiquinone] iron-sulfur protein 2, mitochondrial (EC 1.6.5.3) (Complex I-49kD) (CI-49kD) (NADH-ubiquinone oxidoreductase (IOP2)

DISEASE: Hyperglycinemia, lactic acidosis, and seizures (HGCLAS) [MIM:614462]: An autosomal recessive disorder characterized in infancy by failure to thrive, feeding difficulty, hypoglycemia, severe convulsions, lactic acidosis, and hyperglycinemia. Affected individuals have severe developmental delay, with mental retardation, autistic-like behavior, and severe growth disturbance. Longitudinal growth fails to keep pace with head growth. Final height is below the third percentile, and adult height is usually below the first percentile. Behavioral abnormalities include severely impaired social and language skills. Synodic movements are present in some affected individuals. Patients die in late infancy or early childhood. ECO:0000269|PubMed:11220739, ECO:0000269|PubMed:11349233.


DISEASE: Multiple mitochondrial dysfunctions syndrome 4 (MMDS4) [MIM:616370]: A syndrome described in infancy by failure to thrive, feeding difficulty, hypoglycemia, severe convulsions, lactic acidosis, and hyperglycinemia. Affected individuals have severe developmental delay, with mental retardation, autistic-like behavior, and severe growth disturbance. Longitudinal growth fails to keep pace with head growth. Final height is below the third percentile, and adult height is usually below the first percentile. Behavioral abnormalities include severely impaired social and language skills. Synodic movements are present in some affected individuals. Patients die in late infancy or early childhood. ECO:0000269|PubMed:18304497.

DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the nuclear prelamin A recognition factor (Iron-only hydrogenase-like protein 2)

Electron transfer 1.6.5.3; 1.6.99.3  Mitochondrion  Yes

NADH dehydrogenase [ubiquinone] iron-sulfur protein 2, mitochondrial (EC 1.6.5.3) (Complex I-49kD) (CI-49kD) (NADH-ubiquinone oxidoreductase (IOP2)

DISEASE: Hyperglycinemia, lactic acidosis, and seizures (HGCLAS) [MIM:614462]: An autosomal recessive disorder characterized in infancy by failure to thrive, feeding difficulty, hypoglycemia, severe convulsions, lactic acidosis, and hyperglycinemia. Affected individuals have severe developmental delay, with mental retardation, autistic-like behavior, and severe growth disturbance. Longitudinal growth fails to keep pace with head growth. Final height is below the third percentile, and adult height is usually below the first percentile. Behavioral abnormalities include severely impaired social and language skills. Synodic movements are present in some affected individuals. Patients die in late infancy or early childhood. ECO:0000269|PubMed:11220739, ECO:0000269|PubMed:11349233.


DISEASE: Multiple mitochondrial dysfunctions syndrome 4 (MMDS4) [MIM:616370]: A syndrome described in infancy by failure to thrive, feeding difficulty, hypoglycemia, severe convulsions, lactic acidosis, and hyperglycinemia. Affected individuals have severe developmental delay, with mental retardation, autistic-like behavior, and severe growth disturbance. Longitudinal growth fails to keep pace with head growth. Final height is below the third percentile, and adult height is usually below the first percentile. Behavioral abnormalities include severely impaired social and language skills. Synodic movements are present in some affected individuals. Patients die in late infancy or early childhood. ECO:0000269|PubMed:18304497.
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- **NADH dehydrogenase [ubiquinone] iron-sulfur protein 7, mitochondrial (EC 1.6.5.3)**
- **NADH dehydrogenase [ubiquinone] flavoprotein 1, mitochondrial (EC 1.6.5.3)**
- **Cytosolic Fe-S cluster assembly factor NUBP1 (Nucleotide-binding protein 1)**
- **DNA N-glycosylase/DNA-(apurinic or apyrimidinic site) lyase**
- **Cysteine desulfurase, mitochondrial (EC 2.8.1.7)**
- **(NADH-ubiquinone oxidoreductase 51 kDa subunit)**
- **(NADH-ubiquinone oxidoreductase 23 kDa subunit)**
- **(NADH-ubiquinone oxidoreductase 20 kDa subunit)**

**DISEASE**:
- **Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]**: A disorder of the mitochondrial respiratory chain complex I.
- **Leigh syndrome (LS) [MIM:256000]**: An early-onset progressive neurodegenerative disorder.
- **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.

**Gene Ontology**
- **cell projection organization [GO:0030030]**
- **iron-sulfur cluster assembly [GO:0016226]**
- **Electron transfer [EC 1.6.5.3; 1.6.99.3]**
- **Mitochondrion**
- **DNA replication, synthesis of RNA primer [GO:0006269]**
- **DNA replication**

**Protein Complexes**
- NADH ubiquinone oxidoreductase
- NADH-ubiquinone oxidoreductase complex 1
- NADH-ubiquinone oxidoreductase complex 2
- NADH-ubiquinone oxidoreductase complex 3

**Molecular Functions**
- **[GO:0032201]**
- **[GO:0016226]**
- **[GO:0010826]**
- **[GO:0072697]**
- **[GO:0006879]**
- **[GO:0051642]**
- **[GO:0006296]**
- **[GO:0045008]**
- **[GO:0006285]**
- **[GO:0000096]**
- **[GO:0044281]**
- **[GO:0006777]**
- **[GO:0006461]**
- **[GO:0032324]**
- **[GO:0024169]**
- **[GO:0027026]**
- **[GO:0042775]**
- **[GO:0042773]**
- **[GO:0006120]**
- **[GO:0006979]**
- **[GO:0014693]**
- **[GO:0015344]**

**Cellular Processes**
- **telomere maintenance via semi-conservative replication [GO:0032201]**
- **mitochondrial respiratory chain complex I assembly [GO:0032981]**
- **mitochondrial respiratory chain complex I [GO:0016226]**
- **mitochondrial respiratory chain complex I assembly [GO:0032981]**

**Pathway**
- Mitochondrial respiratory chain complex I assembly [GO:0032981]
- Mitochondrial respiratory chain complex I [GO:0016226]
- Mitochondrial respiratory chain complex I assembly [GO:0032981]

**Small Molecule Metabolism**
- **[GO:0048738]**
- **[GO:0042775]**
- **[GO:0042773]**
- **[GO:0014693]**
- **[GO:0015344]**

**Degradation**
- **[GO:0032324]**
- **[GO:0024169]**
- **[GO:0027026]**

**Transport**
- **NADH ubiquinone oxidoreductase complex 1**
- **NADH ubiquinone oxidoreductase complex 3**

**Genetic Disorder**
- **Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]**
- **Leigh syndrome (LS) [MIM:256000]**
| P47989 | IDH_HUMAN | IDH1 | IDH1 | Xanthine dehydrogenase/oxidase [Includes: Xanthine dehydrogenase (XD) (EC 1.17.1.4); Xanthine oxidase (XO) (EC 1.17.3.2) (Xanthine oxidoreductase) (XOR)] | 2 × Fe₂S₂ | Electron transfer | 1.17.1.4; 1.17.3.2 | Cytoplasm, Extracellular space, Peroxisome | No | 670-679: Xanthinuria I (XAN1) [MIM:278300]: A disorder characterized by excretion of very large amounts of xanthine in the urine and a tendency to form xanthine stones. Serum uric acid is strikingly diminished in severe and acute 670-679-673 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:0060177]; negative regulation of xanthine dehydrogenase/oxidase [GO:1900694]; positive regulation of mitochondrial respiration [GO:0045791]; positive regulation of reactive oxygen species metabolic process [GO:0045791]; putative reductase activity [GO:0045791]; sulfite oxidase activity [GO:0045791]; xanthine oxidase activity [GO:0045791]; xanthine dehydrogenase activity [GO:0045791]; xanthine dehydrogenase/oxidase activity [GO:0045791] |