|  |  |  |
| --- | --- | --- |
| [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%2CXYY_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%2C_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%2C_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%2C_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%2C_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 arteriopathywith subcortical infarcts and leukoencephalopathy*see* [CADASIL syndrome](http://en.wikipedia.org/wiki/CADASIL_syndrome) | P | 3 |
| Cerebral autosomal dominant ateriopathywith 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%2C_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%2C_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%2C_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%2C_type_4) |  |  |
| Duchenne/Becker muscular dystrophy*see* [Muscular dystrophy, Duchenne and Becker type](http://en.wikipedia.org/wiki/Muscular_dystrophy%2C_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%2C_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%2C_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%2C_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%2C_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%2C_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%2C_primary) |  |  |
| Glycine encephalopathy*see* [Nonketotic hyperglycinemia](http://en.wikipedia.org/wiki/Nonketotic_hyperglycinemia) |  |  |
| Glycolic aciduria*see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria%2C_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%2C_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 dysfunctionusually 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%2C_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%2C_primary) |  |  |
| HP2*see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria%2C_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%2C_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%2C_familial) |  |  |
| [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria%2C_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%2C_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%2C_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%2C_type_II) |  |  |
| Langer-Saldino dysplasia*see* [achondrogenesis, type II](http://en.wikipedia.org/wiki/Achondrogenesis%2C_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%2C_familial) |  |  |
| LIPD deficiency*see* [lipoprotein lipase deficiency, familial](http://en.wikipedia.org/wiki/Lipoprotein_lipase_deficiency%2C_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%2C_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%2C_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_%28Gene%29)) |
| 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%2C_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%2C_primary) |  |  |
| Oxaluria, primary*see* [hyperoxaluria, primary](http://en.wikipedia.org/wiki/Hyperoxaluria%2C_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%2C_familial) |  |  |
| Periodic peritonitis*see* [Mediterranean fever, familial](http://en.wikipedia.org/wiki/Mediterranean_fever%2C_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%2C_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%2C_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%2C_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%2C_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%2C_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%2C_Strudwick_type) |  |  |
| SMED, type I*see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia%2C_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%2C_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%2C_types_II_and_XI) |  |  |
| spondylometaepiphyseal dysplasia congenita, Strudwick type*see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia%2C_Strudwick_type) |  |  |
| spondylometaphyseal dysplasia (SMD)*see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia%2C_Strudwick_type) |  |  |
| spondylometaphyseal dysplasia, Strudwick type*see* [spondyloepimetaphyseal dysplasia, Strudwick type](http://en.wikipedia.org/wiki/Spondyloepimetaphyseal_dysplasia%2C_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%2C_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%2C_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%2C_XXXX) |  |  |
| XXXXX syndrome*see* [49, XXXXX](http://en.wikipedia.org/wiki/49%2C_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%2CXYY_syndrome) |  |  |
| XYY syndrome*see* [47,XYY syndrome](http://en.wikipedia.org/wiki/47%2CXYY_syndrome) |  |  |
| YY syndrome*see* [47,XYY syndrome](http://en.wikipedia.org/wiki/47%2CXYY_syndrome) |  |  |