

**Table S4 - List of lethal phenotypes**

Phenotype	Severity Level	Clinical Category	Reasoning Note
3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh-like
ACHONDROGENESIS, LANGER-SALDINO TYPE	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: achondrogenesis
ANO1-related fatal neonatal disease due to impaired chloride currents	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Achondrogenesis	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: achondrogenesis
Achondrogenesis type II	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: achondrogenesis
Achondrogenesis, type IA	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: achondrogenesis
Achondrogenesis, type IB	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: achondrogenesis
Alpers Syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: alpers syndrome
Alpers-Huttenlocher Syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: alpers-huttenlocher
Alveolar capillary dysplasia with pulmonary venous misalignment	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: alveolar capillary dysplasia
Amish lethal microcephaly	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Atypical teratoid rhabdoid tumor	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: rhabdoid tumor
CLOVERLEAF SKULL WITH THANATOPHORIC DWARFISM	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
Canavan Disease, Familial Form	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: canavan disease
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Cardiomyopathy, fatal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal

Cardiomyopathy, fatal infantile	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Complex lethal osteochondrodysplasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Early-onset myopathy with fatal cardiomyopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Ethylmalonic encephalopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: ethylmalonic encephalopathy
Fatal Infantile Cardioencephalomyopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Fatal familial insomnia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Fatal infantile hypertonic myofibrillar myopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Fatal multiple mitochondrial dysfunctions syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
Fowler syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fowler syndrome
GRACILE syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: gracile syndrome
Gaucher disease perinatal lethal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Glycine encephalopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: glycine encephalopathy
Glycine encephalopathy 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: glycine encephalopathy
Glycine encephalopathy 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: glycine encephalopathy
Glycogen storage disease due to glycogen branching enzyme deficiency, fatal perinatal neuromuscular form	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: fatal
HEMOGLOBIN H HYDROPS FETALIS SYNDROME	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: hydrops fetalis
Hydroletharus syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Hydroletharus syndrome 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Hydroletharus syndrome 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Hydrops fetalis	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: hydrops fetalis
Infantile hypophosphatasia	Confirmed Lethal	Infantile-Lethal Syndrome	lethal type
Infantile neuroaxonal dystrophy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: infantile neuroaxonal dystrophy
Junctional epidermolysis bullosa gravis of Herlitz	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: herlitz
Junctional epidermolysis bullosa, non-Herlitz type	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: herlitz

Krabbe disease due to saposin A deficiency	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: krabbe disease
LEIGH SYNDROME, NUCLEAR	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
LETHAL SHORT-LIMBED PLATYSPONDYLIC DWARFISM, SAN DIEGO TYPE	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Leigh Syndrome (mtDNA mutation)	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
Leigh syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
Leigh syndrome due to mitochondrial complex I deficiency	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
Leigh syndrome due to mitochondrial complex IV deficiency	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
Leigh syndrome, mitochondrial	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
Lethal Encephalopathy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal Kniest-like syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal acantholytic epidermolysis bullosa	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal arthrogryposis-anterior horn cell disease syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contractural syndrome Finnish type	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 11	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 3	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 4	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 6	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 7	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 8	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital contracture syndrome 9	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal congenital glycogen storage disease of heart	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal fetal cerebrenogenitourinary agenesis/hypoplasia syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal left ventricular non-compaction-seizures-hypotonia- cataract-developmental delay syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal multiple pterygium syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal

Lethal occipital encephalocele-skeletal dysplasia syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal osteosclerotic bone dysplasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal polymalformative syndrome, Boissel type	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal short-limb skeletal dysplasia, Al Gazali type	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal short-limbed short stature	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal skeletal dysplasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Lethal tight skin contracture syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
MECKEL-GRUBER SYNDROME, TYPE 3	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel-gruber
MYBPC1-related autosomal recessive non-lethal arthrogryposis multiplex congenita syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Meckel syndrome 13	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome 14	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 10	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 11	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 3	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 4	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 5	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 6	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 8	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel syndrome, type 9	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel syndrome
Meckel-Gruber syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: meckel-gruber

Metatropic dysplasia, nonlethal dominant	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Mild Canavan disease	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: canavan disease
Mitochondrial DNA-Associated Leigh Syndrome and NARP	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: leigh syndrome
NEK9-related lethal skeletal dysplasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Neu-Laxova syndrome 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: neu-laxova
Neu-Laxova syndrome 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: neu-laxova
Niemann-Pick disease, type A	Confirmed Lethal	Infantile-Lethal Syndrome	type a
Non-immune hydrops fetalis	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: hydrops fetalis
OSTEOGENESIS IMPERFECTA, TYPE II	Confirmed Lethal	Infantile-Lethal Syndrome	lethal type
Osteogenesis imperfecta type III	Confirmed Lethal	Infantile-Lethal Syndrome	lethal type
Osteogenesis imperfecta, dominant perinatal lethal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Osteogenesis imperfecta, perinatal lethal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Osteogenesis imperfecta, type III/IV	Confirmed Lethal	Infantile-Lethal Syndrome	lethal type
PHARC syndrome	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: arc syndrome
PIEZO1-related generalized lymphatic dysplasia with non-immune hydrops fetalis	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: hydrops fetalis
PLATYSPONDYLIC LETHAL SKELETAL DYSPLASIA, SAN DIEGO TYPE	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Pena-Shokeir syndrome type I	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: pena-shokeir
Perinatal lethal hypophosphatasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Pontocerebellar hypoplasia, type 2F	Confirmed Lethal	Infantile-Lethal Syndrome	type 2
Preimplantation embryonic lethality 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Preimplantation embryonic lethality 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
RESTRICTIVE DERMOPATHY 2, LETHAL	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Restrictive dermatopathy 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: restrictive dermatopathy
Restrictive dermatopathy 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: restrictive dermatopathy
SLEEP APNEA, LETHAL CENTRAL	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
Sandhoff disease	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: sandhoff disease

Sandhoff disease, adult form	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: sandhoff disease
Sandhoff disease, chronic	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: sandhoff disease
Sandhoff disease, infantile form	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: sandhoff disease
Sandhoff disease, juvenile form	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: sandhoff disease
Severe hydrops fetalis	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: hydrops fetalis
Severe progressive deforming recessive osteogenesis imperfecta (type III)	Confirmed Lethal	Infantile-Lethal Syndrome	lethal type
Skeletal dysplasia and progressive central nervous system degeneration, lethal	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: lethal
THANATOPHORIC DYSPLASIA WITH KLEEBLATTSCHAEDEL	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
THANATOPHORIC DYSPLASIA WITH STRAIGHT FEMURS AND CLOVERLEAF SKULL	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
Tay-Sachs disease	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: tay-sachs
Tay-Sachs disease, B1 variant	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: tay-sachs
Tay-Sachs disease, variant AB	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: tay-sachs
Tay-sachs disease, juvenile	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: tay-sachs
Tay-sachs disease, juvenile/adult	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: tay-sachs
Thanatophoric dysplasia	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
Thanatophoric dysplasia type 1	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
Thanatophoric dysplasia, type 2	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: thanatophoric
Walker-Warburg congenital muscular dystrophy	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: walker-warburg
Wolman disease	Confirmed Lethal	Infantile-Lethal Syndrome	Matched top-tier keyword: wolman disease
Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Congenital microcephaly - severe encephalopathy - progressive cerebral atrophy syndrome	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic

Deafness, congenital, and adult-onset progressive leukoencephalopathy	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
EPIDERMOLYSIS BULLOSA, JUNCTIONAL 5B, WITH PYLORIC ATRESIA	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Encephalopathy, neonatal severe, with lactic acidosis and brain abnormalities	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Epidermolysis bullosa junctionalis with pyloric atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Epidermolysis bullosa simplex 5C, with pyloric atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Epidermolysis bullosa simplex with pyloric atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Epidermolysis bullosa, junctional 6, with pyloric atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Epidermolysis bullosa, junctional, with pyloric atresia and aplasia cutis congenita	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Junctional epidermolysis bullosa with pyloric atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
LEUKOENCEPHALOPATHY, PROGRESSIVE, INFANTILE-ONSET, WITH DEAFNESS	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Leukoencephalopathy, progressive, infantile-onset, with or without deafness	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
Necrotizing encephalopathy infantile subacute of Leigh	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
PLEC1-Related Epidermolysis Bullosa with Pyloric Atresia	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Critical System + Catastrophic Finding
PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic
Severe neonatal-onset encephalopathy with microcephaly	Confirmed Lethal	Lethal Clinical Pattern	Matched pattern: Severe+Early+Systemic

Spinal muscular atrophy, type II	Confirmed Lethal	Progressive Neuromuscular D	lethal type
Spinal muscular atrophy, type IV	Confirmed Lethal	Progressive Neuromuscular D	lethal type
Neonatal Marfan syndrome	Confirmed Lethal	Severe Connective Tissue Dis	neonatal
Infantile osteopetrosis	Confirmed Lethal	Severe Skeletal Dysplasia	malignant infantile
Infantile osteopetrosis with neuroaxonal dysplasia	Confirmed Lethal	Severe Skeletal Dysplasia	malignant infantile
Atypical Leigh syndrome	High risk of lethality	Conflicting Modifiers	'atypical' conflicts with a top-tier lethal term
Atypical glycine encephalopathy	High risk of lethality	Conflicting Modifiers	'atypical' conflicts with a top-tier lethal term
ATM-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
ATM-related cancer predisposition syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
BARD1-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
BRCA1-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
BRCA2-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
BRIP1-associated familial cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Basal cell carcinoma, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Basal cell carcinoma, susceptibility to, 7	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast and colorectal cancer, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast cancer, invasive, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast cancer, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast cancer, susceptibility to, in BRCA1 and BRCA2 carriers	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast-ovarian cancer, familial, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast-ovarian cancer, familial, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast-ovarian cancer, familial, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast-ovarian cancer, familial, susceptibility to, 4	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Breast-ovarian cancer, familial, susceptibility to, 5	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
CHEK2-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
CONGENITAL MYOPATHY 1A, AUTOSOMAL DOMINANT, WITH SUSCEPTIBILITY TO MALIGNANT HYPERTHERMIA	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colon cancer, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colorectal cancer, sporadic, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colorectal cancer, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colorectal cancer, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colorectal cancer, susceptibility to, 10	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.



Colorectal cancer, susceptibility to, 12	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Colorectal cancer, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
ESOPHAGEAL CANCER, ALCOHOL-RELATED, SUSCEPTIBILITY TO	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Erythroleukemia, familial, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Family history of cancer	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
HOXB13-Related Cancer Predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Hereditary thrombocytopenia and hematologic cancer predisposition syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Hereditary thrombocytopenia and hematological cancer predisposition syndrome associated with RUNX1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Hodgkin lymphoma, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Increased risk to develop myelodysplastic syndrome, acute myeloid leukemia, or chronic myelomonocytic leukemia	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Kaposi sarcoma, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
LUNG CANCER, SUSCEPTIBILITY TO	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Leukemia, acute lymphoblastic, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Leukemia, acute lymphocytic, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Leukemia, acute myeloid, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Leukemia, chronic lymphocytic, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
MEDULLOBLASTOMA PREDISPOSITION SYNDROME	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Malignant hyperthermia, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Malignant hyperthermia, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Malignant hyperthermia, susceptibility to, 5	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 5	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 6	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 8	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, cutaneous malignant, susceptibility to, 9	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Melanoma, uveal, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.

Melanoma, uveal, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
NEUROBLASTOMA, SUSCEPTIBILITY TO, 1, INCLUDED	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Nasopharyngeal carcinoma, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Neuroblastoma, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Neuroblastoma, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Neuroblastoma, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Oligodontia-cancer predisposition syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Ovarian cancer, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
PALB2-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Pancreatic cancer, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Pancreatic cancer, susceptibility to, 1	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Pancreatic cancer, susceptibility to, 2	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Pancreatic cancer, susceptibility to, 3	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Pancreatic cancer, susceptibility to, 4	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Predisposition to cancer	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Predisposition to medulloblastoma	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Predisposition to neuroblastoma	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Progeroid features-hepatocellular carcinoma predisposition syndrome	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Prostate cancer, hereditary, 2, susceptibility to	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
RAD51D-related cancer predisposition	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
THYROID CANCER, NONMEDULLARY, 2, SUSCEPTIBILITY TO	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
Triple-Negative Breast Cancer Finding	High risk of lethality	Heritable Cancer Syndrome	High-penetrance cancer predisposition.
ADRENOCORTICAL CARCINOMA, PEDIATRIC	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
APC-mutation negative familial colorectal cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Acute granulocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute lymphoid leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute megakaryoblastic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute megakaryoblastic leukemia in down syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute megakaryoblastic leukemia without down syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute myeloblastic leukemia with maturation	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute myelogenous leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia

Acute myeloid leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute myeloid leukemia, adult	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute non-lymphocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Acute promyelocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Adenoid cystic carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Adrenal cortex carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Adrenocortical carcinoma, hereditary	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Alveolar rhabdomyosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Angioimmunoblastic T-cell lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Astroblastoma, MN1-altered	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
B Lymphoblastic Leukemia/Lymphoma with t(9;22)(q34.1;q11.2)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B Lymphoblastic Leukemia/Lymphoma with t(v;11q23.3)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B Lymphoblastic Leukemia/Lymphoma, Not Otherwise Specified	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B lymphoblastic leukemia lymphoma with hyperdiploidy	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B lymphoblastic leukemia lymphoma with t(12;21)(p13;q22)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B lymphoblastic leukemia lymphoma, no ICD-O subtype	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B-cell childhood acute lymphoblastic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B-cell chronic lymphocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
B-cell non-Hodgkin lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
B-lymphoblastic leukemia/lymphoma with hypodiploidy	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
BAP1 Cancer Syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
BARRETT ESOPHAGUS/ESOPHAGEAL ADENOCARCINOMA	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
BREAST CANCER, FAMILIAL	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Basal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Bile duct cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Bladder cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Blast phase chronic myelogenous leukemia, BCR-ABL1 positive	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Bone osteosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Breast adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Breast and Ovarian Cancer Susceptibility	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer

Breast and colorectal cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Breast and/or ovarian cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Breast cancer, early-onset	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Breast carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Breast ductal adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Breast lobular carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Burkitt lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
CDH1-related diffuse gastric and lobular breast cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
CIC-rearranged sarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
CUP (carcinoma unknown primary) syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Cancer of cervix	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Carcinoma of colon	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Carcinoma of esophagus	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Carcinoma of male breast	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Carcinoma of pancreas	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Carcinoma, adrenocortical, androgen-secreting	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Cerebellar hemangioblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Cerebellar medulloblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Cervical cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Cervical cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Cervix cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Childhood ganglioglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Childhood kidney cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Cholangiocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Chondrosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Choroid plexus carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Chromophobe renal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Chronic myelogenous leukemia, BCR-ABL1 positive	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Classic Hodgkin lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Classic medulloblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Clear cell carcinoma of kidney	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Colon adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Colon cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer

Colon carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Colonic carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Colorectal / endometrial cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer susceptibility 12	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer, hereditary nonpolyposis, type 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer, hereditary nonpolyposis, type 6	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer, hereditary nonpolyposis, type 7	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer, non-polyposis	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Colorectal carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Congenital fibrosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Cutaneous Malignant Melanoma, Dominant	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Cutaneous malignant melanoma 9	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
DIFFUSE GASTRIC AND LOBULAR BREAST CANCER SYNDROME	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Deafness-lymphedema-leukemia syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Dermatofibrosarcoma protuberans	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Desmoplastic/nodular medulloblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Diffuse gastric and lobular breast cancer syndrome with cleft lip and with or without cleft palate	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Diffuse hemispheric glioma, H3 G34-mutant	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Diffuse intrinsic pontine glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Diffuse midline glioma, H3 K27-altered	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Diffuse midline glioma, H3 K27M-mutant	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Diffuse pediatric-type high-grade glioma, H3-wildtype and IDH-wildtype	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Duodenal adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
EBV-Positive Inflammatory Follicular Dendritic Cell Sarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
EBV-positive nodal T- and NK-cell lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
EGFR-related lung cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
EOSINOPHILS, MALIGNANT PROLIFERATION OF	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Early T cell progenitor acute lymphoblastic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Eccrine porocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Endometrial cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer

Endometrial carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Endometrial carcinoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Epidermal nevus with urothelial cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Esophageal squamous cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Esophageal squamous cell carcinoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Ewing sarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Exocrine pancreatic carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Extraskeletal myxoid chondrosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
FAMILIAL ATYPICAL MOLE-MALIGNANT MELANOMA SYNDROME	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Familial breast and ovarian cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Familial cancer of breast	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Familial colorectal cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Familial colorectal cancer type X	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Familial medullary thyroid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Familial melanoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Familial ovarian cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Familial ovarian carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Familial pancreatic carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Familial papillary thyroid carcinoma with renal papillary neoplasia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Familial prostate cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
GEN1-related prostate cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Gallbladder cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Ganglioglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Gastric adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Gastric adenocarcinoma and proximal polyposis of the stomach	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Gastric cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Gastrointestinal carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Glioblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Glioblastoma 3	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Glioblastoma multiforme	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Glioblastoma multiforme, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Glioblastoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma

Glioma susceptibility 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Glioma susceptibility 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Glioma susceptibility 3	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Head and neck cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Head and neck squamous cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hepatoblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Hepatocellular carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hereditary Prostate Cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary breast cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary breast cancer, ABRAXAS1-related	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary breast ovarian cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary cancer-predisposing syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary diffuse gastric adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hereditary leiomyomatosis and renal cell cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary nonpolyposis colon cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary nonpolyposis colorectal carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hereditary papillary renal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hereditary pheochromocytoma-paraganglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Hereditary renal cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Hereditary renal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Hereditary retinoblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Hereditary site-specific ovarian cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
High grade surface osteosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Hirschsprung disease-ganglioneuroblastoma syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Hurthle cell carcinoma of thyroid	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Infant-type hemispheric glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Infiltrating duct carcinoma of breast	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Inherited acute myeloid leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Inherited breast cancer and ovarian cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Inherited ovarian cancer (without breast cancer)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Inherited polyposis and early onset colorectal cancer - germline testing	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Inherited prostate cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Intrahepatic cholangiocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Invasive medullary breast carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma

Joint contractures, osteochondromas, and B-cell lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Juvenile myelomonocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
LEUKEMIA, ACUTE MYELOID, M0 SUBTYPE	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
LEUKEMIA, CHRONIC LYMPHOCYTIC, SOMATIC	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
LIVER CELL CARCINOMA	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Leiomyosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Leukemia, Philadelphia chromosome-positive, resistant to imatinib	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Leukemia, acute myelogenous, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Leukemia, acute myeloid, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Lip and oral cavity carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Lung adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Lung cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Lung cancer susceptibility 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Lung cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Lung carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Lung sarcomatoid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Lymphoma, non-Hodgkin, familial	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
MELANOMA, MALIGNANT	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Malignant Colorectal Neoplasm	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant Melanoma Susceptibility	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Malignant eosinophil proliferation	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant fibrous histiocytoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant germ cell tumor of ovary	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Malignant hyperpyrexia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant hyperthermia and exertional rhabdomyolysis	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant hyperthermia of anesthesia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant hyperthermia susceptibility 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant hypothermia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant lymphoma, large B-cell, diffuse	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Malignant melanoma of skin	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Malignant mesothelioma (disease)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant neoplasm of brain	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant



Malignant neoplastic disease	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of ascending colon	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of breast	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of esophagus	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of lung	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of pancreas	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of prostate	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of stomach	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of testis	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of thyroid gland	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Malignant tumor of urinary bladder	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Mantle cell lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Medullary thyroid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Medullary thyroid gland carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Medulloblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Medulloblastoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Melanoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Melanoma and neural system tumor syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Melanoma-pancreatic cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Mesothelioma, malignant	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: malignant
Metastatic Prostate Small Cell Carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Mismatch repair cancer syndrome 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Mismatch repair cancer syndrome 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Mismatch repair cancer syndrome 3	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Mismatch repair cancer syndrome 4	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Mixed Phenotype Acute Leukemia, T/Myeloid, Not Otherwise Specified	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Mixed phenotype acute leukemia with t(v;11q23.3)	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Monosomy 7 myelodysplasia and leukemia syndrome 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Monosomy 7 myelodysplasia and leukemia syndrome 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Mucosa-associated lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Multifocal breast carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Myelodysplastic syndrome progressed to acute myeloid leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Nasopharyngeal carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Nasopharyngeal carcinoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma

Nephroblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Neuroblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Neuroblastoma Susceptibility	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Non-Hodgkin lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
Non-small cell lung carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Nonpapillary renal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Nut midline carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
OVARIAN CANCER, EPITHELIAL, SOMATIC	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
OVARIAN CANCER, SOMATIC	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Oligodendroglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Optic nerve glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Osteoporosis with pseudoglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Osteosarcoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Ovarian Cancers	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Ovarian cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Ovarian carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Ovarian serous surface papillary adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
PANCREATIC ACINAR CARCINOMA	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
PANCREATIC CARCINOMA	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
PMS1-related breast cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
PMS2-related cancer disorders	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
POLE-related polyposis and colorectal cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Palmoplantar keratoderma-esophageal carcinoma syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pancreatic adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pancreatic cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Pancreatic carcinoma, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pancreatic ductal adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Papillary carcinoma of the corpus uteri	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Papillary renal cell carcinoma type 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Papillary thyroid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Paranglioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma

Parangliomas 3	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 4	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 5	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 6	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas 7	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parangliomas with sensorineural hearing loss	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Parathyroid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pediatric high-grade glioma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Peritoneal Gliomatosis	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: glioma
Peritoneum cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Pheochromocytoma and amyloid producing medullary thyroid carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pineoblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Pituitary carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Pleuropulmonary blastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Precursor B-cell acute lymphoblastic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
Primary carcinoma of liver	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Primary intracranial sarcoma, DICER1-mutant	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Primary low grade serous adenocarcinoma of ovary	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Primary peritoneal carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Prostate cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer susceptibility	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, hereditary, 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, hereditary, 12	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, hereditary, 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, hereditary, 9	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, hereditary, X-linked 3	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Prostate cancer/brain cancer susceptibility	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
RENAL CELL CARCINOMA WITH PARANEOPLASTIC ERYTHROCYTOSIS	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Rectum adenocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Renal cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Renal cell carcinoma, Xp11-associated	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Renal transitional cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Retinoblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma

Rhabdomyosarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Rhabdomyosarcoma, embryonal, 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Sarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Sessile serrated polyposis cancer syndrome	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Sigmoid colon cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Small cell carcinoma of the ovary, hypercalcemic type	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Small cell lung carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Spinal hemangioblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Spindle cell sarcoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: sarcoma
Squamous cell carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Squamous cell carcinoma of the head and neck	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Squamous cell carcinoma, head and neck, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Squamous cell lung carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Stomach cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Subcutaneous panniculitis-like T-cell lymphoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: lymphoma
T-cell prolymphocytic leukemia	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: leukemia
THYROID CARCINOMA, FOLLICULAR	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Teratocarcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Testicular cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid cancer, familial medullary	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid cancer, nonmedullary, 1	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid cancer, nonmedullary, 2	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid cancer, nonmedullary, 4	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid cancer, nonmedullary, 5	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Thyroid carcinoma, follicular, somatic	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Thyroid carcinoma, sporadic medullary	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Thyroid gland carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Thyroid gland undifferentiated (anaplastic) carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Trilateral retinoblastoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: blastoma
Triple-negative breast cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Triple-negative breast carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
Urinary bladder cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Uterine corpus cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer
Uveal melanoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: melanoma
Vulvar adenocarcinoma of mammary gland type	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
bilateral breast cancer	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: cancer

hereditary breast carcinoma	High risk of lethality	Heritable Malignancy	Matched high-risk keyword: carcinoma
ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Acute aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Arrhythmogenic Right Ventricular Dysplasia/Cardiomyopathy 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic cardiomyopathy	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic cardiomyopathy with variable ectodermal abnormalities	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic cardiomyopathy with woolly hair and keratoderma	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic cardiomyopathy with woolly hair and keratoderma	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular cardiomyopathy	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 10	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 11	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 12	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 13	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 2	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic

Arrhythmogenic right ventricular dysplasia 5	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia 9	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic right ventricular dysplasia, familial, 14	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Arrhythmogenic ventricular cardiomyopathy	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Ascending aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Brugada syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome (shorter-than-normal QT interval)	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 2	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 3	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 4	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 5	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 6	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 7	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome
Brugada syndrome 9	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: brugada syndrome

Catecholaminergic polymorphic ventricular tachycardia	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Catecholaminergic polymorphic ventricular tachycardia 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Catecholaminergic polymorphic ventricular tachycardia 2	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Catecholaminergic polymorphic ventricular tachycardia 3	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Catecholaminergic polymorphic ventricular tachycardia 4	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Catecholaminergic polymorphic ventricular tachycardia 5	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: catecholaminergic polymorphic ventricular tachycardia
Congenital long QT syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
DSP-related arrhythmogenic cardiomyopathy	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Descending aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Early age onset of sporadic thoracic aortic dissections	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Familial isolated arrhythmogenic right ventricular dysplasia	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: arrhythmogenic
Familial thoracic aortic aneurysm and aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
Heritable pulmonary arterial hypertension	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Idiopathic and/or familial pulmonary arterial hypertension	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Loeys-Dietz syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome

Loeys-Dietz syndrome 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome
Loeys-Dietz syndrome 2	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome
Loeys-Dietz syndrome 4	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome
Loeys-Dietz syndrome 6	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome
Long QT syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 1, recessive	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 1/2, digenic	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 10	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 11	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 12	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 13	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 14	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 15	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 16	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 2	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 2/5, digenic	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 3	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome



Long QT syndrome 3/6, digenic	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 4	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 5	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 6	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long QT syndrome 9	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
Long qt syndrome 8	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: long qt syndrome
MARFAN SYNDROME, AUTOSOMAL RECESSIVE	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
MARFAN SYNDROME, TYPE I	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Marfan Syndrome/Loeys-Dietz Syndrome/Familial Thoracic Aortic Aneurysms and Dissections	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Marfan syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Marfan syndrome type 1	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Marfan syndrome, atypical	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Marfan syndrome, classic	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: marfan syndrome
Pulmonary arterial hypertension	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Pulmonary arterial hypertension associated with another disease	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Pulmonary arterial hypertension associated with congenital heart disease	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Pulmonary arterial hypertension associated with connective tissue disease	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension

Rare disease with thoracic aortic aneurysm and aortic dissection	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: aortic dissection
SMAD2- related Loeys-Dietz syndrome	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: loeys-dietz syndrome
SOX17-related pulmonary arterial hypertension	High risk of lethality	High Risk Cardiac	Matched high-risk keyword: pulmonary arterial hypertension
Adams-Oliver syndrome	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 1	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 2	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 3	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 4	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 5	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Adams-Oliver syndrome 6	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: adams-oliver syndrome
Bilateral renal agenesis	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal agenesis
Congenital diaphragmatic hernia	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: diaphragmatic hernia
Diaphragmatic hernia 3	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: diaphragmatic hernia
Diaphragmatic hernia 4, with cardiovascular defects	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: diaphragmatic hernia
Gaucher Disease, Type 3	High risk of lethality	High Variability Syndrome	type 3
HEREDITARY RENAL APLASIA	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal aplasia
Hypoplastic left heart syndrome	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: hypoplastic left heart syndrome
Hypoplastic left heart syndrome 1	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: hypoplastic left heart syndrome
Hypoplastic left heart syndrome 2	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: hypoplastic left heart syndrome

Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal agenesis
Neurofacioskeletal syndrome with or without renal agenesis	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal agenesis
Niemann-Pick disease, type C	High risk of lethality	High Variability Syndrome	type c
Niemann-Pick disease, type C1	High risk of lethality	High Variability Syndrome	type c
Niemann-Pick disease, type C1, juvenile form	High risk of lethality	High Variability Syndrome	type c
Niemann-Pick disease, type C2	High risk of lethality	High Variability Syndrome	type c
Posterolateral diaphragmatic hernia	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: diaphragmatic hernia
RENAL APLASIA	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal aplasia
Renal agenesis	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal agenesis
Renal agenesis and hypodysplasia	High risk of lethality	High Variability Syndrome	Matched high-risk keyword: renal agenesis
Diffuse cerebral and cerebellar atrophy - intractable seizures - progressive microcephaly syndrome	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Encephalopathy, familial progressive necrotizing	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 1	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Encephalopathy, progressive, early-onset, with episodic rhabdomyolysis	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Encephalopathy, progressive, with amyotrophy and optic atrophy	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Global developmental delay-visual anomalies-progressive cerebellar atrophy-truncal hypotonia syndrome	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Leukoencephalopathy, progressive, with ovarian failure	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Myopathy, epilepsy, and progressive cerebral atrophy	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.
Progressive childhood encephalopathy	High risk of lethality	Progressive Critical System D	Progressive nature implies high risk.

Progressive distal muscular atrophy	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Progressive encephalopathy	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Progressive encephalopathy with leukodystrophy due to DECR deficiency	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Progressive spinal muscular atrophy	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome	High risk of lethality	Progressive Critical System D Progressive nature implies high risk.
Ataxia-telangiectasia syndrome	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: ataxia-telangiectasia
Ataxia-telangiectasia without immunodeficiency	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: ataxia-telangiectasia
Ataxia-telangiectasia-like disorder	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: ataxia-telangiectasia
Ataxia-telangiectasia-like disorder 1	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: ataxia-telangiectasia
Ataxia-telangiectasia-like disorder 2	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: ataxia-telangiectasia
Autosomal recessive dyskeratosis congenita	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Autosomal recessive dyskeratosis congenita 4	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Cystic fibrosis	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: cystic fibrosis
Cystic fibrosis-gastritis-megaloblastic anemia syndrome	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: cystic fibrosis
Duchenne muscular dystrophy	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: duchenne muscular dystrophy
Dyskeratosis Congenita, Dominant	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis Congenita, Recessive	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita

Dyskeratosis congenita Scoggins type	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, X-linked	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal dominant 1	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal dominant 2	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal dominant 3	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal dominant 4	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal dominant 6	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 1	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 2	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 3	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 5	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 6	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 7	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, autosomal recessive 8	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Dyskeratosis congenita, digenic	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: dyskeratosis congenita
Fanconi Anemia, X-Linked	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: fanconi anemia
Fanconi anemia	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: fanconi anemia
Fanconi anemia complementation group A	High risk of lethality	Progressive Systemic Disorder: Matched high-risk keyword: fanconi anemia

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Fanconi anemia, complementation group S	High risk of lethality	Progressive Systemic Disorder	Matched high-risk keyword: fanconi anemia
Fanconi anemia, complementation group W	High risk of lethality	Progressive Systemic Disorder	Matched high-risk keyword: fanconi anemia
Athabaskan severe combined immunodeficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
FHL1-related disorder	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: fhl
FHL2-related disorder	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: fhl
Familial hemophagocytic lymphohistiocytosis	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Familial hemophagocytic lymphohistiocytosis 2	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Familial hemophagocytic lymphohistiocytosis 3	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Familial hemophagocytic lymphohistiocytosis 4	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Familial hemophagocytic lymphohistiocytosis 5	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Familial hemophagocytic lymphohistiocytosis type 1	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, 5, WITHOUT MICROVILLUS INCLUSION DISEASE	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Hemophagocytic lymphohistiocytosis, familial, 6	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: hemophagocytic lymphohistiocytosis
Severe combined immunodeficiency disease	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to CARD11 deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to CARMIL2 deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to CORO1A deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to CTPS1 deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency

Severe combined immunodeficiency due to DCLRE1C deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to DNA-PKcs deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to IKK2 deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to LAT deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency due to LCK deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency, B cell-negative	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-negative, due to adenosine deaminase deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-negative, NK cell-positive	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Short-limb skeletal dysplasia with severe combined immunodeficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
T-B+ severe combined immunodeficiency due to JAK3 deficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
X-linked severe combined immunodeficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
ZAP70-Related Severe Combined Immunodeficiency	High risk of lethality	Severe Immunodeficiency	Matched high-risk keyword: severe combined immunodeficiency
Combined malonic and methylmalonic acidemia	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
LCHAD deficiency with maternal acute fatty liver of pregnancy	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: lchad deficiency
Maple syrup urine disease	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: maple syrup urine disease
Maple syrup urine disease type 1A	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: maple syrup urine disease



Maple syrup urine disease type 1B	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: maple syrup urine disease
Maple syrup urine disease type 2	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: maple syrup urine disease
Maple syrup urine disease, mild variant	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: maple syrup urine disease
Methylmalonic acidemia	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Methylmalonic acidemia due to transcobalamin receptor defect	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Methylmalonic acidemia with homocystinuria cblC	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Methylmalonic acidemia with homocystinuria, type cblJ	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Methylmalonic acidemia with homocystinuria, type cblX	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: methylmalonic acidemia
Mitochondrial trifunctional protein deficiency	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: mitochondrial trifunctional protein deficiency
Mitochondrial trifunctional protein deficiency 1	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: mitochondrial trifunctional protein deficiency
Mitochondrial trifunctional protein deficiency 2	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: mitochondrial trifunctional protein deficiency
Mitochondrial trifunctional protein deficiency 2 with myopathy and neuropathy	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: mitochondrial trifunctional protein deficiency
Propionic acidemia	High risk of lethality	Severe Metabolic Crisis Risk	Matched high-risk keyword: propionic acidemia

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