|  |  |  |
| --- | --- | --- |
| [18p deletion syndrome](http://en.wikipedia.org/wiki/18p_deletion_syndrome) | D | 18p |
| [21-hydroxylase deficiency](http://en.wikipedia.org/wiki/21-hydroxylase_deficiency) |  | 6p21.3 |
| 45,X *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) | C | X |
| 47,XX,+21 *see* [Down syndrome](http://en.wikipedia.org/wiki/Down_syndrome) | C | 21 |
| 47,XXX *see* [triple X syndrome](http://en.wikipedia.org/wiki/Triple_X_syndrome) | C | X |
| 47,XXY *see* [Klinefelter's syndrome](http://en.wikipedia.org/wiki/Klinefelter%27s_syndrome) | C | X |
| 47,XY,+21 *see* [Down syndrome](http://en.wikipedia.org/wiki/Down_syndrome) | C | 21 |
| [47,XYY syndrome](http://en.wikipedia.org/wiki/47,XYY_syndrome) | C | Y |
| 5-ALA dehydratase-deficient porphyria *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| 5-aminolaevulinic dehydratase deficiency porphyria *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| 5p deletion syndrome *see* [Cri du chat](http://en.wikipedia.org/wiki/Cri_du_chat) | D | 5p |
| 5p- syndrome *see* [Cri du chat](http://en.wikipedia.org/wiki/Cri_du_chat) | D | 5p |
| A-T *see* [ataxia telangiectasia](http://en.wikipedia.org/wiki/Ataxia_telangiectasia) |  |  |
| AAT *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  |  |
| Absence of vas deferens *see* [congenital absence of the vas deferens](http://en.wikipedia.org/wiki/Congenital_absence_of_the_vas_deferens) |  |  |
| Absent vasa *see* [congenital absence of the vas deferens](http://en.wikipedia.org/wiki/Congenital_absence_of_the_vas_deferens) |  |  |
| [aceruloplasminemia](http://en.wikipedia.org/wiki/Aceruloplasminemia) |  |  |
| ACG2 *see* [achondrogenesis type II](http://en.wikipedia.org/wiki/Achondrogenesis_type_II) |  |  |
| ACH *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  |  |
| [Achondrogenesis type II](http://en.wikipedia.org/wiki/Achondrogenesis_type_II) |  |  |
| [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) | substitution | 4p16.3 |
| Acid beta-glucosidase deficiency *see* [Gaucher disease type 1](http://en.wikipedia.org/wiki/Gaucher_disease_type_1) |  |  |
| Acrocephalosyndactyly (Apert) *see* [Apert syndrome](http://en.wikipedia.org/wiki/Apert_syndrome) |  |  |
| acrocephalosyndactyly, type V *see* [Pfeiffer syndrome](http://en.wikipedia.org/wiki/Pfeiffer_syndrome) |  |  |
| Acrocephaly *see* [Apert syndrome](http://en.wikipedia.org/wiki/Apert_syndrome) |  |  |
| Acute cerebral Gaucher's disease *see* [Gaucher disease type 2](http://en.wikipedia.org/wiki/Gaucher_disease_type_2) |  |  |
| [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| ACY2 deficiency *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| AD *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| Adelaide-type craniosynostosis *see* [Muenke syndrome](http://en.wikipedia.org/wiki/Muenke_syndrome) |  |  |
| Adenomatous Polyposis Coli *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| Adenomatous Polyposis of the Colon *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| ADP *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| [adenylosuccinate lyase deficiency](http://en.wikipedia.org/wiki/Adenylosuccinate_lyase_deficiency) |  |  |
| Adrenal gland disorders *see* [21-hydroxylase deficiency](http://en.wikipedia.org/wiki/21-hydroxylase_deficiency) |  |  |
| Adrenogenital syndrome *see* [21-hydroxylase deficiency](http://en.wikipedia.org/wiki/21-hydroxylase_deficiency) |  |  |
| [Adrenoleukodystrophy](http://en.wikipedia.org/wiki/Adrenoleukodystrophy) |  |  |
| AIP *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| AIS *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  |  |
| AKU *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| ALA dehydratase porphyria *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| ALA-D porphyria *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| Alcaptonuria *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| Alkaptonuric ochronosis *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  |  |
| alpha-1 proteinase inhibitor *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  | 14q32.1 |
| alpha-1 related emphysema *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  | 14q32.1 |
| Alpha-galactosidase A deficiency *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) | P | Xq22.1 |
| ALS *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| [Alström syndrome](http://en.wikipedia.org/wiki/Alstr%C3%B6m_syndrome) |  |  |
| ALX *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| [Amelogenesis imperfecta](http://en.wikipedia.org/wiki/Amelogenesis_imperfecta) |  |  |
| Amino levulinic acid dehydratase deficiency *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| Aminoacylase 2 deficiency *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| Anderson-Fabry disease *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) | P | Xq22.1 |
| [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  |  |
| [Anemia](http://en.wikipedia.org/wiki/Anemia) |  |  |
| Anemia, hereditary sideroblastic *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  | X |
| Anemia, sex-linked hypochromic sideroblastic *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  | X |
| Anemia, splenic, familial *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| [Angelman syndrome](http://en.wikipedia.org/wiki/Angelman_syndrome) |  |  |
| Angiokeratoma Corporis Diffusum *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) | P | Xq22.1 |
| Angiokeratoma diffuse *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) |  |  |
| Angiomatosis retinae *see* [von Hippel–Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel%E2%80%93Lindau_disease) |  |  |
| ANH1 *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  | X |
| APC resistance, Leiden type *see* [factor V Leiden thrombophilia](http://en.wikipedia.org/wiki/Factor_V_Leiden_thrombophilia) |  |  |
| [Apert syndrome](http://en.wikipedia.org/wiki/Apert_syndrome) |  |  |
| AR deficiency *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  |  |
| AR-CMT2 *see* [Charcot-Marie-Tooth disease, type 2](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease,_type_2) |  |  |
| Arachnodactyly *see* [Marfan syndrome](http://en.wikipedia.org/wiki/Marfan_syndrome) |  |  |
| ARNSHL *see* [Nonsyndromic deafness#autosomal recessive](http://en.wikipedia.org/wiki/Nonsyndromic_deafness#autosomal_recessive) |  |  |
| Arthro-ophthalmopathy, hereditary progressive *see* [Stickler syndrome#COL2A1](http://en.wikipedia.org/wiki/Stickler_syndrome#COL2A1) |  |  |
| Arthrochalasis multiplex congenita *see* [Ehlers–Danlos syndrome#arthrochalasia type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#arthrochalasia_type) |  |  |
| AS *see* [Angelman syndrome](http://en.wikipedia.org/wiki/Angelman_syndrome) |  |  |
| Asp deficiency *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| Aspa deficiency *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| Aspartoacylase deficiency *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| [ataxia telangiectasia](http://en.wikipedia.org/wiki/Ataxia_telangiectasia) |  |  |
| Autism-Dementia-Ataxia-Loss of Purposeful Hand Use syndrome *see* [Rett syndrome](http://en.wikipedia.org/wiki/Rett_syndrome) |  |  |
| autosomal dominant juvenile ALS *see* [amyotrophic lateral sclerosis, type 4](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis,_type_4) |  |  |
| Autosomal dominant opitz G/BBB syndrome *see* [22q11.2 deletion syndrome](http://en.wikipedia.org/wiki/22q11.2_deletion_syndrome) | D | 22q |
| autosomal recessive form of juvenile ALS type 3 *see* [Amyotrophic lateral sclerosis#type 2](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis#type_2) |  |  |
| Autosomal recessive nonsyndromic hearing loss *see* [Nonsyndromic deafness#autosomal recessive](http://en.wikipedia.org/wiki/Nonsyndromic_deafness#autosomal_recessive) |  |  |
| Autosomal Recessive Sensorineural Hearing Impairment and Goiter *see* [Pendred syndrome](http://en.wikipedia.org/wiki/Pendred_syndrome) |  |  |
| AxD *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| Ayerza syndrome *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| B variant of the Hexosaminidase GM2 gangliosidosis *see* [Sandhoff disease](http://en.wikipedia.org/wiki/Sandhoff_disease) |  |  |
| BANF *see* [neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II) |  |  |
| [Beare-Stevenson cutis gyrata syndrome](http://en.wikipedia.org/w/index.php?title=Beare-Stevenson_cutis_gyrata_syndrome&action=edit&redlink=1) |  | 10q26 |
| Benign paroxysmal peritonitis *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| [Benjamin syndrome](http://en.wikipedia.org/wiki/Benjamin_syndrome) |  |  |
| [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| BH4 Deficiency *see* [tetrahydrobiopterin deficiency](http://en.wikipedia.org/wiki/Tetrahydrobiopterin_deficiency) |  |  |
| Bilateral Acoustic Neurofibromatosis *see* [neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II) |  |  |
| [biotinidase deficiency](http://en.wikipedia.org/wiki/Biotinidase_deficiency) |  |  |
| [bladder cancer](http://en.wikipedia.org/wiki/Bladder_cancer) |  |  |
| Bleeding disorders *see* [factor V Leiden thrombophilia](http://en.wikipedia.org/wiki/Factor_V_Leiden_thrombophilia) |  |  |
| Bloch-Sulzberger syndrome *see* [incontinentia pigmenti](http://en.wikipedia.org/wiki/Incontinentia_pigmenti) |  |  |
| [Bloom syndrome](http://en.wikipedia.org/wiki/Bloom_syndrome) |  | 15q26.1 |
| [Bone diseases](http://en.wikipedia.org/wiki/Bone_diseases) |  |  |
| Bone marrow diseases *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| Bonnevie-Ullrich syndrome *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| Bourneville disease *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| Bourneville phakomatosis *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| Brain diseases *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) |  |  |
| [Birt–Hogg–Dubé syndrome](http://en.wikipedia.org/wiki/Birt%E2%80%93Hogg%E2%80%93Dub%C3%A9_syndrome) |  | 17 |
| Brittle bone disease *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| Broad Thumb-Hallux syndrome *see* [Rubinstein-Taybi syndrome](http://en.wikipedia.org/wiki/Rubinstein-Taybi_syndrome) |  |  |
| Bronze Diabetes *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| Bronzed cirrhosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| Bulbospinal muscular atrophy, X-linked *see* [Kennedy's disease](http://en.wikipedia.org/wiki/Kennedy%27s_disease) |  |  |
| Burger-Grutz syndrome *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) | P | 3 |
| CGD [Chronic granulomatous disorder](http://en.wikipedia.org/wiki/Chronic_granulomatous_disorder) |  |  |
| [Campomelic dysplasia](http://en.wikipedia.org/wiki/Campomelic_dysplasia) | C | 17q24.3-q25.1 |
| [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| [Cancer](http://en.wikipedia.org/wiki/Cancer) |  |  |
| Cancer Family syndrome *see* [hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) |  |  |
| Cancer of breast *see* [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) [[1]](http://en.wikipedia.org/wiki/List_of_genetic_disorders#cite_note-0) |  |  |
| Cancer of the bladder *see* [bladder cancer](http://en.wikipedia.org/wiki/Bladder_cancer) |  |  |
| Carboxylase Deficiency, Multiple, Late-Onset *see* [biotinidase deficiency](http://en.wikipedia.org/wiki/Biotinidase_deficiency) | P | 3 |
| Cardiomyopathy *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Cat cry syndrome *see* [Cri du chat](http://en.wikipedia.org/wiki/Cri_du_chat) |  |  |
| CAVD *see* [congenital absence of the vas deferens](http://en.wikipedia.org/wiki/Congenital_absence_of_the_vas_deferens) |  |  |
| Caylor cardiofacial syndrome *see* [22q11.2 deletion syndrome](http://en.wikipedia.org/wiki/22q11.2_deletion_syndrome) | D | 22q |
| CBAVD *see* [congenital absence of the vas deferens](http://en.wikipedia.org/wiki/Congenital_absence_of_the_vas_deferens) |  |  |
| [Celiac Disease](http://en.wikipedia.org/wiki/Coeliac_disease) |  |  |
| CEP *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| Ceramide trihexosidase deficiency *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) |  | X |
| Cerebelloretinal Angiomatosis, familial *see* [von Hippel-Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel-Lindau_disease) | P | 3 (p26-p25) |
| Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) | P | 3 |
| Cerebral autosomal dominant ateriopathy with subcortical infarcts and leukoencephalopathy *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) | P | 3 |
| Cerebral sclerosis *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  | 9 (q34), 16 (p13.3) |
| Cerebroatrophic Hyperammonemia *see* [Rett syndrome](http://en.wikipedia.org/wiki/Rett_syndrome) |  | X |
| Cerebroside Lipidosis syndrome *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) | P | 1(q21) |
| CF *see* [cystic fibrosis](http://en.wikipedia.org/wiki/Cystic_fibrosis) [[2]](http://en.wikipedia.org/wiki/List_of_genetic_disorders#cite_note-1) | D (most common); or substitution | CFTR (7q31.2) |
| CH *see* [congenital hypothyroidism](http://en.wikipedia.org/wiki/Congenital_hypothyroidism) |  |  |
| Charcot disease *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| Chondrodystrophia *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  |  |
| Chondrodystrophy syndrome *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  |  |
| Chondrodystrophy with sensorineural deafness *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| Chondrogenesis imperfecta *see* [achondrogenesis, type II](http://en.wikipedia.org/wiki/Achondrogenesis,_type_II) |  |  |
| Choreoathetosis self-mutilation hyperuricemia syndrome *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) | P | X |
| Classic Galactosemia *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) | P | 9 (p13) |
| Classical Ehlers–Danlos syndrome *see* [Ehlers–Danlos syndrome#classical type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#classical_type) |  |  |
| Classical Phenylketonuria *see* [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) |  |  |
| Cleft lip and palate *see* [Stickler syndrome](http://en.wikipedia.org/wiki/Stickler_syndrome) |  |  |
| Cloverleaf skull with thanatophoric dwarfism *see* [Thanatophoric dysplasia#type 2](http://en.wikipedia.org/wiki/Thanatophoric_dysplasia#type_2) |  |  |
| CLS *see* [Coffin-Lowry syndrome](http://en.wikipedia.org/wiki/Coffin-Lowry_syndrome) |  |  |
| CMT *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) |  |  |
| [Coffin-Lowry syndrome](http://en.wikipedia.org/wiki/Coffin-Lowry_syndrome) |  |  |
| [collagenopathy, types II and XI](http://en.wikipedia.org/wiki/Collagenopathy,_types_II_and_XI) |  |  |
| Colon Cancer, familial Nonpolyposis *see* [hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) |  |  |
| Colon cancer, familial *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| [Colorectal cancer](http://en.wikipedia.org/wiki/Colorectal_cancer) [[3]](http://en.wikipedia.org/wiki/List_of_genetic_disorders#cite_note-2) |  |  |
| Complete HPRT deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| Complete hypoxanthine-guanine phosphoribosyltransferase deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| Compression neuropathy *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| Congenital adrenal hyperplasia *see* [21-hydroxylase deficiency](http://en.wikipedia.org/wiki/21-hydroxylase_deficiency) |  |  |
| congenital bilateral absence of vas deferens *see* [Congenital absence of the vas deferens](http://en.wikipedia.org/wiki/Congenital_absence_of_the_vas_deferens) |  |  |
| [Congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| [Congenital heart disease](http://en.wikipedia.org/wiki/Congenital_heart_disease) |  |  |
| Congenital hypomyelination *see* [Charcot-Marie-Tooth disease#Type 1](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease#Type_1) *see* [Charcot-Marie-Tooth disease#Type 4](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease#Type_4) |  |  |
| [Congenital hypothyroidism](http://en.wikipedia.org/wiki/Congenital_hypothyroidism) |  |  |
| Congenital methemoglobinemia *see* [Methemoglobinemia#Congenital methaemoglobinaemia](http://en.wikipedia.org/wiki/Methemoglobinemia#Congenital_methaemoglobinaemia) |  |  |
| Congenital osteosclerosis *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  |  |
| Congenital sideroblastic anaemia *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  | X |
| [Connective tissue disease](http://en.wikipedia.org/wiki/Connective_tissue_disease) |  |  |
| Conotruncal anomaly face syndrome *see* [22q11.2 deletion syndrome](http://en.wikipedia.org/wiki/22q11.2_deletion_syndrome) | D | 22q |
| Cooley's Anemia *see* [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| Copper storage disease *see* [Wilson's disease](http://en.wikipedia.org/wiki/Wilson%27s_disease) |  | 13 (q14.3) |
| Copper transport disease *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| Coproporphyria, hereditary *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| Coproporphyrinogen oxidase deficiency *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| [Cowden syndrome](http://en.wikipedia.org/wiki/Cowden_syndrome) |  |  |
| CPO deficiency *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| CPRO deficiency *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| CPX deficiency *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| Craniofacial dysarthrosis *see* [Crouzon syndrome](http://en.wikipedia.org/wiki/Crouzon_syndrome) |  |  |
| Craniofacial Dysostosis *see* [Crouzon syndrome](http://en.wikipedia.org/wiki/Crouzon_syndrome) |  |  |
| Cretinism *see* [congenital hypothyroidism](http://en.wikipedia.org/wiki/Congenital_hypothyroidism) |  |  |
| Creutzfeldt-Jakob disease *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| [Cri du chat](http://en.wikipedia.org/wiki/Cri_du_chat) | D | 5p |
| [Crohn's disease](http://en.wikipedia.org/wiki/Crohn%27s_disease), [fibrostenosing](http://en.wikipedia.org/w/index.php?title=Fibrostenosing&action=edit&redlink=1) | P | 16q12 |
| [Crouzon syndrome](http://en.wikipedia.org/wiki/Crouzon_syndrome) |  | FGFR2 (10q25.3-q26) |
| Crouzon syndrome with acanthosis nigricans *see* [Crouzonodermoskeletal syndrome](http://en.wikipedia.org/wiki/Crouzonodermoskeletal_syndrome) |  |  |
| [Crouzonodermoskeletal syndrome](http://en.wikipedia.org/wiki/Crouzonodermoskeletal_syndrome) |  |  |
| CS *see* [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) *see* [Cowden syndrome](http://en.wikipedia.org/wiki/Cowden_syndrome) |  |  |
| Curschmann-Batten-Steinert syndrome *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| cutis gyrata syndrome of Beare-Stevenson *see* [Beare-Stevenson cutis gyrata syndrome](http://en.wikipedia.org/w/index.php?title=Beare-Stevenson_cutis_gyrata_syndrome&action=edit&redlink=1) |  |  |
| D-glycerate dehydrogenase deficiency *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Primary_hyperoxaluria) |  |  |
| Dappled metaphysis syndrome *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| DAT - Dementia Alzheimer's type *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| Genetic hypercalciuria *see* [Dent's disease](http://en.wikipedia.org/wiki/Dent%27s_disease) |  | Xp11.22 |
| DBMD *see* [muscular dystrophy, Duchenne and Becker types](http://en.wikipedia.org/wiki/Muscular_dystrophy) |  |  |
| Deafness with goiter *see* [Pendred syndrome](http://en.wikipedia.org/wiki/Pendred_syndrome) |  |  |
| Deafness-retinitis pigmentosa syndrome *see* [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| Deficiency disease, Phenylalanine Hydroxylase *see* [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) | P | 12q |
| [Degenerative nerve diseases](http://en.wikipedia.org/wiki/Degenerative_nerve_diseases) |  |  |
| de Grouchy syndrome 1 *see* [De Grouchy syndrome](http://en.wikipedia.org/wiki/De_Grouchy_syndrome) | D | 18p |
| Dejerine-Sottas syndrome *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| Delta-aminolevulinate dehydratase deficiency porphyria *see* [ALA dehydratase deficiency](http://en.wikipedia.org/wiki/ALA_dehydratase_deficiency) |  |  |
| Dementia *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) |  |  |
| demyelinogenic leukodystrophy *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| Dermatosparactic type of Ehlers–Danlos syndrome *see* [Ehlers–Danlos syndrome#dermatosparaxis type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#dermatosparaxis_type) |  |  |
| Dermatosparaxis *see* [Ehlers–Danlos syndrome#dermatosparaxis type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#dermatosparaxis_type) |  |  |
| [developmental disabilities](http://en.wikipedia.org/wiki/Developmental_disabilities) |  |  |
| dHMN *see* [Amyotrophic lateral sclerosis#type 4](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis#type_4) |  |  |
| DHMN-V *see* [distal spinal muscular atrophy, type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy,_type_V&action=edit&redlink=1) |  |  |
| DHTR deficiency *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  | X |
| Diffuse Globoid Body Sclerosis *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| [Di George's syndrome](http://en.wikipedia.org/wiki/Di_George%27s_syndrome) | D | 22q |
| Dihydrotestosterone receptor deficiency *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  | X |
| [distal spinal muscular atrophy, type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy,_type_V&action=edit&redlink=1) |  |  |
| DM1 *see* [Myotonic dystrophy#type 1](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_1) | T | 19 |
| DM2 *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) | T | 3 |
| [Down syndrome](http://en.wikipedia.org/wiki/Down_syndrome) |  | 21 |
| DSMAV *see* [distal spinal muscular atrophy, type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy,_type_V&action=edit&redlink=1) |  |  |
| DSN *see* [Charcot-Marie-Tooth disease#type 4](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease#type_4) |  |  |
| DSS *see* [Charcot-Marie-Tooth disease, type 4](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease,_type_4) |  |  |
| Duchenne/Becker muscular dystrophy *see* [Muscular dystrophy, Duchenne and Becker type](http://en.wikipedia.org/wiki/Muscular_dystrophy,_Duchenne_and_Becker_type) |  |  |
| Dwarf, achondroplastic *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  | 3 |
| Dwarf, thanatophoric *see* [thanatophoric dysplasia](http://en.wikipedia.org/wiki/Thanatophoric_dysplasia) |  |  |
| [Dwarfism](http://en.wikipedia.org/wiki/Dwarfism) |  |  |
| Dwarfism-retinal atrophy-deafness syndrome *see* [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) |  |  |
| dysmyelinogenic leukodystrophy *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| Dystrophia myotonica *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) | T | 19 |
| dystrophia retinae pigmentosa-dysostosis syndrome *see* [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| Early-Onset familial alzheimer disease (EOFAD) *see* [Alzheimer disease#type 1](http://en.wikipedia.org/wiki/Alzheimer_disease#type_1) *see* [Alzheimer disease#type 3](http://en.wikipedia.org/wiki/Alzheimer_disease#type_3) *see* [Alzheimer disease#type 4](http://en.wikipedia.org/wiki/Alzheimer_disease#type_4) |  |  |
| EDS *see* [Ehlers–Danlos syndrome](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome) |  |  |
| [Ehlers–Danlos syndrome](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome) |  |  |
| Ekman-Lobstein disease *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| Entrapment neuropathy *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| Epiloia *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| EPP *see* [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| Erythroblastic anemia *see* [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| Erythrohepatic protoporphyria *see* [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| Erythroid 5-aminolevulinate synthetase deficiency *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| Erythropoietic porphyria *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| Erythropoietic uroporphyria *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| Eye cancer *see* [retinoblastoma FA - Friedreich ataxia](http://en.wikipedia.org/w/index.php?title=Retinoblastoma_FA_-_Friedreich_ataxia&action=edit&redlink=1) *see* [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| FA *see* [fanconi anemia](http://en.wikipedia.org/wiki/Fanconi_anemia) |  |  |
| [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) | P | Xq22.1 |
| [Facial injuries and disorders](http://en.wikipedia.org/wiki/Facial_injuries_and_disorders) |  |  |
| [factor V Leiden thrombophilia](http://en.wikipedia.org/wiki/Factor_V_Leiden_thrombophilia) |  |  |
| FALS *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| familial acoustic neuroma *see* [neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II) |  |  |
| [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| familial Alzheimer disease (FAD) *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| familial amyotrophic lateral sclerosis *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| familial fat-induced hypertriglyceridemia *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| familial hemochromatosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| familial LPL deficiency *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| familial nonpolyposis colon cancer *see* [hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) |  |  |
| familial paroxysmal polyserositis *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| familial PCT *see* [porphyria cutanea tarda](http://en.wikipedia.org/wiki/Porphyria_cutanea_tarda) |  |  |
| familial pressure sensitive neuropathy *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| familial primary pulmonary hypertension (FPPH) *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| Familial Turner syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| familial vascular leukoencephalopathy *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) |  |  |
| FAP *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| FD *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| Female pseudo-Turner syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Ferrochelatase deficiency *see* [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| ferroportin disease *see* [Haemochromatosis#type 4](http://en.wikipedia.org/wiki/Haemochromatosis#type_4) |  |  |
| Fever *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| [FG syndrome](http://en.wikipedia.org/wiki/FG_syndrome) |  |  |
| FGFR3-associated coronal synostosis *see* [Muenke syndrome](http://en.wikipedia.org/wiki/Muenke_syndrome) |  |  |
| Fibrinoid degeneration of astrocytes *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| Fibrocystic disease of the pancreas *see* [cystic fibrosis](http://en.wikipedia.org/wiki/Cystic_fibrosis) |  |  |
| FMF *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| Folling disease *see* [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) |  |  |
| fra(X) syndrome *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  | Xq27.3 |
| [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  | Xq27.3 |
| Fragilitas ossium *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| FRAXA syndrome *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  | Xq27.3 |
| FRDA *see* [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) *see* [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| FXS *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  | Xq27.3 |
| [G6PD deficiency](http://en.wikipedia.org/wiki/G6PD_deficiency) |  |  |
| Galactokinase deficiency disease *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| Galactose-1-phosphate uridyl-transferase deficiency disease *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| Galactosylceramidase deficiency disease *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| Galactosylceramide lipidosis *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| galactosylcerebrosidase deficiency *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| galactosylsphingosine lipidosis *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| GALC deficiency *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| GALT deficiency *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Gaucher-like disease *see* [pseudo-Gaucher disease](http://en.wikipedia.org/wiki/Pseudo-Gaucher_disease) |  |  |
| GBA deficiency *see* [Gaucher disease type 1](http://en.wikipedia.org/wiki/Gaucher_disease_type_1) |  |  |
| GD *see* [Gaucher's disease](http://en.wikipedia.org/wiki/Gaucher%27s_disease) |  |  |
| [Genetic brain disorders](http://en.wikipedia.org/wiki/Genetic_brain_disorders) |  |  |
| genetic emphysema *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  |  |
| genetic hemochromatosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| Giant cell hepatitis, neonatal *see* [Neonatal hemochromatosis](http://en.wikipedia.org/wiki/Neonatal_hemochromatosis) |  |  |
| GLA deficiency *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) |  |  |
| Glioblastoma, retinal *see* [retinoblastoma](http://en.wikipedia.org/wiki/Retinoblastoma) |  |  |
| Glioma, retinal *see* [retinoblastoma](http://en.wikipedia.org/wiki/Retinoblastoma) |  |  |
| globoid cell leukodystrophy (GCL, GLD) *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| globoid cell leukoencephalopathy *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| Glucocerebrosidase deficiency *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glucocerebrosidosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glucosyl cerebroside lipidosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glucosylceramidase deficiency *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glucosylceramide beta-glucosidase deficiency *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glucosylceramide lipidosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Glyceric aciduria *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| Glycine encephalopathy *see* [Nonketotic hyperglycinemia](http://en.wikipedia.org/wiki/Nonketotic_hyperglycinemia) |  |  |
| Glycolic aciduria *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| GM2 gangliosidosis, type 1 *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| Goiter-deafness syndrome *see* [Pendred syndrome](http://en.wikipedia.org/wiki/Pendred_syndrome) |  |  |
| Graefe-Usher syndrome *see* [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| Gronblad-Strandberg syndrome *see* [pseudoxanthoma elasticum](http://en.wikipedia.org/wiki/Pseudoxanthoma_elasticum) |  |  |
| Guenther porphyria *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| Gunther disease *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| Haemochromatosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| Hallgren syndrome *see* [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| [Harlequin type ichthyosis](http://en.wikipedia.org/wiki/Harlequin_type_ichthyosis) |  |  |
| Hb S disease *see* [sickle cell anemia](http://en.wikipedia.org/wiki/Sickle_cell_anemia) |  |  |
| HCH *see* [hypochondroplasia](http://en.wikipedia.org/wiki/Hypochondroplasia) |  |  |
| HCP *see* [hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) |  |  |
| [Head and brain malformations](http://en.wikipedia.org/w/index.php?title=Head_and_brain_malformations&action=edit&redlink=1) |  |  |
| [Hearing disorders and deafness](http://en.wikipedia.org/w/index.php?title=Hearing_disorders_and_deafness&action=edit&redlink=1) |  |  |
| [Hearing problems in children](http://en.wikipedia.org/w/index.php?title=Hearing_problems_in_children&action=edit&redlink=1) |  |  |
| HEF2A *see* [hemochromatosis#type 2](http://en.wikipedia.org/wiki/Hemochromatosis#type_2) |  |  |
| HEF2B *see* [hemochromatosis#type 2](http://en.wikipedia.org/wiki/Hemochromatosis#type_2) |  |  |
| Hematoporphyria *see* [porphyria](http://en.wikipedia.org/wiki/Porphyria) |  |  |
| Heme synthetase deficiency *see* [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| Hemochromatoses *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| hemoglobin M disease *see* [methemoglobinemia#beta-globin type](http://en.wikipedia.org/wiki/Methemoglobinemia#beta-globin_type) |  |  |
| Hemoglobin S disease *see* [sickle cell anemia](http://en.wikipedia.org/wiki/Sickle_cell_anemia) |  |  |
| [hemophilia](http://en.wikipedia.org/wiki/Hemophilia) |  |  |
| HEP *see* [hepatoerythropoietic porphyria](http://en.wikipedia.org/wiki/Hepatoerythropoietic_porphyria) |  |  |
| hepatic AGT deficiency *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| [hepatoerythropoietic porphyria](http://en.wikipedia.org/wiki/Hepatoerythropoietic_porphyria) |  |  |
| Hepatolenticular degeneration syndrome *see* [Wilson disease](http://en.wikipedia.org/wiki/Wilson_disease) |  |  |
| Hereditary arthro-ophthalmopathy *see* [Stickler syndrome](http://en.wikipedia.org/wiki/Stickler_syndrome) |  |  |
| [Hereditary coproporphyria](http://en.wikipedia.org/wiki/Hereditary_coproporphyria) | P | 3q12 |
| Hereditary dystopic lipidosis *see* [Fabry disease](http://en.wikipedia.org/wiki/Fabry_disease) |  |  |
| Hereditary hemochromatosis (HHC) *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) [[4]](http://en.wikipedia.org/wiki/List_of_genetic_disorders#cite_note-3) |  |  |
| [Hereditary hemorrhagic telangiectasia](http://en.wikipedia.org/wiki/Hereditary_hemorrhagic_telangiectasia) (HHT) |  |  |
| Hereditary Inclusion Body Myopathy *see* [skeletal muscle regeneration](http://en.wikipedia.org/w/index.php?title=Skeletal_muscle_regeneration&action=edit&redlink=1) |  |  |
| Hereditary iron-loading anemia *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| Hereditary motor and sensory neuropathy *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| Hereditary motor neuronopathy *see* [spinal muscular atrophy](http://en.wikipedia.org/wiki/Spinal_muscular_atrophy) |  |  |
| Hereditary motor neuronopathy, type V *see* [distal spinal muscular atrophy, type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy,_type_V&action=edit&redlink=1) |  |  |
| [Hereditary multiple exostoses](http://en.wikipedia.org/wiki/Hereditary_multiple_exostoses) |  |  |
| [Hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) | DNA mismatch repair dysfunction usually in [MSH2](http://en.wikipedia.org/wiki/MSH2) and MLH1 genes | usually chromosomes 2 and 3 |
| Hereditary periodic fever syndrome *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| Hereditary Polyposis Coli *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| Hereditary pulmonary emphysema *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  |  |
| Hereditary resistance to activated protein C *see* [factor V Leiden thrombophilia](http://en.wikipedia.org/wiki/Factor_V_Leiden_thrombophilia) |  |  |
| Hereditary sensory and autonomic neuropathy type III *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| Hereditary spastic paraplegia *see* [infantile-onset ascending hereditary spastic paralysis](http://en.wikipedia.org/w/index.php?title=Infantile-onset_ascending_hereditary_spastic_paralysis&action=edit&redlink=1) |  |  |
| Hereditary spinal ataxia *see* [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| Hereditary spinal sclerosis *see* [Friedreich's ataxia](http://en.wikipedia.org/wiki/Friedreich%27s_ataxia) |  |  |
| Herrick's anemia *see* [sickle cell anemia](http://en.wikipedia.org/wiki/Sickle_cell_anemia) |  |  |
| Heterozygous OSMED *see* [Weissenbacher-Zweymüller syndrome](http://en.wikipedia.org/wiki/Weissenbacher-Zweym%C3%BCller_syndrome) |  |  |
| Heterozygous otospondylomegaepiphyseal dysplasia *see* [Weissenbacher-Zweymüller syndrome](http://en.wikipedia.org/wiki/Weissenbacher-Zweym%C3%BCller_syndrome) |  |  |
| HexA deficiency *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| Hexosaminidase A deficiency *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| Hexosaminidase alpha-subunit deficiency (variant B) *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| HFE-associated hemochromatosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| HGPS *see* [Progeria](http://en.wikipedia.org/wiki/Progeria) |  |  |
| Hippel-Lindau disease *see* [von Hippel-Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel-Lindau_disease) |  |  |
| HLAH *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| HMN V *see* [distal spinal muscular atrophy, type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy,_type_V&action=edit&redlink=1) |  |  |
| HMSN *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| HNPCC *see* [hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) |  |  |
| HNPP *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| [homocystinuria](http://en.wikipedia.org/wiki/Homocystinuria) |  |  |
| Homogentisic acid oxidase deficiency *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| Homogentisic acidura *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| Homozygous porphyria cutanea tarda *see* [hepatoerythropoietic porphyria](http://en.wikipedia.org/wiki/Hepatoerythropoietic_porphyria) |  |  |
| HP1 *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| HP2 *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| HPA *see* [hyperphenylalaninemia](http://en.wikipedia.org/wiki/Hyperphenylalaninemia) |  |  |
| HPRT - Hypoxanthine-guanine phosphoribosyltransferase deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| HSAN type III *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| HSAN3 *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| HSN-III *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| Human dermatosparaxis *see* [Ehlers–Danlos syndrome#dermatosparaxis type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#dermatosparaxis_type) |  |  |
| [Huntington's disease](http://en.wikipedia.org/wiki/Huntington%27s_disease) | T | 4p16.3 |
| Hutchinson-Gilford progeria syndrome *see* [progeria](http://en.wikipedia.org/wiki/Progeria) |  |  |
| Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency *see* [21-hydroxylase deficiency](http://en.wikipedia.org/wiki/21-hydroxylase_deficiency) |  |  |
| Hyperchylomicronemia, familial *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| Hyperglycinemia with ketoacidosis and leukopenia *see* [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| Hyperlipoproteinemia type I *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| hyperphenylalaninaemia *see* [hyperphenylalaninemia](http://en.wikipedia.org/wiki/Hyperphenylalaninemia) |  |  |
| [hyperphenylalaninemia](http://en.wikipedia.org/wiki/Hyperphenylalaninemia) |  |  |
| Hypochondrodysplasia *see* [hypochondroplasia](http://en.wikipedia.org/wiki/Hypochondroplasia) |  |  |
| [Hypochondrogenesis](http://en.wikipedia.org/wiki/Hypochondrogenesis) |  |  |
| [Hypochondroplasia](http://en.wikipedia.org/wiki/Hypochondroplasia) |  | 4p16.3 |
| Hypochromic anemia *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| Hypocupremia, congenital *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| Hypoxanthine phosphoribosyltransferse (HPRT) deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| IAHSP *see* [infantile-onset ascending hereditary spastic paralysis](http://en.wikipedia.org/w/index.php?title=Infantile-onset_ascending_hereditary_spastic_paralysis&action=edit&redlink=1) |  |  |
| ICF syndrome *see* [Immunodeficiency, centromere instability and facial anomalies syndrome](http://en.wikipedia.org/wiki/Immunodeficiency,_centromere_instability_and_facial_anomalies_syndrome) |  | 20q11.2 |
| Idiopathic hemochromatosis *see* [hemochromatosis, type 3](http://en.wikipedia.org/w/index.php?title=Hemochromatosis,_type_3&action=edit&redlink=1) |  |  |
| Idiopathic neonatal hemochromatosis *see* [hemochromatosis, neonatal](http://en.wikipedia.org/w/index.php?title=Hemochromatosis,_neonatal&action=edit&redlink=1) |  |  |
| Idiopathic pulmonary hypertension *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| Immune system disorders *see* [X-linked severe combined immunodeficiency](http://en.wikipedia.org/wiki/X-linked_severe_combined_immunodeficiency) |  |  |
| [Incontinentia pigmenti](http://en.wikipedia.org/wiki/Incontinentia_pigmenti) | P | Xq28 |
| Infantile cerebral Gaucher's disease *see* [Gaucher disease type 2](http://en.wikipedia.org/wiki/Gaucher_disease_type_2) |  |  |
| Infantile Gaucher disease *see* [Gaucher disease type 2](http://en.wikipedia.org/wiki/Gaucher_disease_type_2) |  |  |
| [infantile-onset ascending hereditary spastic paralysis](http://en.wikipedia.org/w/index.php?title=Infantile-onset_ascending_hereditary_spastic_paralysis&action=edit&redlink=1) |  |  |
| [Infertility](http://en.wikipedia.org/wiki/Infertility) |  |  |
| inherited emphysema *see* [alpha 1-antitrypsin deficiency](http://en.wikipedia.org/wiki/Alpha_1-antitrypsin_deficiency) |  |  |
| Inherited human transmissible spongiform encephalopathies *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| inherited tendency to pressure palsies *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| Insley-Astley syndrome *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| Intermittent acute porphyria syndrome *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| Intestinal polyposis-cutaneous pigmentation syndrome *see* [Peutz–Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz%E2%80%93Jeghers_syndrome) |  |  |
| IP *see* [incontinentia pigmenti](http://en.wikipedia.org/wiki/Incontinentia_pigmenti) |  |  |
| Iron storage disorder *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| Isodicentric 15 *see* [isodicentric 15](http://en.wikipedia.org/wiki/Isodicentric_15) | Inv dup | 15q11-14 |
| Isolated deafness *see* [nonsyndromic deafness](http://en.wikipedia.org/wiki/Nonsyndromic_deafness) |  |  |
| [Jackson-Weiss syndrome](http://en.wikipedia.org/wiki/Jackson-Weiss_syndrome) |  |  |
| JH *see* [Haemochromatosis#type 2](http://en.wikipedia.org/wiki/Haemochromatosis#type_2) |  |  |
| [Joubert syndrome](http://en.wikipedia.org/wiki/Joubert_syndrome) |  |  |
| JPLS *see* [Juvenile Primary Lateral Sclerosis](http://en.wikipedia.org/wiki/Juvenile_Primary_Lateral_Sclerosis) |  | [ALS2](http://en.wikipedia.org/wiki/ALS2) |
| juvenile amyotrophic lateral sclerosis *see* [Amyotrophic lateral sclerosis#type 2](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis#type_2) |  |  |
| Juvenile gout, choreoathetosis, mental retardation syndrome *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| juvenile hyperuricemia syndrome *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| JWS *see* [Jackson-Weiss syndrome](http://en.wikipedia.org/wiki/Jackson-Weiss_syndrome) |  |  |
| KD *see* [X-linked spinal-bulbar muscle atrophy](http://en.wikipedia.org/wiki/X-linked_spinal-bulbar_muscle_atrophy) |  |  |
| Kennedy disease *see* [X-linked spinal-bulbar muscle atrophy](http://en.wikipedia.org/wiki/X-linked_spinal-bulbar_muscle_atrophy) |  |  |
| Kennedy spinal and bulbar muscular atrophy *see* [X-linked spinal-bulbar muscle atrophy](http://en.wikipedia.org/wiki/X-linked_spinal-bulbar_muscle_atrophy) |  |  |
| Kerasin histiocytosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Kerasin lipoidosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Kerasin thesaurismosis *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| ketotic glycinemia *see* [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| ketotic hyperglycinemia *see* [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| Kidney diseases *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| [Klinefelter's syndrome](http://en.wikipedia.org/wiki/Klinefelter%27s_syndrome) |  |  |
| Klinefelter's syndrome *see* [Klinefelter's syndrome](http://en.wikipedia.org/wiki/Klinefelter%27s_syndrome) |  |  |
| [Kniest dysplasia](http://en.wikipedia.org/wiki/Kniest_dysplasia) |  |  |
| [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| Lacunar dementia *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) |  |  |
| Langer-Saldino achondrogenesis *see* [achondrogenesis, type II](http://en.wikipedia.org/wiki/Achondrogenesis,_type_II) |  |  |
| Langer-Saldino dysplasia *see* [achondrogenesis, type II](http://en.wikipedia.org/wiki/Achondrogenesis,_type_II) |  |  |
| Late-onset Alzheimer disease *see* [Alzheimer disease#type 2](http://en.wikipedia.org/wiki/Alzheimer_disease#type_2) |  |  |
| Late-onset familial Alzheimer disease (AD2) *see* [Alzheimer disease#type 2](http://en.wikipedia.org/wiki/Alzheimer_disease#type_2) |  |  |
| late-onset Krabbe disease (LOKD) *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| [Learning Disorders](http://en.wikipedia.org/wiki/Learning_disorders) *see* [Learning disability](http://en.wikipedia.org/wiki/Learning_disability) |  |  |
| Lentiginosis, perioral *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| [Leukodystrophies](http://en.wikipedia.org/wiki/Leukodystrophies) |  |  |
| leukodystrophy with Rosenthal fibers *see* [Alexander disease](http://en.wikipedia.org/wiki/Alexander_disease) |  |  |
| Leukodystrophy, spongiform *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| LFS *see* [Li-Fraumeni syndrome](http://en.wikipedia.org/wiki/Li-Fraumeni_syndrome) |  |  |
| [Li-Fraumeni syndrome](http://en.wikipedia.org/wiki/Li-Fraumeni_syndrome) |  |  |
| Lipase D deficiency *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| LIPD deficiency *see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| Lipidosis, cerebroside *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| Lipidosis, ganglioside, infantile *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| Lipoid histiocytosis (kerasin type) *see* [Gaucher disease](http://en.wikipedia.org/wiki/Gaucher_disease) |  |  |
| [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency,_familial) |  |  |
| Liver diseases *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| Lou Gehrig disease *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| Louis-Bar syndrome *see* [ataxia telangiectasia](http://en.wikipedia.org/wiki/Ataxia_telangiectasia) |  |  |
| Lynch syndrome *see* [hereditary nonpolyposis colorectal cancer](http://en.wikipedia.org/wiki/Hereditary_nonpolyposis_colorectal_cancer) |  |  |
| Lysyl-hydroxylase deficiency *see* [Ehlers–Danlos syndrome#kyphoscoliosis type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#kyphoscoliosis_type) |  |  |
| Machado-Joseph disease *see* [Spinocerebellar ataxia#type 3](http://en.wikipedia.org/wiki/Spinocerebellar_ataxia#type_3) |  |  |
| Male breast cancer *see* [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) |  |  |
| [Male genital disorders](http://en.wikipedia.org/w/index.php?title=Male_genital_disorders&action=edit&redlink=1) |  |  |
| Male Turner syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Malignant neoplasm of breast *see* [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) |  |  |
| malignant tumor of breast *see* [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) |  |  |
| Malignant tumor of urinary bladder *see* [bladder cancer](http://en.wikipedia.org/wiki/Bladder_cancer) |  |  |
| Mammary cancer *see* [breast cancer](http://en.wikipedia.org/wiki/Breast_cancer) |  |  |
| [Marfan syndrome](http://en.wikipedia.org/wiki/Marfan_syndrome) |  | 15 |
| Marker X syndrome *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  |  |
| Martin-Bell syndrome *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  |  |
| [McCune–Albright syndrome](http://en.wikipedia.org/wiki/McCune%E2%80%93Albright_syndrome) |  | 20 q13.2-13.3 |
| [McLeod syndrome](http://en.wikipedia.org/wiki/McLeod_syndrome) |  | X |
| MEDNIK [[5]](http://en.wikipedia.org/wiki/List_of_genetic_disorders#cite_note-4) | D | [AP1S1](http://en.wikipedia.org/wiki/AP1S1) |
| Mediterranean Anemia *see* [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| Mega-epiphyseal dwarfism *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| Menkea syndrome *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| Mental retardation with osteocartilaginous abnormalities *see* [Coffin-Lowry syndrome](http://en.wikipedia.org/wiki/Coffin-Lowry_syndrome) |  |  |
| [Metabolic disorders](http://en.wikipedia.org/wiki/Metabolic_disorders) |  |  |
| Metatropic dwarfism, type II *see* [Kniest dysplasia](http://en.wikipedia.org/wiki/Kniest_dysplasia) |  |  |
| Metatropic dysplasia type II *see* [Kniest dysplasia](http://en.wikipedia.org/wiki/Kniest_dysplasia) |  |  |
| [Methemoglobinemia#beta-globin type](http://en.wikipedia.org/wiki/Methemoglobinemia#beta-globin_type) |  |  |
| [methylmalonic acidemia](http://en.wikipedia.org/wiki/Methylmalonic_acidemia) |  |  |
| MFS *see* [Marfan syndrome](http://en.wikipedia.org/wiki/Marfan_syndrome) |  |  |
| MHAM *see* [Cowden syndrome](http://en.wikipedia.org/wiki/Cowden_syndrome) |  |  |
| MK *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| [Micro syndrome](http://en.wikipedia.org/wiki/Micro_syndrome) |  | 2q21.3 |
| [Microcephaly](http://en.wikipedia.org/wiki/Microcephaly) | P | 1q31 ([ASPM](http://en.wikipedia.org/wiki/ASPM_(Gene))) |
| MMA *see* [methylmalonic acidemia](http://en.wikipedia.org/wiki/Methylmalonic_acidemia) |  |  |
| MNK *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| Monosomy 1p36 syndrome *see* [1p36 deletion syndrome](http://en.wikipedia.org/wiki/1p36_deletion_syndrome) | D | 1p36 |
| monosomy X *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| Motor neuron disease, amyotrophic lateral sclerosis *see* [amyotrophic lateral sclerosis](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis) |  |  |
| [Movement disorders](http://en.wikipedia.org/wiki/Movement_disorder) |  |  |
| [Mowat-Wilson syndrome](http://en.wikipedia.org/wiki/Mowat-Wilson_syndrome) |  |  |
| [Mucopolysaccharidosis](http://en.wikipedia.org/wiki/Mucopolysaccharidosis) (MPS I) |  |  |
| Mucoviscidosis *see* [cystic fibrosis](http://en.wikipedia.org/wiki/Cystic_fibrosis) |  |  |
| [Muenke syndrome](http://en.wikipedia.org/wiki/Muenke_syndrome) |  |  |
| Multi-Infarct dementia *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) |  |  |
| Multiple carboxylase deficiency, late-onset *see* [biotinidase deficiency](http://en.wikipedia.org/wiki/Biotinidase_deficiency) |  |  |
| Multiple hamartoma syndrome *see* [Cowden syndrome](http://en.wikipedia.org/wiki/Cowden_syndrome) |  |  |
| Multiple neurofibromatosis *see* [neurofibromatosis](http://en.wikipedia.org/wiki/Neurofibromatosis) |  |  |
| [Muscular dystrophy](http://en.wikipedia.org/wiki/Muscular_dystrophy) |  |  |
| [Muscular dystrophy, Duchenne and Becker type](http://en.wikipedia.org/wiki/Muscular_dystrophy,_Duchenne_and_Becker_type) |  |  |
| Myotonia atrophica *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| Myotonia dystrophica *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| Myxedema, congenital *see* [congenital hypothyroidism](http://en.wikipedia.org/wiki/Congenital_hypothyroidism) |  |  |
| Nance-Insley syndrome *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| Nance-Sweeney chondrodysplasia *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| NBIA1 *see* [pantothenate kinase-associated neurodegeneration](http://en.wikipedia.org/wiki/Pantothenate_kinase-associated_neurodegeneration) |  |  |
| Neill-Dingwall syndrome *see* [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) |  |  |
| Neuroblastoma, retinal *see* [retinoblastoma](http://en.wikipedia.org/wiki/Retinoblastoma) |  |  |
| Neurodegeneration with brain iron accumulation type 1 *see* [pantothenate kinase-associated neurodegeneration](http://en.wikipedia.org/wiki/Pantothenate_kinase-associated_neurodegeneration) |  |  |
| [Neurofibromatosis type I](http://en.wikipedia.org/wiki/Neurofibromatosis_type_I) |  | 17q11.2 |
| [Neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II) |  |  |
| [Neurologic diseases](http://en.wikipedia.org/wiki/Neurologic_diseases) |  |  |
| [Neuromuscular disorders](http://en.wikipedia.org/wiki/Neuromuscular_disorders) |  |  |
| neuronopathy, distal hereditary motor, type V *see* [Distal spinal muscular atrophy#type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy&action=edit&redlink=1) |  |  |
| neuronopathy, distal hereditary motor, with pyramidal features *see* [Amyotrophic lateral sclerosis#type 4](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis#type_4) |  |  |
| Niemann-Pick *see* [Niemann–Pick disease](http://en.wikipedia.org/wiki/Niemann%E2%80%93Pick_disease) | [NPA](http://en.wikipedia.org/wiki/Niemann%E2%80%93Pick_disease#Classification), [NPB](http://en.wikipedia.org/wiki/Niemann%E2%80%93Pick_disease#Classification), [NPC1](http://en.wikipedia.org/wiki/NPC1), [NPC2](http://en.wikipedia.org/wiki/NPC2), [Sphingomyelin phosphodiesterase 1](http://en.wikipedia.org/wiki/Sphingomyelin_phosphodiesterase_1) | [SMPD1](http://en.wikipedia.org/wiki/SMPD1) |
| Noack syndrome *see* [Pfeiffer syndrome](http://en.wikipedia.org/wiki/Pfeiffer_syndrome) |  |  |
| Nonketotic hyperglycinemia *see* [Glycine encephalopathy](http://en.wikipedia.org/wiki/Glycine_encephalopathy) |  |  |
| Non-neuronopathic Gaucher disease *see* [Gaucher disease type 1](http://en.wikipedia.org/wiki/Gaucher_disease_type_1) |  |  |
| Non-phenylketonuric hyperphenylalaninemia *see* [tetrahydrobiopterin deficiency](http://en.wikipedia.org/wiki/Tetrahydrobiopterin_deficiency) |  |  |
| [nonsyndromic deafness](http://en.wikipedia.org/wiki/Nonsyndromic_deafness) |  |  |
| [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Norrbottnian Gaucher disease *see* [Gaucher disease type 3](http://en.wikipedia.org/wiki/Gaucher_disease_type_3) |  |  |
| Ochronosis *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| Ochronotic arthritis *see* [alkaptonuria](http://en.wikipedia.org/wiki/Alkaptonuria) |  |  |
| OI *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| Osler-Weber-Rendu disease *see* [Hereditary hemorrhagic telangiectasia](http://en.wikipedia.org/wiki/Hereditary_hemorrhagic_telangiectasia) |  |  |
| OSMED *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| Osteopsathyrosis *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| Osteosclerosis congenita *see* [achondroplasia](http://en.wikipedia.org/wiki/Achondroplasia) |  |  |
| Oto-spondylo-megaepiphyseal dysplasia *see* [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| [otospondylomegaepiphyseal dysplasia](http://en.wikipedia.org/wiki/Otospondylomegaepiphyseal_dysplasia) |  |  |
| Oxalosis *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| Oxaluria, primary *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| [pantothenate kinase-associated neurodegeneration](http://en.wikipedia.org/wiki/Pantothenate_kinase-associated_neurodegeneration) |  |  |
| [Patau Syndrome (Trisomy 13)](http://en.wikipedia.org/wiki/Patau_syndrome) |  |  |
| PBGD deficiency *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| PCC deficiency *see* [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| PCT *see* [porphyria cutanea tarda](http://en.wikipedia.org/wiki/Porphyria_cutanea_tarda) |  |  |
| PDM *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) |  |  |
| [Pendred syndrome](http://en.wikipedia.org/wiki/Pendred_syndrome) |  |  |
| Periodic disease *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| Periodic peritonitis *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| Periorificial lentiginosis syndrome *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| Peripheral nerve disorders *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| Peripheral neurofibromatosis *see* [neurofibromatosis type I](http://en.wikipedia.org/wiki/Neurofibromatosis_type_I) |  |  |
| Peroneal muscular atrophy *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| peroxisomal alanine:glyoxylate aminotransferase deficiency *see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria,_primary) |  |  |
| [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| [Pfeiffer syndrome](http://en.wikipedia.org/wiki/Pfeiffer_syndrome) |  |  |
| Phenylalanine hydroxylase deficiency disease *see* [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) |  |  |
| [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) |  |  |
| [Pheochromocytoma](http://en.wikipedia.org/wiki/Pheochromocytoma) *see* [von Hippel-Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel-Lindau_disease) |  |  |
| Pierre Robin syndrome with fetal chondrodysplasia *see* [Weissenbacher-Zweymüller syndrome](http://en.wikipedia.org/wiki/Weissenbacher-Zweym%C3%BCller_syndrome) |  |  |
| [Pigmentary cirrhosis](http://en.wikipedia.org/w/index.php?title=Pigmentary_cirrhosis&action=edit&redlink=1) *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| PJS *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| PKAN *see* [pantothenate kinase-associated neurodegeneration](http://en.wikipedia.org/wiki/Pantothenate_kinase-associated_neurodegeneration) |  |  |
| PKU *see* [phenylketonuria](http://en.wikipedia.org/wiki/Phenylketonuria) |  |  |
| Plumboporphyria *see* [ALA deficiency porphyria](http://en.wikipedia.org/wiki/ALA_deficiency_porphyria) |  |  |
| PMA *see* [Charcot-Marie-tooth disease](http://en.wikipedia.org/w/index.php?title=Charcot-Marie-tooth_disease&action=edit&redlink=1) |  |  |
| [Polycystic kidney disease](http://en.wikipedia.org/wiki/Polycystic_kidney_disease) | P | [16](http://en.wikipedia.org/wiki/Chromosome_16) ([PKD1](http://en.wikipedia.org/wiki/PKD1)) or [4](http://en.wikipedia.org/wiki/Chromosome_4) ([PKD2](http://en.wikipedia.org/wiki/PKD2)) |
| polyostotic fibrous dysplasia *see* [McCune–Albright syndrome](http://en.wikipedia.org/wiki/McCune%E2%80%93Albright_syndrome) |  | 20 q13.2-13.3 |
| polyposis coli *see* [familial adenomatous polyposis](http://en.wikipedia.org/wiki/Familial_adenomatous_polyposis) |  |  |
| polyposis, hamartomatous intestinal *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| polyposis, intestinal, II *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| polyps-and-spots syndrome *see* [Peutz-Jeghers syndrome](http://en.wikipedia.org/wiki/Peutz-Jeghers_syndrome) |  |  |
| Porphobilinogen synthase deficiency *see* [ALA deficiency porphyria](http://en.wikipedia.org/wiki/ALA_deficiency_porphyria) |  |  |
| [porphyria](http://en.wikipedia.org/wiki/Porphyria) |  |  |
| porphyrin disorder *see* [porphyria](http://en.wikipedia.org/wiki/Porphyria) |  |  |
| PPH *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| PPOX deficiency *see* [variegate porphyria](http://en.wikipedia.org/wiki/Variegate_porphyria) |  |  |
| Prader-Labhart-Willi syndrome *see* [Prader-Willi syndrome](http://en.wikipedia.org/wiki/Prader-Willi_syndrome) |  |  |
| [Prader-Willi syndrome](http://en.wikipedia.org/wiki/Prader-Willi_syndrome) |  |  |
| presenile and senile dementia *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| primary hemochromatosis *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| primary hyperuricemia syndrome *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| primary senile degenerative dementia *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| procollagen type EDS VII, mutant *see* [Ehlers–Danlos syndrome#arthrochalasia type](http://en.wikipedia.org/wiki/Ehlers%E2%80%93Danlos_syndrome#arthrochalasia_type) |  |  |
| progeria *see* [Hutchinson Gilford Progeria Syndrome](http://en.wikipedia.org/wiki/Hutchinson_Gilford_Progeria_Syndrome) |  |  |
| Progeria-like syndrome *see* [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) |  |  |
| progeroid nanism *see* [Cockayne syndrome](http://en.wikipedia.org/wiki/Cockayne_syndrome) |  |  |
| progressive chorea, chronic hereditary (Huntington) *see* [Huntington's disease](http://en.wikipedia.org/wiki/Huntington%27s_disease) |  |  |
| progressive muscular atrophy *see* [spinal muscular atrophy](http://en.wikipedia.org/wiki/Spinal_muscular_atrophy) |  |  |
| progressively deforming osteogenesis imperfecta with normal sclerae *see* [Osteogenesis imperfecta#Type III](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta#Type_III) |  |  |
| PROMM *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) |  |  |
| [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| propionyl-CoA carboxylase deficiency *see* [propionic acidemia](http://en.wikipedia.org/wiki/Propionic_acidemia) |  |  |
| [protein C deficiency](http://en.wikipedia.org/wiki/Protein_C_deficiency) |  |  |
| [protein S deficiency](http://en.wikipedia.org/wiki/Protein_S_deficiency) |  |  |
| protoporphyria *see* [erythropoietic protoporphyria](http://en.wikipedia.org/wiki/Erythropoietic_protoporphyria) |  |  |
| protoporphyrinogen oxidase deficiency *see* [variegate porphyria](http://en.wikipedia.org/wiki/Variegate_porphyria) |  |  |
| proximal myotonic dystrophy *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) |  |  |
| proximal myotonic myopathy *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) |  |  |
| [pseudo-Gaucher disease](http://en.wikipedia.org/wiki/Pseudo-Gaucher_disease) |  |  |
| pseudo-Ullrich-Turner syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| [pseudoxanthoma elasticum](http://en.wikipedia.org/wiki/Pseudoxanthoma_elasticum) |  |  |
| psychosine lipidosis *see* [Krabbe disease](http://en.wikipedia.org/wiki/Krabbe_disease) |  |  |
| pulmonary arterial hypertension *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| pulmonary hypertension *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| PWS *see* [Prader-Willi syndrome](http://en.wikipedia.org/wiki/Prader-Willi_syndrome) |  |  |
| PXE - pseudoxanthoma elasticum *see* [pseudoxanthoma elasticum](http://en.wikipedia.org/wiki/Pseudoxanthoma_elasticum) |  |  |
| Rb *see* [retinoblastoma](http://en.wikipedia.org/wiki/Retinoblastoma) |  |  |
| Recklinghausen disease, nerve *see* [neurofibromatosis type I](http://en.wikipedia.org/wiki/Neurofibromatosis_type_I) |  |  |
| Recurrent polyserositis *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| [Retinal disorders](http://en.wikipedia.org/wiki/Retinal_disorders) |  |  |
| Retinitis pigmentosa-deafness syndrome *see* [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| [Retinoblastoma](http://en.wikipedia.org/wiki/Retinoblastoma) |  |  |
| [Rett syndrome](http://en.wikipedia.org/wiki/Rett_syndrome) |  |  |
| RFALS type 3 *see* [Amyotrophic lateral sclerosis#type 2](http://en.wikipedia.org/wiki/Amyotrophic_lateral_sclerosis#type_2) |  |  |
| Ricker syndrome *see* [Myotonic dystrophy#type 2](http://en.wikipedia.org/wiki/Myotonic_dystrophy#type_2) |  |  |
| Riley-Day syndrome *see* [familial dysautonomia](http://en.wikipedia.org/wiki/Familial_dysautonomia) |  |  |
| Roussy-Levy syndrome *see* [Charcot-Marie-Tooth disease](http://en.wikipedia.org/wiki/Charcot-Marie-Tooth_disease) |  |  |
| RSTS *see* [Rubinstein-Taybi syndrome](http://en.wikipedia.org/wiki/Rubinstein-Taybi_syndrome) |  |  |
| RTS *see* [Rett syndrome](http://en.wikipedia.org/wiki/Rett_syndrome) *see* [Rubinstein-Taybi syndrome](http://en.wikipedia.org/wiki/Rubinstein-Taybi_syndrome) |  |  |
| RTT *see* [Rett syndrome](http://en.wikipedia.org/wiki/Rett_syndrome) |  |  |
| [Rubinstein-Taybi syndrome](http://en.wikipedia.org/wiki/Rubinstein-Taybi_syndrome) |  |  |
| Sack-Barabas syndrome *see* [Ehlers–Danlos syndrome, vascular type](http://en.wikipedia.org/wiki/Ehlers-Danlos_syndrome,_vascular_type) |  |  |
| [SADDAN](http://en.wikipedia.org/wiki/SADDAN) |  |  |
| sarcoma family syndrome of Li and Fraumeni *see* [Li-Fraumeni syndrome](http://en.wikipedia.org/wiki/Li-Fraumeni_syndrome) |  |  |
| sarcoma, breast, leukemia, and adrenal gland (SBLA) syndrome *see* [Li-Fraumeni syndrome](http://en.wikipedia.org/wiki/Li-Fraumeni_syndrome) |  |  |
| SBLA syndrome *see* [Li-Fraumeni syndrome](http://en.wikipedia.org/wiki/Li-Fraumeni_syndrome) |  |  |
| SBMA *see* [X-linked spinal-bulbar muscle atrophy](http://en.wikipedia.org/wiki/X-linked_spinal-bulbar_muscle_atrophy) |  |  |
| SCD *see* [sickle cell anemia](http://en.wikipedia.org/wiki/Sickle_cell_anemia) |  |  |
| Schwannoma, acoustic, bilateral *see* [neurofibromatosis type II](http://en.wikipedia.org/wiki/Neurofibromatosis_type_II) |  |  |
| SCIDX1 *see* [X-linked severe combined immunodeficiency](http://en.wikipedia.org/wiki/X-linked_severe_combined_immunodeficiency) |  |  |
| sclerosis tuberosa *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| SDAT *see* [Alzheimer's disease](http://en.wikipedia.org/wiki/Alzheimer%27s_disease) |  |  |
| SED congenita *see* [spondyloepiphyseal dysplasia congenita](http://en.wikipedia.org/wiki/Spondyloepiphyseal_dysplasia_congenita) |  |  |
| SED Strudwick *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| SEDc *see* [spondyloepiphyseal dysplasia congenita](http://en.wikipedia.org/wiki/Spondyloepiphyseal_dysplasia_congenita) |  |  |
| SEMD, Strudwick type *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| senile dementia *see* [Alzheimer disease#type 2](http://en.wikipedia.org/wiki/Alzheimer_disease#type_2) |  |  |
| severe achondroplasia with developmental delay and acanthosis nigricans *see* [SADDAN](http://en.wikipedia.org/wiki/SADDAN) |  |  |
| Shprintzen syndrome *see* [22q11.2 deletion syndrome](http://en.wikipedia.org/wiki/22q11.2_deletion_syndrome) | D | 22q |
| [sickle cell anemia](http://en.wikipedia.org/wiki/Sickle_cell_anemia) | D | 18q |
| Siderius X-linked mental retardation syndrome *caused by mutations in the*[*PHF8*](http://en.wikipedia.org/wiki/PHF8)*gene* | PD | Xp11.22 |
| skeleton-skin-brain syndrome *see* [SADDAN](http://en.wikipedia.org/wiki/SADDAN) |  |  |
| [Skin pigmentation disorders](http://en.wikipedia.org/w/index.php?title=Skin_pigmentation_disorders&action=edit&redlink=1) |  |  |
| SMA *see* [spinal muscular atrophy](http://en.wikipedia.org/wiki/Spinal_muscular_atrophy) |  |  |
| SMED, Strudwick type *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| SMED, type I *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| [Smith-Lemli-Opitz syndrome](http://en.wikipedia.org/wiki/Smith-Lemli-Opitz_syndrome) |  |  |
| South-African genetic porphyria *see* [variegate porphyria](http://en.wikipedia.org/wiki/Variegate_porphyria) |  |  |
| spastic paralysis, infantile onset ascending *see* [infantile-onset ascending hereditary spastic paralysis](http://en.wikipedia.org/w/index.php?title=Infantile-onset_ascending_hereditary_spastic_paralysis&action=edit&redlink=1) |  |  |
| [Speech and communication disorders](http://en.wikipedia.org/w/index.php?title=Speech_and_communication_disorders&action=edit&redlink=1) |  |  |
| sphingolipidosis, Tay-Sachs *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| [spinal-bulbar muscular atrophy](http://en.wikipedia.org/w/index.php?title=Spinal-bulbar_muscular_atrophy&action=edit&redlink=1) |  |  |
| [spinal muscular atrophy](http://en.wikipedia.org/wiki/Spinal_muscular_atrophy) |  |  |
| spinal muscular atrophy, distal type V *see* [Distal spinal muscular atrophy#type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy&action=edit&redlink=1) |  |  |
| spinal muscular atrophy, distal, with upper limb predominance *see* [Distal spinal muscular atrophy#type V](http://en.wikipedia.org/w/index.php?title=Distal_spinal_muscular_atrophy&action=edit&redlink=1) |  |  |
| [spinocerebellar ataxia](http://en.wikipedia.org/wiki/Spinocerebellar_ataxia) |  |  |
| [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| [spondyloepiphyseal dysplasia congenita](http://en.wikipedia.org/wiki/Spondyloepiphyseal_dysplasia_congenita) |  |  |
| spondyloepiphyseal dysplasia *see* [collagenopathy, types II and XI](http://en.wikipedia.org/wiki/Collagenopathy,_types_II_and_XI) |  |  |
| spondylometaepiphyseal dysplasia congenita, Strudwick type *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| spondylometaphyseal dysplasia (SMD) *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| spondylometaphyseal dysplasia, Strudwick type *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| spongy degeneration of central nervous system *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| spongy degeneration of the brain *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| spongy degeneration of white matter in infancy *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| sporadic primary pulmonary hypertension *see* [primary pulmonary hypertension](http://en.wikipedia.org/wiki/Primary_pulmonary_hypertension) |  |  |
| SSB syndrome *see* [SADDAN](http://en.wikipedia.org/wiki/SADDAN) |  |  |
| steely hair syndrome *see* [Menkes disease](http://en.wikipedia.org/wiki/Menkes_disease) |  |  |
| Steinert disease *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| Steinert myotonic dystrophy syndrome *see* [myotonic dystrophy](http://en.wikipedia.org/wiki/Myotonic_dystrophy) |  |  |
| [Stickler syndrome](http://en.wikipedia.org/wiki/Stickler_syndrome) |  |  |
| stroke *see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) |  |  |
| Strudwick syndrome *see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia,_Strudwick_type) |  |  |
| subacute neuronopathic Gaucher disease *see* [Gaucher disease type 3](http://en.wikipedia.org/wiki/Gaucher_disease_type_3) |  |  |
| Swedish genetic porphyria *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| Swedish porphyria *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| Swiss cheese cartilage dysplasia *see* [Kniest dysplasia](http://en.wikipedia.org/wiki/Kniest_dysplasia) |  |  |
| [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| TD - thanatophoric dwarfism *see* [thanatophoric dysplasia](http://en.wikipedia.org/wiki/Thanatophoric_dysplasia) |  |  |
| TD with straight femurs and cloverleaf skull *see* [thanatophoric dysplasia#Type 2](http://en.wikipedia.org/wiki/Thanatophoric_dysplasia#Type_2) |  |  |
| Telangiectasia, cerebello-oculocutaneous *see* [ataxia telangiectasia](http://en.wikipedia.org/wiki/Ataxia_telangiectasia) |  |  |
| Testicular feminization syndrome *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  |  |
| [tetrahydrobiopterin deficiency](http://en.wikipedia.org/wiki/Tetrahydrobiopterin_deficiency) |  |  |
| TFM - testicular feminization syndrome *see* [androgen insensitivity syndrome](http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome) |  |  |
| thalassemia intermedia *see* [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| Thalassemia Major *see* [beta-thalassemia](http://en.wikipedia.org/wiki/Beta-thalassemia) |  |  |
| [thanatophoric dysplasia](http://en.wikipedia.org/wiki/Thanatophoric_dysplasia) |  |  |
| Thrombophilia due to deficiency of cofactor for activated protein C, Leiden type *see* [factor V Leiden thrombophilia](http://en.wikipedia.org/wiki/Factor_V_Leiden_thrombophilia) |  |  |
| [Thyroid disease](http://en.wikipedia.org/wiki/Thyroid_disease) |  |  |
| Tomaculous neuropathy *see* [hereditary neuropathy with liability to pressure palsies](http://en.wikipedia.org/wiki/Hereditary_neuropathy_with_liability_to_pressure_palsies) |  |  |
| Total HPRT deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| Total hypoxanthine-guanine phosphoribosyl transferase deficiency *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| Tourette syndrome |  |  |
| Transmissible dementias *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| Transmissible spongiform encephalopathies *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| [Treacher Collins syndrome](http://en.wikipedia.org/wiki/Treacher_Collins_syndrome) |  | 5q32-q33.1 |
| Trias fragilitis ossium *see* [osteogenesis imperfecta#Type I](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta#Type_I) |  |  |
| [triple X syndrome](http://en.wikipedia.org/wiki/Triple_X_syndrome) |  |  |
| Triplo X syndrome *see* [triple X syndrome](http://en.wikipedia.org/wiki/Triple_X_syndrome) |  |  |
| Trisomy 21 *see* [Down syndrome](http://en.wikipedia.org/wiki/Down_syndrome) |  |  |
| Trisomy X *see* [triple X syndrome](http://en.wikipedia.org/wiki/Triple_X_syndrome) |  |  |
| Troisier-Hanot-Chauffard syndrome *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| TS *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| TSD *see* [Tay-Sachs disease](http://en.wikipedia.org/wiki/Tay-Sachs_disease) |  |  |
| TSEs *see* [prion disease](http://en.wikipedia.org/wiki/Prion_disease) |  |  |
| tuberose sclerosis *see* [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| [tuberous sclerosis](http://en.wikipedia.org/wiki/Tuberous_sclerosis) |  |  |
| [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| Turner syndrome in female with X chromosome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Turner's phenotype, karyotype normal *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Turner's syndrome *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| Turner-like syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Type 2 Gaucher disease *see* [Gaucher disease type 2](http://en.wikipedia.org/wiki/Gaucher_disease_type_2) |  |  |
| Type 3 Gaucher disease *see* [Gaucher disease type 3](http://en.wikipedia.org/wiki/Gaucher_disease_type_3) |  |  |
| UDP-galactose-4-epimerase deficiency disease *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| UDP glucose 4-epimerase deficiency disease *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| UDP glucose hexose-1-phosphate uridylyltransferase deficiency *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| Ullrich-Noonan syndrome *see* [Noonan syndrome](http://en.wikipedia.org/wiki/Noonan_syndrome) |  |  |
| Ullrich-Turner syndrome *see* [Turner syndrome](http://en.wikipedia.org/wiki/Turner_syndrome) |  |  |
| Undifferentiated deafness *see* [nonsyndromic deafness](http://en.wikipedia.org/wiki/Nonsyndromic_deafness) |  |  |
| UPS deficiency *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| Urinary bladder cancer *see* [bladder cancer](http://en.wikipedia.org/wiki/Bladder_cancer) |  |  |
| UROD deficiency *see* [porphyria cutanea tarda](http://en.wikipedia.org/wiki/Porphyria_cutanea_tarda) |  |  |
| Uroporphyrinogen decarboxylase deficiency *see* [porphyria cutanea tarda](http://en.wikipedia.org/wiki/Porphyria_cutanea_tarda) |  |  |
| Uroporphyrinogen synthase deficiency *see* [acute intermittent porphyria](http://en.wikipedia.org/wiki/Acute_intermittent_porphyria) |  |  |
| UROS deficiency *see* [congenital erythropoietic porphyria](http://en.wikipedia.org/wiki/Congenital_erythropoietic_porphyria) |  |  |
| [Usher syndrome](http://en.wikipedia.org/wiki/Usher_syndrome) |  |  |
| UTP hexose-1-phosphate uridylyltransferase deficiency *see* [galactosemia](http://en.wikipedia.org/wiki/Galactosemia) |  |  |
| Van Bogaert-Bertrand syndrome *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| Van der Hoeve syndrome *see* [osteogenesis imperfecta#Type I](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta#Type_I) |  |  |
| [variegate porphyria](http://en.wikipedia.org/wiki/Variegate_porphyria) |  |  |
| Velocardiofacial syndrome *see* [22q11.2 deletion syndrome](http://en.wikipedia.org/wiki/22q11.2_deletion_syndrome) | D | 22q |
| VHL syndrome *see* [von Hippel-Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel-Lindau_disease) |  |  |
| Vision impairment and blindness *see* [Alström syndrome](http://en.wikipedia.org/wiki/Alstr%C3%B6m_syndrome) |  |  |
| Von Bogaert-Bertrand disease *see* [Canavan disease](http://en.wikipedia.org/wiki/Canavan_disease) |  |  |
| [von Hippel-Lindau disease](http://en.wikipedia.org/wiki/Von_Hippel-Lindau_disease) |  |  |
| Von Recklenhausen-Applebaum disease *see* [hemochromatosis](http://en.wikipedia.org/wiki/Hemochromatosis) |  |  |
| von Recklinghausen disease *see* [neurofibromatosis type I](http://en.wikipedia.org/wiki/Neurofibromatosis_type_I) |  |  |
| VP *see* [variegate porphyria](http://en.wikipedia.org/wiki/Variegate_porphyria) |  |  |
| Vrolik disease *see* [osteogenesis imperfecta](http://en.wikipedia.org/wiki/Osteogenesis_imperfecta) |  |  |
| [Waardenburg syndrome](http://en.wikipedia.org/wiki/Waardenburg_syndrome) |  |  |
| Warburg Sjo Fledelius Syndrome *see* [Micro syndrome](http://en.wikipedia.org/wiki/Micro_syndrome) |  | 2q21.3 |
| WD *see* [Wilson disease](http://en.wikipedia.org/wiki/Wilson_disease) |  |  |
| [Weissenbacher-Zweymüller syndrome](http://en.wikipedia.org/wiki/Weissenbacher-Zweym%C3%BCller_syndrome) |  |  |
| [Williams Syndrome](http://en.wikipedia.org/wiki/Williams_Syndrome) |  |  |
| [Wilson disease](http://en.wikipedia.org/wiki/Wilson_disease) |  |  |
| Wilson's disease *see* [Wilson disease](http://en.wikipedia.org/wiki/Wilson_disease) |  |  |
| [Wolf–Hirschhorn syndrome](http://en.wikipedia.org/wiki/Wolf%E2%80%93Hirschhorn_syndrome) | D | 4p |
| Wolff Periodic disease *see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever,_familial) |  |  |
| WZS *see* [Weissenbacher-Zweymüller syndrome](http://en.wikipedia.org/wiki/Weissenbacher-Zweym%C3%BCller_syndrome) |  |  |
| [Xeroderma pigmentosum](http://en.wikipedia.org/wiki/Xeroderma_pigmentosum) | ERCC4 | 15 |
| X-linked mental retardation and macroorchidism *see* [fragile X syndrome](http://en.wikipedia.org/wiki/Fragile_X_syndrome) |  |  |
| X-linked primary hyperuricemia *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| [X-linked severe combined immunodeficiency](http://en.wikipedia.org/wiki/X-linked_severe_combined_immunodeficiency) |  |  |
| [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| X-linked spinal-bulbar muscle atrophy *see* [Kennedy's disease](http://en.wikipedia.org/wiki/Kennedy%27s_disease) |  |  |
| X-linked uric aciduria enzyme defect *see* [Lesch-Nyhan syndrome](http://en.wikipedia.org/wiki/Lesch-Nyhan_syndrome) |  |  |
| X-SCID *see* [X-linked severe combined immunodeficiency](http://en.wikipedia.org/wiki/X-linked_severe_combined_immunodeficiency) |  |  |
| XLSA *see* [X-linked sideroblastic anemia](http://en.wikipedia.org/wiki/X-linked_sideroblastic_anemia) |  |  |
| XSCID *see* [X-linked severe combined immunodeficiency](http://en.wikipedia.org/wiki/X-linked_severe_combined_immunodeficiency) |  |  |
| XXX syndrome *see* [triple X syndrome](http://en.wikipedia.org/wiki/Triple_X_syndrome) |  |  |
| XXXX syndrome *see* [48, XXXX](http://en.wikipedia.org/wiki/48,_XXXX) |  |  |
| XXXXX syndrome *see* [49, XXXXX](http://en.wikipedia.org/wiki/49,_XXXXX) |  |  |
| XXY syndrome *see* [Klinefelter's syndrome](http://en.wikipedia.org/wiki/Klinefelter%27s_syndrome) |  |  |
| XXY trisomy *see* [Klinefelter's syndrome](http://en.wikipedia.org/wiki/Klinefelter%27s_syndrome) |  |  |
| XYY karyotype *see* [47,XYY syndrome](http://en.wikipedia.org/wiki/47,XYY_syndrome) |  |  |
| XYY syndrome *see* [47,XYY syndrome](http://en.wikipedia.org/wiki/47,XYY_syndrome) |  |  |
| YY syndrome *see* [47,XYY syndrome](http://en.wikipedia.org/wiki/47,XYY_syndrome) |  |  |