Tests Reported by DNA to Z

1- Medicines

5-fluorouracil
6-Mercaptopurine
Abacavir
Abiraterone acetate
Amlodipine
Aspirin
Atomoxetine
Azathioprine
Bupropion
Capecitabine
Carbamazepine
Cetuximab
Chloroquine
Chlorpropamide
Chlorthalidone
Cisplatin
Clopidogrel
Clozapine
Codeine
Disulfiram
Docetaxel
Dofetilide
Escitalopram
Esomeprazole
Flucloxacillin
Fluoropyrimidines
Fluticasone propionate
Gefitinib
Glimepiride
Glipizide
Glyburide
Herceptin
Ibuprofen
Infliximab
Interferon
Iressa
Irinotecan
Ivacaftor
Kalydeco

Lansoprazole
Lidocaine
Losec
Mafenide
Metformin
Methadone
Methylene blue
Morphine
Moviprep
Naproxen
Nexium
Nitrofurantoin
Nortriptyline
Olanzapine
Omeprazole
Paclitaxel
Pegloticase
Phenytoin
Plavix
Pravastatin
Prevacid
Prilosec
Primaquine
Proguanil
Rasburicase
Remicade
Risperidone
Rituximab
Rosuvastatin
Sedatives
Sertraline
Statins
Sulfamethoxazole
Tamoxifen
Taxotere
Tegafur
Venlafaxine
Warfarin
Ziagen

2- Medical Conditions

17-Beta-Hydroxysteroid Dehydrogenase III Deficiency
2-aminoadipic 2-oxoadipic aciduria
2-methyl-3-hydroxybutyric aciduria

21-hydroxylase deficiency
22q13.3 deletion syndrome
3 Methylcrotonyl-CoA carboxylase 1 deficiency
3 beta-Hydroxysteroid dehydrogenase deficiency
3-MCC Deficiency
3-Methylglutaconic aciduria
3-Methylglutaconic aciduria type 2
3-Methylglutaconic aciduria type 3
3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency
3-methylcrotonyl CoA carboxylase 2 deficiency
3-methylglutaconic aciduria type V
3-methylglutaconic aciduria with cataracts
3-methylglutaconic aciduria with deafness
4-Alpha-hydroxyphenylpyruvate hydroxylase deficiency
4-Hydroxyphenylpyruvate dioxygenase deficiency

5-Oxoprolinase deficiency
6-pyruvoyl-tetrahydropterin synthase deficiency
ABCA1-Related Disorders
ABCA4-Related Disorders
ABCB4-Related Intrahepatic Cholestasis
ABCD syndrome
ABO blood group system
ACTH deficiency
ACTH resistance
ACTININ
ACTN3 deficiency
ADRENAL INSUFFICIENCY
ADULT syndrome
AICAR transformylase/IMP cyclohydrolase deficiency
AIPL1-Related Disorders
ALBUMIN ASOLA
ALBUMIN B
ALBUMIN BLENHEIM
ALBUMIN CASTEL DI SANGRO
ALBUMIN DUBLIN
ALBUMIN FUKUOKA 1
ALBUMIN HAWKES BAY
ALBUMIN HERBORN
ALBUMIN HIROSHIMA 1
ALBUMIN HIROSHIMA 2
ALBUMIN IOWA CITY 1
ALBUMIN KOMAGOME 2
ALBUMIN MALMO-10
ALBUMIN MALMO-95
ALBUMIN MERSIN
ALBUMIN NAGOYA

ALBUMIN NASKAPI
ALBUMIN ORTONOVO
ALBUMIN OSAKA 1
ALBUMIN PARKLANDS
ALBUMIN ROMA
ALBUMIN RUGBY PARK
ALBUMIN SONDRIA
ALBUMIN TORINO
ALBUMIN TRADATE 2
ALBUMIN VANCOUVER
ALBUMIN VIBO VALENTIA
ALDH9A1*2 POLYMORPHISM
ALPHA-2-MACROGLOBULIN POLYMORPHISM
AMINOLEVULINATE DEHYDRATASE
AMYLOIDOSIS
ANEMIA
ANO5-Related Disorders
ANTERIOR SEGMENT DYSGENESIS 5
ANTICHYMOTRYPSIN BONN 1
ANTICHYMOTRYPSIN ISEHARA 1
APC-Associated Polyposis Disorders
APOE2-DUNEDIN
APOE3 VARIANT
APOE3(-)-FREIBURG
APOE4 VARIANT
APOE4(-)-FREIBURG
APOLIPOPROTEIN A-I (BALTIMORE)
APOLIPOPROTEIN A-I (GIESSEN)
APOLIPOPROTEIN A-I (MARBURG)
APOLIPOPROTEIN A-I (MUNSTER3B)
APOLIPOPROTEIN A-I (MUNSTER4)
APOLIPOPROTEIN A-I (NORWAY)
APOLIPOPROTEIN A-IV POLYMORPHISM
APOLIPOPROTEIN A-IV RARE VARIANT
APOLIPOPROTEIN C-II (AFRICAN)
APOLIPOPROTEIN C-II (AUCKLAND)
APOLIPOPROTEIN C-II (PARIS)
APOLIPOPROTEIN C-II (SAN FRANCISCO)
APOLIPOPROTEIN C-II (WAKAYAMA)
APRT deficiency
ARYLSULFATASE A POLYMORPHISM
ATR-X syndrome
AUBERGER BLOOD GROUP POLYMORPHISM Au(a)/Au(b)
Aarskog syndrome
Aase syndrome
Abdominal obesity-metabolic syndrome 3
Abetalipoproteinaemia

Abetalipoproteinemia
Abnormal blistering of the skin
Abnormal electroretinogram
Abnormal facial shape
Abnormal glycosylation (CDG IIa)
Abnormality of brain morphology
Abnormality of cardiovascular system morphology
Abnormality of dental enamel
Abnormality of earlobe
Abnormality of neuronal migration
Abnormality of the corpus callosum
Abnormality of the macula
Abnormality of the teeth
Abortive cerebellar ataxia
Abruzzo Erickson syndrome
Absent corpus callosum cataract immunodeficiency
Absent or delayed speech development
Absent speech
Acampomelic campomelic dysplasia
Acanthocytosis due to band 3 ht
Achalasia-alacrima syndrome
Achondrogenesis
Achondroplasia
Achromatopsia
Achromatopsia 2
Achromatopsia 3
Achromatopsia 4
Achromatopsia 5
Achromatopsia 6
Acid alpha-glucosidase
Acid-labile subunit deficiency
Acquired hemoglobin H disease
Acquired long QT syndrome
Acquired susceptibility to long QT syndrome 1
Acrocallosal syndrome
Acrocapitofemoral dysplasia
Acrocephalosyndactyly type I
Acrodysostosis 1 with or without hormone resistance
Acrodysostosis 2
Acroerythrokeratoderma
Acrokeratosis verruciformis of Hopf
Acromelic frontonasal dysostosis
Acromesomelic dysplasia Maroteaux type
Acromicric dysplasia
Acth-independent macronodular adrenal hyperplasia 2
Acute Recurrent Myoglobinuria
Acute alcohol sensitivity

Acute intermittent porphyria
Acute lymphoid leukemia
Acute megakaryoblastic leukemia
Acute myeloid leukemia
Acute neuronopathic Gaucher's disease
Acyl-CoA dehydrogenase family
Adams-Oliver syndrome 1
Adams-Oliver syndrome 2
Adams-Oliver syndrome 3
Adams-Oliver syndrome 4
Adams-Oliver syndrome 5
Addison disease
Addison's disease
Adenine phosphoribosyltransferase deficiency
Adenocarcinoma of lung
Adenocarcinoma of prostate
Adenocarcinoma of stomach
Adenoid cystic carcinoma
Adenoma of the adrenal gland
Adenomatous polyposis coli
Adenosine deaminase 2 allozyme
Adenosine triphosphate
Adenylate kinase deficiency
Adenylosuccinate lyase deficiency
Adolescent nephronophthisis
Adrenal insufficiency
Adrenocortical carcinoma
Adrenocortical tumor
Adrenoleukodystrophy
Adult hypophosphatasia
Adult junctional epidermolysis bullosa
Adult neuronal ceroid lipofuscinosis
Adult onset ataxia with oculomotor apraxia
Adult polyglucosan body neuropathy
Adult-onset night blindness
Advanced sleep phase syndrome
Afibrinogenemia
Agammaglobulinemia 2
Agammaglobulinemia 6
Agammaglobulinemia 7
Age-related cortical cataract
Age-related macular degeneration 12
Age-related macular degeneration 14
Age-related macular degeneration 2
Age-related macular degeneration 3
Age-related macular degeneration 4
Age-related macular degeneration 6

Age-related macular degeneration 7
Age-related macular degeneration 8
Agenesis of cerebellar vermis
Agenesis of corpus callosum
Aicardi Goutieres syndrome
Aicardi Goutieres syndrome 1
Aicardi Goutieres syndrome 2
Aicardi Goutieres syndrome 3
Aicardi Goutieres syndrome 4
Aicardi Goutieres syndrome 5
Aicardi-goutieres syndrome 6
Aicardi-goutieres syndrome 7
Alacrima
Alagille syndrome 1
Alagille syndrome 2
Alazami-Yuan syndrome
Albinism
Alcohol dependence
Aldosterone Producing Adrenal Cortex Adenoma
Aldosterone-producing adrenal adenoma
Alexander's disease
Alkaptonuria
Allan-Herndon-Dudley syndrome
Alloalbuminemia
Alopecia
Alopecia of scalp
Alopecia universalis
Alopecia universalis congenita
Alpers encephalopathy
Alpha-1-antitrypsin deficiency
Alpha-B crystallinopathy
Alpha-fetoprotein
Alpha-methylacyl-CoA racemase deficiency
Alpha-thalassemia-2
Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion
Alport syndrome
Alstrom syndrome
Alternating hemiplegia of childhood 1
Alternating hemiplegia of childhood 2
Alveolar capillary dysplasia with misalignment of pulmonary veins
Alzheimer disease
Alzheimer disease 19
Alzheimer disease 2
Alzheimer disease familial 3
Alzheimer's disease
Amelogenesis Imperfecta
Amelogenesis imperfecta

Amelogenesis imperfecta - hypoplastic autosomal dominant - local
Aminoacylase 1 deficiency
Aminoglycoside-induced deafness
Amish infantile epilepsy syndrome
Amish lethal microcephaly
Amyloid Cardiomyopathy
Amyloidogenic transthyretin amyloidosis
Amyloidosis
Amyotrophic Lateral Sclerosis
Amyotrophic lateral sclerosis
Amyotrophic lateral sclerosis 1
Amyotrophic lateral sclerosis 14
Amyotrophic lateral sclerosis 14 without frontotemporal dementia
Amyotrophic lateral sclerosis 16
Amyotrophic lateral sclerosis 17
Amyotrophic lateral sclerosis 18
Amyotrophic lateral sclerosis 19
Amyotrophic lateral sclerosis 20
Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia
Amyotrophic lateral sclerosis 6
Amyotrophic lateral sclerosis type 1
Amyotrophic lateral sclerosis type 10
Amyotrophic lateral sclerosis type 11
Amyotrophic lateral sclerosis type 12
Amyotrophic lateral sclerosis type 2
Amyotrophic lateral sclerosis type 4
Amyotrophic lateral sclerosis type 5
Amyotrophic lateral sclerosis type 6
Amyotrophic lateral sclerosis type 8
Amyotrophic lateral sclerosis type 9
Amyotrophic lateral sclerosis-parkinsonism/dementia complex 1
Amyotrophy
Anal atresia
Analbuminemia
Analbuminemia baghdad
Andermann syndrome
Andersen Tawil syndrome
Androgen resistance syndrome
Anemia
Anemia sideroblastic and spinocerebellar ataxia
Angelman syndrome
Angelman syndrome-like
Angiofibroma
Angiopathy
Angiotensin i-converting enzyme
Anhaptoglobinemia
Aniridia

Aniridia 1
Anonychia
Anophthalmia
Anophthalmia - microphthalmia
Anophthalmia/Microphthalmia
Anorexia nervosa 2
Antenatal Bartter Syndrome
Anterior segment anomalies
Anterior segment dysgenesis 6
Anterior segment dysgenesis 7
Anterior segment mesenchymal dysgenesis
Anti-plasmin deficiency
Antigen in Scianna blood group system
Antiinflammatory agents
Antithrombin III deficiency
Antithrombin deficiency
Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
Aortic aneurysm
Aortic root dilatation
Aortic valve disease 2
Aortic valve disorder
Aphakia
Aphasia
Aplasia cutis congenita
Aplastic anemia
Apolipoprotein A-I deficiency
Apolipoprotein A-II deficiency
Apolipoprotein C2 deficiency
Apolipoprotein c-ii variant
Apolipoprotein c-iii
Apolipoproteinemia E1
Apparent mineralocorticoid excess
Aquaporin 1 deficiency
Arachnodactyly
Arginase deficiency
Arginine:glycine amidinotransferase deficiency
Argininosuccinate lyase deficiency
Aromatase deficiency
Arrhythmia
Arrhythmogenic right ventricular cardiomyopathy
Arrhythmogenic right ventricular dysplasia
Arrhythmogenic right ventricular dysplasia/cardiomyopathy
Arterial calcification of infancy
Arterial tortuosity syndrome
Arthrogryposis
Arthrogryposis multiplex congenita

Arthrogryposis multiplex congenita distal type 1
Arthrogryposis renal dysfunction cholestasis syndrome
Arthyrgryposis
Arts syndrome
Arylsulfatase A pseudodeficiency
Arylsulfatase a
Arylsulfatase a pseudodeficiency
Asparagine synthetase deficiency
Aspartylglucosaminuria
Aspartylglycosaminuria
Aspergillosis
Asphyxiating thoracic dystrophy 2
Asphyxiating thoracic dystrophy 4
Asphyxiating thoracic dystrophy 5
Asplenia
Asthma
Astrocytoma
Ataxia
Ataxia Neuropathy Spectrum Disorders
Ataxia and retinitis pigmentosa with isolated vitamin e deficiency
Ataxia with vitamin E deficiency
Ataxia-telangiectasia syndrome
Ataxia-telangiectasia variant
Ataxia-telangiectasia without immunodeficiency
Ataxia-telangiectasia-like disorder 1
Ataxia-telangiectasia-like disorder 2
Ateleiotic dwarfism
Atelosteogenesis type 1
Atelosteogenesis type 2
Atelosteogenesis type 3
Atopic asthma
Atransferrinemia
Atrial fibrillation
Atrial septal defect
Atrial septal defect 2
Atrial septal defect 4
Atrial septal defect 5
Atrial septal defect 6
Atrial septal defect 7 with or without atrioventricular conduction defects
Atrial standstill 1
Atrial standstill 2
Atrichia with papular lesions
Atrioventricular block
Atrioventricular canal defect
Atrioventricular septal defect
Atrioventricular septal defect 2
Atrioventricular septal defect 4

Atrioventricular septal defect and common atrioventricular junction
Atrophia bulborum hereditaria
Attention deficit hyperactivity disorder
Attention deficit-hyperactivity disorder 7
Atypical Rett syndrome
Atypical hemolytic uremic syndrome
Atypical hemolytic-uremic syndrome 1
Atypical hemolytic-uremic syndrome 3
Atypical hemolytic-uremic syndrome 6
Atypical mycobacteriosis
Auditory neuropathy
Aural atresia
Auriculocondylar syndrome 1
Auriculocondylar syndrome 2
Auriculocondylar syndrome 3
Autism
Autism 1
Autism 10
Autism 15
Autism 16
Autistic spectrum disorder with isolated skills
Autoimmune disease
Autoimmune disease 6
Autoimmune lymphoproliferatiVe syndrome
Autoimmune lymphoproliferative syndrome
Autoimmune polyglandular syndrome type 1
Autoinflammation
Autoinflammation with infantile enterocolitis
Autosomal dominant hypohidrotic ectodermal dysplasia
Autosomal dominant hypophosphatemic rickets
Autosomal dominant optic atrophy plus syndrome
Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 1
Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 2
Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 3
Autosomal dominant progressive external ophthalmoplegia with mitochondrial DNA deletions 4
Autosomal dominant torsion dystonia 4
Autosomal recessive Dejerine-Sottas syndrome
Autosomal recessive centronuclear myopathy
Autosomal recessive congenital ichthyosis 1
Autosomal recessive congenital ichthyosis 2
Autosomal recessive congenital ichthyosis 3
Autosomal recessive congenital ichthyosis 4A
Autosomal recessive congenital ichthyosis 4B
Autosomal recessive congenital ichthyosis 5
Autosomal recessive congenital ichthyosis 9
Autosomal recessive cutis laxa type 1B
Autosomal recessive cutis laxa type 2B

Autosomal recessive cutis laxa type 3B
Autosomal recessive cutis laxa type IA
Autosomal recessive hearing impairment with normal menstrual cycles
Autosomal recessive hypohidrotic ectodermal dysplasia syndrome
Autosomal recessive hypophosphatemic bone disease
Autosomal recessive hypophosphatemic vitamin D refractory rickets
Autosomal recessive non-syndromic sensorineural deafness type DFNB
Autosomal recessive polycystic kidney disease
Autosomal recessive syndrome of syndactyly
Autosomal recessive woolly hair 1
Avascular necrosis of the head of femur
Axenfeld-Rieger Syndrome
Axenfeld-Rieger syndrome type 1
Axenfeld-Rieger syndrome type 3
Ayme-gripp syndrome
Azathioprine response
B-cell expansion with NFKB and T-cell anergy
B-cell non-Hodgkin lymphoma
BCHE
BCS1L-Related Disorders
BEST1-Related Disorders
BETHLEM MYOPATHY 1
BLOOD GROUP ERIK
BLOOD GROUP--FROESE
BLOOD GROUP--LUTHERAN INHIBITOR
BLOOD GROUP--LUTHERAN NULL
BLOOD GROUP--OK
BLOOD GROUP--WALDNER TYPE
BLOOD GROUP--WRIGHT ANTIGEN
BRCA2-Related Disorders
Babinski sign
Bacteremia
Bainbridge-Ropers syndrome
Baller-Gerold syndrome
Bamforth syndrome
Band 3 memphis
Band-like calcification with simplified gyration and polymicrogyria
Bannayan-Riley-Ruvalcaba syndrome
Baraitser-Winter Syndrome 2
Baraitser-Winter syndrome 1
Barakat syndrome
Bardet-Biedl syndrome
Bardet-Biedl syndrome 1
Bardet-Biedl syndrome 1/7
Bardet-Biedl syndrome 10
Bardet-Biedl syndrome 11
Bardet-Biedl syndrome 12

Bardet-Biedl syndrome 13
Bardet-Biedl syndrome 14
Bardet-Biedl syndrome 15
Bardet-Biedl syndrome 16
Bardet-Biedl syndrome 17
Bardet-Biedl syndrome 18
Bardet-Biedl syndrome 19
Bardet-Biedl syndrome 2
Bardet-Biedl syndrome 21
Bardet-Biedl syndrome 3
Bardet-Biedl syndrome 4
Bardet-Biedl syndrome 5
Bardet-Biedl syndrome 6
Bardet-Biedl syndrome 7
Bardet-Biedl syndrome 8
Bardet-Biedl syndrome 9
Bardet-biedl syndrome 1/2
Bardet-biedl syndrome 2/4
Bardet-biedl syndrome 2/6
Bare Lymphocyte Syndrome
Bare lymphocyte syndrome
Bare lymphocyte syndrome type 2
Barrett esophagus/esophageal adenocarcinoma
Bartter syndrome
Bartter syndrome type 3
Bartter syndrome type 4
Basal cell carcinoma
Basal ganglia calcification
Basal ganglia disease
Basal laminar drusen
Bche
Beaded hair
Beaulieu-Boycott-Innes syndrome
Becker muscular dystrophy
Beckwith-Wiedemann syndrome
Bell-shaped thorax
Benign Neonatal Epilepsy
Benign familial hematuria
Benign familial neonatal seizures
Benign familial neonatal seizures 1
Benign familial neonatal seizures 2
Benign familial neonatal-infantile seizures
Benign hereditary chorea
Benign recurrent intrahepatic cholestasis 2
Benign scapuloperoneal muscular dystrophy with cardiomyopathy
Bent bone dysplasia syndrome
Berger disease

Bernard Soulier syndrome
Bernard-Soulier syndrome
Bernard-Soulier syndrome type C
Bestrophinopathy
Beta thalassemia intermedia
Beta thalassemia major
Beta-2-adrenoreceptor agonist
Beta-D-mannosidosis
Beta-aminoisobutyric aciduria
Beta-glycopyranoside tasting
Beta-hydroxyisobutyryl-CoA deacylase deficiency
Beta-malay-thalassemia
Beta-plus-thalassemia
Beta-showa-yakushiji thalassemia
Bethlem myopathy 1
Bietti crystalline corneoretinal dystrophy
Bifid nose with or without anorectal and renal anomalies
Bifunctional peroxisomal enzyme deficiency
Bilateral cleft lip and palate
Bilateral squint
Bilateral undescended testicles
Bile acid malabsorption
Bile acid synthesis defect
Bilirubin
Biotinidase deficiency
Bipolar affective disorder
Birbeck granule deficiency
Birk Barel mental retardation dysmorphism syndrome
Bisphosphonates response - Efficacy
Bjornstad syndrome with mild mitochondrial complex III deficiency
Bladder cancer
Bladder carcinoma
Blau syndrome
Blepharophimosis
Blepharophimosis syndrome type 1
Blepharophimosis syndrome type 2
Blindness
Blood group
Bloom syndrome
Body mass index
Body mass index quantitative trait locus 12
Body mass index quantitative trait locus 9
Bombay phenotype
Bone fragility with contractures
Bone marrow failure syndrome 1
Boomerang dysplasia
Borjeson-Forssman-Lehmann syndrome

Borrone Di Rocco Crovato syndrome
Bosch-Boonstra-Schaaf optic atrophy syndrome
Bosley-Salih-Alorainy syndrome
Bothnia retinal dystrophy
Boucher Neuhauser syndrome
Bowed humerus
Bowen-Conradi syndrome
Bowing of the long bones
Brachydactyly
Brachydactyly syndrome
Brachydactyly type A1
Brachydactyly type A2
Brachydactyly type B1
Brachydactyly type B2
Brachydactyly type C
Brachydactyly type D
Brachydactyly type E1
Brachydactyly type E2
Brachyolmia
Brachyrachia (short spine dysplasia)
Brain atrophy
Brain iron accummulation
Brain malformation
Brain small vessel disease with hemorrhage
Brain tumor-polyposis syndrome 2
Brainstem glioma
Branched-chain keto acid dehydrogenase kinase deficiency
Branchiooculofacial syndrome
Branchiootic syndrome
Branchiootic syndrome 3
Branchiootorenal Spectrum Disorders
Branchiootorenal syndrome 2
Branchiootorenal syndrome with cataract
Breast adenocarcinoma
Breast and colorectal cancer
Breast and/or ovarian cancer
Breast cancer
Breast carcinoma
Breast-ovarian cancer
British HPFH
Brittle cornea syndrome 2
Broad thumb
Brody myopathy
Bronchiectasis with or without elevated sweat chloride 1
Bronchiectasis with or without elevated sweat chloride 2
Bronchiectasis with or without elevated sweat chloride 3
Brown-Vialetto-Van Laere syndrome 1

Brown-Vialetto-Van Laere syndrome 2
Bruck syndrome 1
Bruck syndrome 2
Brugada syndrome
Brugada syndrome 1
Brugada syndrome 2
Brugada syndrome 3
Brugada syndrome 4
Brugada syndrome 5
Brugada syndrome 6
Brugada syndrome 7
Brugada syndrome 8
Bruising susceptibility
Budd-Chiari syndrome
Bulimia nervosa 2
Bull's eye macular dystrophy
Bull's eye maculopathy
Bullous ichthyosiform erythroderma
Burkitt lymphoma
Burn-McKeown syndrome
Butyrylcholinesterase deficiency
C-like syndrome
C1q deficiency
C3 deficiency
C6 A/B POLYMORPHISM
C7 and C6 deficiency
CAPN3-Related Disorders
CARBONIC ANHYDRASE II VARIANT
CCR5 POLYMORPHISM
CDH23-Related Disorders
CEP152-Related Disorders
CEP290-Related Disorders
CEREBRAL AMYLOID ANGIOPATHY
CHARGE association
CHEK2-Related Cancer Susceptibility
CHOLECYSTOKININ A RECEPTOR POLYMORPHISM
CIP1/WAF1 TUMOR-ASSOCIATED POLYMORPHISM 1
CLCN4-related disorder
CNS hypomyelination
COACH syndrome
CODAS syndrome
CODON 72 POLYMORPHISM
COLTON BLOOD GROUP POLYMORPHISM
CONGENITAL DISORDER OF GLYCOSYLATION
CYBA POLYMORPHISM
CYP2E1*5B ALLELE
CYP3A4 PROMOTER POLYMORPHISM

Cachexia
Calcaneovalgus deformity
Calcification of joints and arteries
Campomelic dysplasia with autosomal sex reversal
Camptocormism
Camptodactyly
Camptodactyly of finger
Camptomelic dysplasia
Canavan disease
Cancer of multiple types
Cancer progression and tumor cell motility
Candidiasis
Cap myopathy 2
Capillary hemangiomas
Capillary malformation without arteriovenous malformation
Capillary malformation-arteriovenous malformation
Capillary malformations
Carbamazepine hypersensitivity
Carbohydrate-deficient glycoprotein syndrome type I
Carbohydrate-deficient glycoprotein syndrome type II
Carbonic anhydrase I
Carbonic anhydrase I deficiency
Carbonic anhydrase VA deficiency
Carboxylesterase 1 deficiency
Carcinoid tumor of intestine
Carcinoma
Carcinoma of cervix
Carcinoma of colon
Carcinoma of gallbladder
Carcinoma of pancreas
Cardiac arrest
Cardiac arrhythmia
Cardiac conduction defect
Cardiac conduction disease with or without dilated cardiomyopathy
Cardiac rhabdomyoma
Cardiac valvular dysplasia
Cardio-facio-cutaneous syndrome
Cardioencephalomyopathy
Cardiofaciocutaneous syndrome 1
Cardiofaciocutaneous syndrome 2
Cardiofaciocutaneous syndrome 3
Cardiofaciocutaneous syndrome 4
Cardiomyopathy
Cardiomyopathy and Deafness
Cardiomyopathy dilated with woolly hair and keratoderma
Cardiomyopathy with or without skeletal myopathy
Cardiovascular phenotype

Carious teeth
Carnevale syndrome
Carney complex
Carney complex variant
Carney triad
Carnitine acylcarnitine translocase deficiency
Carnitine palmitoyltransferase I deficiency
Carnitine palmitoyltransferase II deficiency
Carotid body paraganglioma
Carpal tunnel syndrome
Carpenter syndrome
Carpenter syndrome 1
Carpenter syndrome 2
Caspase-8 deficiency
Cataplexy
Cataplexy and narcolepsy
Cataract
Cataract 1
Cataract 16
Cataract 2
Cataract 20
Cataract 23
Cataract 30
Cataract 4
Cataract 41
Cataract 43
Cataract 6
Cataract 9
Cataract and cardiomyopathy
Cataracts
Catecholaminergic polymorphic ventricular tachycardia
Catel Manzke syndrome
Caudal dysgenesis syndrome
Caudal regression syndrome
Cd8 deficiency
Central core disease
Central hypoventilation syndrome
Central precocious puberty
Central scotoma
Centromeric instability of chromosomes 1
Cerebellar ataxia
Cerebellar ataxia and mental retardation with quadrupedal locomotion 1
Cerebellar ataxia infantile with progressive external ophthalmoplegia
Cerebellar atrophy
Cerebellar hypoplasia
Cerebellar hypoplasia with endosteal sclerosis
Cerebello-oculo-renal syndrome (nephronophthisis

Cerebellofaciodental syndrome
Cerebral amyloid angiopathy
Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy
Cerebral cavernous malformation
Cerebral cavernous malformations 1
Cerebral cavernous malformations 2
Cerebral folate deficiency
Cerebral infarction
Cerebral ischemia
Cerebral palsy
Cerebro-oculo-facio-skeletal syndrome
Cerebrooculofacioskeletal Syndrome
Cerebrooculofacioskeletal syndrome 2
Cerebrooculofacioskeletal syndrome 4
Cerebroretinal microangiopathy with calcifications and cysts 1
Ceroid lipofuscinosis
Ceroid lipofuscinosis neuronal 1
Ceroid lipofuscinosis neuronal 10
Ceroid lipofuscinosis neuronal 2
Ceroid lipofuscinosis neuronal 4B autosomal dominant
Ceroid lipofuscinosis neuronal 5
Ceroid lipofuscinosis neuronal 6
Ceroid lipofuscinosis neuronal 7
Ceroid lipofuscinosis neuronal 8
Char syndrome
Charcot-Marie-Tooth
Charcot-Marie-Tooth Neuropathy X
Charcot-Marie-Tooth disease
Charcot-Marie-Tooth disease and deafness
Charcot-Marie-Tooth disease dominant intermediate 3
Charcot-Marie-Tooth disease type 2B1
Charcot-Marie-Tooth disease type 2B2
Charcot-Marie-Tooth disease type 2C
Charcot-Marie-Tooth disease type 2D
Charcot-Marie-Tooth disease type 2E
Charcot-Marie-Tooth disease type 2F
Charcot-Marie-Tooth disease type 2I
Charcot-Marie-Tooth disease type 2J
Charcot-Marie-Tooth disease type 2K
Charcot-Marie-Tooth disease type 2P
Charcot-Marie-Tooth disease type 4B2 with early-onset glaucoma
Chediak-Higashi syndrome
Chilblain Lupus
Chilblain lupus 1
Chilblain lupus 2
Child syndrome

Childhood hepatocellular carcinoma
Childhood hypophosphatasia
Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia
Chitotriosidase deficiency
Chloramphenicol resistance
Cholecystitis
Cholestanol storage disease
Cholestasis
Cholestasis of pregnancy
Cholesterol monooxygenase (side-chain cleaving) deficiency
Chondrocalcinosis 2
Chondrodysplasia Blomstrand type
Chondrodysplasia punctata 1
Chondrodysplasia punctata 2 X-linked dominant
Chondrodysplasia with joint dislocations
Chondrodysplasia with platyspondyly
Chondroectodermal dysplasia
Choreoacanthocytosis
Choreoathetosis
Choroid plexus carcinoma
Choroidal Dystrophy
Choroidal dystrophy
Choroideremia
Christianson syndrome
Chromophobe renal cell carcinoma
Chromosome 2q32-q33 deletion syndrome
Chromosome 9q deletion syndrome
Chromosome Xq28 deletion syndrome
Chronic and progressive ataxia
Chronic atrial and intestinal dysrhythmia
Chronic granuloma and hemolytic anemia
Chronic granulomatous disease
Chronic infantile neurological
Chronic intestinal pseudoobstruction
Chronic kidney disease
Chronic lung disease
Chronic lymphocytic leukemia
Chronic myeloid leukemia
Chronic progressive multiple sclerosis
Chudley-McCullough syndrome
Chylomicron retention disease
Chédiak-Higashi syndrome
Ciliary dyskinesia
Cirrhosis
Citrin deficiency
Citrullinemia
Citrullinemia type I

Citrullinemia type II
Classical primary microcephaly
Clear cell carcinoma of kidney
Cleft Lip +/- Cleft Palate
Cleft lip/palate-ectodermal dysplasia syndrome
Cleft palate with ankyloglossia
Cleidocranial dysostosis
Cleidocranial dysplasia
Clopidogrel response
Coarse facial features
Cockayne syndrome
Cockayne syndrome B
Cockayne syndrome type A
Coenzyme Q10 deficiency
Coffin Siris/Intellectual Disability
Coffin-Lowry syndrome
Coffin-Siris syndrome 1
Coffin-Siris syndrome 5
Cognitive impairment
Cogwheel rigidity
Cohen syndrome
Colchicine resistance
Cold-induced sweating syndrome 1
Cold-induced sweating syndrome 2
Cole disease
Cole-Carpenter syndrome 2
Collagen VI-related myopathy
Coloboma
Coloboma of optic disc
Colobomatous microphthalmia
Colon cancer
Colonic adenocarcinoma
Colorectal / endometrial cancer
Colorectal Neoplasms
Colorectal adenoma
Colorectal cancer
Colorectal cancer 3
Colorectal cancer with chromosomal instability
Colton-null
Combined cellular and humoral immune defects with granulomas
Combined d-2- and l-2-hydroxyglutaric aciduria
Combined deficiency of factor V and factor VIII
Combined deficiency of sialidase AND beta galactosidase
Combined immunodeficiency
Combined malonic and methylmalonic aciduria
Combined oxidative phosphorylation deficiency 1
Combined oxidative phosphorylation deficiency 10

Combined oxidative phosphorylation deficiency 11
Combined oxidative phosphorylation deficiency 12
Combined oxidative phosphorylation deficiency 13
Combined oxidative phosphorylation deficiency 14
Combined oxidative phosphorylation deficiency 15
Combined oxidative phosphorylation deficiency 16
Combined oxidative phosphorylation deficiency 17
Combined oxidative phosphorylation deficiency 18
Combined oxidative phosphorylation deficiency 20
Combined oxidative phosphorylation deficiency 21
Combined oxidative phosphorylation deficiency 23
Combined oxidative phosphorylation deficiency 24
Combined oxidative phosphorylation deficiency 27
Combined oxidative phosphorylation deficiency 3
Combined oxidative phosphorylation deficiency 4
Combined oxidative phosphorylation deficiency 5
Combined oxidative phosphorylation deficiency 8
Combined oxidative phosphorylation deficiency 9
Combined partial 17-alpha-hydroxylase/17
Combined saposin deficiency
Common Variable Immune Deficiency
Common variable immunodeficiency 1
Common variable immunodeficiency 10
Common variable immunodeficiency 11
Common variable immunodeficiency 2
Common variable immunodeficiency 6
Common variable immunodeficiency 7
Common variable immunodeficiency 8
Complement 1s deficiency
Complement component 2 deficiency
Complement component 4
Complement component 7 deficiency
Complement component 8 deficiency type 2
Complement component 9 deficiency
Complement factor B deficiency
Complement factor d deficiency
Complete combined 17-alpha-hydroxylase/17
Cone dystrophy 3
Cone dystrophy 4
Cone monochromatism
Cone-Rod Dystrophy
Cone-rod dystrophy
Cone-rod dystrophy 10
Cone-rod dystrophy 11
Cone-rod dystrophy 12
Cone-rod dystrophy 13
Cone-rod dystrophy 16

Cone-rod dystrophy 18
Cone-rod dystrophy 19
Cone-rod dystrophy 2
Cone-rod dystrophy 20
Cone-rod dystrophy 3
Cone-rod dystrophy 5
Cone-rod dystrophy 6
Cone-rod dystrophy 7
Cone-rod dystrophy 9
Cone-rod dystrophy amelogenesis imperfecta
Congenital Bile Acid Synthesis Defect
Congenital Cataracts
Congenital Indifference to Pain
Congenital Muscular Dystrophy
Congenital Myasthenic Syndrome
Congenital Stationary Night Blindness
Congenital absence of salivary gland
Congenital adrenal hyperplasia
Congenital adrenal hypoplasia
Congenital amegakaryocytic thrombocytopenia
Congenital anomalies of kidney and urinary tract 1
Congenital bilateral absence of the vas deferens
Congenital cataract
Congenital cataracts
Congenital central hypoventilation
Congenital contractural arachnodactyly
Congenital contracture
Congenital cystic disease of liver
Congenital defect of folate absorption
Congenital diaphragmatic hernia
Congenital disorder of deglycosylation
Congenital disorder of glycosylation
Congenital disorder of glycosylation type 1B
Congenital disorder of glycosylation type 1C
Congenital disorder of glycosylation type 1D
Congenital disorder of glycosylation type 1E
Congenital disorder of glycosylation type 1F
Congenital disorder of glycosylation type 1G
Congenital disorder of glycosylation type 1H
Congenital disorder of glycosylation type 1I
Congenital disorder of glycosylation type 1J
Congenital disorder of glycosylation type 1K
Congenital disorder of glycosylation type 1M
Congenital disorder of glycosylation type 1N
Congenital disorder of glycosylation type 1P
Congenital disorder of glycosylation type 1Q
Congenital disorder of glycosylation type 1t

Congenital disorder of glycosylation type 1w
Congenital disorder of glycosylation type 1x
Congenital disorder of glycosylation type 2B
Congenital disorder of glycosylation type 2C
Congenital disorder of glycosylation type 2H
Congenital disorder of glycosylation type 2J
Congenital disorder of glycosylation type 2L
Congenital disorder of glycosylation type 2k
Congenital dyserythropoietic anemia
Congenital ectodermal dysplasia of face
Congenital erythropoietic porphyria
Congenital finger flexion contractures
Congenital generalized lipodystrophy type 1
Congenital generalized lipodystrophy type 2
Congenital glucose-galactose malabsorption
Congenital heart defects
Congenital heart defects 1
Congenital heart disease
Congenital hyperammonemia
Congenital hypomyelinating neuropathy
Congenital hypothyroidism
Congenital ichthyosiform erythroderma
Congenital lactase deficiency
Congenital lipomatous overgrowth
Congenital long QT syndrome
Congenital microcephaly
Congenital microvillous atrophy
Congenital muscular dystrophy
Congenital muscular dystrophy due to partial LAMA2 deficiency
Congenital muscular dystrophy-dystroglycanopathy (with or without mental retardation) type B5
Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies
Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5
Congenital muscular dystrophy-dystroglycanopathy with mental retardation
Congenital muscular dystrophy-dystroglycanopathy without mental retardation
Congenital muscular hypertrophy-cerebral syndrome
Congenital myasthenic syndrome
Congenital myasthenic syndrome 13
Congenital myasthenic syndrome 1B
Congenital myasthenic syndrome with tubular aggregates 1
Congenital myopathy
Congenital myopathy with fiber type disproportion
Congenital myotonia
Congenital ocular coloboma
Congenital order of glycosylation type 1r
Congenital secretory diarrhea
Congenital sensorineural hearing impairment
Congenital sensory neuropathy with selective loss of small myelinated fibers

Congenital short bowel syndrome
Congenital stationary night blindness
Connective tissue disorder
Conotruncal anomaly face syndrome/velocardiofacial syndrome
Constipation
Coproporphyria
Cornea plana 2
Corneal Dystrophy
Corneal dystrophy
Corneal dystrophy and perceptive deafness
Corneal endothelial dystrophy
Corneal epithelial dystrophy
Corneal fragility keratoglobus
Cornelia de Lange syndrome 1
Cornelia de Lange syndrome 3
Cornelia de Lange syndrome 4
Cornelia de Lange syndrome 5
Coronary artery disease
Coronary artery disease/myocardial infarction
Coronary artery spasm 3
Coronary heart disease
Corpus callosum
Corpus callosum abnormalities
Corpus callosum agenesis
Cortical dysplasia
Cortical dysplasia-focal epilepsy syndrome
Cortical malformations
Cortical senile cataract
Corticosteroid-binding globulin deficiency
Corticosterone methyloxidase type 1 deficiency
Corticosterone methyloxidase type 2 deficiency
Cortisone reductase deficiency 1
Cortisone reductase deficiency 2
Costello syndrome
Cowchock syndrome
Cowden syndrome
Cowden syndrome 1
Cowden syndrome 2
Cowden syndrome 3
Cowden syndrome 5
Cowden syndrome 6
Cowden syndrome 7
Coxa plana
Craniodiaphyseal dysplasia
Cranioectodermal dysplasia 1
Cranioectodermal dysplasia 2
Cranioectodermal dysplasia 3

Cranioectodermal dysplasia 4
Craniofacial deafness hand syndrome
Craniofacial dysmorphism
Craniofrontonasal dysplasia
Craniolenticulosutural dysplasia
Craniometaphyseal dysplasia
Cranioosteoarthropathy
Craniosynostosis
Craniosynostosis 1
Craniosynostosis 2
Craniosynostosis 3
Craniosynostosis 4
Craniosynostosis and dental anomalies
Craniosynostosis syndrome
Creatine deficiency
Creatine phosphokinase
Crigler Najjar syndrome
Crigler-Najjar syndrome
Crohn disease
Cromer blood group system
Crouzon syndrome
Crouzon syndrome with acanthosis nigricans
Cryptophthalmos syndrome
Cryptorchidism
Culler-Jones syndrome
Currarino triad
Curry-Hall syndrome
Cushing's symphalangism
Cushing's syndrome
Cutaneous malignant melanoma 1
Cutaneous malignant melanoma 3
Cutaneous malignant melanoma 5
Cutaneous melanoma
Cutaneous telangiectasia and cancer syndrome
Cutis Gyrata syndrome of Beare and Stevenson
Cutis Laxa
Cutis laxa
Cutis laxa with osteodystrophy
Cutis laxa with severe pulmonary
Cutis laxa-corneal clouding-oligophrenia syndrome
Cyanosis
Cyclical neutropenia
Cyclical vomiting syndrome
Cylindromatosis
Cyp3a4-v
Cystathioninuria
Cystic Fibrosis-Like Syndrome

Cystic fibrosis
Cystinosis
Cystinuria
Cytochrome c oxidase i deficiency
Cytochrome-c oxidase deficiency
Czech dysplasia metatarsal type
D-2-hydroxyglutaric aciduria 1
D-2-hydroxyglutaric aciduria 2
DFNA 2 Nonsyndromic Hearing Loss
DFNA6/14/38 Nonsyndromic Low-Frequency Sensorineural Hearing Loss
DICER1-related pleuropulmonary blastoma cancer predisposition syndrome
DIEGO BLOOD GROUP ANTIGEN
DLD-Related Disorders
DNA ligase I deficiency
DNA topoisomerase I
DNM2-related intermediate Charcot-Marie-Tooth neuropathy
DOPAMINE RECEPTOR D4 POLYMORPHISM
DUFFY BLOOD GROUP SYSTEM
DYSF-Related Disorders
DYSPROTHROMBINEMIA PROTHROMBIN HIMI-II
Dandy-Walker like malformation with atrioventricular septal defect
Dandy-Walker malformation
Danon disease
Darier disease
Deafness
Deafness and myopia
Deafness with labyrinthine aplasia microtia and microdontia (LAMM)
Death in early adulthood
Death in infancy
Debrisoquine
Decreased activity of cytochrome C oxidase in muscle tissue
Decreased activity of mitochondrial complex I
Decreased activity of the pyruvate dehydrogenase complex
Decreased body weight
Decreased muscle mass
Deep venous thrombosis
Deeply set eye
Deficiency of 2-methylbutyryl-CoA dehydrogenase
Deficiency of 3-hydroxyacyl-CoA dehydrogenase
Deficiency of UDPglucose-hexose-1-phosphate uridylyltransferase
Deficiency of acetyl-CoA acetyltransferase
Deficiency of alpha-mannosidase
Deficiency of aromatic-L-amino-acid decarboxylase
Deficiency of beta-ureidopropionase
Deficiency of bisphosphoglycerate mutase
Deficiency of butyryl-CoA dehydrogenase
Deficiency of butyrylcholine esterase

Deficiency of ferroxidase
Deficiency of galactokinase
Deficiency of glycerate kinase
Deficiency of glycerol kinase
Deficiency of guanidinoacetate methyltransferase
Deficiency of hyaluronoglucosaminidase
Deficiency of hydroxymethylglutaryl-CoA lyase
Deficiency of iodide peroxidase
Deficiency of isobutyryl-CoA dehydrogenase
Deficiency of malonyl-CoA decarboxylase
Deficiency of phosphoserine phosphatase
Deficiency of ribose-5-phosphate isomerase
Deficiency of steroid 11-beta-monooxygenase
Deficiency of steroid 17-alpha-monooxygenase
Deficiency of xanthine oxidase
Dejerine-Sottas disease
Dejerine-Sottas syndrome
Dejerine-sottas neuropathy
Dejerine-sottas syndrome
Delayed gross motor development
Delayed puberty
Delayed reflexes
Delayed speech and language development
Delta-0-thalassemia
Delta-plus-thalassemia
Delta-zero-thalassemia
Dementia
Dementia familial British
Dendritic cell
Dent disease 1
Dent disease 2
Dental enamel pits
Dentinogenesis imperfecta - Shield's type II
Dentinogenesis imperfecta - Shield's type III
Deoxygalactonojirimycin response
Depressed nasal bridge
Dermatitis
Dermatofibrosis lenticularis disseminata
Desbuquois dysplasia 1
Desbuquois dysplasia 2
Desmosterolosis
Developmental delay
Diabetes Mellitus
Diabetes insipidus
Diabetes mellitus
Diabetes mellitus AND insipidus with optic atrophy AND deafness
Diabetes mellitus type 1

Diabetes mellitus type 2
Diabetes-deafness syndrome maternally transmitted
Diamond-Blackfan anemia
Diamond-Blackfan anemia 1
Diamond-Blackfan anemia 10
Diamond-Blackfan anemia 3
Diamond-Blackfan anemia 5
Diamond-Blackfan anemia 7
Diamond-Blackfan anemia 8
Diamond-Blackfan anemia 9
Diaphanospondylodysostosis
Diaphragmatic hernia 3
Diaphyseal dysplasia
Diaphyseal medullary stenosis with malignant fibrous histiocytoma
Diarrhea 3
Diarrhea 4
Diarrhea 5
Diarrhea 6
Diarrhea 7
Diastrophic dysplasia
Dicarboxylic aminoaciduria
Diffuse mesangial sclerosis
Diffuse palmoplantar keratoderma
Digital arthropathy-brachydactyly
Digital clubbing
Digitorenocerebral syndrome
Dihydropteridine reductase deficiency
Dihydropyrimidinase deficiency
Dihydropyrimidine dehydrogenase deficiency
Dilated Cardiomyopathy
Dilated cardiomyopathy
Dilated cardiomyopathy 1A
Dilated cardiomyopathy 1AA
Dilated cardiomyopathy 1BB
Dilated cardiomyopathy 1C
Dilated cardiomyopathy 1CC
Dilated cardiomyopathy 1DD
Dilated cardiomyopathy 1E
Dilated cardiomyopathy 1EE
Dilated cardiomyopathy 1F
Dilated cardiomyopathy 1FF
Dilated cardiomyopathy 1G
Dilated cardiomyopathy 1HH
Dilated cardiomyopathy 1I
Dilated cardiomyopathy 1II
Dilated cardiomyopathy 1JJ
Dilated cardiomyopathy 1KK

Dilated cardiomyopathy 1L
Dilated cardiomyopathy 1LL
Dilated cardiomyopathy 1M
Dilated cardiomyopathy 1N
Dilated cardiomyopathy 1O
Dilated cardiomyopathy 1P
Dilated cardiomyopathy 1R
Dilated cardiomyopathy 1S
Dilated cardiomyopathy 1T
Dilated cardiomyopathy 1V
Dilated cardiomyopathy 1W
Dilated cardiomyopathy 1X
Dilated cardiomyopathy 1Y
Dilated cardiomyopathy 1Z
Dilated cardiomyopathy 3B
Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome
Dimethylglycine dehydrogenase deficiency
Disordered steroidogenesis due to cytochrome p450 oxidoreductase deficiency
Disorders of Intracellular Cobalamin Metabolism
Disproportionate short-limb short stature
Disproportionate short-trunk short stature
Dissecting aortic aneurysm
Disseminated atypical mycobacterial infection
Distal Renal Tubular Acidosis
Distal arthrogryposis
Distal arthrogryposis type 1B
Distal arthrogryposis type 2B
Distal hereditary motor neuronopathy 2D
Distal hereditary motor neuronopathy type 2A
Distal hereditary motor neuronopathy type 2B
Distal hereditary motor neuronopathy type 2C
Distal hereditary motor neuronopathy type 5
Distal hereditary motor neuronopathy type 5B
Distal hereditary motor neuronopathy type 7B
Distal muscle weakness
Distal myopathy
Distal myopathy Markesbery-Griggs type
Distal spinal muscular atrophy
Distichiasis-lymphedema syndrome
Dna topoisomerase II
Dominant hereditary optic atrophy
Donnai Barrow syndrome
Dopamine beta hydroxylase deficiency
Dopamine receptor d2
Double outlet right ventricle
Dowling-Degos disease 1
Dowling-Degos disease 2

Dowling-degos disease 4
Downslanted palpebral fissures
Doyne honeycomb retinal dystrophy
Drash syndrome
Duane syndrome type 2
Duane-radial ray syndrome
Dubin-Johnson syndrome
Duchenne muscular dystrophy
Ductal breast carcinoma
Dursun syndrome
Dyggve-Melchior-Clausen syndrome
Dyschromatosis universalis hereditaria 3
Dyserythropoietic anemia with thrombocytopenia
Dysfibrinogenemia
Dysgnathia complex
Dyskeratosis Congenita
Dyskeratosis congenita
Dyskeratosis congenita X-linked
Dyskeratosis congenita autosomal dominant
Dyskeratosis congenita autosomal recessive 1
Dysmetria
Dysmorphism
Dysplasminogenemia
Dyssegmental Dysplasia
Dystonia
Dystonia 1
Dystonia 10
Dystonia 12
Dystonia 16
Dystonia 2
Dystonia 23
Dystonia 24
Dystonia 25
Dystonia 3
Dystonia 5
Dystonia 6
Dystonia 9
Dystransthyretinemic euthyroidal hyperthyroxinemia
Dystrophic epidermolysis bullosa
EEM syndrome
EHLERS-DANLOS SYNDROME
EMG abnormality
EPIDERMODYSPLASIA VERRUCIFORMIS
EPOXIDE HYDROLASE POLYMORPHISM
ERCC6-Related Disorders
Early Infantile Epileptic Encephalopathy
Early T cell progenitor acute lymphoblastic leukemia

Early infantile epileptic encephalopathy
Early infantile epileptic encephalopathy 10
Early infantile epileptic encephalopathy 11
Early infantile epileptic encephalopathy 13
Early infantile epileptic encephalopathy 14
Early infantile epileptic encephalopathy 16
Early infantile epileptic encephalopathy 17
Early infantile epileptic encephalopathy 18
Early infantile epileptic encephalopathy 2
Early infantile epileptic encephalopathy 21
Early infantile epileptic encephalopathy 4
Early infantile epileptic encephalopathy 5
Early infantile epileptic encephalopathy 7
Early infantile epileptic encephalopathy 8
Early infantile epileptic encephalopathy 9
Early myoclonic encephalopathy
Early-Onset Familial Alzheimer Disease
Ectodermal dysplasia
Ectodermal dysplasia 10a
Ectodermal dysplasia 11a
Ectodermal dysplasia 11b
Ectodermal dysplasia 9
Ectodermal dysplasia skin fragility syndrome
Ectodermal dysplasia-syndactyly syndrome 1
Ectodermal dysplasia/short stature syndrome
Ectopia lentis
Ectrodactyly
Eculizumab
Efavirenz response
Ehlers-Danlos syndrome
Ehlers-Danlos syndrome progeroid type
Ehlers-Danlos-like syndrome due to tenascin-X deficiency
Eichsfeld type congenital muscular dystrophy
Eiken skeletal dysplasia
Elevated alkaline phosphatase
Elevated serum creatine phosphokinase
Elliptocytosis
Elliptocytosis 2
Elliptocytosis 3
Ellis-van Creveld Syndrome
Emery-Dreifuss muscular dystrophy
Emery-Dreifuss muscular dystrophy 3
Emery-Dreifuss muscular dystrophy 4
Emery-Dreifuss muscular dystrophy 5
Emery-Dreifuss muscular dystrophy 6
Emery-Dreifuss muscular dystrophy 7
Enamel-renal syndrome

Encephalopathy
Encephalopathy due to defective mitochondrial and peroxisomal fission 1
Encephalopathy due to defective mitochondrial and peroxisomal fission 2
Endocrine-cerebroosteodysplasia
Endometrial Endometrioid Adenocarcinoma
Endometrial carcinoma
Endometrial neoplasm
Endplate acetylcholinesterase deficiency
Enhanced s-cone syndrome
Enlarged cisterna magna
Enlarged vestibular aqueduct syndrome
Enterokinase deficiency
Eosinophil peroxidase deficiency
Epicanthus
Epidermal nevus
Epidermolysa bullosa simplex and limb girdle muscular dystrophy
Epidermolysis bullosa
Epidermolysis bullosa dystrophica
Epidermolysis bullosa dystrophica inversa
Epidermolysis bullosa herpetiformis
Epidermolysis bullosa junctionalis with pyloric atresia
Epidermolysis bullosa pruriginosa
Epidermolysis bullosa simplex
Epidermolysis bullosa simplex with mottled pigmentation
Epidermolysis bullosa simplex with pyloric atresia
Epidermolytic palmoplantar keratoderma
Epilepsy
Epilepsy juvenile absence
Epilepsy with grand mal seizures on awakening
Epileptic encephalopathy
Epiphyseal chondrodysplasia
Epiphyseal dysplasia
Episodic ataxia
Episodic ataxia type 1
Episodic ataxia type 2
Episodic hemiplegia
Episodic pain syndrome
Erythema
Erythrocyte AMP deaminase deficiency
Erythrocyte lactate transporter defect
Erythrocytosis
Erythroderma
Erythrokeratodermia variabilis
Erythrokeratodermia variabilis et progressiva 2
Erythropoietic protoporphyria
Esophageal cancer
Esophageal squamous cell carcinoma

Essential thrombocythemia
Essential tremor
Estrogen resistance
Ethylmalonic encephalopathy
Exercise intolerance
Exercise intolerance and complex III deficiency
Exercise stress response
Exercise-induced hyperinsulinemic hypoglycemia
Exfoliative ichthyosis
Exocrine pancreatic insufficiency
Exudative vitreoretinopathy
Exudative vitreoretinopathy 1
Exudative vitreoretinopathy 4
Exudative vitreoretinopathy 5
F12-Related Disorders
FACTOR IX
FACTOR IX POLYMORPHISM
FACTOR VII-ACTIVATING PROTEASE MARBURG I POLYMORPHISM
FACTOR VIII (EAST HARTFORD)
FACTOR VIII (OKAYAMA)
FACTOR XII (LOCARNO)
FACTOR XII (TENRI)
FG syndrome
FG syndrome 2
FG syndrome 4
FIBRINOGEN AARHUS 1
FIBRINOGEN ASAHI
FIBRINOGEN BALTIMORE 1
FIBRINOGEN BALTIMORE 3
FIBRINOGEN CANTERBURY
FIBRINOGEN CARACAS 2
FIBRINOGEN CHRISTCHURCH 2
FIBRINOGEN DUSART
FIBRINOGEN HAIFA 1
FIBRINOGEN HILLSBOROUGH
FIBRINOGEN ISE
FIBRINOGEN KEOKUK
FIBRINOGEN KYOTO 2
FIBRINOGEN KYOTO 3
FIBRINOGEN LILLE 1
FIBRINOGEN LONGMONT
FIBRINOGEN MATSUMOTO 1
FIBRINOGEN MILANO 1
FIBRINOGEN NAGOYA 1
FIBRINOGEN NAPLES
FIBRINOGEN NIJMEGEN
FIBRINOGEN PONTOISE 2

FIBRINOGEN TOKYO 2
FOVEAL HYPOPLASIA 2 WITH OPTIC NERVE MISROUTING
FOVEAL HYPOPLASIA 2 WITH OPTIC NERVE MISROUTING AND ANTERIOR SEGMENT DYSGENESIS
FRAXE
FRONTOTEMPORAL DEMENTIA WITH TDP43 INCLUSIONS
FU1/FU2 POLYMORPHISM
Fabry disease
Facioscapulohumeral muscular dystrophy 2
Factor H deficiency
Factor V Hong Kong
Factor V deficiency
Factor VII Marburg I Variant Thrombophilia
Factor VII deficiency
Factor X deficiency
Factor XII deficiency disease
Factor v and factor viii
Factor xiii
Failure of tooth eruption
Failure to thrive
Familial Atypical Mycobacteriosis
Familial Candidiasis
Familial Febrile Seizures
Familial High Density Lipoprotein Deficiency
Familial Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome
Familial Intrahepatic Cholestasis
Familial Isolated Pituitary Adenomas
Familial Mediterranean fever
Familial X-linked hypophosphatemic vitamin D refractory rickets
Familial acne inversa 1
Familial adenomatous polyposis 1
Familial advanced sleep phase syndrome 1
Familial amyloid nephropathy with urticaria AND deafness
Familial amyloid polyneuropathy
Familial aortopathy
Familial atrial fibrillation
Familial benign pemphigus
Familial cancer of breast
Familial cardiomyopathy
Familial chronic mucocutaneous candidiasis
Familial cold autoinflammatory syndrome
Familial cold autoinflammatory syndrome 4
Familial cold urticaria
Familial colorectal cancer
Familial dysautonomia
Familial episodic pain syndrome 1
Familial erythrocytosis
Familial exudative vitreoretinopathy

Familial febrile seizures 8
Familial hemiplegic migraine type 1
Familial hemiplegic migraine type 2
Familial hemiplegic migraine type 3
Familial hemophagocytic lymphohistiocytosis
Familial hyperaldosteronism
Familial hyperaldosteronism type 3
Familial hypercholesterolemia
Familial hyperinsulinism
Familial hyperkalemic periodic paralysis
Familial hypertrophic cardiomyopathy 1
Familial hypertrophic cardiomyopathy 10
Familial hypertrophic cardiomyopathy 11
Familial hypertrophic cardiomyopathy 12
Familial hypertrophic cardiomyopathy 13
Familial hypertrophic cardiomyopathy 14
Familial hypertrophic cardiomyopathy 15
Familial hypertrophic cardiomyopathy 16
Familial hypertrophic cardiomyopathy 17
Familial hypertrophic cardiomyopathy 18
Familial hypertrophic cardiomyopathy 19
Familial hypertrophic cardiomyopathy 2
Familial hypertrophic cardiomyopathy 20
Familial hypertrophic cardiomyopathy 22
Familial hypertrophic cardiomyopathy 23
Familial hypertrophic cardiomyopathy 24
Familial hypertrophic cardiomyopathy 3
Familial hypertrophic cardiomyopathy 4
Familial hypertrophic cardiomyopathy 6
Familial hypertrophic cardiomyopathy 7
Familial hypertrophic cardiomyopathy 8
Familial hypertrophic cardiomyopathy 9
Familial hypoalphalipoproteinemia
Familial hypobetalipoproteinemia
Familial hypocalciuric hypercalcemia
Familial hypokalemia-hypomagnesemia
Familial hypoplastic
Familial infantile myasthenia
Familial juvenile gout
Familial mediterranean fever
Familial medullary thyroid carcinoma
Familial multiple polyposis syndrome
Familial multiple trichoepitheliomata
Familial partial lipodystrophy
Familial partial lipodystrophy 2
Familial partial lipodystrophy 3
Familial partial lipodystrophy 5

Familial periodic paralysis
Familial platelet disorder with associated myeloid malignancy
Familial porphyria cutanea tarda
Familial progressive hyperpigmentation with or without hypopigmentation
Familial pulmonary capillary hemangiomatosis
Familial renal glucosuria
Familial renal hypouricemia
Familial restrictive cardiomyopathy
Familial restrictive cardiomyopathy 1
Familial restrictive cardiomyopathy 3
Familial type 3 hyperlipoproteinemia
Familial type 5 hyperlipoproteinemia
Familial visceral amyloidosis
Fanconi anemia
Fanconi renotubular syndrome 3
Fanconi renotubular syndrome 4 with maturity-onset diabetes of the young
Fanconi-Bickel syndrome
Farber's lipogranulomatosis
Fatal familial insomnia
Favism
Febrile seizures
Fechtner syndrome
Feeding difficulties
Feingold syndrome 1
Femoral bowing
Fetal akinesia sequence
Fetal hemoglobin quantitative trait locus 1
Fetal hemoglobin quantitative trait locus 2
Fetal hemoglobin quantitative trait locus 6
Fibrinogen Milano XII
Fibrochondrogenesis
Fibrosis of extraocular muscles
Fibrous dysplasia of jaw
Fibular hypoplasia and complex brachydactyly
Filippi syndrome
Finnish congenital nephrotic syndrome
Fish-eye disease
Fleck corneal dystrophy
Fleck retina
Flexed deformity
Floating-Harbor syndrome
Fluorouracil response
Focal cortical dysplasia of Taylor
Focal cortical dysplasia of Taylor type 2B
Focal dermal hypoplasia
Focal epilepsy
Focal segmental glomerulosclerosis

Focal segmental glomerulosclerosis 1
Focal segmental glomerulosclerosis 2
Focal segmental glomerulosclerosis 3
Focal segmental glomerulosclerosis 4
Focal segmental glomerulosclerosis 5
Focal segmental glomerulosclerosis 6
Focal segmental glomerulosclerosis 7
Focal segmental glomerulosclerosis 8
Focal segmental glomerulosclerosis 9
Focal segmental glomerulosclerosis and dilated cardiomyopathy
Follicle-stimulating hormone deficiency
Forebrain defects
Foveal hypoplasia 1 with cataract
Foveal hypoplasia 1 with or without anterior segment anomalies
Foveal hypoplasia 2
Foveal hypoplasia 2 and optic nerve misrouting with or without anterior segment dysgenesis
Foveal hypoplasia and presenile cataract syndrome
Fragile X syndrome
Fragile skin
Frank Ter Haar syndrome
Frasier syndrome
Freeman-Sheldon syndrome
Friedreich's ataxia
Frontometaphyseal dysplasia
Frontonasal dysplasia 1
Frontonasal dysplasia 2
Frontonasal dysplasia 3
Frontotemporal Dementia
Frontotemporal dementia
Frontotemporal dementia and/or amyotrophic lateral sclerosis 2
Frontotemporal dementia and/or amyotrophic lateral sclerosis 3
Fructose-biphosphatase deficiency
Fructosuria
Fucosidosis
Fucosyltransferase 6 deficiency
Fuhrmann syndrome
Fukuyama congenital muscular dystrophy
Fumarase deficiency
Fumarylacetoacetase pseudodeficiency
Fundus albipunctatus
G6PD A+
G6PD ALHAMBRA
G6PD ANAHEIM
G6PD ANANT
G6PD ANDALUS
G6PD ASAHI
G6PD AVEIRO

G6PD BETICA
G6PD BEVERLY HILLS
G6PD CAGLIARI
G6PD CASTILLA
G6PD CHATHAM
G6PD COIMBRA
G6PD DHON
G6PD DISTRITO FEDERAL
G6PD GAOHE
G6PD GASTONIA
G6PD GUADALAJARA
G6PD HARILAOU
G6PD IERAPETRA
G6PD ILESHA
G6PD IOWA
G6PD IOWA CITY
G6PD JAMMU
G6PD JAPAN
G6PD KAIPING
G6PD LOMA LINDA
G6PD MAHIDOL
G6PD MAHIDOL-LIKE
G6PD MALAGA
G6PD MARION
G6PD MEDITERRANEAN
G6PD METAPONTO
G6PD MINNESOTA
G6PD MODENA
G6PD NAMORU
G6PD NANKANG
G6PD NASHVILLE
G6PD NEAPOLIS
G6PD ORISSA
G6PD PAWNEE
G6PD PETRICH-LIKE
G6PD PORTICI
G6PD QUING YUAN
G6PD RIVERSIDE
G6PD SANTAMARIA
G6PD SANTIAGO DE CUBA
G6PD SAPPORO-LIKE
G6PD SASSARI
G6PD SEATTLE-LIKE
G6PD SERRES
G6PD SPLIT
G6PD SPRINGFIELD
G6PD TAIWAN-HAKKA 2

G6PD TEPIC
G6PD TOMAH
G6PD VIANGCHAN
G6PD WALTER REED
GALT POLYMORPHISM
GALT POLYMORPHISM (DUARTE
GALT POLYMORPHISM (LOS ANGELES
GATA-1-related thrombocytopenia with dyserythropoiesis
GC1/GC2 POLYMORPHISM
GLUT1 deficiency syndrome 1
GLUT1 deficiency syndrome 2
GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY
GLUTAMIC PYRUVATE TRANSAMINASE POLYMORPHISM
GM1 gangliosidosis
GM1 gangliosidosis type 2
GM1-gangliosidosis
GMP REDUCTASE POLYMORPHISM
GNPTAB-Related Disorders
GRACILE syndrome
GTP cyclohydrolase I deficiency
Gait ataxia
Gait imbalance
Galactose epimerase deficiency
Galactosemia
Galactosialidosis
Galactosylceramide beta-galactosidase deficiency
Galloway-Mowat syndrome
Gamma-aminobutyric acid transaminase deficiency
Gamma-glutamylcysteine synthetase deficiency
Ganglioside sialidase deficiency
Gangliosidosis GM1 type 3
Gardner syndrome
Gastroesophageal reflux
Gastrointestinal stromal tumor
Gaucher disease
Gaucher disease type 3C
Gaucher's disease
Gaze palsy
Geleophysic dysplasia
Geleophysic dysplasia 1
Geleophysic dysplasia 2
Generalized arterial calcification of infancy 2
Generalized dominant dystrophic epidermolysis bullosa
Generalized epilepsy
Generalized epilepsy and paroxysmal dyskinesia
Generalized epilepsy with febrile seizures plus
Generalized epilepsy with febrile seizures plus 3

Generalized epilepsy with febrile seizures plus type 5
Generalized hypotonia
Generalized joint laxity
Genetic prion diseases
Genitopatellar syndrome
Geroderma osteodysplastica
Gerstmann-Straussler-Scheinker syndrome
Ghosal hematodiaphyseal syndrome
Giant axonal neuropathy
Giant axonal neuropathy 2
Gilbert's syndrome
Gingival fibromatosis with hypertrichosis
Gingival overgrowth
Glabellar hemangioma
Glanzmann thrombasthenia
Glaucoma
Glaucoma 1
Glaucoma 3
Glioblastoma
Glioblastoma multiforme
Glioma susceptibility 1
Glioma susceptibility 2
Glioma susceptibility 3
Glipizide poor metabolizer
Global developmental delay
Globozoospermia
Glomerulocystic kidney disease with hyperuricemia and isosthenuria
Glomerulopathy
Glomerulopathy with fibronectin deposits 2
Glucocorticoid deficiency 2
Glucocorticoid deficiency 4 with or without mineralocorticoid deficiency
Glucocorticoid deficiency with achalasia
Glucocorticoid resistance
Glucose 6 phosphate dehydrogenase deficiency
Glucose-6-phosphate transport defect
Glutamine deficiency
Glutaric acidemia
Glutaric acidemia IIA
Glutaric acidemia IIB
Glutaric acidemia IIC
Glutaric acidemia iic
Glutaric aciduria
Glutaryl-CoA oxidase deficiency
Glutathione synthetase deficiency
Glutathione synthetase deficiency of erythrocytes
Gluthathione synthetase deficiency
Glycine N-methyltransferase deficiency

Glycogen content in skeletal muscle
Glycogen storage disease
Glycogen storage disease II
Glycogen storage disease IIIa
Glycogen storage disease IIIb
Glycogen storage disease IV
Glycogen storage disease IXa2
Glycogen storage disease IXb
Glycogen storage disease IXc
Glycogen storage disease IXd
Glycogen storage disease XI
Glycogen storage disease XV
Glycogen storage disease of heart
Glycogen storage disease type 13
Glycogen storage disease type 1A
Glycogen storage disease type II
Glycogen storage disease type III
Glycogen storage disease type IXa1
Glycogen storage disease type X
Glycosylphosphatidylinositol deficiency
Gm2-gangliosidosis
Gnathodiaphyseal dysplasia
Goiter
Goldberg-Shprintzen megacolon syndrome
Goldmann-Favre syndrome
Gonadal dysgenesis with auditory dysfunction
Gonadotropin-independent familial sexual precocity
Gordon Holmes syndrome
Gordon's syndrome
Gorlin syndrome
Gracile bone dysplasia
Granulomatous disease
Granulosa cell tumor of the ovary
Gray platelet syndrome
Grebe syndrome
Greenberg dysplasia
Greig cephalopolysyndactyly syndrome
Griscelli syndrome type 2
Griscelli syndrome type 3
Groenouw corneal dystrophy type I
Growth and mental retardation
Growth delay
Growth hormone deficiency with pituitary anomalies
Growth hormone insensitivity with immunodeficiency
Growth retardation
Gyrate atrophy of choroid and retina with pyridoxine-responsive ornithinemia
HBB-Related Disorders

HEMOGLOBIN A(2) AGRINIO
HEMOGLOBIN A(2) BABINGA
HEMOGLOBIN A(2) CANADA
HEMOGLOBIN A(2) CAPRI
HEMOGLOBIN A(2) COBURG
HEMOGLOBIN A(2) CORFU
HEMOGLOBIN A(2) FITZROY
HEMOGLOBIN A(2) FLATBUSH
HEMOGLOBIN A(2) GROVETOWN
HEMOGLOBIN A(2) LUCANIA
HEMOGLOBIN A(2) MANZANARES
HEMOGLOBIN A(2) MELBOURNE
HEMOGLOBIN A(2) METAPONTO
HEMOGLOBIN A(2) MONREALE
HEMOGLOBIN A(2) NIIGATA
HEMOGLOBIN A(2) NINIVE
HEMOGLOBIN A(2) PELENDRI
HEMOGLOBIN A(2) PUGLIA
HEMOGLOBIN A(2) ROOSEVELT
HEMOGLOBIN A(2) TROODOS
HEMOGLOBIN A(2) WRENS
HEMOGLOBIN A(2) YIALOUSA
HEMOGLOBIN A(2) ZAGREB
HEMOGLOBIN A(2)-PRIME
HEMOGLOBIN ALAMO
HEMOGLOBIN ALESHA
HEMOGLOBIN ALTDORF
HEMOGLOBIN ARLINGTON PARK
HEMOGLOBIN ATTLEBORO
HEMOGLOBIN B(2)
HEMOGLOBIN BARBIZON
HEMOGLOBIN BOLOGNA
HEMOGLOBIN BRESCIA
HEMOGLOBIN BRISTOL
HEMOGLOBIN BURKE
HEMOGLOBIN C
HEMOGLOBIN CAEN
HEMOGLOBIN CALAIS
HEMOGLOBIN CHANDIGARH
HEMOGLOBIN CHAROLLES
HEMOGLOBIN CHICAGO
HEMOGLOBIN CHONGQING
HEMOGLOBIN CLEVELAND
HEMOGLOBIN CLINICO-MADRID
HEMOGLOBIN CONTALDO
HEMOGLOBIN CRETE
HEMOGLOBIN DEBROUSSE

HEMOGLOBIN DUARTE
HEMOGLOBIN DURHAM-N.C.
HEMOGLOBIN ETHIOPIA
HEMOGLOBIN F (CALTECH)
HEMOGLOBIN F (DICKINSON)
HEMOGLOBIN F (IWATA)
HEMOGLOBIN F (JAMAICA)
HEMOGLOBIN F (MACEDONIA II)
HEMOGLOBIN F (ONODA)
HEMOGLOBIN F (PORTO TORRES)
HEMOGLOBIN FLATBUSH (GEORGIA)
HEMOGLOBIN FUKUI
HEMOGLOBIN FUKUOKA
HEMOGLOBIN G (HSI-TSOU)
HEMOGLOBIN G (SAN JOSE)
HEMOGLOBIN G (TAIWAN-AMI)
HEMOGLOBIN GAINESVILLE-GA
HEMOGLOBIN HANAMAKI
HEMOGLOBIN HAZEBROUCK
HEMOGLOBIN HEATHROW
HEMOGLOBIN HOPKINS 1
HEMOGLOBIN INDIANAPOLIS
HEMOGLOBIN J (AUCKLAND)
HEMOGLOBIN J (BALTIMORE)
HEMOGLOBIN J (CORDOBA)
HEMOGLOBIN J (EUROPA)
HEMOGLOBIN J (LENS)
HEMOGLOBIN JAMAICA PLAIN
HEMOGLOBIN JENKINS
HEMOGLOBIN JIANGHUA
HEMOGLOBIN K (IBADAN)
HEMOGLOBIN KENWOOD
HEMOGLOBIN KOCHI
HEMOGLOBIN LIMASSOL
HEMOGLOBIN LOMBARD
HEMOGLOBIN LUFKIN
HEMOGLOBIN MACHIDA
HEMOGLOBIN MALAY
HEMOGLOBIN MASUDA
HEMOGLOBIN MATERA
HEMOGLOBIN MCKEES ROCKS
HEMOGLOBIN MOLFETTA
HEMOGLOBIN MONT SAINT-AIGNAN
HEMOGLOBIN N (BALTIMORE)
HEMOGLOBIN N (JENKINS)
HEMOGLOBIN NATAL
HEMOGLOBIN OEGSTGEEST

HEMOGLOBIN OKAZAKI
HEMOGLOBIN OLMSTED
HEMOGLOBIN PHILLY
HEMOGLOBIN QUIN-HAI
HEMOGLOBIN RAINIER
HEMOGLOBIN RENERT
HEMOGLOBIN RIVERDALE-BRONX
HEMOGLOBIN ROCKFORD
HEMOGLOBIN ROSEAU-POINTE A PITRE
HEMOGLOBIN ROUEN
HEMOGLOBIN SAALE
HEMOGLOBIN SAINT JACQUES
HEMOGLOBIN SHOWA-YAKUSHIJI
HEMOGLOBIN SILVER SPRINGS
HEMOGLOBIN SOUTH MILWAUKEE
HEMOGLOBIN STANMORE
HEMOGLOBIN TIANSHUI
HEMOGLOBIN TIZI-OUZOU
HEMOGLOBIN TOKONAME
HEMOGLOBIN TSUKUMI
HEMOGLOBIN TUNIS
HEMOGLOBIN VALLETTA
HEMOGLOBIN WIEN
HEMOGLOBIN WINDSOR
HEMOGLOBIN YATSUSHIRO
HEMOGLOBIN ZENGCHENG
HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN
HEXA
HEXB POLYMORPHISM
HEXOSAMINIDASE B
HFE POLYMORPHISM
HIS2*1/HIS2*2 POLYMORPHISM
HMG CoA reductase inhibitors response - Efficacy
HNSHA due to aldolase A deficiency
HPRT ANN ARBOR
HPRT ARLINGTON
HPRT ASHVILLE
HPRT DETROIT
HPRT EDINBURGH
HPRT LONDON
HPRT MIDLAND
HPRT MILWAUKEE
HPRT MONTREAL
HPRT MOOSE JAW
HPRT MUNICH
HPRT NEW BRITON
HPRT NEW HAVEN

HPRT PARIS
HPRT SWAN
HPRT TOKYO
HPRT TOOWONG
HPRT URANGAN
HPRT YALE
HYPERHOMOCYSTEINEMIA
HYPOGONADOTROPIC HYPOGONADISM 2 WITH ANOSMIA
Haemorrhagic telangiectasia 1
Haemorrhagic telangiectasia 2
Haim-Munk syndrome
Hair morphology 1
Hajdu-Cheney syndrome
Hamartoma
Hand foot uterus syndrome
Harderoporphyria
Hartsfield syndrome
Hashimoto thyroiditis
Hay-Wells syndrome of ectodermal dysplasia
Hb SS disease
Head and Neck Neoplasms
Headache
Hearing impairment
Heart
Heart failure
Heart-hand syndrome
Hecht syndrome
Heinz body anemia
Hemangioblastoma
Hemangioma
Hematologic neoplasm
Hemihypertrophy
Hemiparesis
Hemiplegia
Hemivertebrae
Hemochromatosis
Hemochromatosis type 1
Hemochromatosis type 2A
Hemochromatosis type 2B
Hemochromatosis type 3
Hemochromatosis type 4
Hemoglobin H disease
Hemoglobinopathy
Hemolytic anemia
Hemolytic anemia due to hexokinase deficiency
Hemolytic uremic syndrome
Hemophagocytic lymphohistiocytosis

Hemophilia B
Hemophilia b(m)
Hemorrhagic destruction of the brain
Hemorrhagic disease due to alpha-1-antitrypsin Pittsburgh mutation
Hemosiderosis
Hennekam lymphangiectasia-lymphedema syndrome
Hennekam lymphangiectasia-lymphedema syndrome 2
Heparin cofactor II deficiency
Hepatic lipase deficiency
Hepatic methionine adenosyltransferase deficiency
Hepatic venoocclusive disease with immunodeficiency
Hepatitis C virus infection
Hepatitis b virus
Hepatoblastoma
Hepatocellular carcinoma
Hepatoerythropoietic porphyria
Hepatomegaly
Hereditary Angioedema
Hereditary C1 esterase inhibitor deficiency - dysfunctional factor
Hereditary Paraganglioma-Pheochromocytoma Syndromes
Hereditary Sideroblastic Anemia with Myopathy and Lactic Acidosis
Hereditary acrodermatitis enteropathica
Hereditary angioedema type 1
Hereditary breast and ovarian cancer syndrome
Hereditary cancer-predisposing syndrome
Hereditary cerebral amyloid angiopathy
Hereditary congenital facial paresis 3
Hereditary coproporphyria
Hereditary cutaneous melanoma
Hereditary diffuse gastric cancer
Hereditary diffuse leukoencephalopathy with spheroids
Hereditary factor II deficiency disease
Hereditary factor IX deficiency disease
Hereditary factor VIII deficiency disease
Hereditary factor XI deficiency disease
Hereditary fructosuria
Hereditary hemochromatosis
Hereditary hemorrhagic telangiectasia type 2
Hereditary insensitivity to pain with anhidrosis
Hereditary leiomyomatosis and renal cell cancer
Hereditary liability to pressure palsies
Hereditary lymphedema type I
Hereditary motor and sensory neuropathy
Hereditary motor and sensory neuropathy with optic atrophy
Hereditary myopathy with early respiratory failure
Hereditary nephrotic syndrome
Hereditary nonpolyposis colorectal cancer type 4

Hereditary nonpolyposis colorectal cancer type 5
Hereditary nonpolyposis colorectal cancer type 6
Hereditary nonpolyposis colorectal cancer type 7
Hereditary pancreatitis
Hereditary pyropoikilocytosis
Hereditary sensory and autonomic neuropathy type II
Hereditary sensory and autonomic neuropathy type IIA
Hereditary sensory and autonomic neuropathy type IIB
Hereditary sensory neuropathy type 1D
Hereditary sensory neuropathy type IE
Hereditary sensory neuropathy type IF
Hereditary sideroblastic anemia
Hermansky Pudlak syndrome 2
Hermansky-Pudlak syndrome
Hermansky-Pudlak syndrome 1
Hermansky-Pudlak syndrome 3
Hermansky-Pudlak syndrome 4
Hermansky-Pudlak syndrome 5
Hermansky-Pudlak syndrome 6
Hermansky-Pudlak syndrome 7
Hermansky-Pudlak syndrome 9
Herpes simplex encephalitis 2
Heterochromia iridis
Heterotaxy
Heterotopia
Hidrotic ectodermal dysplasia syndrome
High CSF lactic acid
High anterior hairline
High bone mass
High density lipoprotein cholesterol level quantitative trait locus 10
High density lipoprotein cholesterol level quantitative trait locus 12
High density lipoprotein cholesterol level quantitative trait locus 13
High forehead
Hirschsprung Disease
Hirschsprung disease
Hirschsprung disease 1
Hirschsprung disease 2
Hirschsprung disease 3
Hirschsprung disease 4
Hirschsprung disease ganglioneuroblastoma
Histidinemia
Histiocytic medullary reticulosis
Histiocytosis-lymphadenopathy plus syndrome
Hodgkin lymphoma
Holocarboxylase synthetase deficiency
Holoprosencephaly
Holoprosencephaly 11

Holoprosencephaly 2
Holoprosencephaly 3
Holoprosencephaly 4
Holoprosencephaly 7
Holoprosencephaly 9
Holoprosencephaly sequence
Holt-Oram syndrome
Homocysteinemia due to MTHFR deficiency
Homocystinuria
Homocystinuria due to CBS deficiency
Homocystinuria due to MTHFR deficiency
Homocystinuria-Megaloblastic anemia due to defect in cobalamin metabolism
Horseshoe kidney
Howel-Evans syndrome
Hoyeraal Hreidarsson syndrome
Human immunodeficiency virus type 1
Hurler syndrome
Hurthle cell carcinoma of thyroid
Hutchinson-Gilford progeria syndrome
Hutchinson-Gilford syndrome
Hyaline fibromatosis syndrome
Hydatidiform mole
Hydranencephaly
Hydranencephaly with abnormal genitalia
Hydrocephalus
Hydrolethalus syndrome
Hydrolethalus syndrome 1
Hydrops
Hydroxykynureninuria
Hyper-IgE syndrome
Hyperactive airways
Hyperaldosteronism
Hyperalphalipoproteinemia
Hyperalphalipoproteinemia 2
Hyperammonaemia
Hyperammonemia
Hyperapobetalipoproteinemia
Hyperbilirubinemia
Hyperbilirubinemia transient familial neonatal
Hyperbiliverdinemia
Hypercalciuria
Hypercalciuric hypercalcemia
Hypercarotenemia and vitamin a deficiency
Hyperchlorhidrosis
Hypercholanemia
Hypercholesterolaemia
Hypercholesterolemia

Hypercholesterolemia and hypertriglyceridemia
Hyperekplexia
Hyperekplexia 2
Hyperekplexia 3
Hyperekplexia hereditary
Hypereosinophilic syndrome
Hyperferritinemia cataract syndrome
Hyperglycinuria
Hypergonadotropic hypogonadism
Hyperimmunoglobulin D with periodic fever
Hyperimmunoglobulin E recurrent infection syndrome
Hyperimmunoglobulin E syndrome
Hyperinsulinemia
Hyperinsulinemic hypoglycemia
Hyperinsulinemic hypoglycemia familial 3
Hyperinsulinemic hypoglycemia familial 5
Hyperinsulinism
Hyperinsulinism-hyperammonemia syndrome
Hyperkalemic Periodic Paralysis Type 1
Hyperlipidemia
Hyperlipoproteinemia
Hyperlysinemia
Hypermanganesemia with dystonia 1
Hypermethioninemia
Hypermethioninemia due to adenosine kinase deficiency
Hypermethioninemia with s-adenosylhomocysteine hydrolase deficiency
Hypermetropia
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome
Hyperparathyroidism 1
Hyperparathyroidism 2
Hyperphenylalaninemia
Hyperphosphatasemia with bone disease
Hyperphosphatasia with mental retardation syndrome 1
Hyperphosphatasia with mental retardation syndrome 2
Hyperphosphatasia with mental retardation syndrome 3
Hyperphosphatasia with mental retardation syndrome 4
Hyperphosphatasia with mental retardation syndrome 5
Hyperpigmentation of the skin
Hyperproinsulinemia
Hyperproreninemia
Hyperreflexia
Hypertelorism
Hypertension
Hyperthyroidism
Hyperthyroxinemia
Hypertrichosis
Hypertrichotic osteochondrodysplasia

Hypertriglyceridemia
Hypertrophic cardiomyopathy
Hypertrypsinemia
Hypertyrosinemia
Hyperuricemia
Hyperuricemic nephropathy
Hyphidrosis
Hypoadiponectinemia
Hypoagammaglobulinemia
Hypobetalipoproteinemia
Hypocalcemia
Hypocalciuric hypercalcemia
Hypoceruloplasminemia
Hypochondrogenesis
Hypochondroplasia
Hypochromic microcytic anemia with iron overload
Hypochromic microcytic anemia with iron overload 2
Hypodysfibrinogenemia
Hypoglycemia
Hypoglycemia with deficiency of glycogen synthetase in the liver
Hypogonadism
Hypogonadism with anosmia
Hypogonadotrophic hypogonadism
Hypogonadotropic hypogonadism 10 without anosmia
Hypogonadotropic hypogonadism 11 with or without anosmia
Hypogonadotropic hypogonadism 13 with or without anosmia
Hypogonadotropic hypogonadism 14 with or without anosmia
Hypogonadotropic hypogonadism 17 with or without anosmia
Hypogonadotropic hypogonadism 18 with anosmia
Hypogonadotropic hypogonadism 19 with or without anosmia
Hypogonadotropic hypogonadism 2 with anosmia
Hypogonadotropic hypogonadism 20 with or without anosmia
Hypogonadotropic hypogonadism 20 without anosmia
Hypogonadotropic hypogonadism 21 with or without anosmia
Hypogonadotropic hypogonadism 22 with anosmia
Hypogonadotropic hypogonadism 7 with or without anosmia
Hypogonadotropic hypogonadism 8 without anosmia
Hypogonadotropic hypogonadism 9 with or without anosmia
Hypohidrotic Ectodermal Dysplasia
Hypohidrotic X-linked ectodermal dysplasia
Hypohidrotic ectodermal dysplasia
Hypoinsulinemic hypoglycemia and hemihypertrophy
Hypokalemic periodic paralysis
Hypokalemic periodic paralysis 1
Hypomagnesemia 1
Hypomagnesemia 2
Hypomagnesemia 4

Hypomagnesemia 5
Hypomagnesemia 6
Hypomyelinating leukodystrophy 7
Hypomyelinating leukodystrophy 8
Hypomyelination
Hypomyelination and Congenital Cataract
Hypomyelination with brainstem and spinal cord involvement and leg spasticity
Hypoparathyroidism familial isolated
Hypoparathyroidism retardation dysmorphism syndrome
Hypophosphatasia
Hypophosphatemic Nephrolithiasis/Osteoporosis
Hypophosphatemic Rickets
Hypophosphatemic rickets
Hypoplasia of the corpus callosum
Hypoplastic enamel-onycholysis-hypohidrosis syndrome
Hypoplastic left heart syndrome
Hypoplastic left heart syndrome 2
Hypoprebetalipoproteinemia
Hypoproteinemia
Hypospadias 1
Hypospadias 2
Hypothyroidism
Hypotonia
Hypotrichosis 12
Hypotrichosis 2
Hypotrichosis 3
Hypotrichosis 4
Hypotrichosis 6
Hypotrichosis 8
Hypotrichosis and recurrent skin vesicles
Hypotrichosis-lymphedema-telangiectasia syndrome
Hystrix-like ichthyosis with deafness
I blood group system
I cell disease
IFAP syndrome with or without BRESHECK syndrome
IL21R immunodeficiency
INDIAN BLOOD GROUP SYSTEM POLYMORPHISM
INFLAMMATORY BOWEL DISEASE 1 (CROHN DISEASE)
IRAK4 deficiency
Ichthyosis
Ichthyosis bullosa of Siemens
Ichthyosis exfoliativa
Ichthyosis prematurity syndrome
Ichthyosis vulgaris
Idiopathic basal ganglia calcification 1
Idiopathic basal ganglia calcification 5
Idiopathic fibrosing alveolitis

Idiopathic hypercalcemia of infancy
Idiopathic livedo reticularis with systemic involvement
IgA nephropathy
Iminoglycinuria
Immune dysfunction with T-cell inactivation due to calcium entry defect 1
Immune dysfunction with T-cell inactivation due to calcium entry defect 2
Immunodeficiency
Immunodeficiency 12
Immunodeficiency 14
Immunodeficiency 16
Immunodeficiency 17
Immunodeficiency 18
Immunodeficiency 19
Immunodeficiency 22
Immunodeficiency 23
Immunodeficiency 24
Immunodeficiency 26 with or without neurologic abnormalities
Immunodeficiency 26 without neurologic abnormalities
Immunodeficiency 28
Immunodeficiency 30
Immunodeficiency 31C
Immunodeficiency 31a
Immunodeficiency 32a
Immunodeficiency 32b
Immunodeficiency 37
Immunodeficiency 39
Immunodeficiency 8
Immunodeficiency due to defect in cd3-zeta
Immunodeficiency due to ficolin 3 deficiency
Immunodeficiency with hyper IgM type 1
Immunodeficiency with hyper IgM type 2
Immunodeficiency with hyper IgM type 3
Immunodeficiency with hyper IgM type 5
Immunodeficiency-centromeric instability-facial anomalies syndrome 2
Immunoglobulin A deficiency 2
Impdh2 enzyme activity
Inability to walk by childhood/adolescence
Inborn genetic diseases
Inclusion body myopathy 2
Inclusion body myopathy 3
Inclusion body myopathy with early-onset paget disease and frontotemporal dementia
Inclusion body myopathy with early-onset paget disease with or without frontotemporal dementia 2
Incontinentia pigmenti syndrome
Increased analgesia from kappa-opioid receptor agonist
Increased body weight
Increased left ventricular wall thickness
Indifference to pain

Infantile GM1 gangliosidosis
Infantile Parkinsonism-dystonia
Infantile convulsions and paroxysmal choreoathetosis
Infantile cortical hyperostosis
Infantile epilepsy
Infantile hypercalcemia
Infantile hypophosphatasia
Infantile liver failure syndrome 1
Infantile myofibromatosis
Infantile myofibromatosis 1
Infantile myofibromatosis 2
Infantile nephronophthisis
Infantile neuroaxonal dystrophy
Infantile nystagmus
Infantile onset
Infantile spasms
Infantile-onset ascending hereditary spastic paralysis
Infections
Infertility
Infertility associated with multi-tailed spermatozoa and excessive DNA
Inflammatory bowel disease 19
Inflammatory bowel disease 25
Inflammatory bowel disease 28
Inflammatory skin and bowel disease
Inherited Erythromelalgia
Inosine triphosphatase deficiency
Insomnia
Insulin resistance
Insulin-Like Growth Factor I Resistance
Insulin-dependent diabetes mellitus secretory diarrhea syndrome
Insulin-like growth factor 1 resistance to
Insulin-like growth factor I deficiency
Insulin-resistant diabetes mellitus AND acanthosis nigricans
Intellectual Disability
Intellectual disability
Interferon gamma receptor deficiency
Interleukin 1 receptor antagonist deficiency
Interleukin 2 receptor
Intermediate maple syrup urine disease type 2
Intermediate muscular dystrophy
Intermittent diarrhea
Interrupted aortic arch
Interstitial lung and liver disease
Interstitial nephritis
Intervertebral disc disease
Intimal medial thickness of internal carotid artery
Intractable seizure

Intrauterine growth retardation
Intrinsic factor deficiency
Invasive pneumococcal disease
Iodotyrosine deiodination defect
Iodotyrosyl coupling defect
Irido-corneo-trabecular dysgenesis
Iridogoniodysgenesis
Iridogoniodysgenesis type1
Iron accumulation in brain
Iron deficiency anemia
Iron deposition in globus pallidus
Ischemic stroke
Ischiopatellar dysplasia
Islet cell hyperplasia
Isolated 17
Isolated GnRH Deficiency
Isolated growth hormone deficiency type 1B
Isolated lutropin deficiency
Isovaleric acidemia
Isovaleryl-CoA dehydrogenase deficiency
Ivacaftor response
JP and JP/HHT
Jackson-Weiss syndrome
Jakob-Creutzfeldt disease
Jankovic Rivera syndrome
Jaundice
Jervell and Lange-Nielsen syndrome
Jervell and Lange-Nielsen syndrome 1
Jervell and Lange-Nielsen syndrome 2
Jeune thoracic dystrophy
Jk-null variant
Johanson-Blizzard syndrome
John Milton Hagen blood group system
Joint hypermobility
Joubert syndrome
Joubert syndrome 1
Joubert syndrome 12/15
Joubert syndrome 13
Joubert syndrome 14
Joubert syndrome 16
Joubert syndrome 17
Joubert syndrome 20
Joubert syndrome 21
Joubert syndrome 22
Joubert syndrome 3
Joubert syndrome 5
Joubert syndrome 6

Joubert syndrome 7
Joubert syndrome 8
Joubert syndrome 9
Joubert syndrome 9/15
Junctional epidermolysis bullosa gravis of Herlitz
Juvenile Polyposis
Juvenile macular degeneration and hypotrichosis
Juvenile myelomonocytic leukemia
Juvenile myoclonic epilepsy
Juvenile myopathy
Juvenile nephropathic cystinosis
Juvenile neuronal ceroid lipofuscinosis
Juvenile polyposis syndrome
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome
Juvenile primary lateral sclerosis
Juvenile retinoschisis
Juvenile-onset dystonia
KCNE2-Related Disorders
KCNQ1-related acquired long QT syndrome
KCNQ2-Related Disorders
KIDD BLOOD POLYMORPHISM Jk(a)/Jk(b)
Kabuki syndrome 1
Kabuki syndrome 2
Kallikrein
Kallmann syndrome 1
Kallmann syndrome 2
Kallmann syndrome 3
Kallmann syndrome 4
Kallmann syndrome 5
Kallmann syndrome 6
Kanzaki disease
Karak syndrome
Kartagener syndrome
Kaufman oculocerebrofacial syndrome
Kawasaki disease
Kearns Sayre syndrome
Kel6 antigen
Kenny-Caffey syndrome type 2
Keratitis
Keratitis-Ichthyosis-Deafness Syndrome
Keratitis-ichthyosis-deafness syndrome
Keratoconus 1
Keratoderma palmoplantar deafness
Keratosis
Keratosis follicularis
Keratosis palmoplantaris striata 1
Keratosis palmoplantaris striata II

Keratosis pilaris decalvans
Keutel syndrome
Kidney Carcinoma
Kindler's syndrome
King Denborough syndrome
Kininogen deficiency
Klein-Waardenberg's syndrome
Klippel Feil syndrome
Klippel-Feil syndrome 1
Klippel-Feil syndrome 2
Klippel-Feil syndrome 3
Kniest dysplasia
Knobloch syndrome 1
Knuckle pads
Kohlschutter's syndrome
Koolen-de Vries syndrome
Kowarski syndrome
L-2-hydroxyglutaric aciduria
L-ferritin deficiency
LEOPARD syndrome 1
LEOPARD syndrome 2
LEOPARD syndrome 3
LEPTIN RECEPTOR POLYMORPHISM
LIPOPROTEIN LIPASE (OLBIA)
LIPOPROTEIN LIPASE POLYMORPHISM
LUNG CANCER
Lactase persistence
Lactate dehydrogenase B deficiency
Lafora disease
Laing distal myopathy
Lamellar ichthyosis
Laminopathy
Landsteiner-Wiener phenotype
Langer mesomelic dysplasia syndrome
Langereis blood group
Laron syndrome with elevated serum GH-binding protein
Laron syndrome with undetectable serum GH-binding protein
Laron-type isolated somatotropin defect
Larsen syndrome
Late-onset retinal degeneration
Lathosterolosis
Lattice corneal dystrophy Type I
Lattice corneal dystrophy Type III
Lattice corneal dystrophy type 3A
Lchad deficiency with maternal acute fatty liver of pregnancy
Leanness
Leber congenital amaurosis

Leber congenital amaurosis 1
Leber congenital amaurosis 10
Leber congenital amaurosis 11
Leber congenital amaurosis 12
Leber congenital amaurosis 13
Leber congenital amaurosis 15
Leber congenital amaurosis 16
Leber congenital amaurosis 17
Leber congenital amaurosis 18
Leber congenital amaurosis 2
Leber congenital amaurosis 3
Leber congenital amaurosis 4
Leber congenital amaurosis 5
Leber congenital amaurosis 6
Leber congenital amaurosis 7
Leber congenital amaurosis 8
Leber congenital amaurosis 9
Leber hereditary optic neuropathy with dystonia
Leber's optic atrophy
Left ventricular noncompaction 1
Left ventricular noncompaction 10
Left ventricular noncompaction 3
Left ventricular noncompaction 4
Left ventricular noncompaction 5
Left ventricular noncompaction 6
Left ventricular noncompaction 7
Left ventricular noncompaction 8
Left ventricular noncompaction 9
Left ventricular noncompaction cardiomyopathy
Left-right axis malformations
Legionellosis
Legius syndrome
Leigh syndrome
Leigh syndrome due to mitochondrial complex I deficiency
Leigh syndrome due to mitochondrial complex III deficiency
Leiner disease
Lenz microphthalmia syndrome
Lenz-Majewski hyperostosis syndrome
Leprechaunism syndrome
Leptin deficiency or dysfunction
Leptin receptor deficiency
Leri Weill dyschondrosteosis
Lesch-Nyhan syndrome
Lesch-nyhan syndrome
Lethal arthrogryposis with anterior horn cell disease
Lethal congenital contractural syndrome 3
Lethal congenital contracture syndrome 1

Lethal congenital contracture syndrome 4
Lethal congenital contracture syndrome 6
Lethal multiple pterygium syndrome
Lethal short-limbed short stature
Lethal tight skin contracture syndrome
Leukemia
Leukocyte adhesion deficiency
Leukocyte adhesion deficiency type 1
Leukodystrophy
Leukoencephalopathy
Leukoencephalopathy with Brainstem and Spinal Cord Involvement and Lactate Elevation
Leukoencephalopathy with ataxia
Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation
Leukoencephalopathy with vanishing white matter
Leukonychia totalis
Leukopenia
Levy-Hollister syndrome
Lewy body dementia
Leydig cell agenesis
Leydig cell hypoplasia
Leydig hypoplasia
Lhermitte-Duclos disease
Li-Fraumeni syndrome
Li-Fraumeni syndrome 1
Li-Fraumeni syndrome 2
Li-Fraumeni-like syndrome
Lig4 syndrome
Limb-Girdle Muscular Dystrophy
Limb-girdle muscle weakness
Limb-girdle muscular dystrophy
Limb-girdle muscular dystrophy-dystroglycanopathy
Linear skin defects with multiple congenital anomalies 1
Linear skin defects with multiple congenital anomalies 2
Lipase deficiency combined
Lipid proteinosis
Lipoatrophy with Diabetes
Lipodystrophy
Lipoprotein glomerulopathy
Lipoprotein(a) deficiency
Lissencephaly
Lissencephaly 1
Lissencephaly 2
Lissencephaly 2 (Norman-Roberts type)
Lissencephaly 3
Lissencephaly 4
Lissencephaly 5
Lissencephaly 6

Liver failure acute infantile
Lobar holoprosencephaly
Loeys-Dietz syndrome
Loeys-Dietz syndrome 1
Loeys-Dietz syndrome 2
Loeys-Dietz syndrome 3
Loeys-Dietz syndrome 4
Loeys-Dietz syndrome 5
Long QT syndrome
Long QT syndrome 1
Long QT syndrome 10
Long QT syndrome 11
Long QT syndrome 12
Long QT syndrome 13
Long QT syndrome 14
Long QT syndrome 15
Long QT syndrome 2
Long QT syndrome 2/3
Long QT syndrome 3
Long QT syndrome 4
Long QT syndrome 5
Long QT syndrome 6
Long QT syndrome 9
Long qt syndrome 3
Long qt syndrome 3/6
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
Low density lipoprotein cholesterol level quantitative trait locus 1
Low-set
Low-set ears
Lowe syndrome
Lower limb amyotrophy
Lower limb muscle weakness
Lower limb undergrowth
Lumbosacral myelomeningocele
Lung cancer
Lung cancer susceptibility 2
Lupus nephritis
Luteinizing hormone resistance
Lymphangiomyomatosis
Lymphedema
Lymphoblastic leukemia
Lymphoma
Lymphoproliferative syndrome 1
Lymphoproliferative syndrome 2
Lynch syndrome
Lynch syndrome I
Lynch syndrome II

Lysinuric protein intolerance
Lysosomal acid lipase deficiency
MACROTHROMBOCYTOPENIA
MACULAR DEGENERATION
MAPLE SYRUP URINE DISEASE
MAPT-Related Spectrum Disorders
MASP2 deficiency
MASS syndrome
MEN2 phenotype: Unclassified
MEN2 phenotype: Unknown
MEN2A and FMTC
MEN2A and Unclassified
MEND syndrome
MERFF syndrome
MERRF/MELAS overlap syndrome
METHYLCOBALAMIN DEFICIENCY
METHYLMALONIC ACIDURIA
METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA
MICROCEPHALY
MLH3-Related Lynch Syndrome
MORM syndrome
MPV17-Related Disorders
MSH2 POLYMORPHISM
MTHFR deficiency
MULTIPLE ENDOCRINE NEOPLASIA
MULTIPLE JOINT DISLOCATIONS
MYBPC3-Related Disorders
MYH-associated polyposis
MYH9 related disorders
MYH9-related disorder
MYO7A-Related Disorders
MYOC-Related Disorders
MYOPATHY
Macrocephaly
Macrocephaly/autism syndrome
Macroglobulinemia
Macular corneal dystrophy
Macular corneal dystrophy Type I
Macular degeneration
Macular dystrophy
Macular dystrophy with central cone involvement
Majeed syndrome
Malaria
Malattia leventinese
Male infertility
Malformation of the heart and great vessels
Malignant Melanoma Susceptibility

Malignant hyperthermia
Malignant hyperthermia susceptibility
Malignant hyperthermia susceptibility type 5
Malignant hypothermia
Malignant lymphoma
Malignant melanoma
Malignant melanoma of skin
Malignant neoplasm of body of uterus
Malignant tumor of esophagus
Malignant tumor of floor of mouth
Malignant tumor of prostate
Malignant tumor of testis
Malignant tumor of urinary bladder
Mandibuloacral dysostosis
Mandibuloacral dysplasia
Mandibuloacral dysplasia with type A lipodystrophy
Mandibuloacral dysplasia with type B lipodystrophy
Mandibulofacial dysostosis
Mandibulofacial dysostosis with mental deficiency
Mannose-binding protein deficiency
Mantle cell lymphoma
Maple syrup urine disease
Maple syrup urine disease type 1A
Maple syrup urine disease type 2
Marden Walker like syndrome
Marden-Walker syndrome
Marfan lipodystrophy syndrome
Marfan syndrome
Marfanoid habitus
Marinesco-Sjögren syndrome
Marked Hypotonia
Marles Greenberg Persaud syndrome
Marshall syndrome
Marshall/Stickler syndrome
Martsolf syndrome
Mast cell disease
Mast cell leukemia
Mastocytosis
Mastocytosis with associated hematologic disorder
Maturity-onset diabetes of the young
May-Hegglin anomaly
McArdle disease
McCune-Albright syndrome
McKusick Kaufman syndrome
McLeod neuroacanthocytosis syndrome
Meacham syndrome
Meckel syndrome

Meckel syndrome type 1
Meckel syndrome type 3
Meckel syndrome type 4
Meckel syndrome type 5
Meckel syndrome type 6
Meckel syndrome type 7
Meckel syndrome type 8
Meckel-Gruber syndrome
Meconium ileus
Medium-chain acyl-coenzyme A dehydrogenase deficiency
Medullary cystic kidney disease 2
Medullary thyroid carcinoma
Medulloblastoma
Meesman's corneal dystrophy
Megacystis
Megalencephalic leukoencephalopathy with subcortical cysts
Megalencephalic leukoencephalopathy with subcortical cysts 1
Megalencephalic leukoencephalopathy with subcortical cysts 2a
Megalencephalic leukoencephalopathy with subcortical cysts 2b
Megalencephaly cutis marmorata telangiectatica congenita
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2
Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3
Megaloblastic anemia
Megaloblastic anemia 1
Megaloblastic anemia due to dihydrofolate reductase deficiency
Megaloblastic anemia due to inborn errors of metabolism
Megalocornea
Meier-Gorlin syndrome
Meier-Gorlin syndrome 1
Meier-Gorlin syndrome 2
Meier-Gorlin syndrome 3
Meier-Gorlin syndrome 4
Meier-Gorlin syndrome 5
Melanoma
Melanoma-pancreatic cancer syndrome
Melioidosis
Melnick-Fraser syndrome
Melnick-Needles syndrome
Memory impairment
Memory quantitative trait locus
Meningeal Neoplasms
Meningioma
Menkes kinky-hair syndrome
Mental deterioration
Mental retardation
Mental retardation 21

Mental retardation 3
Mental retardation 30
Mental retardation 49
Mental retardation 58
Mental retardation 63
Mental retardation X-linked with cerebellar hypoplasia and distinctive facial appearance
Mental retardation and microcephaly with pontine and cerebellar hypoplasia
Mental retardation with language impairment and with or without autistic features
Mental retardation-hypotonic facies syndrome
Mental retardation-hypotonic facies syndrome X-linked
Mephenytoin
Merkel cell carcinoma
Merosin deficient congenital muscular dystrophy
Mesangiocapillary glomerulonephritis
Metabolic syndrome
Metacarpal 4-5 fusion
Metachondromatosis
Metachromatic leukodystrophy
Metaphyseal anadysplasia
Metaphyseal anadysplasia 1
Metaphyseal anadysplasia 2
Metaphyseal chondrodysplasia
Metaphyseal dysplasia without hypotrichosis
Metatrophic dysplasia
Methemoglobinemia
Methemoglobinemia type 2
Methylmalonate semialdehyde dehydrogenase deficiency
Methylmalonic acidemia
Methylmalonic acidemia with homocystinuria
Methylmalonic acidemia with homocystinuria cblD
Methylmalonic aciduria
Methylmalonic aciduria cblA type
Methylmalonic aciduria cblB type
Methylmalonic aciduria due to methylmalonyl-CoA mutase deficiency
Methylmalonyl-CoA epimerase deficiency
Mevalonic aciduria
Michels syndrome
Microcephalic osteodysplastic primordial dwarfism type 2
Microcephaly
Microcephaly and chorioretinopathy
Microcephaly with chorioretinopathy
Microcephaly with mental retardation and digital anomalies
Microcephaly with or without chorioretinopathy
Microcephaly-capillary malformation syndrome
Microcornea
Microcytic anemia
Micrognathia

Micropenis
Microphthalmia
Microphthalmia syndromic 3
Microphthalmia syndromic 5
Microphthalmia syndromic 6
Microphthalmia syndromic 9
Microphthalmia/coloboma and skeletal dysplasia syndrome
Microspherophakia
Microtia
Microvascular complications of diabetes 7
Migraine
Mild non-PKU hyperphenylalanemia
Mild proteinuria
Miller syndrome
Mineralocorticoid deficiency
Minicore myopathy with external ophthalmoplegia
Mirror movements 3
Missing ribs
Mitchell-Riley syndrome
Mitochondrial DNA depletion syndrome
Mitochondrial DNA depletion syndrome 1 (MNGIE type)
Mitochondrial DNA depletion syndrome 11
Mitochondrial DNA depletion syndrome 12b (cardiomyopathic type)
Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type)
Mitochondrial DNA depletion syndrome 2
Mitochondrial DNA depletion syndrome 4B
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)
Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)
Mitochondrial DNA depletion syndrome 8B (MNGIE type)
Mitochondrial DNA depletion syndrome 9 (encephalomyopathic with methylmalonic aciduria)
Mitochondrial DNA-depletion syndrome 3
Mitochondrial complex I deficiency
Mitochondrial complex II deficiency
Mitochondrial complex III deficiency
Mitochondrial diseases
Mitochondrial encephalomyopathy
Mitochondrial myopathy
Mitochondrial myopathy and sideroblastic anemia
Mitochondrial phosphate carrier deficiency
Mitochondrial pyruvate carrier deficiency
Mitochondrial short-chain enoyl-coa hydratase 1 deficiency
Mitochondrial trifunctional protein deficiency
Miyoshi muscular dystrophy 1
Miyoshi muscular dystrophy 3
Miyoshi myopathy
Mo ALLOANTIGEN POLYMORPHISM
Mohr-Tranebjaerg syndrome

Molybdenum cofactor deficiency
Monoamine oxidase A deficiency
Monocarboxylate transporter 1 deficiency
Monochromacy
Monogenic Non-Syndromic Obesity
Monogenic diabetes
Mononeuropathy of the median nerve
Morbid obesity
Morbid obesity and spermatogenic failure
Morquio syndrome
Mosaic variegated aneuploidy syndrome
Mosaic variegated aneuploidy syndrome 2
Motor delay
Motor neuron disease
Mowat-Wilson syndrome
Moyamoya disease 5
Moyamoya disease 6 with achalasia
Mucolipidosis
Mucolipidosis III Gamma
Mucolipidosis III alpha/beta
Mucopolysaccharidosis
Mucopolysaccharidosis type I
Mucopolysaccharidosis type VI
Mucopolysaccharidosis type VII
Muenke syndrome
Muir-Torré syndrome
Mulibrey nanism syndrome
Mullerian aplasia and hyperandrogenism
Multi-minicore disease and atypical periodic paralysis
Multicentric osteolysis
Multicentric osteolysis nephropathy
Multiminicore Disease
Multiple Cutaneous and Mucosal Venous Malformations
Multiple Cutaneous and Uterine Leiomyomas
Multiple Epiphyseal Dysplasia
Multiple congenital anomalies
Multiple congenital anomalies-hypotonia-seizures syndrome 1
Multiple congenital anomalies-hypotonia-seizures syndrome 2
Multiple congenital anomalies-hypotonia-seizures syndrome 3
Multiple congenital exostosis
Multiple endocrine neoplasia
Multiple endocrine neoplasia IIA
Multiple epiphyseal dysplasia 1
Multiple epiphyseal dysplasia 4
Multiple epiphyseal dysplasia 5
Multiple exostoses type 2
Multiple fibrofolliculomas

Multiple gastrointestinal atresias
Multiple joint dislocations
Multiple mitochondrial dysfunctions syndrome
Multiple mitochondrial dysfunctions syndrome 1
Multiple mitochondrial dysfunctions syndrome 3
Multiple mitochondrial dysfunctions syndrome 4
Multiple myeloma
Multiple pterygium syndrome Escobar type
Multiple sulfatase deficiency
Multiple synostoses syndrome 2
Multiple synostoses syndrome 3
Multisystem disorder
Multisystemic smooth muscle dysfunction syndrome
Muscle AMP deaminase deficiency
Muscle cramps
Muscle eye brain disease
Muscle hypertrophy
Muscle stiffness
Muscle weakness
Muscular dystrophy
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies)
Muscular dystrophy-dystroglycanopathy (congenital with mental retardation)
Muscular dystrophy-dystroglycanopathy (limb-girdle)
Muscular hypotonia
Muscular hypotonia of the trunk
Mutilating keratoderma
Myasthenia
Myasthenic syndrome
Mycobacterial and viral infections
Mycobacterium tuberculosis
Myd88 deficiency
Myelodysplastic syndrome
Myelofibrosis
Myelokathexis
Myeloperoxidase deficiency
Myeloproliferative Neoplasm
Myeloproliferative disorder
Myhre syndrome
Myocardial infarction
Myocardial infarction 1
Myoclonic dystonia
Myoclonic epilepsy
Myoclonic epilepsy myopathy sensory ataxia
Myoclonus
Myoclonus with epilepsy with ragged red fibers
Myofibrillar Myopathy
Myofibrillar myopathy

Myofibrillar myopathy 1
Myoglobinuria
Myokymia 1
Myokymia 1 with hypomagnesemia
Myopathy
Myopathy with lactic acidosis
Myopathy with postural muscle atrophy
Myopathy with tubular aggregates
Myopia
Myopia 21
Myopia 24
Myopia 6
Myosclerosis
Myosin storage myopathy
Myostatin-related muscle hypertrophy
Myotilinopathy
Myotonia
Myotonia congenita
Ménière's disease
N-terminal acetyltransferase deficiency
NADH-CYTOCHROME b5 REDUCTASE POLYMORPHISM
NAT1*17 ALLELE
NEUROPATHY
NR2E3-Related Disorders
NUCLEOSIDE PHOSPHORYLASE POLYMORPHISM
Nager syndrome
Nail disease
Nail disorder
Nail dystrophy
Nail-patella syndrome
Nance-Horan syndrome
Nanophthalmos 2
Nanophthalmos 4
Narcolepsy 1
Narcolepsy 7
Narrow chest
Nasopharyngeal Neoplasms
Native American myopathy
Navajo neurohepatopathy
Naxos disease
Nemaline Myopathy
Nemaline myopathy 1
Nemaline myopathy 10
Nemaline myopathy 2
Nemaline myopathy 3
Nemaline myopathy 4
Nemaline myopathy 5

Nemaline myopathy 6
Nemaline myopathy 7
Nemaline myopathy 8
Nemaline myopathy 9
Neonatal adrenoleucodystrophy
Neonatal death
Neonatal diabetes mellitus
Neonatal hypotonia
Neonatal insulin-dependent diabetes mellitus
Neonatal intrahepatic cholestasis caused by citrin deficiency
Neonatal respiratory distress
Neonatal severe hyperparathyroidism
Neoplasm
Neoplasm of brain
Neoplasm of breast
Neoplasm of ovary
Neoplasm of stomach
Neoplasm of the thyroid gland
Nephrogenic diabetes insipidus
Nephrolithiasis
Nephrolithiasis/osteoporosis
Nephronophthisis
Nephronophthisis 1
Nephronophthisis 11
Nephronophthisis 12
Nephronophthisis 13
Nephronophthisis 14
Nephronophthisis 15
Nephronophthisis 16
Nephronophthisis 18
Nephronophthisis 19
Nephronophthisis 4
Nephronophthisis 7
Nephronophthisis 9
Nephronophthisis-like nephropathy 1
Nephropathic cystinosis
Nephrotic syndrome
Netherton syndrome
Neu-Laxova syndrome 1
Neu-laxova syndrome 2
Neural tube defect
Neural tube defects
Neuroblastoma
Neuroblastoma 1
Neuroblastoma 2
Neuroblastoma 3
Neuroblastoma Susceptibility

Neurodegeneration
Neurodegeneration with brain iron accumulation 1
Neurodegeneration with brain iron accumulation 2b
Neurodegeneration with brain iron accumulation 4
Neurodegeneration with brain iron accumulation 5
Neurodegeneration with brain iron accumulation 6
Neurodegenerative illness progressing to crippling dystonia and death with relentless cerebral atrophy
Neurodegeration
Neuroferritinopathy
Neurofibromatosis
Neurofibromatosis-Noonan syndrome
Neurofibrosarcoma
Neurogastrointestinal syndrome
Neurohypophyseal diabetes insipidus
Neurologic
Neurological speech impairment
Neuromuscular Diseases
Neuromuscular disease
Neuromyotonia and axonal neuropathy
Neuronal Ceroid-Lipofuscinosis
Neuronal ceroid lipofuscinosis
Neuropathy
Neuropathy hereditary sensory and autonomic type 1
Neuropsychiatric disorder and early-onset cataract
Neutral 1 amino acid transport defect
Neutral lipid storage disease with myopathy
Neutropenia
Neutrophil immunodeficiency syndrome
Nicolaides-Baraitser syndrome
Nicotine
Niemann-Pick disease
Niemann-Pick disease type C1
Niemann-Pick disease type C2
Niemann-pick disease
Nijmegen breakage syndrome-like disorder
No MEN2 disease
Nodal rhythm
Non-immune hydrops fetalis
Non-ketotic hyperglycinemia
Non-small cell lung cancer
Non-syndromic X-linked intellectual disability
Nonaka myopathy
Nonsmall cell lung cancer
Nonsyndromic Deafness
Nonsyndromic Hearing Loss
Nonsyndromic Trigonocephaly
Nonsyndromic hearing loss and deafness

Noonan syndrome
Noonan syndrome 1
Noonan syndrome 3
Noonan syndrome 4
Noonan syndrome 5
Noonan syndrome 6
Noonan syndrome 7
Noonan syndrome 8
Noonan syndrome with multiple lentigines
Noonan syndrome-like disorder with juvenile myelomonocytic leukemia
Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia
Noonan-like facies
Noonan-like syndrome with loose anagen hair
Normokalemic periodic paralysis
North american indian childhood cirrhosis
Norum disease
Nuclearly-encoded mitochondrial complex V (ATP synthase) deficiency 1
Nuclearly-encoded mitochondrial complex V (ATP synthase) deficiency 2
Nuclearly-encoded mitochondrial complex V (ATP synthase) deficiency 3
Nystagmus 6
OAT POLYMORPHISM
OBESITY (BMIQ14)
OCULOCUTANEOUS ALBINISM
ORNITHINE TRANSCARBAMYLASE POLYMORPHISM
OSTEOGENESIS IMPERFECTA
Obesity
Obsessive-compulsive disorder
Occult macular dystrophy
Ochoa syndrome
Ocular albinism
Ocular coloboma
Oculocutaneous albinism
Oculocutaneous albinism type 1
Oculocutaneous albinism type 1B
Oculocutaneous albinism type 3
Oculocutaneous albinism type 4
Oculodentodigital dysplasia
Oculofaciocardiodental syndrome
Oculomaxillofacial dysostosis
Oculomelic amyoplasia
Oculopharyngeal muscular dystrophy
Odontohypophosphatasia
Odontoonychodermal dysplasia
Odontotrichomelic syndrome
Oesophageal carcinoma
Oguchi's disease
Ohdo syndrome

Okt4 epitope deficiency
Oligodendroglioma
Oligodontia-colorectal cancer syndrome
Oligohydramnios
Olivopontocerebellar hypoplasia
Omodysplasia 1
Omphalocele
Opitz-Frias syndrome
Opsismodysplasia
Optic Atrophy
Optic atrophy
Optic atrophy 7
Optic atrophy 9
Optic nerve aplasia
Optic nerve hypoplasia
Optic nerve hypoplasia and abnormalities of the central nervous system
Optic neuropathy
Oral-facial-digital syndrome
Ornithine aminotransferase deficiency
Ornithine carbamoyltransferase deficiency
Orofacial cleft 11
Orofacial cleft 5
Orofacial cleft 7
Orofacial cleft 8
Orofacial-digital syndrome IV
Orofaciodigital syndrome 6
Orofaciodigital syndrome xiv
Orolaryngeal cancer
Orotic aciduria
Orstavik Lindemann Solberg syndrome
Orthostatic intolerance
Osler hemorrhagic telangiectasia syndrome
Osteoarthritis susceptibility 1
Osteoarthritis with mild chondrodysplasia
Osteochondritis dissecans
Osteodysplastic primordial dwarfism
Osteogenesis Imperfecta
Osteogenesis imperfecta
Osteogenesis imperfecta type 1
Osteogenesis imperfecta type 12
Osteogenesis imperfecta type 5
Osteogenesis imperfecta type 7
Osteogenesis imperfecta type 8
Osteogenesis imperfecta type 9
Osteogenesis imperfecta type I
Osteogenesis imperfecta type III
Osteogenesis imperfecta with normal sclerae

Osteoglophonic dysplasia
Osteomyelitis
Osteopathia striata with cranial sclerosis
Osteopenia
Osteopetrosis
Osteopetrosis autosomal dominant type 1
Osteopetrosis autosomal dominant type 2
Osteopetrosis autosomal recessive 1
Osteopetrosis autosomal recessive 2
Osteopetrosis autosomal recessive 4
Osteopetrosis autosomal recessive 7
Osteopetrosis with renal tubular acidosis
Osteoporosis with pseudoglioma
Osteosarcoma
Oto-palato-digital syndrome
Otofaciocervical syndrome
Otofaciocervical syndrome 2
Otospondylomegaepiphyseal dysplasia
Ovarian Adenocarcinoma
Ovarian Dysgenesis
Ovarian Neoplasms
Ovarian Serous Cystadenocarcinoma
Ovarian cancer
Ovarian dysgenesis 1
Ovarian dysgenesis 2
Ovarian dysgenesis 4
Ovarian hyperstimulation syndrome
Ovarian neoplasm
Ovarioleukodystrophy
Overgrowth
PC-K6a
PEHO syndrome
PEN(a)/PEN(b) ALLOANTIGEN POLYMORPHISM
PEPTIDE TRANSPORTER PSF1 POLYMORPHISM
PEPTIDE TRANSPORTER PSF2 POLYMORPHISM
PEX7-Related Disorders
PI
PI CHRISTCHURCH
PI F
PI I
PI M(MINERAL SPRINGS)
PI M(PROCIDA)
PI M1-ALA213
PI M2
PI M3
PI M4
PI NULL(CARDIFF)

PI NULL(DEVON)
PI NULL(LUDWIGSHAFEN)
PI NULL(NEWPORT)
PI P(DUARTE)
PI P(LOWELL)
PI Q0(CARDIFF)
PI Q0(DEVON)
PI Q0(LUDWIGSHAFEN)
PI Q0(NEWPORT)
PI S
PI S(IIYAMA)
PI W(BETHESDA)
PI Z
PI Z(AUGSBURG)
PI Z(TUN)
PIK3CA related overgrowth spectrum
PITX2-Related Eye Abnormalities
POLG-Related Spectrum Disorders
PRKAG2 cardiac syndrome
PROTHROMBIN TYPE 3
PSEUDOHYPOPARATHYROIDISM
PTEN hamartoma tumor syndrome
PULMONARY ALVEOLAR MICROLITHIASIS
Pachydermoperiostosis syndrome
Pachyonychia congenita
Pachyonychia congenita 4
Pachyonychia congenita type 2
Paget disease of bone
Pallister-Hall syndrome
Palmoplantar carcinoma
Palmoplantar keratoderma
Palmoplantar keratoderma and woolly hair
Pancreatic adenocarcinoma
Pancreatic agenesis
Pancreatic agenesis and congenital heart disease
Pancreatic cancer
Pancreatic cancer 1
Pancreatic cancer 2
Pancreatic cancer 3
Pancreatic cancer 4
Pancreatitis
Papillary renal cell carcinoma
Papillon-Lefèvre syndrome
Papillorenal syndrome with macular abnormalities
Papule
Para-Bombay phenotype
Paraganglioma and gastric stromal sarcoma

Paragangliomas 1
Paragangliomas 1 with sensorineural hearing loss
Paragangliomas 2
Paragangliomas 3
Paragangliomas 4
Paragangliomas 5
Paramyotonia congenita of von Eulenburg
Paramyotonia congenita/hyperkalemic periodic paralysis
Parastremmatic dwarfism
Parathyroid adenoma
Parathyroid carcinoma
Parietal foramina 1
Parietal foramina 2
Parkinson Disease
Parkinson disease
Parkinson disease 1
Parkinson disease 11
Parkinson disease 13
Parkinson disease 14
Parkinson disease 15
Parkinson disease 17
Parkinson disease 18
Parkinson disease 19a
Parkinson disease 2
Parkinson disease 20
Parkinson disease 21
Parkinson disease 6
Parkinson disease 7
Parkinson disease 8
Parkinson disease 9
Parkinsonism
Parkinsonism with spasticity
Paroxysmal atrial fibrillation
Paroxysmal choreoathetosis
Paroxysmal dyskinesia
Paroxysmal dystonia
Paroxysmal extreme pain disorder
Paroxysmal familial ventricular fibrillation
Paroxysmal familial ventricular fibrillation 1
Paroxysmal nocturnal hemoglobinuria 1
Partial adenosine deaminase deficiency
Partial albinism
Partial hypoxanthine-guanine phosphoribosyltransferase deficiency
Patterned dystrophy of retinal pigment epithelium
Pectus excavatum
Peeling skin syndrome
Peeling skin syndrome 3

Pelger-Huët anomaly
Pelizaeus-Merzbacher disease
Pelvic girdle muscle weakness
Pena-Shokeir syndrome type I
Pendred's syndrome
Periodic paralysis
Periodontitis
Peripheral arterial occlusive disease 1
Peripheral axonal neuropathy
Peripheral demyelinating neuropathy
Peripheral neuropathy
Peripheral visual field loss
Periportal fibrosis
Periventricular nodular heterotopia 1
Permanent neonatal diabetes mellitus
Peroxisomal biogenesis disorder 3b
Peroxisomal fatty acyl-coa reductase 1 disorder
Peroxisome biogenesis disorder 11A
Peroxisome biogenesis disorder 11B
Peroxisome biogenesis disorder 13A
Peroxisome biogenesis disorder 14B
Peroxisome biogenesis disorder 1B
Peroxisome biogenesis disorder 2a (zellweger)
Peroxisome biogenesis disorder 3A
Peroxisome biogenesis disorder 4B
Peroxisome biogenesis disorder 5B
Peroxisome biogenesis disorder 5a (zellweger)
Peroxisome biogenesis disorder 6A
Peroxisome biogenesis disorder 6B
Peroxisome biogenesis disorder 7A
Peroxisome biogenesis disorder 7B
Peroxisome biogenesis disorder 8B
Peroxisome biogenesis disorder 9B
Peroxisome biogenesis disorders
Perrault Syndrome
Perrault syndrome 2
Perrault syndrome 3
Perrault syndrome 4
Perrault syndrome 5
Perry syndrome
Persistent hyperinsulinemic hypoglycemia of infancy
Persistent mullerian duct syndrome
Persistent truncus arteriosus
Peters anomaly
Peters plus syndrome
Pettigrew syndrome
Peutz-Jeghers syndrome

Pfeiffer syndrome
Phenylketonuria
Phenylthiocarbamide tasting
Phenytoin response
Pheochromocytoma
Phosphate transport defect
Phosphoenolpyruvate carboxykinase (GTP) deficiency
Phosphoglycerate dehydrogenase deficiency
Phosphoglycerate kinase 1 deficiency
Phosphoglycerate kinase electrophoretic variant PGK II
Phosphohydroxylysinuria
Phospholipase a2
Phosphoribosylpyrophosphate synthetase superactivity
Phosphoserine aminotransferase deficiency
Phytanic acid storage disease
Pick's disease
Piebaldism
Piebaldism with sensorineural deafness
Pierson syndrome
Pigmentary pallidal degeneration
Pigmentary retinal dystrophy
Pigmentary retinopathy and sensorineural deafness
Pigmented nodular adrenocortical disease
Pigmented paravenous chorioretinal atrophy
Pili torti-deafness syndrome
Pilomatrixoma
Pineal hyperplasia AND diabetes mellitus syndrome
Pitt-Hopkins syndrome
Pitt-Hopkins-like syndrome
Pitt-Hopkins-like syndrome 1
Pitt-Hopkins-like syndrome 2
Pituitary adenoma predisposition
Pituitary dependent hypercortisolism
Pituitary hormone deficiency
Pituitary stalk interruption syndrome
Pityriasis rubra pilaris
Plasma triglyceride level quantitative trait locus
Plasminogen activator inhibitor type 1 deficiency
Plasminogen deficiency
Plasmodium vivax
Platelet glycoprotein IV deficiency
Platelet-activating factor acetylhydrolase deficiency
Platelet-type bleeding disorder 13
Platelet-type bleeding disorder 15
Platelet-type bleeding disorder 16
Platelet-type bleeding disorder 17
Platelet-type bleeding disorder 19

Platelet-type bleeding disorder 8
Platinum compounds response - Efficacy
Platinum compounds response - Toxicity/ADR
Platyspondylic lethal skeletal dysplasia Torrance type
Pleural effusion
Pleuropulmonary blastoma
Pneumothorax
Poikiloderma
Poikiloderma with neutropenia
Pointed chin
Poliosis
Polyagglutinable erythrocyte syndrome
Polyarteritis nodosa
Polycystic kidney disease
Polycystic kidney disease 2
Polycystic kidney dysplasia
Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy
Polycystic liver disease 2
Polydactyly
Polyglandular autoimmune syndrome
Polyglucosan body disease
Polyglucosan body myopathy 1 with or without immunodeficiency
Polyglucosan body myopathy 2
Polymicrogyria
Polyneuropathy
Polysubstance abuse
Pontocerebellar hypoplasia
Pontocerebellar hypoplasia type 1A
Pontocerebellar hypoplasia type 2A
Pontocerebellar hypoplasia type 2B
Pontocerebellar hypoplasia type 2D
Pontocerebellar hypoplasia type 4
Pontocerebellar hypoplasia type 5
Pontocerebellar hypoplasia type 6
Pontocerebellar hypoplasia type 8
Poor speech
Popliteal pterygium syndrome
Popliteal pterygium syndrome lethal type
Porencephaly 1
Porencephaly 2
Poretti-boltshauser syndrome
Porokeratosis
Porokeratosis 8
Porphobilinogen synthase deficiency
Porphyria
Porphyria cutanea tarda
Possible: Alpha-1-antitrypsin deficiency, ANCA Vasculitis

Postanesthetic apnea
Postaxial foot polydactyly
Postaxial hand polydactyly
Postaxial polydactyly
Postaxial polydactyly type A6
Posterior Polymorphous Corneal Dystrophy
Posterior column ataxia with retinitis pigmentosa
Posterior polar cataract
Postmenopausal osteoporosis
Postnatal growth retardation
Posttransfusion purpura
Postural instability
Potassium aggravated myotonia
Preaxial polydactyly 4
Preeclampsia
Preeclampsia/eclampsia 4
Preeclampsia/eclampsia 5
Pregnancy loss
Prekallikrein deficiency
Prelingual sensorineural hearing impairment
Premature birth
Premature chromatid separation trait
Premature ovarian failure
Premature ovarian failure 1
Premature ovarian failure 10
Premature ovarian failure 2b
Premature ovarian failure 3
Premature ovarian failure 4
Premature ovarian failure 5
Premature ovarian failure 7
Premature ovarian failure 9
Pretibial epidermolysis bullosa
Primary Microcephaly
Primary aldosteronism
Primary autosomal recessive microcephaly 1
Primary autosomal recessive microcephaly 12
Primary autosomal recessive microcephaly 13
Primary autosomal recessive microcephaly 2
Primary autosomal recessive microcephaly 3
Primary autosomal recessive microcephaly 5
Primary autosomal recessive microcephaly 6
Primary autosomal recessive microcephaly 7
Primary autosomal recessive microcephaly 9
Primary ciliary dyskinesia
Primary ciliary dyskinesia 23
Primary ciliary dyskinesia 24
Primary ciliary dyskinesia 25

Primary congenital glaucoma
Primary dilated cardiomyopathy
Primary erythromelalgia
Primary familial hypertrophic cardiomyopathy
Primary hyperoxaluria
Primary hypertrophic osteoarthropathy
Primary hypomagnesemia
Primary localized cutaneous amyloidosis 1
Primary microcephaly
Primary open angle glaucoma
Primary open angle glaucoma juvenile onset 1
Primary progressive aphasia
Primary pulmonary hypertension
Primary pulmonary hypertension 2
Primary pulmonary hypertension 4
Primordial dwarfism
Primrose syndrome
Prion disease
Progressive External Ophthalmoplegia with Mitochondrial DNA Deletions
Progressive distal muscle weakness
Progressive external ophthalmoplegia
Progressive external ophthalmoplegia with mitochondrial DNA deletions
Progressive familial heart block
Progressive familial heart block type 1A
Progressive familial heart block type 1B
Progressive familial intrahepatic cholestasis 2
Progressive familial intrahepatic cholestasis 3
Progressive familial intrahepatic cholestasis 4
Progressive intrahepatic cholestasis
Progressive myoclonic epilepsy
Progressive myoclonus epilepsy with ataxia
Progressive myositis ossificans
Progressive proximal muscle weakness
Progressive pseudorheumatoid dysplasia
Progressive sclerosing poliodystrophy
Progressive supranuclear ophthalmoplegia
Progressive visual loss
Proguanil
Prolactinoma
Prolidase deficiency
Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome
Proline dehydrogenase deficiency
Prolonged electroretinal response suppression
Prominent metopic ridge
Proopiomelanocortin deficiency
Properdin deficiency
Propionic acidemia

Proprotein convertase 1/3 deficiency
Proptosis
Prostate cancer
Prostate cancer susceptibility
Prostate cancer/brain cancer susceptibility
Prostatic Neoplasms
Protection against Creutzfeldt-Jakob disease
Protein S deficiency
Protein s heerlen
Proteinuria
Proteus syndrome
Prothrombin deficiency
Protoporphyria
Proud Levine Carpenter syndrome
Proximal muscle weakness
Pseudo von Willebrand disease
Pseudo-Hurler polydystrophy
Pseudoachondroaplasia
Pseudoachondroplasia
Pseudoachondroplastic spondyloepiphyseal dysplasia syndrome
Pseudoexfoliation glaucoma
Pseudohermaphroditism
Pseudohypoaldosteronism
Pseudohypoaldosteronism type 1 autosomal dominant
Pseudohypoaldosteronism type 1 autosomal recessive
Pseudohypoaldosteronism type 2B
Pseudohypoaldosteronism type 2D
Pseudohypoaldosteronism type 2E
Pseudohypoparathyroidism type 1A
Pseudohypoparathyroidism type 1C
Pseudoinflammatory fundus dystrophy
Pseudomonas aeruginosa
Pseudoneonatal adrenoleukodystrophy
Pseudoprimary hyperaldosteronism
Pseudopseudohypoparathyroidism
Pseudoxanthoma elasticum
Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency
Psoriasis 2
Psoriasis susceptibility 2
Psychomotor retardation
Pulmonary Surfactant Metabolism Dysfunction
Pulmonary alveolar proteinosis
Pulmonary arterial hypertension related to hereditary hemorrhagic telangiectasia
Pulmonary fibrosis
Pulmonary fibrosis and/or bone marrow failure
Pulmonary hypertension
Pulmonary venoocclusive disease 1

Pulmonic stenosis
Purine-nucleoside phosphorylase deficiency
Pustular psoriasis
Pyknodysostosis
Pyloric stenosis
Pyogenic arthritis
Pyridoxal 5'-phosphate-dependent epilepsy
Pyridoxine-dependent epilepsy
Pyrimidine analogues response - Toxicity/ADR
Pyruvate carboxylase deficiency
Pyruvate dehydrogenase E1-alpha deficiency
Pyruvate dehydrogenase E1-beta deficiency
Pyruvate dehydrogenase E2 deficiency
Pyruvate dehydrogenase E3-binding protein deficiency
Pyruvate dehydrogenase lipoic acid synthetase deficiency
Pyruvate dehydrogenase phosphatase deficiency
Pyruvate kinase deficiency of red cells
Question mark ears
RECOMBINATION ACTIVATING GENE 1 POLYMORPHISM
RLBP1-Related Disorders
RPE65-Related Disorders
RPGRIP1L-Related Disorders
RRM2B-related mitochondrial disease
Radial aplasia-thrombocytopenia syndrome
Radin blood group
Radiohumeral fusions with other skeletal and craniofacial anomalies
Rapadilino syndrome
Rapp-Hodgkin ectodermal dysplasia syndrome
Rasopathy
Recessive dystrophic epidermolysis bullosa
Recurrent abortion
Recurrent aphthous stomatitis
Recurrent respiratory infections
Recurrent subcortical infarcts
Refsum disease
Reifenstein syndrome
Reis-Bucklers' corneal dystrophy
Relapsing remitting multiple sclerosis
Renal Hypomagnesemia
Renal adysplasia
Renal carnitine transport defect
Renal cell carcinoma
Renal cell carcinoma with paraneoplastic erythrocytosis
Renal cyst
Renal dysplasia
Renal dysplasia and retinal aplasia
Renal hamartomas nephroblastomatosis and fetal gigantism

Renal hypouricemia 2
Renal tubular acidosis
Renal tubular acidosis with progressive nerve deafness
Renal-hepatic-pancreatic dysplasia
Renal-hepatic-pancreatic dysplasia 2
Renpenning syndrome 1
Reticular dysgenesis
Reticulate acropigmentation of Kitamura
Retinal Macular Dystrophy
Retinal arteries
Retinal atrophy
Retinal cone dystrophy 3A
Retinal cone dystrophy 3B
Retinal cone dystrophy 4
Retinal degeneration
Retinal dystrophy
Retinitis Pigmentosa
Retinitis Pigmentosa 23
Retinitis pigmentosa
Retinitis pigmentosa 1
Retinitis pigmentosa 10
Retinitis pigmentosa 11
Retinitis pigmentosa 12
Retinitis pigmentosa 13
Retinitis pigmentosa 14
Retinitis pigmentosa 15
Retinitis pigmentosa 17
Retinitis pigmentosa 18
Retinitis pigmentosa 19
Retinitis pigmentosa 2
Retinitis pigmentosa 20
Retinitis pigmentosa 25
Retinitis pigmentosa 26
Retinitis pigmentosa 27
Retinitis pigmentosa 28
Retinitis pigmentosa 33
Retinitis pigmentosa 35
Retinitis pigmentosa 36
Retinitis pigmentosa 37
Retinitis pigmentosa 38
Retinitis pigmentosa 39
Retinitis pigmentosa 4
Retinitis pigmentosa 40
Retinitis pigmentosa 41
Retinitis pigmentosa 42
Retinitis pigmentosa 43
Retinitis pigmentosa 44

Retinitis pigmentosa 45
Retinitis pigmentosa 46
Retinitis pigmentosa 48
Retinitis pigmentosa 49
Retinitis pigmentosa 50
Retinitis pigmentosa 51
Retinitis pigmentosa 53
Retinitis pigmentosa 54
Retinitis pigmentosa 56
Retinitis pigmentosa 58
Retinitis pigmentosa 59
Retinitis pigmentosa 60
Retinitis pigmentosa 62
Retinitis pigmentosa 64
Retinitis pigmentosa 66
Retinitis pigmentosa 68
Retinitis pigmentosa 69
Retinitis pigmentosa 7
Retinitis pigmentosa 70
Retinitis pigmentosa 74
Retinitis pigmentosa 76
Retinitis pigmentosa without situs inversus
Retinitis pigmentosa-deafness syndrome
Retinitis punctata albescens
Retinoblastoma
Retinopathy of prematurity
Rett syndrome
Revesz syndrome
Reynolds syndrome
Rh-mod syndrome
Rh-null hemolytic anemia
Rhabdoid tumor predisposition syndrome 2
Rhabdomyosarcoma
Rhd category d-vii
Rhegmatogenous retinal detachment
Rheumatoid arthritis
Rhizomelic chondrodysplasia punctata
Rhizomelic chondrodysplasia punctata type 1
Rhizomelic chondrodysplasia punctata type 2
Rhizomelic chondrodysplasia punctata type 3
Richieri Costa Pereira syndrome
Riddle syndrome
Rieger anomaly
Right ventricular cardiomyopathy
Rigidity
Rigidity and multifocal seizure syndrome
Ring dermoid of cornea

Rippling muscle disease
Rippling muscle disease 2
Roberts-SC phocomelia syndrome
Robinow Sorauf syndrome
Robinow syndrome
Roifman syndrome
Rolandic epilepsy with mental retardation and speech dyspraxia
Romano-Ward syndrome
Rothmund-Thomson syndrome
Rotor syndrome
Roussy-Lévy syndrome
Rubinstein-Taybi syndrome
Rubinstein-Taybi syndrome 2
Ruijs-Aalfs syndrome
SCHIZENCEPHALY
SCIANNA BLOOD GROUP SYSTEM
SCID due to ADA deficiency
SECRETOR/NONSECRETOR POLYMORPHISM
SELECTIN P POLYMORPHISM
SEROTONIN 5-HT-2C RECEPTOR POLYMORPHISM
SEVERE ACHONDRODYSPLASIA WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS
SH3TC2-Related Disorders
SHORT syndrome
SLC26A2-Related Disorders
SLC26A4-Related Disorders
SN-38 response - Other
SPATA7-Related Disorders
SQUAMOUS CELL CARCINOMA
STEROID 5-ALPHA-REDUCTASE POLYMORPHISM
SUDDEN INFANT DEATH SYNDROME
SWANN BLOOD GROUP ANTIGEN
Sacral agenesis with vertebral anomalies
Saethre-Chotzen syndrome
Saethre-Chotzen syndrome with eyelid anomalies
Salla disease
Sandhoff disease
Sanfilippo syndrome
Sarcoidosis
Sarcoma
Sarcosine dehydrogenase deficiency
Sarcotubular myopathy
Scalp ear nipple syndrome
Scaphocephaly
Scaphocephaly and axenfeld-rieger anomaly
Scapuloperoneal myopathy
Scapuloperoneal spinal muscular atrophy
Scapuloperoneal weakness

Scarring
Scarring alopecia of scalp
Schaaf-yang syndrome
Schimke immunoosseous dysplasia
Schindler disease
Schinzel-Giedion syndrome
Schizophrenia
Schizophrenia 15
Schizophrenia 4
Schneckenbecken dysplasia
Schnyder crystalline corneal dystrophy
Schopf-Schulz-Passarge syndrome
Schwannomatosis 1
Schwannomatosis 2
Schwartz Jampel syndrome type 1
Sclerosing cholangitis
Sclerosteosis
Sclerosteosis 2
Scoliosis
Scrotal hypoplasia
SeSAME syndrome
Sebaceous tumors
Sebastian syndrome
Seckel syndrome
Seckel syndrome 1
Seckel syndrome 2
Seckel syndrome 5
Seckel syndrome 7
Secondary hypothyroidism
Segawa syndrome
Seizure Disorders
Seizure disorder
Seizures
Selective tooth agenesis
Selective tooth agenesis 1
Senior-Loken syndrome 4
Senior-Loken syndrome 5
Senior-Loken syndrome 8
Sensorimotor neuropathy
Sensory ataxic neuropathy
Sensory neuropathy
Sepiapterin reductase deficiency
Sepsis
Septo-optic dysplasia sequence
Septooptic dysplasia
Serkal syndrome
Serum amyloid a variant

Severe Combined Immune Deficiency
Severe Myopia
Severe X-linked myotubular myopathy
Severe autosomal recessive muscular dystrophy of childhood - North African type
Severe brain malformation
Severe cerebellar hypoplasia
Severe combined immunodeficiency
Severe combined immunodeficiency disease
Severe combined immunodeficiency due to ADA deficiency
Severe combined immunodeficiency with microcephaly
Severe combined immunodeficiency with sensitivity to ionizing radiation
Severe congenital neutropenia
Severe congenital neutropenia 2
Severe congenital neutropenia 3
Severe congenital neutropenia 4
Severe congenital neutropenia 6
Severe congenital neutropenia X-linked
Severe congenital neutropenia autosomal dominant
Severe cystic degeneration of the brain
Severe hydrocephalus
Severe immunodeficiency
Severe intellectual deficiency
Severe myoclonic epilepsy in infancy
Severe neonatal-onset encephalopathy with microcephaly
Sex cord-stromal tumor
Shaheen syndrome
Short QT syndrome 2
Short QT syndrome 3
Short Rib Polydactyly Syndrome
Short foot
Short lower limbs
Short palpebral fissure
Short rib polydactyly syndrome 5
Short rib-polydactyly syndrome
Short ribs
Short stature
Short-rib thoracic dysplasia 10 with or without polydactyly
Short-rib thoracic dysplasia 10 with polydactyly
Short-rib thoracic dysplasia 10 without polydactyly
Short-rib thoracic dysplasia 11 with or without polydactyly
Short-rib thoracic dysplasia 13 with or without polydactyly
Short-rib thoracic dysplasia 3 with or without polydactyly
Short-rib thoracic dysplasia 8 with or without polydactyly
Short-rib thoracic dysplasia without polydactyly
Shprintzen syndrome
Shprintzen-Goldberg syndrome
Shwachman syndrome

Sialic acid storage disease
Sialidosis
Sialidosis type I
Sialuria
Sick sinus syndrome
Sick sinus syndrome 1
Sick sinus syndrome 2
Siderius X-linked mental retardation syndrome
Sideroblastic anemia
Sideroblastic anemia with B-cell immunodeficiency
Silver spastic paraplegia syndrome
Simpson-Golabi-Behmel syndrome
Single upper central incisor
Singleton-Merten syndrome 1
Sinus node disease
Sitosterolemia
Sjögren-Larsson syndrome
Skeletal defects
Skeletal dysplasia
Skeletal dysplasia with acanthosis nigricans
Skin fragility woolly hair syndrome
Skin/hair/eye pigmentation
Skin/hair/eye pigmentation 2
Slow acetylator due to N-acetyltransferase enzyme variant
Slowed nerve conduction velocity
Small cell lung cancer
Small fiber neuropathy
Small for gestational age
Small hand
Small intestine carcinoid
Smith-Lemli-Opitz syndrome
Smith-Magenis Syndrome-like
Smith-Magenis syndrome
Smith-McCort dysplasia 1
Smith-McCort dysplasia 2
Smoking as a quantitative trait locus 3
Snowflake vitreoretinal degeneration
Snyder Robinson syndrome
Sodium serum level quantitative trait locus 1
Somatostatin analog
Somatotroph adenoma
Sorsby fundus dystrophy
Sotos Syndrome
Sotos syndrome 1
Sotos syndrome 2
Spastic Paraplegia
Spastic ataxia 5

Spastic ataxia Charlevoix-Saguenay type
Spastic paraparesis
Spastic paraplegia
Spastic paraplegia 1
Spastic paraplegia 10
Spastic paraplegia 11
Spastic paraplegia 12
Spastic paraplegia 13
Spastic paraplegia 15
Spastic paraplegia 18
Spastic paraplegia 2
Spastic paraplegia 26
Spastic paraplegia 3
Spastic paraplegia 30
Spastic paraplegia 31
Spastic paraplegia 33
Spastic paraplegia 35
Spastic paraplegia 39
Spastic paraplegia 4
Spastic paraplegia 42
Spastic paraplegia 43
Spastic paraplegia 45
Spastic paraplegia 46
Spastic paraplegia 50
Spastic paraplegia 54
Spastic paraplegia 55
Spastic paraplegia 56
Spastic paraplegia 5A
Spastic paraplegia 7
Spastic paraplegia 72
Spastic paraplegia 75
Spastic paraplegia 79
Spastic paraplegia 8
Spastic tetraparesis
Spastic tetraplegia
Speech apraxia
Speech-language disorder 1
Spermatocytic seminoma
Spermatogenic failure 11
Spermatogenic failure 13
Spermatogenic failure 3
Spermatogenic failure 8
Spermatogenic failure 9
Spherocytosis
Spherocytosis type 1
Spherocytosis type 2
Spherocytosis type 4

Spherocytosis type 5
Spheroid body myopathy
Sphingolipid activator protein 1 deficiency
Sphingomyelin/cholesterol lipidosis
Spiegler-Brooke syndrome
Spinal muscular atrophy
Spinocerebellar Ataxia
Spinocerebellar ataxia
Spinocerebellar ataxia 13
Spinocerebellar ataxia 14
Spinocerebellar ataxia 15
Spinocerebellar ataxia 19
Spinocerebellar ataxia 21
Spinocerebellar ataxia 23
Spinocerebellar ataxia 26
Spinocerebellar ataxia 27
Spinocerebellar ataxia 28
Spinocerebellar ataxia 29
Spinocerebellar ataxia 35
Spinocerebellar ataxia 38
Spinocerebellar ataxia 40
Spinocerebellar ataxia 5
Spinocerebellar ataxia 6
Spinocerebellar ataxia autosomal recessive 1
Spinocerebellar ataxia autosomal recessive with axonal neuropathy
Splenomegaly
Split-hand/foot malformation 1
Split-hand/foot malformation 1 with sensorineural hearing loss
Split-hand/foot malformation 4
Split-hand/foot malformation 6
Spondylocarpotarsal synostosis syndrome
Spondylocheirodysplasia
Spondylocostal dysostosis 1
Spondylocostal dysostosis 2
Spondylocostal dysostosis 3
Spondylocostal dysostosis 5
Spondyloenchondrodysplasia with immune dysregulation
Spondyloepimetaphyseal dysplasia
Spondyloepimetaphyseal dysplasia Strudwick type
Spondyloepimetaphyseal dysplasia with joint laxity
Spondyloepimetaphyseal dysplasia with multiple dislocations
Spondyloepiphyseal dysplasia Maroteaux type
Spondyloepiphyseal dysplasia congenita
Spondyloepiphyseal dysplasia tarda
Spondyloepiphyseal dysplasia with congenital joint dislocations
Spondylometaepiphyseal dysplasia short limb-hand type
Spondylometaphyseal dysplasia

Spondylometaphyseal dysplasia with cone-rod dystrophy
Spondyloperipheral dysplasia
Spongiform encephalopathy with neuropsychiatric features
Spongy degeneration of central nervous system
Spontaneous pneumothorax
Sprinting performance
Squamous cell carcinoma
Squamous cell carcinoma of lung
Squamous cell carcinoma of the head and neck
Squamous cell carcinoma of the skin
Stapes ankylosis with broad thumb and toes
Stargardt Disease
Stargardt Disease 3
Stargardt disease
Stargardt disease 1
Stargardt disease 4
Steatocystoma multiplex
Steel syndrome
Stenosis of the external auditory canal
Steroid-resistant nephrotic syndrome
Stickler Syndrome
Stickler syndrome
Stickler syndrome type 1
Stiff skin syndrome
Sting-associated vasculopathy
Stocco dos Santos syndrome
Stormorken syndrome
Strabismus
Striatal necrosis
Striatonigral degeneration
Striatonigral degeneration infantile
Stromme syndrome
Sturge-Weber syndrome
Stuttering
Stuve-Wiedemann syndrome
Subacute neuronopathic Gaucher's disease
Subcortical band heterotopia
Subcortical laminar heterotopia
Sublingual nitroglycerin
Succinate-semialdehyde dehydrogenase deficiency
Succinyl-CoA acetoacetate transferase deficiency
Sucrase-isomaltase deficiency
Sudden cardiac death
Sudden unexplained death
Sulfite oxidase deficiency
Supernumerary ribs
Supravalvar aortic stenosis

Supravalvular aortic stenosis
Surfactant metabolism dysfunction
Susceptibility to Nonalcoholic Fatty Liver Disease
Susceptibility to hangover
Susceptibility to malaria
Susceptibility to neovascular type of age-related macular degeneration
Sveinsson choreoretinal atrophy
Sweat chloride elevation without cystic fibrosis
Symmetrical dyschromatosis of extremities
Symphalangism
Symphalangism-brachydactyly syndrome
Syncope
Syndactyly Cenani Lenz type
Syndactyly type 3
Syndactyly type 9
Syndrome of entercolitis and autoinflmmation caused by mutation of NLRC4 (SCAN4)
Syndromic X-linked mental retardation
Syndromic intellectual disability
Syndromic mental retardation
Synpolydactyly 1
Systemic lupus erythematosus
Systemic lupus erythematosus 11
Systemic lupus erythmatosus
T-cell acute lymphoblastic leukemia
T-cell immunodeficiency
T-cell prolymphocytic leukemia
TAP1 deficiency
TARP syndrome
TCN2 POLYMORPHISM
TCTN2-Related Disorders
THYROID CANCER
THYROID CARCINOMA WITH THYROTOXICOSIS
TLR4 POLYMORPHISM
TMEM67-Related Disorders
TNF receptor binding
TNF receptor-associated periodic fever syndrome (TRAPS)
TRANSTHYRETIN POLYMORPHISM
TRUNCUS ARTERIOSUS
Talipes equinovarus
Tall stature
Tangier disease
Tarsal carpal coalition syndrome
Tatton-Brown-rahman syndrome
Tay-Sachs disease
Tay-sachs disease
Tegafur response
Telangiectasia

Temple-Baraitser syndrome
Temtamy preaxial brachydactyly syndrome
Temtamy syndrome
Tenorio syndrome
Terminal osseous dysplasia
Testicular anomalies with or without congenital heart disease
Testosterone 17-beta-dehydrogenase deficiency
Tetraamelia
Tetralogy of Fallot
Thanatophoric dysplasia type 1
Thecoma
Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type)
Thick vermilion border
Thiel-Behnke corneal dystrophy
Thiopurine methyltransferase deficiency
Thoracic aortic aneurysm and aortic dissection
Three M syndrome 1
Three M syndrome 2
Thrombocytopenia
Thrombocytopenia 2
Thrombocytopenia 4
Thrombocytopenia 5
Thrombocytosis
Thrombophilia
Thrombophilia due to activated protein C resistance
Thrombophilia due to factor V Leiden
Thrombophilia due to protein S deficiency
Thrombophilia due to thrombomodulin defect
Thyroid Hormonogenesis Defect
Thyroid adenoma
Thyroid agenesis
Thyroid cancer
Thyroid carcinoma
Thyroid dyshormonogenesis
Thyroid dyshormonogenesis 1
Thyroid dyshormonogenesis 6
Thyroid hormone metabolism
Thyroid hormone resistance
Thyrotoxic periodic paralysis
Thyrotropin-releasing hormone resistance
Thyroxine-binding globulin
Thyroxine-binding globulin deficiency
Thyroxine-binding globulin quantitative trait locus
Tibia
Tietz syndrome
Timothy syndrome
Tobacco use disorder

Tolbutamide response
Tooth agenesis
Torsades de pointes
Tourette Syndrome
Townes syndrome
Townes-Brocks-branchiootorenal-like syndrome
Transcobalamin II deficiency
Transcolabamin II deficiency
Transferrin variant b2
Transferrin variant bv
Transferrin variant c1/c2
Transferrin variant chi
Transferrin variant d1
Transient Neonatal Diabetes
Transient bullous dermolysis of the newborn
Transient myeloproliferative disorder of Down syndrome
Transient neonatal diabetes mellitus 1
Transient neonatal diabetes mellitus 2
Transient neonatal diabetes mellitus 3
Transitional cell carcinoma of the bladder
Transposition of the great arteries
Treacher Collins Syndrome
Treacher Collins syndrome 1
Treacher Collins syndrome 2
Tremor
Trichohepatoenteric syndrome
Trichohepatoenteric syndrome 1
Trichohepatoenteric syndrome 2
Trichomegaly with mental retardation
Trichorhinophalangeal dysplasia type I
Trichorhinophalangeal syndrome type 3
Trichothiodystrophy
Trichothiodystrophy 1
Trichothiodystrophy 2
Trichothiodystrophy 3
Triglyceride storage disease with ichthyosis
Trigonocephaly 1
Trigonocephaly 2
Trimethylaminuria
Triosephosphate isomerase deficiency
Triphalangeal thumb
Tritanopia
Truncal obesity
Tryptophan 5-monooxygenase deficiency
Tuberous sclerosis 1
Tuberous sclerosis 2
Tuberous sclerosis syndrome

Tumor necrosis factor alpha (TNF-alpha) inhibitors response - Efficacy
Tumor susceptibility linked to germline BAP1 mutations
Tumoral calcinosis
Turcot syndrome
Type II Collagenopathies
Typical Joubert syndrome MRI findings
Tyrosinase-negative oculocutaneous albinism
Tyrosinase-positive oculocutaneous albinism
Tyrosine kinase inhibitor response
Tyrosinemia type 2
Tyrosinemia type I
UCP3 POLYMORPHISM G/A
UDPglucose-4-epimerase deficiency
USH2A-Related Disorders
USHER SYNDROME
UV-sensitive syndrome
UV-sensitive syndrome 3
Ullrich congenital muscular dystrophy 1
Ulna and fibula absence of with severe limb deficiency
Ulnar deviation of the wrist
Ulnar-mammary syndrome
Unipolar depression
Unsteady gait
Unverricht-Lundborg syndrome
Upper limb undergrowth
Upshaw-Schulman syndrome
Uric acid concentration
Uridine 5-prime monophosphate hydrolase deficiency
Urocanate hydratase deficiency
Urticaria
Usher syndrome
Uterine Carcinosarcoma
Uterine cervical neoplasms
Uv-induced skin damage
VACTERL association with hydrocephaly
Van Buchem disease type 2
Van Maldergem syndrome 1
Van Maldergem syndrome 2
Van der Woude syndrome
Variant of unknown significance
Variegate porphyria
Vasculopathy
Venous thromboembolism
Venous thrombosis
Ventral septal defect
Ventricular extrasystoles
Ventricular fibrillation

Ventricular hypertrophy
Ventricular septal defect
Ventricular septal defect 1
Ventricular septal defect 3
Ventricular tachycardia
Ventriculomegaly
Ventriculomegaly with cystic kidney disease
Verheij syndrome
Verloes Bourguignon syndrome
Very long chain acyl-CoA dehydrogenase deficiency
Vesicoureteral reflux 2
Vesicoureteral reflux 3
Vesicoureteral reflux 8
Visceral heterotaxy 5
Visceral myopathy
Visual impairment
Visual loss
Vitamin D-Dependent Rickets
Vitamin D-dependent rickets
Vitamin K-Dependent Clotting Factors
Vitamin d hydroxylation-deficient rickets
Vitamin k-dependent clotting factors
Vitelliform macular dystrophy
Vitelliform macular dystrophy type 2
Vitreoretinochoroidopathy
Vitreoretinopathy
Vitreoretinopathy with phalangeal epiphyseal dysplasia
Von Hippel-Lindau syndrome
WDR35-Related Disorders
WFS1-Related Disorders
Waardenburg syndrome
Waardenburg syndrome type 1
Waardenburg syndrome type 2A
Waardenburg syndrome type 2E
Waardenburg syndrome type 4A
Waardenburg syndrome type 4B
Waardenburg syndrome type 4C
Waddling gait
Wagner syndrome
Walker-Warburg congenital muscular dystrophy
Warburg micro syndrome 1
Warburg micro syndrome 2
Warburg micro syndrome 3
Warburg micro syndrome 4
Warfarin response
Warsaw breakage syndrome
Warts

Weaver syndrome
Webbed neck
Weill-Marchesani syndrome
Weill-Marchesani syndrome 1
Weill-Marchesani syndrome 3
Weissenbacher-Zweymuller syndrome
Werdnig-Hoffmann disease
Werner syndrome
White blood cell count quantitative trait locus 1
White sponge nevus 2
White sponge nevus of cannon
Wide intermamillary distance
Wieacker syndrome
Wiedemann-Steiner syndrome
Wilms Tumor
Wilms tumor
Wilms tumor 1
Wilson disease
Wiskott-Aldrich syndrome
Wolcott-Rallison dysplasia
Wolff-Parkinson-White pattern
Wolff-Parkinson-White syndrome
Wolfram syndrome
Wolfram syndrome 2
Wolfram-like syndrome
Wolman disease
Woolly hair
Worth disease
X-Linked Mental Retardation 41
X-Linked mental retardation 90
X-linked agammaglobulinemia
X-linked agammaglobulinemia with growth hormone deficiency
X-linked hereditary motor and sensory neuropathy
X-linked hydrocephalus syndrome
X-linked ichthyosis with steryl-sulfatase deficiency
X-linked mental retardation with marfanoid habitus syndrome
X-linked severe combined immunodeficiency
XFE progeroid syndrome
Xanthelasmas
Xerocytosis
Xeroderma pigmentosum
Xeroderma pigmentosum group g/Cockayne syndrome
YAO SYNDROME
YT BLOOD GROUP POLYMORPHISM
Young Simpson syndrome
Yunis Varon syndrome
ZNF711-Related X-linked Mental Retardation

Zellweger syndrome
Zimmermann-Laband syndrome 1
Zinc deficiency
Zonular Pulverulent Cataract
Zonular pulverulent cataract 3
Zunich neuroectodermal syndrome
acenocoumarol response - Dosage
adalimumab response - Efficacy
allopurinol response - Efficacy
alterations of great arteries and veins
aminoglycoside antibacterials response - Toxicity/ADR
amitriptyline response - Efficacy
anthracyclines and related substances response - Toxicity/ADR
antineoplastic agents response - Efficacy
antipsychotics response - Toxicity/ADR
aspirin response - Efficacy
aspirin response - Toxicity/ADR
ataluren response - Efficacy
autistic features
beta Thalassemia
beta^0^ Thalassemia
boceprevir response - Efficacy
buprenorphine response - Dosage
bupropion response - Efficacy
capecitabine response - Toxicity/ADR
carbamazepine response - Dosage
carboplatin
carboplatin response - Efficacy
celecoxib response - Dosage
celecoxib response - Toxicity/ADR
cerebellar-facial-dental syndrome
cetuximab response - Efficacy
chlorproguanil and dapsone response - Toxicity/ADR
cisplatin response - Efficacy
cisplatin response - Toxicity/ADR
citalopram response - Efficacy
clomipramine response - Efficacy
clopidogrel response - Efficacy
clozapine response - Toxicity/ADR
congenital neutropenia
cyclophosphamide response - Efficacy
de la Chapelle dysplasia
delta Thalassemia
diclofenac response - Toxicity/ADR
dyschromatosis
dystrophia
efavirenz response - Metabolism/PK

epileptic encephalopathy
erlotinib response - Efficacy
etanercept response - Efficacy
ethambutol
ethanol response - Toxicity/ADR
etoposide response - Toxicity/ADR
fentanyl response - Dosage
fluorouracil
fluorouracil response - Efficacy
fluorouracil response - Toxicity/ADR
furosemide and spironolactone response - Efficacy
gefitinib response - Efficacy
growth hormone deficiency with short stature
hormonal contraceptives for systemic use response - Toxicity/ADR
hydrochlorothiazide response - Efficacy
infliximab response - Efficacy
interferons
irinotecan response - Other
ivacaftor / lumacaftor response - Efficacy
ivacaftor response - Efficacy
meperidine response - Dosage
metformin response - Efficacy
mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
morphine response - Dosage
nevirapine response - Other
nevirapine response - Toxicity/ADR
nicotine response - Efficacy
nicotine response - Toxicity/ADR
not provided
not specified
olanzapine response - Toxicity/ADR
opioids response - Dosage
oxaliplatin response - Efficacy
p phenotype
paclitaxel response - Efficacy
partial sensorineural deafness
peginterferon alfa-2a
peginterferon alfa-2b and ribavirin response - Toxicity/ADR
peginterferon alfa-2b response - Efficacy
peginterferon alfa-2b response - Toxicity/ADR
pentazocine response - Dosage
phenprocoumon response - Dosage
platinum response - Efficacy
platinum response - Toxicity/ADR
radiotherapy response - Toxicity/ADR
ribavirin response - Efficacy
ribavirin response - Toxicity/ADR

risperidone response - Efficacy
risperidone response - Toxicity/ADR
rosiglitazone response - Dosage
rosuvastatin response - Efficacy
short QT syndrome
spino-cellular carcinoma
sporadic abdominal aortic aneurysm
sulfonamides
tacrolimus response - Dosage
tegafur response - Toxicity/ADR
trastuzumab response - Efficacy
ungueal dystrophy
von Willebrand disease
von Willebrand disease type 1
von Willebrand disease type 2
von Willebrand disease type 2M
von Willebrand disease type 2N
von Willebrand disease type 3
von Willebrand disorder
warfarin response - Dosage