11 beta hydroxylase deficiency
11 beta hydroxysteroid dehydrogenase type 2 deficiency
17 alpha hydroxylase deficiency
17 beta hydroxysteroide dehydrogenase deficiency
2,8 dihydroxy-adenine urolithiasis
2-hydroxyglutaricaciduria
21 hydroxylase deficiency
3 beta hydroxysteroid dehydrogenase deficiency
3 hydroxyisobutyric aciduria
3 methylcrotonic aciduria
3 methylglutaconyl coa hydratase deficiency
3-hydroxy 3-methyl glutaryl-coa lyase deficiency
3-hydroxyacyl-coa dehydrogenase deficiency
3-methyl crotonyl-coa carboxylase deficiency
3-methyl glutaconic aciduria
3-methylcrotonylglycinuria
3c syndrome
3m syndrome
4 alpha hydroxyphenylpyruvate hydroxylase deficiency
46 xx gonadal dysgenesis epibulbar dermoid
47 XXY syndrome
47 xyy syndrome
48 xxxx syndrome
48 xxyy syndrome
49 xxxxx syndrome
49 xxxxy syndrome
5 alpha reductase 2 deficiency
6-pyruvoyltetrahydropterin synthase deficiency
7-dehydrocholesterol reductase deficiency
aagenaes syndrome
aarskog like syndrome
aarskog ose pande syndrome
aarskog syndrome
aase smith syndrome
aase syndrome
abcd syndrome
abdallat davis farrage syndrome
abdominal aortic aneurysm
abdominal cystic lymphangioma
abdominal musculature absent microphthalmia joint laxity
abetalipoproteinemia
ablepharon macrostomia syndrome
abnormal systemic veinous return
abruzzo erickson syndrome
absent corpus callosum cataract immunodeficiency
absent hands and feet
abuelo-forman-rubin syndrome
acalvaria
acanthocytosis chorea
acanthocytosis neurologic disorder
acanthosis nigricans
acanthosis nigricans muscle cramps acral enlargement
acatalasemia
accessory pancreas
acetyl coa alpha glucosaminide n acetyl transferase deficiency
achalasia addisonianism alacrimia syndrome
achalasia alacrimia syndrome
achalasia familial esophageal
achalasia microcephaly
acheiropodia
achondrogenesis grebe type
achondrogenesis kozlowski type
achondrogenesis type 1
achondrogenesis type 1b
achondrogenesis type 2
achondroplasia
achondroplasia swiss type agammaglobulinemia
achromatopsia incomplete x linked
acid maltase deficiency

acitretine antenatal infection
ackerman syndrome
acoustic neurinoma
acquired autoimmune haemolytic anemia
acquired hypertrichosis lanuginosa
acquired ichtyosis
acquired progressive kinking of the hair
acquired prothrombin deficiency
acquired willebrand disease
acral dysostosis dyserythropoiesis
acral renal mandibular syndrome
acrania
acro cephalo synostosis
acro coxo mesomelic dysplasia
acro fronto facio nasal dysostosis
acrocallosal syndrome
acrocephalopolydactyly
acrocephalopolysyndactyly type 2
acrocephalopolysyndactyly type 3
acrocephalopolysyndactyly type 4
acrocephalosyndactyly jackson weiss type
acrocephalosyndactyly type 1
acrocephalosyndactyly type 3
acrocephalosyndactyly type 5
acrocephaly pulmonary stenosis mental retardation
acrocraniofacial dysostosis
acrodermatitis enteropathica zinc deficiency type
acrodysostosis
acrodysplasia scoliosis
acrofacial dysostosis ambiguous genitalia
acrofacial dysostosis atypical postaxial
acrofacial dysostosis catania form
acrofacial dysostosis nager type
acrofacial dysostosis preis type
acrofacial dysostosis rodriguez type
acrofacial dysostosis weyers type
acrofaciale dysostosis, palagonia type
Acrokeratoelastoidosis of Costa
acromegaloid changes cutis verticis gyrata corneal leukoma
acromegaloid facial appearance syndrome
acromegaloid hypertrichosis syndrome
acromegaly
acromesomelic dwarfism campailla martinelli type
acromesomelic dysplasia brahimi bacha type
acromesomelic dysplasia campailla martinelli type
acromesomelic dysplasia hunter thompson type
acromesomelic dysplasia maroteaux type
acromesomelic syndrome pfeiffer type
acromicric dysplasia
acroosteolysis dominant type
acroosteolysis neurogenic
acroosteolysis osteoporosis skull and mandible changes
acropectorenal field defect
acropectorovertebral dysplasia
acropigmentation of Dohi
acrorenal field defect ectodermal dysplasia diabetes
acrorenal syndrome recessive
acrorenoocular syndrome
acth resistance
acutane embryopathy
acute articular rheumatism
acute eosinophilic pneumonia
acute erythroblastic leukemia
acute febrile neutrophilic dermatosis
acute lymphoblastic leukemia
acute lymphoblastic leukemia congenital sporadic aniridia
acute megacaryoblastic leukemia
acute monoblastic leukemia
acute myeloblastic leukemia type 1

acute myeloblastic leukemia type 2
acute myeloblastic leukemia type 3
acute myeloblastic leukemia type 4
acute myeloblastic leukemia type 5
acute myeloblastic leukemia type 6
acute myeloblastic leukemia type 7
acute myeloblastic leukemia with maturation
acute myeloblastic leukemia without maturation
acute myelomonocytic leukemia
acute promyelocytic leukemia
acyl coa deshydrogenase deficiency
acyl coa deshydrogenase medium chain deficiency of
acyl coa oxydase deficiency
acyl-coa dehydrogenase short chain deficiency
acyl-coa dehydrogenase very long chain deficiency
adactylia unilateral dominant
adam complex familial
adams nance syndrome
adams oliver syndrome
adducted thumb syndrome recessive form
adducted thumbs dundar type
adenine phosphoribosyltransferase deficiency
adenosine deaminase deficiency
adenosine monophosphate deaminase deficiency
adenosine triphosphatase deficiency anemia due to
adenylosuccinase deficiency
adenylosuccinate lyase deficiency
adolescent benign focal crisis
adolescent idiopathic scoliosis
adrenal adenoma familial
adrenal hypoplasia congenital x linked
adrenal incidentaloma
adrenal macropolyadenomatosis
adrenocortical carcinoma
adrenoleukodystrophy autosomal neonatal form
adrenoleukodystrophy x-linked
adrenomyeloneuropathy (amn)
adrenomyodystrophy
adult idiopathic neutropenia
adult spinal muscular atrophy
adult syndrome
aec syndrome
afibrinogenemia, familial
african trypanosomiasis
agammaglobulinemia alymphocytotic type
agammaglobulinemia x linked
aganglionosis total intestinal
aganthia holoprosencephaly situs inversus
aggressive fibromatosis
aglossia adactylia
agonadism dextrocardia diaphragmatic hernia
agonadism mental retardation delayed bone age
agyria pachygyria polymicrogyria
agyria pachygyria type 1
aicardi goutieres syndrome
aicardi syndrome
akaba hayasaka syndrome
akesson syndrome
aksu stckhausen syndrome
al awadi farag teebi syndrome
al awadi teebi farag syndrome
al frayh facharzt haque syndrome
al gazali al talabani syndrome
al gazali aziz salem syndrome
al gazali donnai muller syndrome
al gazali hirschsprung syndrome
al gazali khidr prem chandran syndrome
al gazali sabrinathan nair syndrome
alagille syndrome

alanine glyoxylate aminotransferase deficiency
alar nasal cartilages coloboma of telecanthus
albers schonberg disease
albinism deafness syndrome
albinism immunodeficiency
albinism ocular
albinism ocular late onset sensorineural deafness
albinism oculocutaneous hermansky pudlak type
albinism yellow mutant type
albrecht schneider belmont syndrome
albright disease
albright hereditary osteodystrophy
albright like syndrome
alcohol antenatal infection
aldolase a deficiency
aldosterone synthase deficiency
alexander disease
alkaptonuria
allain babin demarquez syndrome
allan herndon syndrome
allanson pantzar mcleod syndrome
allergic bronchopulmonary aspergillosis
allgrove syndrome
aloi tomasini isaia syndrome
alopecia anosmia deafness hypogonadism syndrome
alopecia antibody deficiency
alopecia congenita keratosis palmoplantaris
alopecia contractures dwarfism mental retardation syndrome
alopecia epilepsy oligophrenia syndrome of moynahan
alopecia epilepsy pyorrhea mental subnormality
alopecia hypogonadism extrapyramidal disorder
alopecia immunodeficiency
alopecia macular degeneration growth retardation
alopecia mental retardation hypogonadism
alopecia mental retardation syndrome
alopecia totalis
alopecia universalis
alopecia universalis onychodystrophy vitiligo
alpers progressive sclerosing poliodystrophy
alpha 1 antitrypsin deficiency
alpha 2 deficient collagen disease
alpha galactosidase a deficiency
alpha ketoglutarate dehydrogenase deficiency
alpha l fucosidase deficiency
alpha l iduronidase deficiency
alpha mannosidosis
alpha thalassemia
alpha thalassemia mental retardation x linked
Alpha-sarcoglycanopathy
alpha-thalassemia-abnormal morphogenesis
alport deafness nephropathy
alport syndrome
alport syndrome dominant type
alport syndrome macrothrombocytopenia
alport syndrome recessive type
alport syndrome with leukocyte inclusions and macrothrombocytopenia
alport syndrome x linked
alstrom syndrome
alternating hemiplegia
alveolar echinococcosis
alves dos santos castello syndrome
alzheimer disease familial
alzheimer disease type 1
alzheimer disease type 2
alzheimer disease type 3
amaurosis congenita of leber
amaurosis congenita of leber type 1
amaurosis congenita of leber type 2
amaurosis hypertrichosis

ambral syndrome
ambras syndrome
amegakaryocytic thrombocytopenia
amelia cleft lip palate hydrocephalus iris coloboma
amelia facial dysmorphism
amelia x linked
amelo cerebro hypohidrotique syndrome
amelogenesis imperfecta local hypoplastic form
amelogenesis imperfecta nephrocalcinosis
amelogenesis imperfecta x linked
ameloonychohypohidrotic syndrome
american trypanosomiasis
aminopterin antenatal infection
aminopterin like syndrome without aminopterin
amniotic bands sequence
Amoebiasis
Amoebiasis due to Entamoeba histolytica
Amoebiasis due to free-living amoebae
ampola syndrome
amylo-1,6-glucosidase deficiency
amyloid polyneuropathy transthyretin related
amyloidosis
amyloidosis
amyloidosis of gingiva and conjunctiva mental retardation
Amylopectinosis
amyoplasia
amyoplasia mandibulofacial dysostosis
amyotrophic lateral sclerosis
amyotrophy fat tissue anomaly
anaplastic large cell lymphoma
anaplastic thyroid cancer
andermann syndrome
Andersen disease
anderson disease
androgen insensitivity partial
androgen resistance syndrome
anemia congenital hypoplastic blackfan diamond type
anemia sideroblastic spinocerebellar ataxia
anemia triphalangeal thumbs
anencephaly
anencephaly recessive type
anencephaly spina bifida x linked
aneurysm of sinus of valsalva
angel shaped phalango epiphyseal dysplasia
angelman syndrome
angiofollicular lymph hyperplasia
angiokeratoma diffuse
angiokeratoma mental retardation coarse face
angioma hereditary neurocutaneous
angiomatosis systemic cystic seip syndrome
angioneurotic edema hereditary due to C1 esterase inhibitor deficiency
angioosteohypertrophy syndrome
Angiostrongyliasis
Anguillulosis
aniridia absent patella
aniridia ataxia renal agenesis psychomotor retardation
aniridia lens luxation mental retardation
aniridia mental retardation syndrome
aniridia ptosis mental retardation obesity familial type
aniridia renal agenesis psychomotor retardation
aniridia sporadic
aniridia type 2
anisakiasis
ankle defects short stature
ankyloblepharon cleft palate ectodermal defects
ankyloblepharon ectodermal defects cleft lip palate
ankyloblepharon filiforme adnatum cleft palate
ankyloblepharon filiforme imperforate anus
ankyloglossia heterochromia clasped thumbs

ankylosing spondylarthritis
ankylosing vertebral hyperostosis with tylosis
ankylosis of teeth
Ankylostomiasis
annular pancreas
annuloaortic ectasia
ano-rectal atresia
anonychia ectrodactyly
anonychia microcephaly
anonychia onychodystrophy
anonychia onychodystrophy brachydactyly type b
anophtalmia syndactyly waardenburg type
anophthalia pulmonary hypoplasia
anophthalmia cleft lip palate hypothalamic disorder
anophthalmia cleft palate micrognathia
anophthalmia esophageal atresia cryptorchidism
anophthalmia megalocornea cardiopathy skeletal anomalies
anophthalmia microcephaly hypogonadism
anophthalmia plus syndrome
anophthalmia short stature obesity
anophthalmia waardenburg syndrome
anophthalmos
anophthalmos clinical
anophthalmos with limb anomalies
anorectal anomalies
anotia
anotia facial palsy cardiac defect
ansell bywaters elderking syndrome
anterior horn disease
anterior pituitary insufficiency, familial
anti hla hyper immunization
anti-plasmin deficiency
antigen-peptide-transporter 2 deficiency
antihypertensive drugs antenatal infection
antinolo nieto borrego syndrome
antiphospholipid syndrome
antisynthetase syndrome
antithrombin deficiency
antley bixler syndrome
anyane yeboa syndrome
aorta-pulmonary artery fistula
aortic arch anomaly peculiar facies mental retardation
aortic arch interruption
aortic arches defect
aortic dissection lentiginosis
aortic supravalvular stenosis
aortic supravalvular stenosis
aortic valves stenosis of the child
aortic window
apeced syndrome
apert like polydactyly syndrome
apert syndrome
aphalangia hemivertebrae
aphalangia syndactyly microcephaly
aplasia cutis autosomal recessive
aplasia cutis cleft palate epidermolysis
aplasia cutis congenita dominant
aplasia cutis congenita epibulbar dermoids
aplasia cutis congenita intestinal lymphangiectasia
aplasia cutis congenita of limbs recessive
aplasia cutis congenita of limbs recessive
aplasia cutis congenita recessive
aplasia cutis myopia
apo a1 deficiency
apolipoproteine c2 deficiency
apparent mineralocorticoid excess
apple peel syndrome
apudoma
arachnodactyly ataxia cataract aminoaciduria mental retardation

arachnodactyly mental retardation dysmorphism
arachnodactyly ossification abnormal mental retardation
arachnoid cyst
arbovirosis
arbovirus fever
arc syndrome
aredyld syndrome
arginase deficiency
argininemia
argininosuccinase deficiency
argininosuccinate synthetase deficiency
argininosuccinicaciduria
arhinia choanal atresia microphthalmia
arnold chiari malformation
arnold stckler bourne syndrome
aromatic l amino acid decarboxylase deficiency
arrhinia
arroyo garcia cimadevilla syndrome
arrythmogenic right ventricular dysplasia, familial
arterial dysplasia
arterial tortuosity
arterio hepatic dysplasia
arthritis short stature deafness
arthrogryposis congenital myopathic seizures
arthrogryposis due to muscular dystrophy
arthrogryposis ectodermal dysplasia other anomalies
arthrogryposis epileptic seizures migrational brain disorder
arthrogryposis iugr thoracic dystrophy
arthrogryposis like disorder
arthrogryposis like hand anomaly sensorineural
arthrogryposis multiplex congenita cns calcifications
arthrogryposis multiplex congenita distal
arthrogryposis multiplex congenita distal type 1
arthrogryposis multiplex congenita distal type 2
arthrogryposis multiplex congenita lissencephaly
arthrogryposis multiplex congenita neurogenic type
arthrogryposis multiplex congenita pulmonary hypoplasia
arthrogryposis multiplex congenita whistling face
arthrogryposis ophtalmoplegia retinopathy
arthrogryposis renal dysfunction cholestasis
arthrogryposis renal dysfunction cholestasis syndrome
arthrogryposis spinal muscular atrophy
arthroophtalmopathy hereditary progressive
arthropathy camptodactyly syndrome
arthropathy progressive pseudorheumatoid of childhood
arylsulfatase a deficiency
arylsulfatase a pseudodeficiency
arylsulfatase b deficiency
asbestos intoxication
asbestosis
ascher syndrome
aspartoacylase deficiency
aspartylglucosaminidase deficiency
aspartylglycosaminuria
asped syndrome
asperger syndrome
aspergillosis
asphyxiating thoracic dystrophy of the newborn
asplenia syndrome
asplenia with cardiovascular anomalies
asplenia with cystic liver kidney and pancreas
assas syndrome
astrocytoma
asymmetric crying facies
ataxia deafness optic atrophy lethal
ataxia deafness reardon type
ataxia deafness retardation syndrome
ataxia diabetes goiter gonadal insufficiency
ataxia hypogonadism choroidal dystrophy

ataxia lactic acidosis 1
ataxia myoclonies macular degeneration
ataxia ocular motor apraxia
ataxia opsoclonus myoclonus
ataxia optic atrophy hearing loss
ataxia pancytopenia syndrome
ataxia periodic vestibulocerebellar
ataxia photosensitivity short stature
ataxia spastic congenital miosis
ataxia tapetoretinal degeneration
ataxia telangiectasia
ataxia tonic upward deviation of eyes
atelencephaly
atelosteogenesis type 1
atelosteogenesis type 2
atherosclerosis epilepsy deafness
atkin flaitz patil smith syndrome
atp synthetase deficiency
atransferrinemia
atresia of small intestin
atresia of urethra
atrial cardiomyopathy with heart block
atrial fibrillation, familial
atrial myxoma, familial
atrial septal defect
atrial septal defect atrioventricular conduction
atrial septal defect dominant form
atrial tachyarrhythmia with short pr interval
atrichia mental and growth retardation
atrio-ventricular and ventriculo-arterial double discordia
atrioventricular defect blepharophimosis radial defects
aughton hufnagle syndrome
aughton sloan milad syndrome
aughton syndrome
aur syndrome
aural atresia multiple congenital anomalies mental retardation
auralcephalosyndactyly
auricular flutter
auriculoosteodysplasia beals type
ausems wittebol post hennekam syndrome
autism
autoimmune enteropathy haemolytic anaemia polyendocrinopathy
autoimmune lymphoproliferative syndrome
autoimmunization anti factor 8
autoimmunization anti factor VIIIc
autosomic dominant cerebellar ataxia
autosomic dominant spinocerebellar ataxia
axenfeld rieger anomaly hydrocephaly skeletal abnormalities
axial mesodermal dysplasia spectrum
axial osteosclerosis
ayazi syndrome
azoospermia sinopulmonary infections
Babesiosis
bader syndrome
baelz syndrome
bagatelle cassidy syndrome
bahemuka brown syndrome
baker vinters syndrome
balantidiasis
ballard syndrome
ballinger-wallace syndrome
bamboo hair syndrome
bamforth syndrome
bangstad syndrome
banki syndrome
bannayan zonana syndrome
baraitser brett piesowicz syndrome
baraitser burn fixen syndrome
baraitser burn fixen syndrome

baraitser rodeck garner syndrome
barakat syndrome
barber say syndrome
bardet biedl syndrome
bardet biedl syndrome type 1
bardet biedl syndrome type 2
bardet biedl syndrome type 3
bardet biedl syndrome type 4
barnicoat baraitser syndrome
barrett esophagus
barrow fitzsimmons syndrome
bart pumphrey syndrome
barth syndrome
bartsocas papa syndrome
bartter syndrome
bartter syndrome antenatal form
bartter syndrome antenatal hypercalciuric form
basal cell nevus anodontia abnormal bone mineralization
basan syndrome
basaran yilmaz syndrome
basilar impression primary
Bassen-Kornzweig syndrome
bassoe syndrome
battaglia neri syndrome
batten syndrome
baughman syndrome
bazex dupre christol syndrome
bazopoulou kyrkanidou syndrome
bbb syndrome
bbb syndrome x linked
bd syndrome
beals hecht syndrome
beals syndrome
bean syndrome
beardwell syndrome
beare stevenson syndrome
becker disease
beckwith wiedemann syndrome
beemer ertbruggen syndrome
beemer langer syndrome
behcet syndrome
behr syndrome
behrens baumann dust syndrome
beighton goldberg hof syndrome
bell's palsy
bellini chiumello rinoldi syndrome
ben ari shuper mimouni syndrome
benallegue lacete syndrome
bencze syndrome
benign autosomal dominant myopathy
benign chronic pemphigus familial of Hailey-Hailey
benign familial infantile convulsions
benign familial infantile epilepsy
bennion patterson syndrome
bentham driessen hanveld syndrome
beradinelli syndrome
berdon syndrome
berger disease
berk tabatznik syndrome
bernard soulier syndrome
besnier-boeck-schaumann disease
best disease
beta galactosidase deficiency
beta glucuronidase deficiency
beta mannosidosis
beta thalassemia
Beta-sarcoglycanopathy
betaketothiolase deficiency
bethlem myopathy

beveridge syndrome
bhaskar jagannathan syndrome
bianchine lewis syndrome
bickel fanconi glycogenosis
bicuspid aortic valve
bids syndrome
biemond syndrome
biemond syndrome type 1
biemond syndrome type 2
biermer disease
bifid nose dominant
bifunctional enzyme deficiency
bilateral renal agenesis
bilateral renal agenesis dominant type
biliary malformation renal tubular insufficiency
bilirubin uridinediphosphate glucuronosylyltransferase deficiency
billard toutain maheut syndrome
billet bear syndrome
binder syndrome
bindewald ulmer muller syndrome
binswanger disease
biotinidase deficiency
bird headed dwarfism montreal type
Birt-Hogg-Dube syndrome
bixler christian gorlin syndrome
bjornstad syndrome
Bjornstadt syndrome
blaichman syndrome
blastogenesis defect
blepharo cheilo dontic syndrome
blepharo facio skeletal syndrome
blepharo naso facial syndrome van maldergem type
blepharonasofacial malformation syndrome
blepharophimosis epicanthus inversus and ptosis
blepharophimosis nasal groove growth retardation
blepharophimosis ptosis esotropia syndactyly short stature
blepharophimosis ptosis syndactyly mental retardation
blepharophimosis radioulnar synostosis
blepharophimosis syndrome ohdo type
blepharophimosis telecanthus microstomia
blepharoptosis aortic anomaly
blepharoptosis cleft palate ectrodactyly dental anomalies
blepharoptosis myopia ectopia lentis
blepharospasm
blethen wenick hawkins syndrome
blomstrand syndrome
bloom syndrome
blount disease
blue cone monochromatism
blue rubber bleb nevus
bod syndrome
boeck sarcoid
bone dysplasia azouz type
bone dysplasia corpus callosum agenesis
bone dysplasia lethal holmgren type
bone dysplasia moore type
bone fragility craniosynostosis proptosis hydrocephalus
bone marrow failure neurologic abnormalities
bonneau beaumont syndrome
bonneman meinecke reich syndrome
bonnemann meinecke syndrome
book syndrome
boomerang dysplasia
booth haworth dilling syndrome
bor syndrome
borjeson forssman lehmann syndrome
bork stender schmidt syndrome
borreliosis
borrone di rocco crovato syndrome

boscherini galasso manca bitti syndrome
bosma henkin christiansen syndrome
Bothriocephalosis
botulism
boucher neuhauser syndrome
boudhina yedes khiari syndrome
bourneville syndrome
bourneville syndrome type 1
bourneville syndrome type 2
bouwes bavinck weaver ellis syndrome
bowen conradi syndrome
bowen hutterite syndrome
bowen syndrome
bowing congenital short bones
bowing of long bones congenital
boylan dew greco syndrome
brachioskeletogenital syndrome
brachman de lange syndrome
brachycephalofrontonasal dysplasia
brachycephaly deafness cataract mental retardation
brachydactylie types b et e combined
brachydactylous dwarfism mseleni type
brachydactyly absence of distal phalanges
brachydactyly anonychia
brachydactyly clinodactyly
brachydactyly deafness skeletal anomalies
brachydactyly dwarfism mental retardation
brachydactyly elbow wrist dysplasia
brachydactyly hypertension
brachydactyly long thumb type
brachydactyly mesomelia mental retardation heart defects
brachydactyly mohr wriedt type
brachydactyly nystagmus cerebellar ataxia
brachydactyly preaxial hallux varus
brachydactyly scoliosis carpal fusion
brachydactyly small stature face anomalies
brachydactyly smorgasbord type
brachydactyly symphalangism syndrome
brachydactyly temtamy type
brachydactyly tibial hypoplasia
brachydactyly type a1
brachydactyly type a2
brachydactyly type a3
brachydactyly type a4
brachydactyly type a5 nail dysplasia
brachydactyly type a6
brachydactyly type a7
brachydactyly type b
brachydactyly type c
brachydactyly type e
brachymesomelia renal syndrome
brachymesophalangy 2 and 5
brachymesophalangy mesomelic short limbs osseous anomalies
brachymesophalangy type 2
brachymetapody anodontia hypotrichosis albinoidism
brachymorphism onychodysplasia dysphalangism syndrome
brachyolmia
brachyolmia recessive hobaek type
brachyolmia toledo type
brachytelephalangy characteristic facies kallmann syndrome
braddock carey syndrome
braddock jones superneau syndrome
brain cavernous angioma
branched chain ketoaciduria
branchial arch defects
branchial arch syndrome x linked
branchial dysplasia mental retardation inguinal hernia
branchio oculo facial syndrome
branchio oculo facial syndrome hing type

branchio oto renal syndrome
braun bayer syndrome
breast and ovarian cancer
breast cancer familial
breast cancer type 1
breast cancer type 2
breast cancer type 3
brittle bone disease
brittle bone syndrome lethal type
brittle cornea syndrome
brittle hair mental deficit
broad-betalipoproteinemia
brodie chole griffin syndrome
bronchiectasis oligospermia
bronchiolitis obliterans organizing pneumonia
bronchiolitis obliterans with obstructive pulmonary disease
bronchogenic cyst
bronchopulmonar amyloidosis
bronspiegel zelnick syndrome
bruce winship syndrome
brucellosis
bruck syndrome
Brugada syndrome
brunner winter syndrome
brunoni syndrome
bruton type agammaglobulinemia
bruyn scheltens syndrome
budd-chiari syndrome
bulbospinal amyotrophy X linked
bull dog syndrome
bull nixon syndrome
bullous dystrophy macular type
bullous ichtyosiform erythroderma congenita
bullous pemphigoid
buntinx lormans martin syndrome
burkitt lymphoma
burn goodship syndrome
burnett schwartz berberian syndrome
buschke fischer brauer syndrome
buschke ollendorff syndrome
bustos simosa pinto cisternas syndrome
buttiens fryns syndrome
butyrylcholinesterase deficiency
byler disease
Bebe collodion syndrome
c syndrome
cacchi ricci disease
cach syndrome
cadasil
cafe au lait spots syndrome
caffey disease
cahmr syndrome
calcinosis raynaud phenomenon sclerodactyly telangiectasis
calderon gonzalez cantu syndrome
calloso genital dysplasia
callus disease
calpainopathy
calvarial hyperostosis
camera lituania cohen syndrome
camera stella syndrome
camfak syndrome
campomelia cumming type
campomelic dysplasia
camptobrachydactyly
camptocormia
camptocormism
camptodactyly fibrous tissue hyperplasia skeletal dysplasia
camptodactyly joint contractures facial skeletal defects
camptodactyly overgrowth unusual facies

camptodactyly syndrome guadalajara type 1
camptodactyly syndrome guadalajara type 2
camptodactyly taurinuria
camptodactyly vertebral fusion
camptomelic dwarfism
camurati engelmann disease
canale-smith syndrome
canavan disease
candidiasis familial chronic
cantalamessa baldini ambrosi syndrome
cantrell haller ravitsch syndrome
cantrell pentalogy
cantu sanchez corona fragoso syndrome
cantu sanchez corona garcia syndrome
cantu sanchez corona hernandes syndrome
Capillary leak syndrome with monoclonal gammopathy
capos syndrome
caratolo cilio pessagno syndrome
carbamoylphosphate synthetase deficiency
carbohydrate deficient glycoprotein syndrome
carbohydrate deficient glycoprotein syndrome type 1a
carbohydrate deficient glycoprotein syndrome type 1b
carbohydrate deficient glycoprotein syndrome type 1c
carbohydrate deficient glycoprotein syndrome type 2
carbohydrate deficient glycoprotein syndrome type 3
carbohydrate deficient glycoprotein syndrome type 4
carbon baby syndrome
carcinoid tumor
cardiac and laterality defects
cardiac conduction defect familial
cardiac diverticulum
cardiac malformation
cardiac valvular dysplasia x-linked
cardiofacial syndrome short limbs
cardiofaciocutaneous syndrome
cardiogenital syndrome
cardiomelic syndrome stratton koehler type
cardiomyopathic lentiginosis
cardiomyopathy cataract hip spine disease
cardiomyopathy diabetes deafness
cardiomyopathy dilated with conduction defect
cardiomyopathy dilated with conduction defect type 1
cardiomyopathy dilated with conduction defect type 2
cardiomyopathy due to anthracyclines
cardiomyopathy familial dilated
cardiomyopathy familial hypertrophic
cardiomyopathy hearing loss type trna lys gene mutation
cardiomyopathy hypogonadism metabolic anomalies
cardiomyopathy infantile fatal x linked
cardiomyopathy spherocytosis
cardioskeletal myopathy neutropenia
carey fineman ziter syndrome
carnevale canun mendoza syndrome
carnevale hernandez castillo syndrome
carnevale krajewska fischetto syndrome
carney syndrome
carnitine deficiency myopathic
carnitine palmitoyl transferase 1 deficiency
carnitine palmitoyl transferase 2 deficiency
carnitine systemic deficiency
carnitine transporter deficiency
carnitine-acylcarnitine translocase deficiency
carnosinase deficiency
carnosinemia
carpal deformity migrognathia microstomia
carpenter hunter type
carpenter syndrome
carpo tarsal osteochondromatosis
carpo tarsal osteolysis recessive

carrington syndrome
cartilage hair hypoplasia like syndrome
cartilage hair hypoplasia syndrome
cartwright nelson fryns syndrome
cassia stocco dos santos syndrome
castleman maladie
castro gago pombo novo syndrome
cat eye syndrome
cat rodrigues syndrome
catalase deficiency
cataract aberrant oral frenula growth retardation
cataract alopecia sclerodactyly
cataract anterior polar dominant
cataract ataxia deafness
cataract cardiomyopathy
cataract congenital autosomal dominant
cataract congenital dominant non nuclear
cataract congenital ichthyosis
cataract congenital volkmann type
cataract congenital with microphthalmia
cataract deafness hypogonadism
cataract hutterite type
cataract hyperostosis frontalis dislocating patella
cataract hypertrichosis mental retardation
cataract mental retardation anal atresia urinary defects
cataract mental retardation hypogonadism
cataract microcornea syndrome
cataract microcornea x linked
cataract microphthalmia septal defect
cataract skeletal anomalies
cataract total congenital
catch 22
catel manzke syndrome
caudal appendage deafness
caudal duplication
caudal dysgenesis familial type
caudal regression sequence
cayler syndrome
cca syndrome
ccge syndrome
cdg syndrome
cdg syndrome type 1a
cdg syndrome type 1b
cdg syndrome type 1c
cdg syndrome type 2
cdg syndrome type 3
cdg syndrome type 4
cdk4 linked melanoma
cecato de lima pinheiro syndrome
celiac disease
celiac disease epilepsy occipital calcifications
cenani lenz syndactylism
cennamo gangemi syndrome
central core myopathy
centromeric instability immunodeficiency syndrome
centrotemporal epilepsy
cephalopolysyndactyly
cephaloskeletal dysplasia
ceramidase deficiency
cerebellar ataxia areflexia pes cavus optic atrophy and sensorineural hearing loss
cerebellar ataxia dominant pure
cerebellar ataxia early onset with retained tendon reflex
cerebellar ataxia ectodermal dysplasia
cerebellar ataxia hypogonadotropic hypogonadism
cerebellar ataxia infantile with progressive external ophtalmoplegia
cerebellar ataxia x linked
cerebellar hypoplasia
cerebellar hypoplasia endosteal sclerosis

cerebellar hypoplasia tapetoretinal degeneration
cerebelloolivary atrophy
cerebelloparenchymal disorder 3
cerebellum agenesis hydrocephaly
cerebral calcification cerebellar hypoplasia
cerebral calcifications opalescent teeth phosphatiuria
cerebral cavernous malformation
cerebral cavernous malformations
cerebral gigantism
cerebral gigantism jaw cysts
cerebral gigantism nevo type
cerebral malformations hypertrichosis claw hands
cerebro costo mandibular syndrome
cerebro facio articular syndrome
cerebro facio thoracic dysplasia
cerebro oculo dento auriculo skeletal syndrome
cerebro oculo genital syndrome
cerebro oculo skeleto renal syndrome
cerebro reno digital syndrome
cerebroarthrodigital syndrome
cerebrohepatorenal syndrome
cerebrooculofacioskeletal syndrome
cerebroretinal vasculopathy
ceroid lipofuscinose neuronal
ceroid lipofuscinose neuronal 1 infantile
ceroid lipofuscinose neuronal 2 late infantile
ceroid lipofuscinose neuronal 3 juvenile
ceroid lipofuscinose neuronal 4 adult type
ceroid lipofuscinose neuronal 5 late infantile finnish variant
ceroid lipofuscinose neuronal 6 late infantile
cervical hypertrichosis neuropathy
cervical hypertrichosis peripheral neuropathy
cervical ribs sprengel anomaly polydactyly
cervical vertebral fusion
cervicooculoacoustic syndrome
cfc syndrome
cfc syndrome
chagas disease
Chanarin disease
chands syndrome
chang davidson carlson syndrome
chaotic atrial tachycardia
char douglas dungan syndrome
charcot disease
charcot marie tooth disease
charcot marie tooth disease deafness dominant type
charcot marie tooth disease deafness mental retardation
charcot marie tooth disease deafness recessive type
charcot marie tooth disease guadalajara neuronal type
charcot marie tooth disease intermediate form
charcot marie tooth disease neuronal type a
charcot marie tooth disease neuronal type b
charcot marie tooth disease neuronal type d
charcot marie tooth disease type 1a
charcot marie tooth disease type 1b
charcot marie tooth disease type 1c
charcot marie tooth disease type 2a
charcot marie tooth disease type 2b
charcot marie tooth disease type 2c
charcot marie tooth disease type 2d
charcot marie tooth neuropathy type 4a
charcot marie tooth neuropathy x linked recessive type 2
charcot marie tooth peroneal muscular atrophy, x linked type 1
charcot marie tooth type 1 aplasia cutis congenita
charcot marie tooth type 4b
charcot marie tooth x linked recessive type 3
charge association
charge like syndrome
charlevoix disease

charlie m syndrome
chediak higashi like syndrome
chediak higashi syndrome
cheilitis glandularis
chemke oliver mallek syndrome
chen kung ho kaufman mcalister syndrome
cherubism
cherubism gingival fibromatosis mental retardation
cherubism optic atrophy short stature
chiari type 1 malformation
CHILD syndrome
childhood ataxia with diffuse central nervous system hypomyelination
chime neuroectodermal dysplasia
chitayat haj chahine syndrome
chitayat meunier hodgkinson syndrome
chitayat moore del bigio syndrome
chitty hall baraitser syndrome
chitty hall webb syndrome
choanal atresia deafness cardiac defects dysmorphism
choledochal cyst hand malformation
cholera
cholestasis lymphedema syndrome
cholestasis pigmentary retinopathy cleft palate
cholestasis progressive familial intrahepatic
cholestasis progressive familial intrahepatic 1
cholestasis progressive familial intrahepatic 2
cholestasis progressive familial intrahepatic 3
cholestatic jaundice renal tubular insufficiency
cholesterol ester storage disease
chondrocalcinosis familiar articular
chondrodysplasia calcificans metaphysealis
chondrodysplasia lethal greenberg rimoin type
chondrodysplasia lethal neonatal
chondrodysplasia lethal recessive
chondrodysplasia pseudohermaphrodism syndrome
chondrodysplasia punctata
chondrodysplasia punctata brachytelephalangic
chondrodysplasia punctata conradi hunermann type
chondrodysplasia punctata ocular colobomata
chondrodysplasia punctata rhizomelic form
chondrodysplasia punctata sheffield type
chondrodysplasia punctata x linked dominant
chondrodysplasia punctata x linked recessive
chondrodysplasia situs inversus imperforate anus polydactyly
chondrodystrophia calcifians punctata
chondrodystrophy advanced carpotarsal ossification
chondrodystrophy sensorineural deafness
chondroectodermal dysplasia
chondromalacia
chondrysplasia punctata humero metacarpal type
chordoma
chorea acanthocytosis
chorea familial benign
choreoacanthocytosis amyotrophic
choreoathetosis familial paroxysmal
chorioretinopathy birdshot type
chorioretinopathy dominant form microcephaly
choroid plexus cyst
choroidal atrophy alopecia
choroideremia
choroideremia deafness obesity
choroideremia hypopituitarism
choroido cerebral calcification syndrome infantile form
christ siemens touraine syndrome
christian demyer franken syndrome
christian johnson angenieta syndrome
christian syndrome
christianson fourie syndrome
Chromomycosis

chromosome 1 ring
chromosome 10 ring
chromosome 12 ring
chromosome 14 ring
chromosome 18 ring
chromosome 19 ring
chromosome 20 ring
chromosome 21 ring
chromosome 22 ring
chromosome 4 ring
chromosome 6 ring
chromosome 7 ring
chromosome 8 ring
chronic autoimmune hepatitis
chronic berylliosis
chronic demyelinizing neuropathy with igm monoclonal gammapathy
chronic fatigue syndrome
chronic hiccup
chronic inflammatory rheumatism juvenile
chronic myeloid leukemia
chronic polyradiculonevritis
chronic, infantile, neurological, cutaneous, articular syndrome
chudley lowry hoar syndrome
chudley rozdilsky syndrome
churg-strauss syndrome
chylous ascites
ciliary discoordination due to random ciliary orientation
ciliary dyskinesia bronchiectasis
ciliary dyskinesia due to transposition of ciliary microtubules
cilliers beighton syndrome
cinca syndrome
circumscribed cutaneous aplasia of the vertex
circumscribed disseminated keratosis jadassohn lewandowsky type
citrullinemia
Clarkson disease
clayton smith donnai syndrome
cleft hand absent tibia
cleft limb heart malformation syndrome
cleft lip and or palate with mucous cysts of lower lip
cleft lip and palate malrotation cardiopathy
cleft lip palate abnormal thumbs microcephaly
cleft lip palate deafness sacral lipoma
cleft lip palate dysmorphism kumar type
cleft lip palate ectrodactyly
cleft lip palate facial eye heart intestinal anomalies
cleft lip palate incisor and finger anoamlies
cleft lip palate lip pits limb deficiency
cleft lip palate mental retardation corneal opacities
cleft lip palate oligodontia syndactyly pili torti
cleft lip palate pituitary deficiency
cleft lip palate tetraphocomelia
cleft lip retinopathy
cleft lip with or without cleft palate
cleft lower lip cleft lateral canthi chorioretinal degeneration
cleft palate cardiac defect ectrodactyly
cleft palate colobomata radial synostosis deafness
cleft palate heart disease polydactyly absent tibiae
cleft palate large ears small head
cleft palate lateral synechia syndrome
cleft palate short stature vertebral anomalies
cleft palate stapes fixation oligodontia
cleft palate x linked
cleft tongue syndrome
cleft upper lip median cutaneous polyps
clefting ectropion conical teeth
cleido rhizomelic syndrome
cleidocranial dysostosis
cleidocranial dysplasia
cleidocranial dysplasia micrognathia absent thumbs

cloacal exstrophy
clouston syndrome
cloverleaf skull bone dysplasia
cloverleaf skull generalised bone dysplasia
cloverleaf skull micromelia thoracic dysplasia
cloverleaf skull syndrome
cmv antenatal infection
coach syndrome
coarctation of aorta dominant
coarse face hypotonia constipation
coats disease
cocaine antenatal infection
cockayne syndrome
cockayne syndrome type 1
cockayne syndrome type 2
cockayne syndrome type 3
cockayne touraine type epidermolysis bullosa simplex
codas syndrome
coenzyme q cytochrome c reductase deficiency of
coffin lowry syndrome
coffin siris syndrome
coffin syndrome
cofs syndrome
cogan's syndrome
cohen hayden syndrome
cohen lockood wyborney syndrome
cohen syndrome
colavita kozlowski syndrome
cole carpenter syndrome
coleman randall syndrome
collins pope syndrome
collins sakati syndrome
coloboma chorioretinal cerebellar vermis aplasia
coloboma hair abnormality
coloboma of choroid and retina
coloboma of eye lens
coloboma of iris
coloboma of lens ala nasi
coloboma of macula
coloboma of macula type b brachydactyly
coloboma of optic papilla
coloboma porencephaly hydronephrosis
coloboma uveal with cleft lip palate and mental retardation
coloboma, ocular
colobomata unilobar lung heart defect
colobomatous microphthalmia
colobomatous microphthalmia heart disease hearing loss
colon cancer familial nonpolyposis
colonic atresia
colver steer godman syndrome
combarros calleja leno syndrome
common mesentery
complement component 2 deficiency
complement component receptor 1
complete atrioventricular canal
complex 1 mitochondrial respiratory chain deficiency of
complex 2 mitochondrial respiratory chain deficiency of
complex 3 mitochondrial respiratory chain deficiency of
complex 4 mitochondrial respiratory chain deficiency of
complex 5 mitochondrial respiratory chain deficiency of
conductive deafness malformed external ear
conductive deafness micrognathia
cone dystrophy x linked
cone rod dystrophy
cone rod dystrophy amelogenesis imperfecta
congenital absence of the uterus and vagina
congenital adrenal hyperplasia
congenital adrenal hyperplasia type 1
congenital adrenal hyperplasia type 2

congenital adrenal hyperplasia type 3
congenital adrenal hyperplasia type 4
congenital adrenal hyperplasia type 5
congenital alopecia x linked
congenital aneurysms of the great vessels
congenital benign spinal muscular atrophy dominant
congenital bronchobiliary fistula
congenital central alveolar hypoventilation
congenital centronuclear myopathy
congenital craniosynostosis maternal hyperthyroidism
congenital cystic eye multiple ocular and intracranial anomalies
congenital dyserythropoietic anemia
congenital dyserythropoietic anemia type 1
congenital dyserythropoietic anemia type 2
congenital dyserythropoietic anemia type 3
congenital erythropoiesis
congenital fiber type disproportion
congenital heart disease ptosis hypodontia craniosynostosis
congenital heart disease radio ulnar synostos mental retardation
Congenital Hemidysplasia with Ichtyosiform erythroderma and Limbs Defects
congenital hypothyroidism
congenital hypotrichosis milia
congenital ichthyosis microcephalus quadriplegia
congenital ichtyosiform erythroderma
congenital intrinsic factor deficiency
congenital lobar emphysema
congenital lymphedema
congenital megalo-ureter
congenital mesoblastic nephroma
congenital microvillous atrophy
congenital mitral malformation
congenital mitral stenosis
congenital muscular dystrophy syringomyelia
congenital nephrotic syndrome finnish type
congenital retinal telangiectasia
congenital short bowel
congenital short femur
congenital stenosis of cervical medullary canal
congenital unilateral pulmonary hypoplasia
congenital vagal hyperreflexivity
congenital wooly hair
connective tissue dysplasia spellacy type
connexin 26 anomaly
conotruncal heart malformation
conradi hunermann syndrome
constrictive bronchiolitis
continuous muscle fiber activity hereditary
continuous spike-wave during slow sleep syndrome
contractural arachnodactyly congenital
contractures ectodermal dysplasia cleft lip palate
contractures hyperkeratosis lethality
contractures of feet muscle atrophy oculomotor apraxia
convulsions benign familial neonatal
convulsions benign familial neonatal dominant form
cooks syndrome
cooley anemia
coper transport disease
copper deficiency familial benign
cormier rustin munnich syndrome de
corneal anesthesia deafness mental retardation
corneal cerebellar syndrome
corneal crystals myopathy neuropathy
corneal dystrophy epithelial short stature
corneal dystrophy ichthyosis microcephaly mental retardation
corneal dystrophy perceptive deafness
corneal dystrophy pigmentary anomaly malabsorption
cornelia de lange syndrome
corneodermatoosseous syndrome
coronal synostosis syndactyly jejunal atresia

coronaro-cardiac fistula
coronary arteries congenital malformation
corpus callosum agenesis
corpus callosum agenesis double urinary collecting system
corpus callosum agenesis neuronopathy
corpus callosum agenesis of blepharophimosis robin
corpus callosum agenesis of with chorioretinal abnormality
corpus callosum agenesis polysyndactyly
corpus callosum dysgenesis cleft spasm
corpus callosum dysgenesis hypopituitarism
corpus callosum dysgenesis x linked recessive
corrected transposition
corsello opitz syndrome
cortada koussef matsumoto syndrome
cortes lacassie syndrome
cortical blindness mental retardation polydactyly
cortical hyperostosis syndactyly
corticobasal degeneration
costello syndrome
costocoracoid ligament congenitally short
costovertebral segmentation defect mesomelia
cote adamopoulos pantelakis syndrome
cote katsantoni syndrome
cousin walbraum cegarra syndrome
covesdem syndrome
cowchock wapner kurtz syndrome
cowden syndrome
coxoauricular syndrome
cramer niederdellmann syndrome
Crandall syndrome
crane heise syndrome
cranio fronto nasal dysplasia poland anomaly
cranio osteoarthropathy
cranioacrofacial syndrome
craniocarpotarsal dystrophy
craniocerebellocardiac dysplasia
craniodiaphyseal dysplasia
craniodigital syndrome mental retardation
cranioectodermal dysplasia
craniofacial and osseous defects mental retardation
craniofacial and skeletal defects
craniofacial deafness hand syndrome
craniofacial digital genital anomalies
craniofacial dysostosis arthrogryposis progeroid appearance
craniofacial dysostosis genital dental cardiac anomalies
craniofacial dyssynostosis
craniofaciocardioskeletal syndrome
craniofaciocervical osteoglyphic dysplasia
craniofrontonasal dysplasia
craniofrontonasal syndrome teebi type
craniometaphyseal dysplasia dominant type
craniometaphyseal dysplasia recessive type
craniomicromelic syndrome
craniostenosis
craniostenosis cataract
craniostenosis with congenital heart disease mental retardation
craniosynostosis
craniosynostosis alopecia brain defect
craniosynostosis arthrogryposis cleft palate
craniosynostosis autosomal dominant
craniosynostosis brachydactyly
craniosynostosis cleft lip palate arthrogryposis
craniosynostosis contractures cleft
craniosynostosis dandy walker hydrocephalus
craniosynostosis exostoses nevus epibulbar dermoids
craniosynostosis fibular aplasia
craniosynostosis fontaine type
craniosynostosis herrmann opitz type
craniosynostosis hydrancephaly thumb aplasia

craniosynostosis maroteaux fonfria type
craniosynostosis mental retardation clefting syndrome
craniosynostosis mental retardation heart defects
craniosynostosis midfacial hypoplasia foot abnormalities
craniosynostosis philadelphia type
craniosynostosis radial aplasia syndrome
craniosynostosis radial aplasia syndrome
craniosynostosis radial aplasia type imaizumi
craniosynostosis synostoses hypertensive nephropathy
craniosynostosis warman type
craniotelencephalic dysplasia
craniotubular syndrome
crash syndrome
crawfurd syndrome
creatine deficiency
Creeping disease
crest syndrome
cretinism athyreotic
creutzfeldt-jakob disease
cri du chat syndrome
crigler najjar syndrome
crisponi syndrome
criss cross
criss cross
criswick schepens syndrome
crohn disease
crome syndrome
cronkhite canada syndrome
cross syndrome
crossed polydactyly type 1
crossed polysyndactyly
crouzon craniofacial dysostosis
crouzon disease
crow fukase syndrome
cryoglobulinemia
cryptococcosis
cryptogenic organized pneumopathy
cryptomicrotia brachydactyly syndrome excess fingertip arch
cryptophthalmos syndrome
cryptorchidism arachnodactyly mental retardation
cryptosporidiosis
culler jones syndrome
curly hair ankyloblepharon nail dysplasia syndrome
currarino triad
curry hall syndrome
curry hall syndrome
curry jones syndrome
Curtaneous larva migrans
Curth-Macklin ichtyosis
curtis rogers stevenson syndrome
cushing syndrome familial
cutaneous albinism hermine phenotype
cutaneous lymphoma
cutaneous photosensitivity colitis lethal
cutaneous vascularitis
cutis gyratum acanthosis nigricans craniosynostosis
cutis laxa
cutis laxa corneal clouding mental retardation
cutis laxa dominant type
cutis laxa joint laxity retarded development
cutis laxa osteoporosis
cutis laxa recessive type 1
cutis laxa recessive type 2
cutis laxa x linked
cutis marmorata telangiectatica congenita
cutis verticis gyrata
cutis verticis gyrata
cutis verticis gyrata mental deficiency
cutis verticis gyrata thyroid aplasia mental retardation

cutler bass romshe syndrome
Cyclosporosis
cypress facial neuromusculosqueletal syndrome
cystathionine beta synthase deficiency
cystathioninuria
cystic adenomatoid malformation of the lung
cystic angiomatosis of bone, diffuse
cystic fibrosis
cystic fibrosis gastritis megaloblastic anaemia
cystic hamartoma of lung and kidney
cystic hygroma lethal cleft palate
cystic medial necrosis of aorta
cysticercosis
cystin transport protein defect of
cystinosis
cystinuria
cystinuria lysinuria
cytochrome c oxidase deficiency of
cytomegalovirus antenatal infection
cytomegalovirus infection congenital
cytoplasmic body myopathy
czeizel brooser syndrome
czeizel losonci syndrome
czeizel syndrome
d 2-hydroxyglutaricaciduria
d ercole syndrome
d-glycerate dehydrogenase deficiency
d-glycerate kinase deficiency
d-glycericacidemia
da silva syndrome
dacryocystitis osteopoikilosis
daentl townsend siegel syndrome
dahlberg borer newcomer syndrome
daish hardman lamont syndrome
dandy walker facial hemangioma
dandy walker macrocephaly
dandy walker malformation
dandy walker malformation postaxial polydactyly
dandy walker syndrome recessive form
dandy walker syndrome recessive x linked
daneman davy mancer syndrome
darier disease
davenport donlan syndrome
david syndrome
davis lafer syndrome
de barsy syndrome
de hauwere leroy adriaenssens syndrome
De la Chapelle syndrome
de morsier syndrome
de sanctis cacchione syndrome
de smet fabry fryns syndrome de
deaf blind hypopigmentation
deafness alopecia hypogonadism
deafness autosomal dominant nonsyndromic sensorineural
deafness autosomal dominant nonsyndromic sensorineural dfna1 linked
deafness autosomal dominant nonsyndromic sensorineural dfna10 linked
deafness autosomal dominant nonsyndromic sensorineural dfna11 linked
deafness autosomal dominant nonsyndromic sensorineural dfna12 linked
deafness autosomal dominant nonsyndromic sensorineural dfna13 linked
deafness autosomal dominant nonsyndromic sensorineural dfna14 linked
deafness autosomal dominant nonsyndromic sensorineural dfna15 linked
deafness autosomal dominant nonsyndromic sensorineural dfna2 linked
deafness autosomal dominant nonsyndromic sensorineural dfna3 linked
deafness autosomal dominant nonsyndromic sensorineural dfna4 linked
deafness autosomal dominant nonsyndromic sensorineural dfna5 linked
deafness autosomal dominant nonsyndromic sensorineural dfna6 linked
deafness autosomal dominant nonsyndromic sensorineural dfna7 linked
deafness autosomal dominant nonsyndromic sensorineural dfna9 linked
deafness blindness dystonia fractures

deafness conductive ptosis skeletal anomalies
deafness conductive stapedial ear malformation facial palsy
deafness congenital onychodystrophy recessive
deafness craniofacial syndrome
deafness dominant indonesian type
deafness enamel hypoplasia nail defects
deafness epiphyseal dysplasia short stature
deafness goiter stippled epiphyses
deafness hyperuricemia neurologic ataxia
deafness hypogonadism syndrome
deafness hypospadias metacarpal and metatarsal synostosis
deafness mesenteric diverticula of small bowel neuropathy
deafness mixed with perilymphatic gusher x linked
deafness nephritis ano rectal malformation
deafness neurosensory pituitary dwarfism
deafness nonsyndromic connexin 26 linked
deafness oligodontia syndrome
deafness onychodystrophy dominant form
deafness optic atrophy syndrome
deafness peripheral neuropathy arterial disease
deafness progressive cataract autosomal dominant
deafness progressive high tone neural
deafness skeletal dysplasia lip granuloma
deafness symphalangism
deafness vitiligo achalasia
deafness white hair contractures papillomas
deafness x linked
deafness x linked (dfn1)
deafness x linked (dfn2)
deafness x linked (dfn3)
deafness x linked (dfn4)
deafness x linked (dfn6)
deafness-tubular acidosis-anemia
deal barratt dillon syndrome
deciduous skin
defect in synthesis of adenosylcobalamin
defective apolipoprotein-b100
defective expression of hla class 2
Degos 'en cocarde' erythrokeratoderma
Degos's malignant atrophic papulosis
dehydratase deficiency
dehydrated hereditary stomatocytosis
dejerine klumpke paralysis
delayed membranous cranial ossification
delayed speech facial asymetry strabismus ear lobe creases
deleted in azoospermia
deletion 10p
deletion 10pter
deletion 10q
deletion 11p
deletion 11p11 p12
deletion 11p13
deletion 11q partielle
deletion 12p12 p11
deletion 12p13
deletion 13q
deletion 13q14
deletion 13q22
deletion 13q32
deletion 14q partial duplication 14p partial
deletion 14q11
deletion 14q31
deletion 14qter
deletion 15q1
deletion 15q25
deletion 17q23 q24
deletion 18p
deletion 18q
deletion 18q23

deletion 1p
deletion 1p22 p13
deletion 1p31 p22
deletion 1p32
deletion 1p34 p32
deletion 1p36
deletion 1q21 q25
deletion 1q25 q32
deletion 1q32 q42
deletion 1q4
deletion 20p
deletion 21q22
deletion 2p22
deletion 2pter p24
deletion 2q
deletion 2q duplication 1p
deletion 2q24
deletion 3p
deletion 3p14 p11
deletion 3p25
deletion 3q13
deletion 3q21 23
deletion 3q27
deletion 4p
deletion 4p14 p16
deletion 4q
deletion 4q32
deletion 5p
deletion 5q35
deletion 6p23
deletion 6q
deletion 6q1
deletion 6q13 q15
deletion 6q16 q21
deletion 6q2
deletion 7
deletion 7q2
deletion 7q21
deletion 7q3
deletion 8p
deletion 8p23 1
deletion 8q
deletion 8q12 21
deletion 8q21 q22
deletion 9p
deletion xp22 pter
deletion xq28
delleman oorthuys syndrome
delta 1 pyrroline 5 carboxylate dehydrogenase deficiency
Delta-sarcoglycanopathy
dementia hereditary multi infarct type
dementia progressive lipomembranous polycysta
Demodicidosis
dengue
dennis cohen syndrome
dennis fairhurst moore syndrome
dent disease
dental aberrations steroid dehydrogenase deficiency
dentatrorubral pallidoluysian atrophy
dentin dysplasia sclerotic bones
denys drash syndrome
der kaloustian jarudi khoury syndrome
der kaloustian mcintosh silver syndrome
dermatitis herpetiformis
dermatocardioskeletal syndrome boronne type
dermatoleukodystrophy
dermatomyositis
dermatoosteolysis kirghizian type
dermochondrocorneal dystrophy of Francois

dermoodontodysplasia
dermopathy restritive lethal
desbuquois grenier michel syndrome
desbuquois syndrome
desmin related myopathy
desmoid disease
developmental delay hypotonia extremities hypertrophy
developmental dysphasia familial
devriendt legius fryns syndrome
devriendt vandenberghe fryns syndrome
dexamethasone sensitive hypertension
dextrocardia
dextrocardia bronchiectasis sinusitis
dextrocardia microphthalmia cleft palate mental retardation
diabete insipide nephrogenique dominant type
diabetes epiphyseal dysplasia
diabetes hypogonadism deafness mental retardation
diabetes insipidus nephrogenic
diabetes insipidus nephrogenic recessive type
diabetes insipidus nephrogenic type 1
diabetes insipidus nephrogenic type 2
diabetes insipidus nephrogenic type 3
diabetes insipidus nephrogenic x linked
diabetes mellitus and insipidus optic atrophy
diabetes persistent mullerian ducts
diabetic embryopathy
diaphragmatic agenesia
diaphragmatic agenesis radial aplasia omphalocele
diaphragmatic defect limb deficiency skull defect
diaphragmatic hernia abnormal face limb
diaphragmatic hernia congenital
diaphragmatic hernia exomphalos corpus callosum agenesis
diaphragmatic hernia upper limb defects
diaphyseal dysplasi anaemia
diarrhea chronic with villous atrophy
diarrhea polyendocrinopathy infections x linked
diastematomyelia
diastrophic dwarfism
diastrophic dysplasia
dibasicaminoaciduria 2
dibasicaminoaciduria type 1
dicarboxylicaminoaciduria
didmoad syndrome
die smulders droog van dijk syndrome de
die smulders vles fryns syndrome
diencephalic syndrome
diethylstilbestrol antenatal infection
diffuse neonatal haemangiomatosis
diffuse palmoplantar keratoderma bothnian type
digeorge syndrome
digestive duplication
digitorenocerebral syndrome
digitotalar dysmorphism
dihydropteridine reductase deficiency
dihydropyrimidine dehydrogenase deficiency
dincsoy salih patel syndrome
dinno shearer weisskopf syndrome
diomedi bernardi placidi syndrome
dionisi vici sabetta gambarara syndrome
Diphallia
diphallus rachischisis imperforate anus
diphosphoglycerate mutase deficiency of erythrocyte
diphtheria
diplegia congenital facial
diprosopia
discoid lupus
dislocation of the hip dysmorphism
disomy 1q12 q21
disomy 9q21

disorder in the hormonal synthesis with or without goiter
disorganization syndrome
dissecting cellulitis of the scalp
dissecting cellulitis of the scalp
distal arthrogryposis moore weaver type
distal myopathy
distal myopathy Markesbery-Griggs type
distal myopathy Nonaka type
distal myopathy welander type, swedish type
distal myopathy with vocal cord weakness
distal primary familial acidosis
distal primary familial acidosis autosomal dominant
distal primary familial acidosis autosomal recessive
distal spinal muscular atrophy vocal cord paralysis
distichiasis heart congenital anomalies
distomatosis
dk phocomelia syndrome
dobrow syndrome
dominant cleft palate
dominant ichtyosis vulgaris
dominant zonular cataract
donnai barrow syndrome
donohue syndrome
door syndrome
dopa responsive dystonia
Dopamine beta-hydroxylase deficiency
double cortex
double discordia
double fingernail of fifth finger
double outlet left ventricule
double outlet right ventricule
double uterus-hemivagina-renal agenesis
double y
drachtman weinblatt sitarz syndrome
dracunculiasis
drash syndrome
duane anomaly mental retardation
duane syndrome
dubin johnson syndrome
dubowitz syndrome
duhring brocq disease
duker weiss siber syndrome
duodenal atresia
duodenal atresia tetralogy of fallot
duplication 1 mosaicism
duplication 10p
duplication 10pter p13
duplication 10q partial
duplication 11q
duplication 11q23
duplication 12 mosaicism
duplication 12p
duplication 12q
duplication 13
duplication 13p
duplication 13q
duplication 14 mosaicism
duplication 14q partial deletion 14p partial
duplication 14qprox
duplication 14qter
duplication 15 mosaicism
duplication 15q
duplication 16 mosaicism
duplication 16p
duplication 16q
duplication 17 mosaicism
duplication 17p
duplication 17p11 2
duplication 18

duplication 18 mosaicism
duplication 18p
duplication 18q
duplication 19q
duplication 1p21 p32
duplication 1q12 q21
duplication 1q32 qter
duplication 1q42 11 q42 12
duplication 1q42 qter
duplication 2 mosaicism
duplication 20 mosaicism
duplication 20p
duplication 22
duplication 22q11 q13
duplication 2p
duplication 2p13 p21
duplication 2pter p24
duplication 2q
duplication 2q37
duplication 3 mosaicism
duplication 3p
duplication 3p25
duplication 3q
duplication 3q13 2 q25
duplication 4p
duplication 4q
duplication 4q21
duplication 4q25 qter
duplication 5p
duplication 5pter p13 3
duplication 5q
duplication 6p
duplication 6q
duplication 7 mosaicism
duplication 7p
duplication 7p13 p12 2
duplication 7q
duplication 8
duplication 8p
duplication 8q
duplication 9 mosaicism
duplication 9p partial
duplication 9q21
duplication 9q32
duplication of leg mirror foot
duplication of the thumb unilateral biphalangeal
duplication of urethra
duplication xp3
duplication xpter xq13
duplication xq
duplication xq13 1 q21 1
duplication xq25
dupont sellier chochillon syndrome
dwarfism bluish sclerae
dwarfism deafness retinitis pigmentosa
dwarfism familial synovial chondromatosis
dwarfism lethal type advanced bone age
dwarfism mental retardation eye abnormality
dwarfism seckel type
dwarfism short limb absent fibulas very short digits
dwarfism stiff joint ocular abnormalities
dwarfism syndesmodysplasic
dwarfism tall vertebrae
dwarfism thin bones multiple fractures
dyggve melchior clausen disease
dykes markes harper syndrome
Dyschondroplasia
dyschondrosteosis
dyschondrosteosis nephritis

dyschromatosis symmetrica hereditaria
dyschromatosis universalis
dysequilibrium syndrome
Dysferlinopathy
dysfibrinogenemia, familial
dysgerminoma
dysharmonic skeletal maturation muscular fibre disproportion
dyskeratosis congenita of zinsser cole engman
dysmorphism abnormal vocalization mental retardation
dysmorphism cleft palate loose skin
dysmorphism corpus callosum agenesis colobomas
dysmorphism multiple structural anomalies
dysosteosclerosis
dysostosis acrofacial postaxial
dysostosis peripheral
dysostosis stanescu type
dysphasic dementia hereditary
dysplasia epiphysealis hemimelica
dysplasia olfactogenitalis of de morsier
dysplastic cortical hyperostosis
dysplastic nevus
dysproconvertinemia
dysprothrombinemia
dysraphism cleft lip palate limb reduction defects
dyssegmental dysplasia glaucoma
dyssegmental dysplasia silverman handmaker type
dystonia musculorum deformans
dystonia musculorum deformans type 1
dystonia musculorum deformans type 2
dystonia progressive with diurnal variation
dystrophic epidermolysis bullosa inversa
Dystrophinopathy
ear patella short stature syndrome
earlobes thickened conductive deafness from incudo
ebola virus disease
ebstein anomaly
ecp syndrome
ectodermal dysplasia absent dermatoglyphics
ectodermal dysplasia adrenal cyst
ectodermal dysplasia alopecia preaxial polydactyly
ectodermal dysplasia anhidrotic
ectodermal dysplasia arthrogryposis diabetes mellitus
ectodermal dysplasia bartalos type
ectodermal dysplasia berlin type
ectodermal dysplasia blindness
ectodermal dysplasia cataracts kyphoscoliosis
ectodermal dysplasia ectrodactyly macular dystrophy
ectodermal dysplasia hydrotic
ectodermal dysplasia hypohidrotic autosomal dominant
ectodermal dysplasia hypohidrotic autosomal recessive
ectodermal dysplasia hypohidrotic hypothyroidism ciliary dyskin
ectodermal dysplasia margarita type
ectodermal dysplasia mental retardation cns malformation
ectodermal dysplasia mental retardation syndactyly
ectodermal dysplasia neurosensory deafness
ectodermal dysplasia osteosclerosis
ectodermal dysplasia tricho odonto onychial type
ectodermic dysplasia anhidrotic cleft lip
ectopia lentis chorioretinal dystrophy myopia
ectopia lentis isolated
ectopic coarctation
ectopic ossification familial type
ectrodactyly cardiopathy dysmorphism
ectrodactyly cleft palate syndrome
ectrodactyly diaphragmatic hernia corpus callosum agenesis
ectrodactyly dominant form
ectrodactyly ectodermal dysplasia cleft lip palate
ectrodactyly ectrodermal dysplasia
ectrodactyly polydactyly

ectrodactyly recessive form
ectrodactyly spina bifida cardiopathy
ectropion inferior cleft lip and or palate
eczema thrombocytopenia immunodeficiency syndrome
edinburgh malformation syndrome
edwards patton dilly syndrome
eec syndrome
eec syndrome without cleft lift palate
eem syndrome
ehlers danlos syndrome classic type
ehlers danlos syndrome hypermobile type
ehlers danlos syndrome type 1
ehlers danlos syndrome type 2
ehlers danlos syndrome type 3
ehlers danlos syndrome type 4 autosomal dominant
ehlers danlos syndrome type 6
ehlers danlos syndrome type 7a
ehlers danlos syndrome type 7b
ehlers danlos syndrome type 7c
ehlers danlos syndrome vascular type
ehlers-danlos syndrome arthrochalasic type
ehlers-danlos syndrome dermatosparaxis type
ehlers-danlos syndrome kyphoscoliotic type
ehrlichiosis
eisenmenger complex
elejalde syndrome
elliott ludman teebi syndrome
elliptocytosis
ellis van creveld syndrome
ellis yale winter syndrome
emery dreifuss muscular dystrophy dominant type
emery dreifuss muscular dystrophy X linked
emery nelson syndrome
emphysema-penoscrotal web-deafness-mental retardation
enamel hypoplasia cataract hydrocephaly
enamel renal syndrome
encephalo cranio cutaneous lipomatosis
encephalocele anterior
encephalocele frontal
encephalopathy basal ganglia calcification
encephalopathy intracerebral calcification retinal degeneration
encephalopathy progressive optic atrophy
encephalopathy subacute spongiform gerstmann-straussler type
encephalophathy recurrent of childhood
Enchondromatosis
enchondromatosis dwarfism deafness
endodermal sinus tumor
endomyocardial fibroelastosis
endosteal hyperostosis worth type
eng strom syndrome
engelhard yatziv syndrome
enolase deficiency
enolase type 1deficiency
enolase type 2 deficiency
enolase type 3 deficiency
enolase type 4 deficiency
enterovirus antenatal infection
envenomization by bothrops lanceolatus
envenomization by the martinique lancehead viper
eosinophilic cellulitis
eosinophilic gastroenteritis
eosinophilic granuloma
eosinophilic idiopathic chronic pneumopathy
ependymoma
epidemic hemorragic fever
epidermal nevus vitamin d resistant rickets
epidermolysa bullosa simplex and limb girdle muscular dystrophy
epidermolysis bullosa dermolytic
epidermolysis bullosa dystrophica bart type

epidermolysis bullosa dystrophica dominant type
epidermolysis bullosa dystrophica hallopeau siemens
epidermolysis bullosa generalized atrophic benign
epidermolysis bullosa herpetiformis Dowling-Meara type
epidermolysis bullosa intraepidermic
epidermolysis bullosa junctional
epidermolysis bullosa junctional with pyloric atresia
epidermolysis bullosa of hands and feet
epidermolysis bullosa pretibial
epidermolysis bullosa simplex anodontia hair nail disorders
epidermolysis bullosa simplex koebner type
epidermolysis bullosa simplex ogna type
epidermolytic hyperkeratosis
epidermolytic palmoplantar keratoderma vorner type
epilepsy benign neonatal
epilepsy benign neonatal dominant form
epilepsy benign neonatal recessive form
epilepsy demantia amelogenesis imperfecta
epilepsy juvenile absence
epilepsy mental deterioration finnish type
epilepsy microcephaly skeletal dysplasia
epilepsy occipital calcifications
epilepsy partial familial
epilepsy progressive myoclonic type 1
epilepsy progressive myoclonic type 2
epilepsy telangiectasia
epilepsy with myoclono-astatic crisis
epilepsy, nocturnal frontal lobe type
epimetaphyseal dysplasia cataract
epimetaphyseal skeletal dysplasia
epiphyseal dysplasia dysmorphism camptodactyly
epiphyseal dysplasia hearing loss dysmorphism
epiphyseal dysplasia microcephaly nystagmus
epiphyseal dysplasia multiple
epiphyseal dysplasia multiple dominant type
epiphyseal dysplasia multiple early onest diabetes mellitus
epiphyseal dysplasia multiple myopia conductive deafness
epiphyseal stippling syndrome osteoclastic hyperplasia
epstein barr virus mononucleosis
epstein syndrome
erdheim disease
eronen somer gustafsson syndrome
erosive pustular dermatosis of the scalp
erythermalgia
erythroderma desquamativa of leiner
erythroderma lethal congenital
erythrokeratodermia ataxia
erythrokeratodermia variabilis mendes da costa type
escher hirt syndrome
escobar syndrome
esophageal atresia coloboma talipes
esthesioneuroblastoma
ethylmalonic aciduria
euhidrotic ectodermal dysplasia
eunuchoidism familial
evans syndrome
ewing sarcoma
exencephaly
exner syndrome
exomphalos macroglossia gigantism syndrome
exostoses anetodermia brachydactyly type e
exostoses multiple
exostoses multiple type 1
exostoses multiple type 2
exostoses multiple type 3
exstrophy of the bladder-epispadias
exstrophy of the bladder-epispadias complex
exsudative retinopathy familial
exsudative retinopathy familial autosomal dominant

exsudative retinopathy familial autosomal recessive
exsudative retinopathy familial x linked recessive
extensor tendons of finger anomalies
extrahepatic biliary atresia
extrasystoles short stature hyperpigmentation microcephaly
eye defects arachnodactyly cardiopathy
eyebrows and eyelashes absence mental retardation
eyebrows duplication syndactyly
eyelashes long mental retardation
Eymerynopathy
fabry disease
faces syndrome
facial asymetry temporal seizures
facial cleft microtia asternia
facial clefting corpus callosum agenesis
facial dysmorphism macrocephaly myopia dandy walker
facial dysmorphism shawl scrotum joint laxity syndrome
facial ectodermal dysplasia
facial paresis partial unilateral
facies unusual arthrogryposis advanced skeletal maturation
facio digito genital syndrome recessive form
facio skeletal genital syndrome rippberger type
facio thoraco genital syndrome
faciocardiomelic dysplasia lethal
faciocardiorenal syndrome
faciodigitogenital syndrome
faciooculoacousticorenal syndrome
facioscapulohumeral muscular dystrophy
faciothoracoskeletal syndrome
factor 10 deficiency
factor 11 deficiency
factor 12 deficiency
factor 13 deficiency
factor 2 deficiency
factor 5 deficiency
factor 7 deficiency
factor 8 deficiency
factor 9 deficiency
fahr syndrome
fallot complex mental growth retardation
familal cholemia
familial amniotic bands
familial amyloid polyneuropathy
familial aortic dissection
familial band heterotopia
familial benign hypercalcemia
familial benign hypercalcemia type 1
familial benign hypercalcemia type 2
familial benign hypercalcemia type 3
familial combined hyperlipidemia
familial conotruncal cardiopathy
familial dysautonomia
familial glucocorticoid deficiency
familial hypertension
familial hypopituitarism
familial intestinal polyatresia syndrome
familial nasal acilia
familial non-immune hyperthyroidism
familial opposable triphalangeal thumbs duplication of the
familial partial epilepsy with variable focus
familial porencephaly
familial supernumerary nipples
familial symmetric lipomatosis
familial temporal epilepsy
familial thyroglossal duct cyst
familial variable immunodeficiency
familial veinous malformations
familial ventricular tachycardia
familial visceral myopathy

famililal adenomatous polyposis
fanconi anemia
fanconi anemia type 1
fanconi anemia type 2
fanconi anemia type 3
fanconi bickel syndrome
fanconi ichthyosis dysmorphism
fanconi like syndrome
fanconi pancytopenia
fanconi pancytopenia type 1
fanconi pancytopenia type 2
fanconi pancytopenia type 3
fanconi syndrome renal with nephrocalcinosis and renal stones
fara chlupackova syndrome
farber lipogranulomatosis
fas deficiency
faulk epstein jones syndrome
faye petersen ward carey syndrome
fechtner syndrome
feigenbaum bergeron richardson syndrome
feigenbaum bergeron syndrome
feingold syndrome
feingold trainer syndrome
female pseudohermaphrodism
female pseudohermaphrodism genuardi type
femoral facial syndrome
femur bifid monodactylous ectrodactyly
femur fibula ulna syndrome
fenton wilkinson toselano syndrome
ferlini ragno calzolari syndrome
fernhoff blackston oakley syndrome
ferrocalcinosis cerebro vascular
fetal acitretin syndrome
fetal akinesia sequence
fetal akinesia syndrome x linked
fetal alcohol syndrome
fetal aminopterin syndrome
fetal and neonatal alloimmune thrombocytopenia
fetal antihypertensive drugs syndrome
fetal brain disruption sequence
fetal cocaine syndrome
fetal cytomegalovirus syndrome
fetal diethylstilbestrol syndrome
fetal edema
fetal enterovirus syndrome
fetal face syndrome
fetal hydantoin syndrome
fetal indomethacin syndrome
fetal iodine syndrome
fetal left ventricular aneurysm
fetal methimazole syndrome
fetal methyl mercury syndrome
fetal minoxidil syndrome
fetal parainfluenza virus type 3 syndrome
fetal parvovirus syndrome
fetal phenothiazine syndrome
fetal prostaglandin syndrome
fetal rubella syndrome
fetal thalidomide syndrome
fetal trimethadione syndrome
fetal valproic syndrome
fetal varicella syndrome
fetal warfarin syndrome
fg syndrome
fibrinogen deficiency
fibrochondrogenesis
fibrodysplasia ossificans progressiva
fibrofolliculomas with trichodiscomas and acrochordons
fibromatosis gingival hepatosplenomegaly other anomalies

fibromatosis gingival hypertrichosis
fibromatosis gingival progressive deafness
fibromatosis juvenile hyaline
fibromatosis multiple non ossifying
fibromuscular dysplasia of arteries
fibrosarcoma
fibrous dysplasia of bone
fibula aplasia complex brachydactyly
fibula ulna duplication tibia radius absence
fibular aplasia ectrodactyly
fibular hypoplasia femoral bowing oligodactyly
fibular hypoplasia scapulo pelvic dysplasia absent 5th fingers
fibulo ulnar hypoplasia renal anomalies
fiessinger-leroy-reiter syndrome
filariasis
filippi syndrome
fine lubinsky syndrome
fingerprints absence syndactyly milia
fingers absence
finnish congenital nephrosis
finnish type amyloidosis
finucane kurtz scott syndrome
fish-eye disease
fistulous vegetative verrucous hydradenoma
fitzsimmons guilbert syndrome
fitzsimmons mclachlan gilbert syndrome
fitzsimmons walson mellor syndrome
flat face microstomia ear anomaly
floating harbor syndrome
flotch syndrome
flynn aird syndrome
focal alopecia congenital megalencephaly
focal dermal hypoplasia
focal dystonia
foix chavany marie syndrome
follicular atrophoderma basal cell carcinoma
follicular hamartoma alopecia cystic fibrosis
follicular ichtyosis
fontaine farriaux blanckaert syndrome
fop
forney robinson pascoe syndrome
fountain syndrome
foveal hypoplasia presenile cataract
fowler christmas chapele syndrome
fra x syndrome
fragile x syndrome
fragile x syndrome type 1
fragile x syndrome type 2
fragile x syndrome type 3
fragoso cid garcia hernandez syndrome
franceschetti klein syndrome
francheschini vardeu guala syndrome
francois dyscephalic syndrome
franek bocker kahlen syndrome
fraser jequier chen syndrome
fraser like syndrome
fraser syndrome
frasier syndrome
fraxa syndrome
fraxe syndrome
fraxf syndrome
free sialic acid storage disease
freeman sheldon syndrome
freiberg's disease
freire maia odontotrichomelic syndrome
freire maia pinheiro opitz syndrome
frenkel russe syndrome
frias syndrome
fried goldberg mundel syndrome

friedel heid grosshans syndrome
friedman goodman syndrome
friedreich ataxia
friedreich ataxia congenital glaucoma
fronto nasal malformation cloacal exstrophy
frontofacionasal dysostosis
frontofacionasal dysplasia type algazali
frontometaphyseal dysplasia
frontonasal dysplasia
frontonasal dysplasia acromelic
frontonasal dysplasia klippel feil syndrome
frontonasal dysplasia phocomelic upper limbs
frontotemporal lobe dementia
froster huch syndrome
froster iskenius waterson syndrome
fructose intolerance
fructose-1,6-biphosphatase deficiency
fructose-1-phosphate aldolase hereditary deficiency
fructosemia hereditary
fructosuria
frydman cohen ashenazi syndrome
frydman cohen karmon syndrome
fryer syndrome
fryns dereymacker haegeman syndrome
fryns fabry remans syndrome
fryns hofkens fabry syndrome
fryns smeets thiry syndrome
fryns syndrome
fucosidosis
fuhrmann rieger de sousa syndrome
fukuda miyanomae nakata syndrome
fumarase deficiency
fumaric aciduria
fumarylacetoacetase deficiency
functioning pancreatic endocrine tumor
fuqua berkovitz syndrome
furlong kurczynski hennessy syndrome
furukawa takagi nakao syndrome
Furunculous myiasis
fused mandibular incisives
g syndrome
g6pd deficiency
gaba transaminase deficiency
galactocerebrosidase deficiency
galactokinase deficiency
galactosamine 6 sulfatase deficiency
galactose-1-phosphate uridyltransferase deficiency
galactosemia
galactosialidosis
galloway syndrome
gamborg nielsen syndrome
game friedman paradice syndrome
gamma aminobutyric acid transaminase deficiency
gamma-cystathionase deficiency
Gamma-sarcoglycanopathy
gamstorp episodic aynamy
gangliosidosis gm1
gangliosidosis gm1 type 1
gangliosidosis gm1 type 2
gangliosidosis gm1 type 3
gapo syndrome
garcia torres guarner syndrome
gardner morrisson abbot syndrome
gardner silengo wachtel syndrome
gardner syndrome
garret tripp syndrome
gastrinoma
gastritis, familial giant hypertrophic
gastro-enteropancreatic neuroendocrine tumor

gastrocutaneous syndrome
gastroschisis
gaucher disease
gaucher disease type 1
gaucher disease type 2
gaucher disease type 3
gaucher ichthyosis restrictive dermopathy
gaucher like disease
gay feinmesser cohen syndrome
geen sandford davison syndrome
geleophysic dwarfism
gelineau disease
gemignani syndrome de
gemss syndrome
generalized resistance to thyroid hormone
genes syndrome
genetic reflex epilepsy
genital anomaly cardiomyopathy
genitopalatocardiac syndrome
gerhardt syndrome
german syndrome
geroderma osteodysplastica
gershinibaruch leibo syndrome
gerstmann-straussler-scheinker syndrome
ghosal syndrome
ghose sachdev kumar syndrome
giant cell arteritis
giant pigmented hairy nevus
giant platelet syndrome
gigantism advanced bone age hoarse cry
gigantism partial nevi hemihypertrophy macrocephaly
gilbert syndrome
gilles de la tourette disease
gillespie syndrome
gingival fibromatosis dominant
gingival fibromatosis facial dysmorphism
gingival hypertrophy corneal dystrophy
girate atrophy of choroid and retina
gitelman syndrome
glanzmann thrombasthenia
glass chapman hockley syndrome de
glastre cochat bouvier syndrome
glaucoma (type 1c)
glaucoma congenital
glaucoma ecopia microspherophakia stiff joints short stature
glaucoma hereditary
glaucoma hereditary adult (type 1a)
glaucoma hereditary juvenile (type 1b)
glaucoma iridogoniodysgenesia
glaucoma primary infantile (type 3a)
glaucoma primary infantile (type 3b)
glaucoma sleep apnea
glioblastoma
glomerulonephritis sparse hair telangiectases
gloomy face syndrome
glossopalatine ankylosis cataracts digital anomalies
glossopalatine ankylosis micrognathia ear anomalies
glucagonoma
glucocerebrosidase deficiency
glucocerebrosidase deficiency type 1
glucocerebrosidase deficiency type 2
glucocerebrosidase deficiency type 3
glucocorticoid resistance
glucocorticoid sensitive hypertension
glucose-6-phosphatase deficiency
glucose-6-phosphate translocase deficiency
glucose-6-phosphate-dehydrogenase deficiency
glucosephosphate isomerase deficiency
glucosidase acid 1,4 alpha deficiency

glut2 deficiency
glutamate decarboxylase deficiency
glutamate-aspartate transport defect
glutaricacidemia
glutaricacidemia type 1
glutaricacidemia type 2a
glutaricacidemia type 2b
glutaricacidemia type 2c
glutaricaciduria
glutaricaciduria type 1
glutaricaciduria type 2a
glutaricaciduria type 2b
glutaricaciduria type 2c
glutaryl coa dehydrogenase deficiency
glutathione synthetase deficiency
gluten intolerance
glyceraldehyde-3-phosphate dehydrogenase deficiency
glycerol kinase deficiency
glycine synthase deficiency
glycinemia ketotic
glycinemia ketotic type 1
glycinemia ketotic type 2
glycogen storage disease type 1
glycogen storage disease type 1a
glycogen storage disease type 1b
glycogen storage disease type 1c
glycogen storage disease type 1d
glycogen storage disease type 2
glycogen storage disease type 3
glycogen storage disease type 4
glycogen storage disease type 5
glycogen storage disease type 6 due to phosphorylase deficiency
glycogen storage disease type 6 due to phosphorylase kinase deficiency
glycogen storage disease type 7
glycogen storage disease type 9
glycogenose type 0
gm2 gangliosidosis (0 variant)
gm2 gangliosidosis (b, b1,ab variant)
gms syndrome
goiter deafness syndrome
golabi rosen syndrome
goldberg bull syndrome
goldberg syndrome
goldblatt behari syndrome
goldblatt carman sprague syndrome
goldblatt viljoen syndrome
goldblatt wallis syndrome
goldblatt wallis zieff syndrome
goldenhar syndrome
goldskag cooks hertz syndrome
goldstein hutt syndrome
gollop coates syndrome
gollop syndrome
gollop wolfgang complex
goltz syndrome
gombo syndrome
gonadal dysgenesis mixed
gonadal dysgenesis xx type
gonadal dysgenesis xy female type
gonadal dysgenesis xy type associated anomalies
gonadotropin deficiency familial
gonadotropin independant familial sexual precocity
goniodysgenesis mental retardation short stature
gonococcal conjonctivitis
gonodal dysgenesis xx type deafness
gonzales del angel syndrome
goodman camptodactyly
goodman syndrome de
goodpasture pneumorenal syndrome

goodpasture syndrome
gordon hyperkaliemia-hypertension syndrome
gordon syndrome
Gorham-Stout disease
gorlin bushkell jensen syndrome
gorlin chaudry moss syndrome de
gorlin goltz syndrome
gorlin syndrome
gorlin syndrome
gougerot sjogren syndrome
graham boyle troxell syndrome
grand kaine fulling syndrome
grant syndrome
granulomatous allergic angiitis
granulomatous disease chronic
gray platelet syndrome
great vessels transposition
grebe chondrodysplasia
greig syndrome
griscelli disease
grix blankenship peterson syndrome
groll hirschowitz syndrome
grosse syndrome
growth deficiency brachydactyly unusual facies
growth mental deficiency syndrome of myhre
growth retardation alopecia pseudoanodontia optic atrophy
growth retardation hydrocephaly lung hypoplasia
growth retardation mental retardation phalangeal hypoplasia
grubben de cock borghgraef syndrome
gtp cyclohydrolase deficiency
guanidinoacetate methyltransferase deficiency
guibaud vainsel syndrome
guillain barre syndrome
guizar vasquez luengas syndrome
guizar vasquez sanchez manzano syndrome
gunal seber basaran syndrome
gupta patton syndrome
gurrieri sammito bellussi syndrome
gusher syndrome
haas chir robinson syndrome
haemorragic proctocolitis
hagemann factor deficiency
hagemoser weinstein bresnick syndrome
hair defect photosensitivity mental retardation
hairy throat syndrome
hajdu cheney syndrome
hal berg rudolph syndrome
halal setton wang syndrome
halal syndrome
hall riggs mental retardation syndrome
hallermam streiff like syndrome
hallermann streiff francois syndrome
hallervorden spatz disease
hallux varus and preaxial polysyndactyly
hamanishi ueba tsuji
hamann zanki schimrigk syndrome
hamano tsukamoto syndrome
hamartoma sebaceus of jadassohn
hand and foot deformity flat facies
hand foot uterus syndrome
hanot syndrome
hans shuller christian disease
hantavirosis
hantavirus fever
hapnes boman skeie syndrome
hard skin syndrome parana type
HARD syndrome
harding ataxia
harrod doman keele syndrome

harrod syndrome
hartnup syndrome
hartsfield bixler demyer syndrome
hashimoto pritzker syndrome
hashimoto struma
haspeslagh fryns muelenaere syndrome
hawkinsinuria
hay wells syndrome dominant form
hay wells syndrome recessive type
hearing loss insensitivity to aldosterone
heart block progressive familial
heart defect round face congenital retarded development
heart defect tongue hamartoma polysyndactyly
heart defects limb shortening
heart hand syndrome spanish type
heart hand syndrome type 2
heart hypertrophy hereditary
heart situs anomaly
heart tumor of the adult
heart tumor of the child
hec syndrome
hecht beals syndrome
hecht scott syndrome
heckenlively syndrome
heide syndrome
helmerhorst heaton crossen syndrome
hemangioma-thrombocytopenia syndrome
hemangiomas cavernous of face supraumbilical midline raphe
hemangiopericytoma
hemeralopia congenital essential
hemeralopia familial
hemi 3 syndrome
hemifacial atrophy agenesis of the caudate nucleus
hemifacial atrophy progressive
hemifacial hyperplasia strabismus
hemifacial microsomia
hemifacial microsomia macrodactyly
hemihypertrophy
hemihypertrophy intestinal web corneal opacity
hemimegalencephaly
hemiplegic migraine familial
hemochromatosis familial
hemoglobin c disease
hemoglobin e disease
hemolytic anemia lethal genital anomalies
hemolytic uremic syndrome
hemophilia
hemophilia a
hemophilia b
hemorragic fever with renal syndrome
hemorrhagiparous thrombocytic dystrophy
hennekam beemer syndrome
hennekam koss de geest syndrome
hennekam syndrome
hennekam van der horst syndrome
henoch-schoenlein purpura
heparane sulfamidase deficiency
heparin-induced thrombopenia
hepatic cystic hamartoma
hepatic fibrosis renal cysts mental retardation
hepatic venoocclusive disease
hepatoblastoma
hepatolenticular degeneration
hepatorenal tyrosinemia
hereditary coproporphyria
hereditary methemoglobinemia recessive
Hereditary myopathy with intranuclear filamentous inclusions
hereditary nodular heterotopia
hereditary primary fanconi disease

hereditary resistance to anti vitamin K
hereditary sensory and autonomic neuropathy 3
hereditary xerocytosis
heredopathia atactica polyneuritiformis
herlitz pearson epidermolysis bullosa junctional
hernandez aguire negrete syndrome
hernandez fragoso syndrome
herpes virus antenatal infection
herpetic embryopathy
herpetic encephalopathy
herrmann opitz arthrogryposis syndrome
herrmann opitz craniosynostosis
hersh podruch weisskopk syndrome
Heterotaxia
heterotaxia autosomal dominant type
heterotaxy visceral x linked
heterotaxy with polysplenia or asplenia
hexosaminidase a deficiency
hexosaminidases a and b deficiency
hhh syndrome
hidradenitis suppurativa familial
hidrotic ectodermal dysplasia halal type
hidrotic ectodermal dysplasia type christianson fourie
high -molecular-weight kininogen deficiency
high scapula
hillig syndrome
hing torack dowston syndrome
hinson-pepys disease
hip dysplasia beukes type
hipo syndrome
hirschsprung disease
hirschsprung disease deafness polydactyly
hirschsprung disease ganglioneuroblastoma
hirschsprung disease polydactyly heart disease
hirschsprung disease type 1
hirschsprung disease type 2
hirschsprung disease type 3
hirschsprung disease type d brachydactyly
hirschsprung disease with pigmentary anomaly
hirschsprung microcephaly cleft palate
hirschsprung nail hypoplasia dysmorphism
hirsutism congenital gingival hyperplasia
hirsutism skeletal dysplasia mental retardation
his bundle tachycardia
histidase deficiency
histidinemia
histidinuria renal tubular defect
histiocytosis x
Histoplasmosis
hittner hirsch kreh syndrome
hm syndrome
hmc syndrome
hmg coa synthetase deficiency
hnpcc
ho kaufman podos syndrome
hodgkin disease
hodgkin lymphoma
hoepffner dreyer reimers syndrome
holmes benacerraf syndrome
holmes borden syndrome
holmes collins syndrome
holmes gang syndrome
holoacardius amorphus
holocarboxylase synthetase deficiency
holoprosencephaly
holoprosencephaly caudal dysgenesis
holoprosencephaly craniosynostosis
holoprosencephaly deletion 2p
holoprosencephaly ectrodactyly cleft lip palate

holoprosencephaly postaxial polydactyly
holoprosencephaly radial heart renal anomalies
holt oram syndrome
holzgreve wagner rehder syndrome
homocarnosinase deficiency
homocarnosinosis
homocystinuria due to cystathionine beta synthase deficiency
homocystinuria due to defect in methylation (cbl e)
homocystinuria due to defect in methylation (cbl g)
homocystinuria due to defect in methylation (mthfr deficiency)
hoon hall syndrome
hordnes engebretsen knudtson syndrome
horn kolb syndrome
hornova dlurosova syndrome
horseshoe kidney
horton disease
houlston ironton temple syndrome de
howard young syndrome
Howell-Evans syndrome
hoyeraal hreidarsson syndrome
hoyeraal syndrome
humero spinal dysostosis congenital heart disease
humeroradial synostosis
humeroradioulnar synostosis
humerus trochlea aplasia of
hunter carpenter mc donald syndrome
hunter jurenka thompson syndrome
hunter macpherson syndrome
hunter mcalpine syndrome
hunter mcdonald syndrome
hunter rudd hoffmann syndrome
hunter syndrome
hunter thomson reed syndrome
huntington chorea
huntington disease
Huriez scleroatrophic syndrome
hurler syndrome
hurst hallam hockey syndrome
hutchinson gilford progeria syndrome
hutteroth spranger syndrome
hyalinosis systemic short stature
hydantoin antenatal infection
Hydatidosis
hyde forster mccarthy berry syndrome
hydranencephaly
hydrocephalus autosomal recessive
hydrocephalus blue sclera nephropathy
hydrocephalus cataract microphthalmos
hydrocephalus cleft palate joint contractures
hydrocephalus costovertebral dysplasia sprengel anomaly
hydrocephalus craniosynostosis bifid nose
hydrocephalus endocardial fibroelastosis cataract
hydrocephalus growth retardation skeletal anomalies
hydrocephalus obesity hypogonadism
hydrocephalus skeletal anomalies
hydrocephalus x linked
hydrocephaly corpus callosum agenesis diaphragmatic hernia
hydrocephaly low insertion umbilicus
hydrocephaly tall stature joint laxity
hydrolethalus syndrome
hydronephrosis congenital
hydronephrosis peculiar facial expression
hydrops ectrodactyly syndactyly
hydrops fetalis
hydrops fetalis anemia immune disorder absent thumb
hydrops fetalis idiopathic
hydroxymethylglutaricaciduria
hygroma cervical
Hymenolepiasis

hyper igd syndrome
hyper igm syndrome x linked
hyperaldosteronism familial type 1
hyperaldosteronism familial type 2
hyperargininemia
hyperbilirubinemia rotor type
hyperbilirubinemia transient familial neonatal
hyperbilirubinemia type 1
hyperbilirubinemia type 2
hypercalciuria idiopathic
hypercalciuria macular coloboma
hypercholesterolemia due to arg3500 mutation of apo b100
hypercholesterolemia due to ldl receptor deficiency
hypercholesterolemia familial
hyperchylomicronemia, familial
hyperekplexia
hypereosinophilic syndrome
hyperferritinemia, hereditary, with congenital cataracts
hyperglycerolemia
hyperglycinemia isolated nonketotic
hyperglycinemia isolated nonketotic type 1
hyperglycinemia isolated nonketotic type 2
hypergonadotropic ovarian failure, familial or sporadic
hyperimidodipeptiduria
hyperimmunoglobinemia d with recurrent fever
hyperimmunoglobulin e - reccurrent infection syndrome
hyperimmunoglobulinemia d with periodic fever
hyperimmunoglobulinemie e
hyperinsulinism familial with pancreatic nesidioblastosis
hyperkaliemic periodic paralysis type 2
Hyperkeratosis lenticularis perstans of Flegel
hyperkeratosis palmoplantar localized acanthokeratolytic
hyperkeratosis palmoplantar localized epidermolytic
hyperkeratosis palmoplantar with palmar crease hyperkeratosis
hyperlipoproteinemia type 1 and 5
hyperlipoproteinemia type 3
hyperlipoproteinemia type 4
hyperlysinemia
hyperornithinemia
hyperornithinemia-hyperammonemia-homocitrullinuria
hyperostosid corticalis deformans juvenilis
hyperostosis cortical infantile
hyperostosis corticalis generalisata
hyperostosis corticalis generalisata
hyperostosis generalisata with striations
hyperoxaluria
hyperoxaluria type 1
hyperoxaluria type 2
hyperparathyroidism familial primary
hyperparathyroidism neonatal severe primary
hyperphalangism dysmorphy bronchomalacia
hyperphenylalalinemia due to dihydropteridine reductase deficiency
hyperphenylalaninemia due to 6-pyruvoyltetrahydropterin synthase deficiency
hyperphenylalaninemia due to dehydratase deficiency
hyperphenylalaninemia due to gtp cyclohydrolase deficiency
hyperphenylalaninemic embryopathy
hyperpipecolatemia
hyperprolinemia
hyperprolinemia type 1
hyperprolinemia type 2
hypertelorism and tetralogy of fallot
hypertelorism hypospadias polysyndactyly syndrome
hypertelorism hypospadias syndrome
hypertelorism microtia facial clefting syndrome
hypertension essential
hypertensive hyperkalemia familial
hypertensive hypokalemia familial
hypertensive hypokalemia recessive
hyperthermia induced defects

hyperthermia of anesthesia
hyperthyroidism due to mutations in tsh receptor
hypertrichosis atrophic skin ectropion macrostomia
hypertrichosis brachydactyly obesity and mental retardation
hypertrichosis congenital generalized x linked
hypertrichosis cubiti short stature
hypertrichosis lanuginosa congenita
hypertrichosis retinopathy dysmorphism
hypertrichosis universalis congenita ambras type
hypertrichotic osteochondrodysplasia
hypertrophic hemangiectasia
hypertrophic osteoarthropathy primary or idiopathic
hypertropic neuropathy of dejerine sottas
hypertryptophanemia
hypo-alphalipoproteinemia primary
hypoadrenocorticism hypoparathyroidism moniliasis
hypoaldosteronism
hypobetalipoproteinaemia ataxia hearing loss
hypobetalipoproteinemia familial
hypocalcemia autosomal dominant
hypocalciuric hypercalcemia familial
hypocalciuric hypercalcemia familial type 1
hypocalciuric hypercalcemia familial type 2
hypocalciuric hypercalcemia familial type 3
hypochondrogenesis
hypochondroplasia
Hypodermyasis
hypodonadotropic hypogonadism alopecia
hypodontia dysplasia of nails
hypodontia of incisors and premolars
hypofibrinogenemia, familial
hypoglycemia with deficiency of glycogen synthetase in the liver
hypogonadism cardiomyopathy
hypogonadism cataract syndrome
hypogonadism hypogonadotropic due to mutations in the gonadotropin-releasing hormone receptor
hypogonadism male mental retardation skeletal anomalies
hypogonadism mitral valve prolapse mental retardation
hypogonadism primary partial alopecia
hypogonadism retinitis pigmentosa
hypogonadism, isolated, hypogonadotropic
hypogonadotropic hypogonadism anosmia
hypogonadotropic hypogonadism anosmia x linked
hypogonadotropic hypogonadism syndactyly
hypogonadotropic hypogonadism without anosmia x linked
hypokalemic alkalosis with hypercalciuria
hypokaliemic periodic paralysis type 1
hypokinetic cardiomyopathy familial dilated
hypomagnesemia primary
hypomandibular faciocranial dysostosis
hypomelanosis of ito
hypomelia mullerian duct anomalies
hypomyelination neuropathy
hypoparathyroidism familial isolated
hypoparathyroidism nerve deafness nephrosis
hypoparathyroidism short stature
hypoparathyroidism short stature mental retardation seizures
hypoparathyroidism x linked
hypophosphatasia infantile
hypophosphatemia x linked
hypopigmentation oculocerebral syndrome cross type
hypopituitarism
hypopituitarism dwarfism skeletal anomalies
hypopituitarism micropenis cleft lip palate
hypopituitarism microphthalmia
hypopituitarism postaxial polydactyly
hypoplastic left heart syndrome
hypoplastic right heart microcephaly
hypoplastic thumb mullerian aplasia

hypoplastic thumbs hydranencephaly
hypoplastic tibiae post axial polydactyly
hypoproconvertinemia
hypoprothrombinemia
hyposmia nasal hypoplasia hypogonadism
hypospadias dysphagia syndrome
hypospadias familial
hypospadias mental retardation goldblatt type
hypotelorism cleft palate hypospadias
hypothalamic dysfunction
hypothalamic hamartoblastoma syndrome
hypothalamic hamartomas
hypothyroidism cleft palate
hypothyroidism dermoid cyst cleft palate
Hypothyroidism due to iodide transport defect
hypothyroidism postaxial polydactyly mental retardation
hypotonic sclerotic muscular dystrophy
hypotrichosis
hypotrichosis mental retardation lopes type
hypoxanthine guanine phosphoribosyltransferase deficiency
i cell disease
ibids syndrome
icf syndrome
ichthyosiform erythroderma corneal involvement deafness
ichthyosis alopecia eclabion ectropion mental retardation
ichthyosis cheek eyebrow syndrome
ichthyosis congenita biliary atresia
ichthyosis congenita collodion fetus type
ichthyosis deafness mental retardation skeletal anomalies
ichthyosis exfoliativa
ichthyosis follicularis atrichia photophobia syndrome
ichthyosis harlequin type
ichthyosis hepatosplenomegaly cerebellar degeneration
ichthyosis male hypogonadism
ichthyosis mental retardation asymptomatic spasticity
ichthyosis mental retardation devriendt type
ichthyosis mental retardation dwarfism renal impairment
ichthyosis microphthalmos
ichthyosis tapered fingers midline groove up
ichthyosis x linked
ichtyosis and male hypogonadism
ichtyosis bullosa of siemens
ichtyosis linearis circumflexa
idaho syndrome
idiopathic congenital nystagmus dominant x linked
idiopathic diffuse interstitial fibrosis
idiopathic facial palsy
idiopathic nephrotic syndrome steroid resistant
idiopathic optic atrophy autosomal recessive
idiopathic orthostatic hypotension
idiopathic thrombocytopenic purpura
idiopathic torsion dystonia
idiopathic ventricular fibrillation
iduronate 2 sulfatase deficiency
ieshima koeda inagaki syndrome
ifap syndrome
iga selective deficiency of
igda syndrome
iida kannari syndrome
illum syndrome
ilyina amoashy grygory syndrome
imaizumi kuroki syndrome
immotile cilia syndrome due to defective radial spokes
immotile cilia syndrome due to excessively long cilia
immotile cilia syndrome kartagener type
immuno osseous dysplasia schimke type
immunodeficiency with short limb dwarfism
imperforate anus
imperforate oropharynx costo vetebral anomalies

impossible syndrome
impuberism and anovulation due to resistance to LH
incisors fused
inclusion body myopathy
inclusion body myositis ibm
incontinentia pigmenti
incontinentia pigmenti type 1
incontinentia pigmenti type 2
indomethacin antenatal infection
infant epilepsy with migrant focal crisis
infantile axonal neuropathy
infantile multisystem inflammatory disease
infantile myofibromatosis
infantile onset spinocerebellar ataxia
infantile recurrent chronic multifocal osteomyolitis
infantile spasms
infantile spasms broad thumbs
infantile spasms x linked
infantile striato thalamic degeneration
infundibulopelvic stenosis multicystic kidney
insentivity to pain with anhidrosis
insomnia familial fatal
instability mitotic non disjunction
insulin resistant acanthosis nigricans type a
insulin-resistance type b
insulin-resistant acanthosis nigricans, type a
insulinoma
interferon gamma receptor 1 deficiency
internal carotid agenesis
intestinal atresia multiple
intestinal lipodystrophy
intestinal malrotation facial anomalies familial type
intestinal pseudoobstruction chronic idiopathic
intracranial aneurysms multiple congenital anomalies
intracranial arterioveinous malformation
intracranial teratoma
intrathoracic kidney vertebral fusion
intrauterine growth retardation mandibular malar hypoplasia
ioan popa fryns syndrome
iodine antenatal infection
iridogoniodysgenesis dominant
iris colobomata cataract cardiopathy
iris dysplasia hypertelorism deafness
irons bhan syndrome
isaacs mertens syndrome
ischiadic hypoplasia renal dysfunction immunodeficiency
ischiopatellar dysplasia
isochromosome 12p syndrome
isochromosome 18p
isolated lissencephaly
Isosporosiasis
isotretinoin embryopathy
isovaleric acid coa dehydrogenase deficiency
isovaleric acidemia
isthmian coarctation
ito hypomelanosis
ivemark syndrome
ivic syndrome
jabs houk bias syndrome
jackson barr syndrome
jackson weiss syndrome
jacobs syndrome
jacobsen syndrome
jadassohn lewandowsky syndrome
jaffe campanacci syndrome
jaffer beighton syndrome
jagell holmgren hofer syndrome
jalili syndrome
jancar syndrome

jankovic rivera syndrome
jansky bielschowsky disease
jarcho lewin syndrome
jejunal atresia
jensen syndrome
jequier kozlowski skeletal dysplasia
jervell lange nielsen syndrome
jeune syndrome
jeune syndrome situs inversus
job syndrome
johanson blizzard syndrome
johnson hall krous syndrome
johnson munson syndrome
johnson neuroectodermal syndrome
johnston aarons schelley syndrome
joint instability syndrome
jones hersh yusk syndrome
jones syndrome
jorgenson lenz syndrome
joubert boltshauser syndrome
joubert syndrome bilateral chorioretinal coloboma
juberg hayward syndrome
juberg marsidi syndrome
judge misch wright syndrome
jung wolff back stahl syndrome
juvenile ankylosing spondylarthritis
juvenile cataract cerebellar atrophy myopathy mental retardation
juvenile chronic arthritis
juvenile gastrointestinal polyposis
juvenile macular degeneration hypotrichosis
kabuti make up syndrome
kalam hafeez syndrome
kaler garrity stern syndrome
kallin syndrome
kallmann syndrome
kallmann syndrome heart disease
kallmann syndrome type 1, X linked
kallmann syndrome type 2 dominant
kallmann syndrome type 3 recessive
kalyanraman syndrome
kantaputra gorlin syndrome
kaplan plauchu fitch syndrome
kaplowitz bodurtha syndrome
kaposiform hemangio-endothelioma
kapur toriello syndrome
karandikar maria kamble syndrome
karsch neugebauer syndrome
kartagener syndrome
kasabach-merritt syndrome
kashani strom utley syndrome
kasznica carlson coppedge syndrome
katsantoni papadakou lagoyanni syndrome
katz syndrome
kaufman mckusick syndrome
kaufman oculocerebrofacial syndrome
kawasaki disease
kawashima tsuji syndrome
kbg syndrome
kearns sayre syndrome
kennedy disease
kennerknecht sorgo oberhoffer syndrome
kennerknecht vogel syndrome
kenny caffey syndrome
kenny syndrome
keratitis ichthyosis deafness
keratitis, hereditary
keratoacanthoma familial
keratoconus posticus circumscriptus
keratoderma ainhumoid and mutilans

keratoderma hypotrichosis leukonychia
keratoderma palmoplantar deafness
keratoderma palmoplantar spastic paralysis
keratoderma palmoplantaris transgrediens
keratodermia palmoplantar periorificial
keratosis focal palmoplantar gingival
keratosis follicularis
keratosis follicularis dwarfism cerebral atrophy
keratosis follicularis spinula decalvans
keratosis palmoplantar periodontopathy
keratosis palmoplantaris adenocarcinoma of the colon
keratosis palmoplantaris oesophageal colon cancer
keratosis palmoplantaris papulosa
keratosis palmoplantaris periodontopathia
keratosis pilaris
keratosis pilaris atrophicans
kerion celsi
kersey syndrome
ketoaciduria mental deficiency ataxia deafness
khalifa graham syndrome
ki 1 cell lymphoma
KID syndrome
Kimura disease
king syndrome
kinsbourne syndrome
kleeblattschaedel syndrome
klein waardenburg syndrome
kleiner holmes syndrome
Klinefelter syndrome
klippel feil deformity conductive deafness absent vagina
klippel feil sequence
klippel feil syndrome
klippel feil syndrome dominant type
klippel feil syndrome recessive type
klippel trenaunay weber syndrome
klumpke paralysis
kniest dysplasia
kniest like dysplasia lethal
knobloch layer syndrome
knuckle pods leuconychia sensorineural deafness
kobberling dunnigan syndrome
kocher debre semelaigne syndrome
kohler's disease
kohlschutter tonz syndrome
kohn elrayyes makadmah syndrome
kok disease
konigsmark knox hussels syndrome
koone rizzo elias syndrome
kopysc barczyk krol syndrome
korula wilson salomon syndrome
kostmann syndrome
kosztolanyi syndrome
kotzot richter syndrome
koussef nichols syndrome
kousseff syndrome
kowarski syndrome
kozlowski brown hardwick syndrome
kozlowski celermajer syndrome
kozlowski massen syndrome
kozlowski ouvrier syndrome
kozlowski rafinski klicharska syndrome
kozlowski tsuruta syndrome
kozlowski tsuruta taki syndrome
kozlowski warren fisher syndrome
kozlowski-krajewska syndrome
krabbe disease
krasnow qazi syndrome
krauss herman holmes syndrome
krieble bixler syndrome

kudo tamura fuse syndrome
kufs disease
kugelberg welander syndrome
kumar levick syndrome
kunze riehm syndrome
kurczynski casperson syndrome
kuskokwim disease
kuster majewski hammerstein syndrome
kuster syndrome
kuzniecky syndrome
kyphosis brachyphalangy optic atrophy
l 2-hydroxyglutaricaciduria
laband syndrome
lachiewicz sibley syndrome
lack of subcutaneous tissue arthritis skeletal dysplasia
lacrimoauriculodentodigital syndrome
lactate dehydrogenase deficiency
lactate dehydrogenase type a deficiency
lactate dehydrogenase type b deficiency
lactate dehydrogenase type c deficiency
lactic acidosis congenital infantile
ladd syndrome
ladda zonana ramer syndrome
lafora disease
lagophthalmia cleft lip palate
LAL
lambdoid synostosis familial
lambert syndrome
lamellar ichtyosis
landau kleffner syndrome
Landau-Kleffner syndrome
landing disease
landouzy dejerine myopathy
landy donnai syndrome
langer giedion syndrome
langer nishino yamaguchi syndrome
langer sadino type achondrogenesis
langerhans cell granulomatosis
langerhans cell histiocytosis
laplane fontaine lagardere syndrome
laron syndrome
larsen like osseous dysplasia dwarfism
larsen like osseous dysplasia dwarfism
larsen like syndrome lethal type
larsen syndrome
larsen syndrome craniosynostosis
larsen syndrome dominant type
larsen syndrome recessive type
laryngeal abductor paralysis
laryngeal abductor paralysis mental retardation
laryngeal and ocular granulation in indian children
laryngeal cleft
laryngeal web congenital heart disease short stature
laryngo onycho cutaneous syndrome
laryngocele
laryngomalacia dominant congenital
laryngotracheoesophageal cleft pulmonary hypoplasia
larynx atresia
Lassueur-Graham-Little syndrome
late onset dominant cone dystrophy
lateral body wall defect
laterality defect
laterality defects dominant
lattice corneal dystrophy type 2
Launois-Bensaude adenolipomatosis
laurence moon syndrome
laurence prosser rocker syndrome
laurin sandrow syndrome
laxova brown hogan syndrome

lbwd syndrome
lcat deficiency
lchad deficiency
le marec bracq picaud syndrome
le merrer syndrome
leao ribeiro da silva syndrome
learman syndrome
leber amaurosis congenita
leber amaurosis congenita type 1
leber amaurosis congenita type 2
leber hereditary optic neuropathy
leber miliary aneurysm
lecithine-cholesterol acyltransferase deficiency (lcat)
lee root fenske syndrome
left ventricule-aorta tunnel
leg absence deformity cataract
legg calve perthes disease
legionellosis
lehman syndrome
leichtman wood rohn syndrome
leifer lai buyse syndrome
leigh disease
leiomyomatose diffuse with alport syndrome
leiomyomatosis familial
leiomyomatosis of oesophagus cataract hematuria
leipala kaitila syndrome
leishmaniasis
leisti hollister rimoin syndrome
lennox gastaut syndrome
lenz majewski hyperostotic dwarfism
lenz microphtalmia
leonard hughes syndrome
leopard syndrome
leprechaunism
leprosy
leptospirosis
leri pleonosteosis
leri weil syndrome
lesch nyhan syndrome
lethal chondrodysplasia moerman type
lethal chondrodysplasia seller type
lethal congenital contracture syndrome
lethal osteosclerotic bone dysplasia
letterer siwe disease
leucinosis
leukocyte adhesion deficiency
leukocyte adhesion deficiency type 1
leukocyte adhesion deficiency type 2
leukodystrophy reunion type
leukoencephalopathy palmoplantar keratoderma
leukoencephalopathy with vanishing white matter
leukomelanoderma mental redardation hypotrichosis
leukonychia totalis multiple sebaceous cysts renal calculi
levic stefanovic nikolic syndrome
levin syndrome
levine crichley syndrome
levy hollister syndrome
lewandowski kikolich syndrome
lewis ocular albinism (type 3)
lewis pashayan syndrome
lewy body dementia
leydig cells hypoplasia
Leydigoma
li fraumeni syndrome
lichen planus follicularis
lichstenstein syndrome
liddle syndrome
limb deficiencies distal micrognathia
limb dystonia

limb reduction defect
limb scalp and skull defects
limb transversal defect cardiac anomaly
lindsay burn syndrome
lindstrom syndrome
linear hamartoma syndrome
linear inflammatory verrucous epidermal nevus
linear nevus syndrome
lip lit syndrome
lipid storage myopathy
lipidosis with triglycerid storage disease
lipoamide dehydrogenase deficiency
lipoatrophic diabetes
lipodystrophy berardinelli type
lipodystrophy familial partial
lipodystrophy rieger anomaly diabetes
lipoid congenital adrenal hyperplasia
lipoid proteinosis of Urbach and Wiethe
lipomatosis central non-encapsulated
lipomatosis familial benign cervical
lipomatosis familial benign cervical
lipomatosis of pancreas congenital
lipoproteine lipase deficiency
lisker garcia ramos syndrome
lison kornbrut feinstein syndrome
lissencephaly immunodeficiency
lissencephaly syndrome miller dieker type
lissencephaly type 1
lissencephaly type 2
lissencephaly x linked
listeriosis
lobar atrophy of brain
lobstein disease
localized epiphyseal dysplasia
locked-in syndrome
lockwood feingold syndrome
loffredo cennamo cecio syndrome
logic syndrome
loiasis
long qt interval-deafness
long qt syndrome familial
long qt syndrome type 1
long qt syndrome type 2
long qt syndrome type 3
loose anagen hair syndrome
loose anagene syndrome
lopes gorlin syndrome
lopes marques de faria syndrome
lopez hernandez syndrome
Lou-Gehrig disease
low birth weight dwarfism dysgammaglobulinemia
lowe kohn cohen syndrome
lowe oculocerebrorenal syndrome
lowe syndrome
lower limb anomaly ureteral obstruction
lower limb deficiency hypospadias
lower limb partial duplication renal agenesis
lower mesodermal defects
lowry maclean syndrome
lowry syndrome
lowry wood syndrome
lowry yong syndrome
lubani al saleh teebi syndrome
lubinsky syndrome
lucey driscoll syndrome
lucky gelehrter syndrome
lujan fryns syndrome
lumbar malsegmentation short stature
lundberg syndrome

lung agenesis heart defect thumb anomalies
lung herniation congenital defect of sternem
lupus anticoagulant familial
lupus erythematosus systemic
lurie kletsky syndrome
luteinizing hormone releasing hormone deficiency of with ataxi
lutz richner landolt syndrome
Lutz-Lewandowsky epidermodysplasia verruciformis
lyell syndrome
lyme disease
lymphangiectasies lymphoedema type hennekam type
lymphangioleiomyomatosis
lymphangioma
lymphangiomatosis
lymphatic filariasis
lymphedema distichiasis
lymphedema hereditary type 1
lymphedema hereditary type 2
lymphedema hydrocele cardiac defects
lymphedema hypoparathyroidism syndrome
lymphedema ptosis
lymphoblastic lymphoma
lynch lee murday syndrome
Lynch syndrome
lynch-bushby syndrome
lyngstadaas syndrome
lysine alpha-ketoglutarate reductase deficiency
lysinuric protein intolerance
lysosomal acid lipase deficiency
lysosomal alpha d mannosidase deficiency
lysosomal beta mannosidase deficiency
lysosomal glycogen storage disease with normal acid maltase activity
maaswinkel mooij stokvis brantsma syndrome
mac dermot patton williams syndrome
mac dermot winter syndrome
maccario mena syndrome
machado joseph disease
macias flores garcia cruz rivera syndrome
mackay shek carr syndrome
macleod fraser syndrome
macrocephaly dominant type
macrocephaly mental retardation facial dysmorphism
macrocephaly mesodermal hamartoma spectrum
macrocephaly mesomelic arms talipes
macrocephaly pigmentation large hands feet
macrocephaly short limbs deafness
macrocephaly short stature paraplegia
macroepiphyseal dysplasia mcalister coe type
macroglossia dominant
macrogyria pseudobulbar palsy
macrophage myofascitis
macrosomia developmental delay dysmorphism
macrosomia microphthalmia cleft palate
macrosomia obesity macrocephaly ocular abnormalities
macrothrombocytopenia progressive deafness
macular degeneration juvenile
macular degeneration, age-related
macular dystrophy vitelliform
macules hereditary congenital hypopigmented hyperpigmented
madokoro ohdo sonoda syndrome
maffucci syndrome
maghazaji syndrome
magnesium defect in renal tubular transport of
magnesium wasting renal
majewxki ozturk syndrome
mal de meleda
malakoplakia
malakoplasia
malaria

male pseudohermaphroditism due to 17 beta hydroxysteroide dehydrogenase deficiency
male pseudohermaphroditism due to 5 alpha reductase 2 deficiency
male pseudohermaphroditism due to androgen insensitivity
male pseudohermaphroditism due to defective lh molecule
male sterility due to y chromosome deletions
male xx syndrome
malignant fibrohistiocytoma
malignant hyperthermia arthrogryposis torticollis
malignant hyperthermia susceptibility
malignant hyperthermia susceptibility type 1
malignant hyperthermia susceptibility type 2
malignant hyperthermia susceptibility type 3
malignant hyperthermia susceptibility type 4
malignant hyperthermia susceptibility type 5
malignant hyperthermia susceptibility type 6
malignant schwannoma
malonic aciduria
malonyl-coa decarboxylase deficiency
malouf syndrome
mandibuloacral dysplasia
mandibulofacial dysostosis
mandibulofacial dysostosis deafness postaxial polydactyly
manic depressive psychosis
manouvrier syndrome
mansonellosis
maple syrup urine disease
marashi gorlin syndrome
marchiafava micheli disease
marden walker like syndrome
marden walker syndrome
marfan like syndrome type boileau
marfan syndrome
marfanoid build spondylolisthesis constricted pelvis
marfanoid craniosynostosis syndrome
marfanoid mental retardation syndrome autosomal
Marie Unna congenital hypotrichosis
marinesco sjogren like syndrome
marinesco sjogren syndrome
marion mayers syndrome
markel vikkula mulliken syndrome
marles greenberg persaud syndrome
maroteaux cohen solal bonaventure syndrome
maroteaux fonfria syndrome
maroteaux lamy syndrome
maroteaux le merrer bensahel syndrome
maroteaux stanescu cousin syndrome
maroteaux verloes stanescu syndrome
marphanoid syndrome type de silva
marsden nyhan sakati syndrome
marshall smith syndrome
marshall syndrome
martinez monasterio pinheiro syndrome
martsolf reed hunter syndrome
martsolf syndrome
masa syndrome
massa casaer ceulemans syndrome
massive osteolysis
mastocytosis, short stature, hearing loss
mastroiacovo de rosa satta syndrome
mastroiacovo gambi segni syndrome
mat deficiency
maternal hyperphenylalaninemia
maternally inherited diabetes and deafness
maternally inherited leigh syndrome
mathieu de broca bony syndrome
matsoukas liarikos giannika syndrome
matthew wood syndrome
maturity onset diabetes of the young

maumenee syndrome
maxillofacial dysostosis
maxillonasal dysplasia binder type
may hegglin thrombocytopenia
mayer rokitanski kuster syndrome
mcad deficiency
mcalister coe white syndrome
mcalister crane syndrome
mccallum macadam johnston syndrome
mccune albright syndrome
mcdonough syndrome
mcdowall syndrome
mcgillivray syndrome
mckusick kaufman syndrome
mclain debakian syndrome
mcph syndrome
mcpherson clemens syndrome
mcpherson robertson cammarano syndrome
meacham winn culler syndrome de
Meadows syndrome
MEB (Muscle-Eye-Brain) syndrome
meckel like syndrome
meckel syndrome
medeira dennis donnai syndrome
median cleft face syndrome
median cleft lip corpus callosum lipoma skin polyp
median nodule of the upper lip
mediterranean fever familial
medrano roldan syndrome
medullary cystic kidney
medullary sponge kidney
medullary thyroid cancer
medulloblastoma
megacystis microcolon intestinal hypoperilstasis hydronephrosis
megaduodenum and/or megacystis
megaepiphyseal dwarfism
megalencephalic leukodystrophy
megalencephaly cystic leukodystrophy syndrome
megalencephaly familial
megalocornea mental retardation syndrome
mehes syndrome
mehta lewis patton syndrome
meier blumberg imahorn syndrome
meier gorlin syndrome
meier rotschild syndrome
meige syndrome
meigel disease
meinecke pepper syndrome
meinecke syndrome
melanocytosis myelomeningocele
melanoma familial
melanoma type 1
melanoma type 2
melanosis neurocutaneous
melas syndrome
melhem fahl syndrome
melkersson rosenthal syndrome
melnick needles osteodysplasty
melorheostosis
menetrier's disease
mengel konigsmark syndrome de
meningeal angiomatosis cleft hypoplastic left heart
meningioma
meningoencephalocele-arthrogryposis-hypoplastic thumb
menkes kinky hair syndrome
mental deficiency epilepsy endocrine disorders
mental mixed retardation deafnes clubbed digits
mental retardatio-polydactyly-uncombable hair
mental retardation anophthalmia craniosynostosis

mental retardation arachnodactyly hypotonia telangiectasia
mental retardation athetosis microphthalmia
mental retardation balding patella luxation acromicria
mental retardation blepharophimosis obesity web neck
mental retardation buenos aires type
mental retardation cataracts calcified pinnae myopathy
mental retardation coloboma slimness
mental retardation contractural arachnodactyly
mental retardation dysmorphism hypogonadism diabetes mellitus
mental retardation epilepsy
mental retardation epilepsy bulbous nose
mental retardation gynecomastia obesity x linked
mental retardation hip luxation g6pd variant
mental retardation hypocupraemia hypobetalipoproteinaemia
mental retardation hypoplastic corpus callosum preauricular tag
mental retardation hypotonia skin hyperpigmentation
mental retardation macrocephaly coarse facies hypotonia
mental retardation microcephaly phalangeal facial abnormalities
mental retardation microcephaly unusual facies
mental retardation multiple nevi
mental retardation myopathy short stature endocrine defect
mental retardation nasal hypoplasia obesity genital hypoplasia
mental retardation nasal papillomata
mental retardation osteosclerosis
mental retardation progressive spasticity
mental retardation psychosis macroorchidism
mental retardation short broad thumbs
mental retardation short stature absent phalanges
mental retardation short stature bombay phenotype
mental retardation short stature cleft palate unusual facies
mental retardation short stature deafness genital
mental retardation short stature hand contractures genital anomalies
mental retardation short stature heart and skeletal defects
mental retardation short stature hypertelorism
mental retardation short stature microcephaly eye
mental retardation short stature ocular and articular anomalies
mental retardation short stature scoliosis
mental retardation short stature unusual facies
mental retardation short stature wedge shaped epip
mental retardation skeletal dysplasia abducens palsy
mental retardation smith fineman myers type
mental retardation sparse hair brachydactyly
mental retardation spasticity ectrodactyly
mental retardation type mietens weber
mental retardation unusual facies
mental retardation unusual facies ampola type
mental retardation unusual facies davis lafer type
mental retardation unusual facies hypothyroidism
mental retardation unusual facies talipes hand anomalies
mental retardation wolff type
mental retardation x linked borderline maoa metabolism anomaly
mental retardation x linked dysmorphism
mental retardation x linked dystonia dysarthria
mental retardation x linked juberg marsidi type
mental retardation x linked marfanoid habitus
mental retardation x linked nonspecific
mental retardation x linked severe gustavson type
mental retardation x linked short stature obesity hypogonadism
mental retardation x linked tranebjaerg type seizures psoriasis
mental retardation x linked type atkin
mental retardation x linked type brunner
mental retardation-unusual facies-intrauterine growth retard.
meretoja syndrome
merlob grunebaum reisner syndrome
merlob syndrome
merrf syndrome
mesangial sclerosis diffuse
mesodermal defects lower type
mesomelia radial hypoplasia bifid thumb unusual facies

mesomelia synostoses
mesomelic dwarfism cleft palate camptodactyly
mesomelic dwarfism langer type
mesomelic dwarfism nievergelt type
mesomelic dwarfism reinhardt pfeiffer type
mesomelic dysplasia skin dimples
mesomelic dysplasia thai type
metacarpals 4 and 5 fusion
metachondromatosis
metachromatic leukodystrophy
metageria
metaphyseal anadysplasia
metaphyseal chondrodysplasia congenital lethal
metaphyseal chondrodysplasia dominant type
metaphyseal chondrodysplasia kaitila type
metaphyseal chondrodysplasia mckusick type
metaphyseal chondrodysplasia recessive type
metaphyseal chondrodysplasia retinitis pigmentosa
metaphyseal chondrodysplasia schmid type
metaphyseal chondrodysplasia shwachman type
metaphyseal chondrodysplasia spahr type
metaphyseal chondrodystrophy sussman type
metaphyseal dysostosis mental retardation conductive deafness
metaphyseal dysplasia hypertelorism hypospadias
metaphyseal dysplasia maxillary hypoplasia brachydacty
metaphyseal dysplasia pyle type
metatropic dwarfism
metatropic dwarfism type 2
metatropic dysplasia 1
methimazole antenatal infection
methionine synthase deficiency
methyl mercury antenatal infection
methylcobalamin deficiency (cbl e)
methylcobalamin deficiency (cbl g)
methylenetetrahydrofolate reductase deficiency
methylmalonic acidemia with homocystinuria
methylmalonic aciduria microcephaly cataract
methylmalonicacidemia with homocystinuria (cbl c)
methylmalonicacidemia with homocystinuria (cbl d)
methylmalonicaciduria vitamin b12 unresponsive mut 0
methylmalonicaciduria with homocystinuria
methylmalonicaciduria with homocystinuria (cbl f)
methylmalonyl coenzyme a mutase deficiency
mevalonate kinase deficiency
mevalonicaciduria
michelin tire baby syndrome
michels caskey syndrome
michels syndrome
mickleson syndrome
micrencephaly corpus callosum agenesis
micrencephaly olivopontocerebellar hypoplasia
micro syndrome
microbrachycephaly ptosis cleft lip
microcephalic osteodysplastic primordial dwarfism taybi linder type
microcephalic osteodysplastic primordial dwarfism type 1
microcephalic osteodysplastic primordial dwarfism type 2
microcephalic osteodysplastic primordial dwarfism type 3
microcephalic primordial dwarfism
microcephalic primordial dwarfism toriello type
microcephaly
microcephaly albinism digital anomalies syndrome
microcephaly autosomal dominant
microcephaly brachydactyly kyphoscoliosis
microcephaly brain defect spasticity hypernatremia
microcephaly cardiac defect lung malsegmentation
microcephaly cardiomyopathy
microcephaly cervical spine fusion anomalies
microcephaly chorioretinopathy recessive form
microcephaly cleft palate autosomal dominant

microcephaly deafness syndrome
microcephaly developmental delay pancytopenia
microcephaly facial clefting preaxial polydactyly
microcephaly glomerulonephritis marfanoid habitus
microcephaly hiatus hernia nephrotic syndrome
microcephaly hypergonadotropic hypogonadism short stature
microcephaly hypogammaglobulinemia abnormal immunity
microcephaly immunodeficiency lymphoreticuloma
microcephaly intracranial calcification
microcephaly lymphoedema chorioretinal dysplasia
microcephaly lymphoedema syndrome
microcephaly mental retardation retinopathy
microcephaly mental retardation spasticity epilepsy
microcephaly mesobrachyphalangy tracheoesophageal fistula
microcephaly microcornea syndrome seemanova type
microcephaly micropenis convulsions
microcephaly microphtalmos blindness
microcephaly pontocerebellar hypoplasia dyskinesia
microcephaly seizures mental retardation heart disease
microcephaly sparse hair mental retardation seizures
microcephaly syndactyly brachymesophalangy
microcoria autosomal dominant
microcoria congenital
microcornea corectopia macular hypoplasia
microcornea glaucoma absent frontal sinuses
microdeletion 22q11
microdontia hypodontia short stature
microgastria limb reduction defect
microgastria short stature diabetes
micromelic dwarfism fryns type
micromelic dysplasia dislocation of radius
microphtalmos bilateral colobomatous orbital cyst
microphthalmia
microphthalmia camptodactyly mental retardation
microphthalmia cataract
microphthalmia diaphragmatic hernia fallot
microphthalmia lentz type
microphthalmia mental deficiency
microphthalmia microtia fetal akinesia
microscopic polyangiitis
microsomia hemifacial radial defects
microspherophakia metaphyseal dysplasia
microsporidiosis
microtia
microtia meatal atresia conductive deafness
microtia meatal atresia deafness dominant
microvillous inclusion disease
midas syndrome
midline cleft of lower lip
midline defects autosomal type
midline defects recessive type
midline developmental field defects
midline field defects
mietens syndrome
mievis verellen dumoulin syndrome
mikati najjar sahli syndrome
miller dieker syndrome
miller fisher syndrome
miller syndrome
milner khallouf gibson syndrome
mils syndrome
Minkowski-Chauffard disease
minoxidil antenatal infection
miosis congenital
mirror hands feet nasal defects
mirror polydactyly segmentation and limbs defects
mitochondrial acetoacetyl-coa thiolase deficiency
mitochondrial diseases of nuclear origin
mitochondrial encephalomyopathy

mitochondrial encephalomyopathy aminoacidopathy
mitochondrial myopathy
mitochondrial myopathy encephalopathy lactic acidosis
mitochondrial myopathy lactic acidosis
mitochondrial trifunctional protein deficiency
mitral atresia
mitral regurgitation deafness skeletal anomalies
mitral valve prolapse familial
mitral valve prolapse familial autosomal dominant
mitral valve prolapse familial X linked
miura syndrome
mixed connective tissue disease
mixed sclerosing bone dystrophy
mls syndrome
mls syndrome
mmep syndrome
mmt syndrome
mngie syndrome
MODY syndrome
moebius axonal neuropathy hypogonadism
moebius syndrome
moerman vandenberghe fryns syndrome
moeschler clarren syndrome
mohr syndrome
Mohr-Tranebjaerg syndrome
molarization of anterior teeth deafness
mollica pavone antener syndrome
moloney syndrome
molybdenum cofactor deficiency
momo syndrome
Monilethrix
monoamine oxidase a deficiency
monodactyly tetramelic
mononen karnes senac syndrome
monosomy 10p
monosomy 10pter
monosomy 10q
monosomy 11p11 p12
monosomy 11q partial
monosomy 12p12 p11
monosomy 12p13
monosomy 13q
monosomy 13q14
monosomy 13q22
monosomy 13q32
monosomy 14q11
monosomy 14q31
monosomy 14qter
monosomy 15q1
monosomy 15q25
monosomy 17q23 q24
monosomy 18 mosaicism
monosomy 18p
monosomy 18q
monosomy 18q23
monosomy 1p
monosomy 1p22 p13
monosomy 1p31 p22
monosomy 1p32
monosomy 1p34 p32
monosomy 1p36
monosomy 1q21 q25
monosomy 1q25 q32
monosomy 1q32 q42
monosomy 1q4
monosomy 20p
monosomy 21
monosomy 21q22
monosomy 2p22

monosomy 2pter p24
monosomy 2q
monosomy 2q duplication 1p
monosomy 2q24
monosomy 2q37
monosomy 2q37
monosomy 3p
monosomy 3p14 p11
monosomy 3p25
monosomy 3q13
monosomy 3q21 23
monosomy 3q27
monosomy 4p
monosomy 4p14 p16
monosomy 4q
monosomy 4q32
monosomy 5p
monosomy 5q35
monosomy 6p23
monosomy 6q
monosomy 6q1
monosomy 6q13 q15
monosomy 6q16 q21
monosomy 6q2
monosomy 7
monosomy 7q21
monosomy 7q3
monosomy 8p
monosomy 8p23 1
monosomy 8q
monosomy 8q12 21
monosomy 8q21 q22
monosomy 9p
monosomy x
monosomy xp22 pter
monosomy xq28
montefiore syndrome
moore federman syndrome
moore smith weaver syndrome
moore weaver syndrome
moreno zachai kaufman syndrome
morhosseini holmes walton syndrome
morillo cucci passarge syndrome
morquio disease
morquio disease type a
morquio disease type b
morrison young syndrome
morse rawnsley sargent syndrome
mosaic variegated aneuplody microcephaly syndrome
motor neuropathy peripheral dysautonomia
motor sensory neuropathy type 1 aplasia cutis congenita
mounier kuhn syndrome
mount reback syndrome
mousa al din al nassar syndrome
moya moya disease
moynahan syndrome
mpo deficiency
mrx35
msbd syndrome
mthfr deficiency
muckle wells syndrome
mucocutaneous lymph node syndrome
mucoepithelial dysplasia
mucolipidosis type 1
mucolipidosis type 2
mucolipidosis type 3
mucolipidosis type 4
mucopolysaccharidosis type 1
mucopolysaccharidosis type 2

mucopolysaccharidosis type 3
mucopolysaccharidosis type 3a
mucopolysaccharidosis type 3b
mucopolysaccharidosis type 3c
mucopolysaccharidosis type 3d
mucopolysaccharidosis type 4
mucopolysaccharidosis type 4a
mucopolysaccharidosis type 4b
mucopolysaccharidosis type 6
mucopolysaccharidosis type 7
mucosulfatidosis
muir torre syndrome
mulibrey dwarfism
muller barth menger syndrome
mullerian derivatives lymphangiectasia polydactyly
mullerian derivatives persistent
mullerian duct abnormalities galactosemia
mulliez roux loterman syndrome
multicentric osteolysis nephropathy
multifocal motor neuropathy with conduction block
multinodular goiter cystic kidney polydactyly
multiple acyl-coa deficiency
multiple carboxylase deficiency
multiple carboxylase deficiency biotin responsive
multiple carboxylase deficiency late onset
multiple congenital anomalies mental retardation growth failure cleft lip palate
multiple congenital anomalies robinow unger type
multiple congenital anomalies ulerythema ophryogenesis
multiple contracture syndrome finnish type
multiple endocrine neoplasia type 1
multiple endocrine neoplasia type 2
multiple epiphyseal dysplasia eiken petersen type
multiple epiphyseal dysplasia ribbing type
multiple epiphyseal dysplasia type 2
multiple fibrofolliculoma familial
multiple hamartoma syndrome
multiple joint dislocations metaphyseal dysplasia
multiple pterygium syndrome
multiple pterygium syndrome lethal type
multiple sclerosis
multiple sclerosis ichthyosis factor 8 deficiency
multiple sulfatase deficiency
multiple synostosis syndrome
multiple system atrophy
multiple vertebral anomalies unusual facies
mulvihill smith syndrome
murcs association
muscle eye brain disease
muscle phosphofructokinase deficiency
muscular atrophy ataxia retinitis pigmentosa diabetes mellitus
muscular dystrophy congenital infantile cataract hypogonadism
muscular dystrophy congenital merosin negative
muscular dystrophy congenital merosin positive
muscular dystrophy duchenne and becker type
muscular dystrophy duchenne and becker type
muscular dystrophy fukuyama type
muscular dystrophy hutterite type
muscular dystrophy limb girdle
muscular dystrophy limb girdle autosomal dominant type 1A, chromosome 5-linked
muscular dystrophy limb girdle autosomal dominant with cardiac involvement type 1B, chromosome 15-linked
muscular dystrophy limb girdle autosomal dominant with caveolin deficiency
muscular dystrophy limb girdle type 2A, Erb type
muscular dystrophy limb girdle type 2B, Myoshi type, chromosome 2-linked
muscular dystrophy limb girdle with sarcoglycan alpha deficiency
muscular dystrophy limb girdle with sarcoglycan beta deficiency
muscular dystrophy limb girdle with sarcoglycan delta deficiency
muscular dystrophy limb girdle with sarcoglycan gamma deficiency
muscular dystrophy white matter spongiosis

muscular fibrosis multifocal obstructed vessels
muscular phosphorylase kinase deficiency
mutation in the aromatase gene
mutations in oestradiol receptor
myalgia eosinophilia associated with tryptophan
myasthenia familial
myasthenia gravis
myasthenia gravis
mycetoma
mycosis fungoides, familial
myelinosis centralis diffusa
myelocerebellar disorder
myelodysplasia facial dysmorphism
myelomeningocele
myeloperoxidase deficiency
myhre ruvalcaba graham syndrome
myhre ruvalcaba kelley syndrome
myhre school syndrome
myoclonic epilepsy juvenile
myoclonus ataxia
myoclonus cerebellar ataxia deafness
myoclonus epilepsy ragged red fibers
myoclonus hereditary progressive distal muscular atrophy
myoclonus progressive epilepsy of unverricht and lundborg
myoglobinuria
myoglobinuria dominant form
myoglobinuria recurrent
myoneurogastrointestinal encephalopathy syndrome
myopathy and diabetes mellitus
myopathy cataract hypogonadism
myopathy congenital multicore with external ophtalmoplegia
myopathy growth and mental retardation hypospadias
myopathy hutterite type
myopathy Mc Ardle type
myopathy mitochondrial cataract
myopathy moebius robin syndrome
myopathy ophthalmoplegia hypoacousia areflexia
myopathy tubular agregates
myopathy with lactic acidosis and sideroblastic anemia
myopathy with lysis of type 1 myofibrils
myophosphorylase deficiency
myopia , infantile severe
myopia severe
myositis ossificans progressiva
myotonia mental retardation skeletal anomalies
myotonic chondrodystrophy
myotubular myopathy
myxoma spotty pigmentation endocrine overactivity
n acetyl alpha glucosaminidase sulfamidase deficiency
n acetyl glucosamine 1 phosphotransferase deficiency
n acetyl glucosamine 6 sulfate sulfatase deficiency
n acetyltransferase deficiency
n syndrome
n-acetyl-alpha-d-galactosaminidase
n-acetylglutamate synthetase deficiency
n5-methylhomocysteine transferase deficiency
nadh coq reductase deficiency of
nadh cytochrome b5 reductase deficiency of
nadh diaphorase deficiency of
nadh methemoglobin reductase deficiency of
naegelli syndrome
nager acrofacial dysostosis
naguib richieri costa syndrome
naguib syndrome
nail patella like renal disease
nail patella syndrome
nakajo nishimura syndrome
nakajo syndrome
nakamura osame syndrome

NAME syndrome
nance horan syndrome
nance insley syndrome
nanism due to combined pituitary hormone deficiency
nanism due to growth hormone isolated deficiency
nanism due to growth hormone isolated deficiency autisomal dominant type
nanism due to growth hormone isolated deficiency autosomal recessive type
nanism due to growth hormone isolated deficiency with x linked hypogammaglobulinemia
nanism due to growth hormone isolated deficiency x linked recessive type
nanism due to growth hormone qualitative anomaly
nanism due to growth hormone resistance
narcolepsy cataplexy
narp syndrome
narrow oral fissure short stature cone shaped epiphyses
nasodigitoacoustic syndrome
nasopalpebral lipoma coloboma syndrome
nasopharyngeal cancer
nasopharyngeal teratoma dandy walker diaphragmatic hernia
natal teeth intestinal pseudoobstruction patent ductus
nathalie syndrome
necrotizing encephalopathy infantile subacute
negative rheumatoid factor polyarthritis
nemaline myopathy
neonatal death immune deficiency
neonatal diabetes
neonatal hemochromatosis
neonatal osseous dysplasia 1
neonatal ovarian cyst
nephritis iga type
nephroblastoma
nephroblastomatosis,fetal ascites,macrosomia and wilms tumor
nephrocalcinosis
nephrolithiasis type 2
nephronophtisis
nephronophtisis familial adult spastic quadriparesis
nephropathy deafness hyperparathyroidism
nephropathy familial with gout
nephropathy familial with hyperuricemia
nephrosis deafness urinary tract digital malformations
nephrosis neuronal dysmigration syndrome
nephrotic syndrome ocular anomalies
nesidioblastosis of pancreas
netherton disease
nettleship falls ocular albinism (type 1a)
neu laxova syndrome
neuhauser daly magnelli syndrome
neuhauser eichner opitz syndrome
neural crest tumour
neural tube defects x linked
neuraminidase beta galactosidase deficiency
neuraminidase deficiency
neuritis with brachial predilection
neuroaxonal dystrophy late infantile
neuroaxonal dystrophy renal tubular acidosis
neuroblastoma
neurocutaneous syndrome,abdallat type
neuroectodermal endocrine syndrome
neuroectodermal syndrome,zunich type
neuroendocrine tumor
neuroepithelioma
neurofaciodigitorenal syndrome
neurofibromatosis noonan syndrome
neurofibromatosis type 1
neurofibromatosis type 2
neurofibromatosis type 3
neurofibromatosis type 6
neuronal heterotopia
neuronal intestinal pseudoobstruction

neuronal intranuclear hyaline inclusion disease
neuronal intranuclear inclusion disease
neuropathy ataxia and retinis pigmentosa
neuropathy congenital sensory neurotrophic keratitis
neuropathy giant axonal
neuropathy hereditary motor and sensory lom type
neuropathy hereditary with liability to pressure palsies
neuropathy motor sensory type 2 deafness mental retardation
neuropathy motor sensory type 2 deafness mental retardation
neuropathy sensory spastic paraplegia
neurosensory nonsyndromic recessive deafness
neurosensory nonsyndromic recessive deafness dfnb1 linked
neurosensory nonsyndromic recessive deafness dfnb10 linked
neurosensory nonsyndromic recessive deafness dfnb11 linked
neurosensory nonsyndromic recessive deafness dfnb12 linked
neurosensory nonsyndromic recessive deafness dfnb13 linked
neurosensory nonsyndromic recessive deafness dfnb14 linked
neurosensory nonsyndromic recessive deafness dfnb15 linked
neurosensory nonsyndromic recessive deafness dfnb16 linked
neurosensory nonsyndromic recessive deafness dfnb17 linked
neurosensory nonsyndromic recessive deafness dfnb18 linked
neurosensory nonsyndromic recessive deafness dfnb19 linked
neurosensory nonsyndromic recessive deafness dfnb2 linked
neurosensory nonsyndromic recessive deafness dfnb20 linked
neurosensory nonsyndromic recessive deafness dfnb3 linked
neurosensory nonsyndromic recessive deafness dfnb4 linked
neurosensory nonsyndromic recessive deafness dfnb5 linked
neurosensory nonsyndromic recessive deafness dfnb6 linked
neurosensory nonsyndromic recessive deafness dfnb7 linked
neurosensory nonsyndromic recessive deafness dfnb8 linked
neurosensory nonsyndromic recessive deafness dfnb9 linked
neutral lipid storage myopathy
neutropenia and hyperlymphocytosis with large granular lymphocytes
neutropenia cyclic
neutropenia intermittent
neutropenia monocytopenia deafness
neutropenia severe congenital
Nevi atrial myxoma melanocytic nevi ephelides
nevi flammei familial multiple
nevo syndrome
nevoid basal cell carcinoma
nevus of ota retinitis pigmentosa
nevus sebaceus of jadassohn
nezelof syndrome
nhl
nicolaides baraitser syndrome
niemann pick a and b disease
niemann pick c disease
niemann pick c1 disease
niemann pick c2 disease
nievergelt syndrome
night blindness congenital stationary with myopia
night blindness skeletal anomalies unusual facies
niikawa kuroki syndrome
nijmegen breakage syndrome
nivelon nivelon mabille syndrome
noack syndrome
noble bass sherman syndrome
nodular erythema digital changes
noma
non hodgkin malignant lymphoma
non-functioning pancreatic endocrine tumor
noninsulin-dependent diabetes mellitus with deafness
nonne milroy syndrome
noonan like contracture myopathy hyperpyrexia
noonan like syndrome
noonan syndrome
norman roberts lissencephaly syndrome
normokaliemic periodic paralysis

norrie disease
northern epilepsy
norum disease
nose agenesia
nose polyposis, familial
nova syndrome
novak syndrome
o doherty syndrome
o donnell pappas syndrome
occipital horn syndrome
occult spinal dysraphism
ochoa syndrome
ocular apraxia cogan type
ocular coloboma imperforate anus
ocular coloboma recessive type
oculo cerebral dysplasia
oculo cerebro acral syndrome
oculo cerebro osseous syndrome
oculo digital syndrome
oculo facio cardio dental syndrome
oculo oto radial syndrome
oculo skeletal renal syndrome
oculo tricho anal syndrome
oculo tricho dysplasia
oculo-urethro-synovial syndrome
oculoauriculofrontonasal syndrome
oculoauriculovertebral dysplasia
oculocerebral hypopigmentation syndrome cross type
oculocerebral hypopigmentation syndrome type preus
oculocerebrocutaneous syndrome
oculocutaneous albinism
oculocutaneous albinism immunodeficiency
oculocutaneous albinism type 1
oculocutaneous albinism type 2
oculocutaneous albinism type 3
oculocutaneous albinism tyrosinase negative
oculocutaneous albinism tyrosinase positive
oculocutaneous tyrosinemia
oculodental syndrome rutherfurd syndrome
oculodentodigital dysplasia dominant
oculodentoosseous dysplasia dominant
oculodentoosseous dysplasia recessive
oculogastrointestinal muscular dystrophy
oculomaxillofacial dysostosis
oculomelic amyoplasia
oculopalatocerebral dwarfism
oculopalatoskeletal syndrome
oculopharyngeal muscular dystrophy
oculorenocerebellar syndrome
odonto onycho dysplasia with alopecia
odontochondrodysplasia
odontomatosis aortae oesophagus stenosis
odontomicronychial dysplasia
odontoonychodermal dysplasia
odontotrichomelic hypohidrotic dysplasia
oeis complex
oerter friedman anderson syndrome
oesophageal atresia
oesophageal atresia associated anomalies
oesophageal duodenal atresia abnormalities of hands and feet
ofd syndrome type 8
ofd syndrome type figuera
ohaha syndrome
ohdo madokoro sonoda syndrome
okamuto satomura syndrome
okihiro syndrome
oligodactyly tetramelic postaxial
oligomeganephronic renal hypoplasia
oligomeganephrony

oliver mcfarlane syndrome
oliver syndrome
olivopontocerebellar atrophy deafness
olivopontocerebellar atrophy type 1
olivopontocerebellar atrophy type 2
olivopontocerebellar atrophy type 3
olivopontocerebellar hypoplasia lethal type
Ollier disease
Olmsted syndrome
omodysplasia
omodysplasia autosomal recessive form
omphalocele
omphalocele cleft palate syndrome lethal
omphalocele exstrophy imperforate anus
omphalomesenteric cyst
onat syndrome
onchocercosis
ondine syndrome
onychonychia hypoplasic distal phalanges
onychoosteodysplasia
onychotrichodysplasia and neutropenia
oochs syndrome
ophtalmic ichtyosis
ophtalmomandibulomelic dysplasia
ophtalmoplegia myalgia tubular aggregates
ophthalmo acromelic syndrome
ophthalmoplegia ataxia hypoacusis
ophthalmoplegia mental retardation lingua scrotalis
ophthalmoplegia progressive external scoliosis
opitz frias syndrome
opitz mollica sorge syndrome
opitz reynolds fitzgerald syndrome
opitz syndrome x-linked
opitz trigonocephaly syndrome
opsismodysplasia
optic atrophy
optic atrophy autosomal dominant
optic atrophy deafness neuropathy
optic atrophy leber type
optic atrophy ophthalmoplegia ptosis deafness myopia
optic atrophy polyneuropathy deafness
optic nerve coloboma with renal disease
optic pathway glioma
opticoacoustic nerve atrophy dementia
oral facial digital syndrome
oral facial digital syndrome type 3
oral facial digital syndrome type 4
oral facial dyskinesia
ornithine aminotransferase deficiency
ornithine carbamoyltransferase deficiency
oro acral syndrome
orocraniodigital syndrome
orofaciodigital syndrome gabrielli type
orofaciodigital syndrome shashi type
orofaciodigital syndrome thurston type
orofaciodigital syndrome type 2
orofaciodigital syndrome type 8
orofaciodigital syndrome type figuera
orofaciodigital syndrome type1
oromandibular limb hypoplasia
oromandibular limb hypoplasia
orotic aciduria hereditary
oroticaciduria
orotidylic decarboxylase deficiency
osebold remondini syndrome
oslam syndrome
osteoarthropathy of fingers familial
osteochondritis deformans
osteochondritis dissecans

osteochondrodysplasia thrombocytopenia hydrocephalus
osteocraniostenosis
osteodysplasia familial anderson type
osteodysplastic dwarfism corsello type
osteoectasia familial
osteogenesis imperfecta
osteogenesis imperfecta congenita microcephaly and cataracts
osteogenesis imperfecta congenita neonatal lethal form
osteogenesis imperfecta congenital joint contractures
osteogenesis imperfecta retinopathy
osteogenesis imperfecta type 1
osteogenesis imperfecta type 2 dominant form
osteogenesis imperfecta type 2 recessive form
osteogenesis imperfecta type 3
osteogenesis imperfecta type 4
osteogenesis imperfecta vrolik type
osteogenesis imperfecta with blue sclerae
osteogenic sarcoma
osteoglophonic dwarfism
osteolysis hereditary multicentric
osteolysis syndrome recessive
osteomesopyknosis
osteopathia condensans disseminata with osteopoikilosis
osteopathia striata cranial sclerosis
osteopathia striata pigmentary dermopathy white forelock
osteopenia mental retardation sparse hair
osteopetrosis
osteopetrosis autosomal dominant type 1
osteopetrosis autosomal dominant type 2
osteopetrosis lethal
osteopetrosis malignant
osteopetrosis mild autosomal recessive form
osteopetrosis renal tubular acidosis
osteopoikilosis
osteoporosis macrocephaly mental retardation blindness
osteoporosis oculocutaneous hypopigmentation syndrome
osteoporosis pseudoglioma syndrome
osteosarcoma
osteosarcoma limb anomalies erythroid macrocytosis
osteosclerose type stanescu
osteosclerosis abnormalities of nervous system and meninge
osteosclerosis autosomal dominant worth type
ostertag type amyloidosis
ostravik lindemann solberg syndrome
ota kawamura ito syndrome
otodental dysplasia
otofaciocervical syndrome
otoonychoperoneal syndrome
otopalatodigital syndrome
otopalatodigital syndrome type 1
otopalatodigital syndrome type 2
otosclerosis
otospondylomegaepiphyseal dysplasia
ouvrier billson syndrome
ovarian insufficiency due to FSH resistance
overfolded helix
overgrowth radial ray defect arthrogryposis
overgrowth syndrome type fryer
overhydrated hereditary stomatocytosis
oxalosis
oxalosis type 1
oxalosis type 2
oxoglutaricaciduria
pachydermoperiostosis
pachygyria joint contractures facial abnormalities
pachygyria mental retardation epilepsy
pachyonychia congenita
pachyonychia congenita jackson lawler type
pacman syndrome

paes whelan modi syndrome
paget disease extramammary
paget disease juvenile type
pagon bird detter syndrome
pagon stephan syndrome
pai levkoff syndrome
pai syndrome
palant cleft palate syndrome
pallister hall syndrome
pallister killian syndrome
pallister w syndrome
palmer pagon syndrome
palmitoyl proteine thioesterase deficiency
palmoplantar porokeratosis of Mantoux
pancreas agenesis
pancreatic carcinoma familial
pancreatic hypoplasia diabetes heart disease
pancreatic lipomatosis duodenal stenosis
pancreatitis hereditary
pancreatoblastoma
pancytopenia multiple congenital anomalies
panhypopituitarism
panostotic fibrous dysplasia
papilloma of choroid plexus
papillon lefevre syndrome
parainfluenza virus type 3 antenatal infection
paramyotonia congenita of von eulenburg
parana hard skin syndrome
paraparesis amyotrophy of hands and feet
paraplegia-brachydactyly-cone shaped epiphysis
paraplegia-mental retardation-hyperkeratosis
parastremmatic dwarfism
parathyroid carcinoma
parc syndrome
parenti fraccaro type achondrogenesis
paris-trousseau thrombopenia
parkes weber syndrome
parkinson dementia steele type
parkinson disease (genetic types)
parkinsonism early onset mental retardation
paroxysmal nocturnal hemoglobinuria
parry romberg syndrome
partial atrioventricular canal
partial deletion of y
partington anderson syndrome
partington mulley syndrome
parvovirus antenatal infection
pascuel castroviejo syndrome
pashayan syndrome
passwell goodman ziprkowski syndrome
patau syndrome
patel bixler syndrome
patella aplasia coxa vara tarsal synostosis
patella hypoplasia mental retardation
patella hypoplasia skeletal malformations
patent ductus arteriosus familial
patterson lowry syndrome
patterson pseudoleprechaunism syndrome
patterson stevenson syndrome
pauciarticular chronic arthritis
pavone fiumara rizzo syndrome
pearson syndrome
pectus excavatum macrocephaly dysplastic nails
peho syndrome
pelizaeus merzbacher disease
pelizaeus merzbacher disease autosomal dominant or late onset type
pelizaeus merzbacher disease recessive acute infantile
pelizaeus merzbacher disease x linked
pellagra like syndrome

pelvi shoulder dysplasia
pelvic dysplasia arthrogryposis of lower limbs
pemphigus vulgaris familial
pena shokeir syndrome type 1
pena shokeir syndrome type 2
penis agenesia
penoscrotal transposition
pentasomy x
pentosuria
penttinen-aula syndrome
pepck deficiency
pepck1 deficiency
pepck2 deficiency
peptidic growth factors deficiency
periarteritis nodosa
pericardial constriction growth failure
pericardial defect diaphragmatic hernia
pericarditis arthropathy camptodactyly syndrome
pericardium absent mental retardation short stature
pericardium congenital anomaly
periodic disease
periventricular nodular heterotopia
perlman syndrome
pernicious anemia
perniola krajewska carnevale syndrome
peroneal atrophy parkinsonism ptosis strabismus
perrault syndrome
persistent mullerian duct syndrome
pertussis
peters anomaly
peters anomaly with short limb dwarfism
peters congenital glaucoma
peters plus syndrome
petit fryns syndrome
petty laxova wiedemann syndrome
peutz jeghers syndrome
peyronie syndrome
pfeiffer cardiocranial syndrome
pfeiffer hirschfelder rott syndrome
pfeiffer kapferer syndrome
pfeiffer mayer syndrome
pfeiffer palm teller syndrome
pfeiffer rockelein syndrome
pfeiffer singer zschiesche syndrome
pfeiffer tietze welte syndrome
pfeiffer type acrocephalosyndactyly
phacomatosis pigmentokeratotica
phacomatosis pigmentovascularis
phaoke sharma agarawal syndrome
phaver syndrome
phenobarbital antenatal infection
phenobarbital embryopathy
phenothiazine antenatal infection
phenylalanine hydroxylase deficiency
phenylketonuria
phenylketonuria type 2
phenylketonuric embryopathy
pheochromocytoma
phocomelia contractures absent thumb
phocomelia ectrodactyly deafness sinus arrhythmia
phocomelia schinzel type
phocomelia thrombocytopenia encephalocele
phosphoenolpyruvate carboxykinase 1 deficiency
phosphoenolpyruvate carboxykinase 2 deficiency
phosphoenolpyruvate carboxykinase deficiency
phosphoethanolaminuria
phosphoglucomutase deficiency
phosphoglucomutase deficiency type 1
phosphoglucomutase deficiency type 2

phosphoglucomutase deficiency type 3
phosphoglucomutase deficiency type 4
phosphoglycerate kinase 1 deficiency
phosphomannoisomerase deficiency
phosphoribosylpyrophosphate synthetase deficiency
phytanic acid oxidase deficiency
phytosterolemia
pibids syndrome
Picardi-Lassueur-Little syndrome
pick disease of brain
piebald trait neurologic defects
piebaldism
piepkorn karp hickoc syndrome
pierre marie cerbellar ataxia
pierre robin sequence congenital heart defect talipes
pierre robin sequence faciodigital anomaly
pierre robin syndrome
pierre robin syndrome fetal chondrodysplasia
pierre robin syndrome hyperphalangy clinodactyly
pierre robin syndrome oligodactyly
pierre robin syndrome skeletal dysplasia polydactyly
pigment anomaly ectrodactyly hypodontia
pignata guarino syndrome
Pili canulati
Pili multigemini
pili torti
pili torti developmental delay neurological abnormalities
pili torti nerve deafness
pili torti onychodysplasia
pillay syndrome
pilo dento ungular dysplasia microcephaly
pilodental dysplasia with refractive errors
pilotto syndrome
pinheiro freire maia miranda syndrome
pinsky di george harley syndrome
pipecolic acidemia
pitt hopkins syndrome
pitt williams brachydactyly
pitt-rogers-danks syndrome
pituitary dwarfism
pityriasis rubra pilaris
piussan lenaerts mathieu syndrome
plagiocephaly x linked mental retardation
plague
plasmalogenes synthesis deficiency isolated
plasminogen activator, tissue type, familial defective release of
plasminogen activitor inhibitor type 1 deficiency
plasminogen deficiency
platyspondylic lethal chondrodysplasia
platyspondyly amelogenesis imperfecta
plott syndrome
plum syndrome
Pneumocystosis
podder-tolmie syndrome
poems syndrome
poikiloderma atrophicans cataract
poikiloderma congenital with bullae weary type
poikiloderma hereditary acrokeratotic weary type
poikiloderma of Kindler
poikiloderma of rothmund thomson
poikilodermatomyositis mental retardation
poikilodermia alopecia retrognathism cleft palate
pointer syndrome
poland anomaly
poliomyelitis
polycystic kidney disease adult type
polycystic kidney disease dominant type
polycystic kidney disease infantile type
polycystic kidney disease recessive type

polycystic kidney disease type 1
polycystic kidney disease type 2
polycystic kidney disease type 3
polycystic liver disease
polycystic ovarian disease, familial
polycystic ovaries urethral sphincter dysfunction
polydactyly
polydactyly alopecia seborrheic dermatitis
polydactyly cleft lip palate psychomotor retardation
polydactyly cleft lip palate psychomotor retardation
polydactyly myopia syndrome
polydactyly neonatal chondrodystrophy
polydactyly postaxial
polydactyly postaxial dental and vertebral
polydactyly postaxial with median cleft of upper lip
polydactyly preaxial type 1
polydactyly syndrome middle ray duplication
polydactyly visceral anomalies cleft lip palate
polymicrogyria turricephaly hypogenitalism
polymorphic catecholinergic ventricular tachycardia
polymyositis
polyneuropathy hand defect
polyneuropathy hepatosplenomegaly hyperpigmentation
polyneuropathy mental retardation acromicria premature menopause
polyostotic fibrous dysplasia
polyposis hamartomatous intestinal
polyposis skin pigmentation alopecia fingernail changes
polysyndactyly cardiac malformation
polysyndactyly microcephaly ptosis
polysyndactyly orofacial anomalies
polysyndactyly overgrowth syndrome
polysyndactyly trigonocephaly agenesis of corpus callosum
polysyndactyly type 4
polysyndactyly type haas
pompe disease
poncet-spiegler's cylindroma
popliteal pterygium syndrome
popliteal pterygium syndrome lethal type
porencephaly cerebellar hypoplasia malformations
porokeratosis of Mibelli
porokeratosis plantaris palmaris and disseminata
porphyria
porphyria acute intermittent
porphyria congenital erythropoietic (gunther disease)
porphyria cutanea tarda familial type
porphyria cutanea tarda sporadic type
porphyria variegata
port wine nevi mega cisterna magna hydrocephalus
portal hypertension due to infrahepatic block
portal thrombosis
portal vein thrombosis
portuguese type amyloidosis
positive rheumatoid factor polyarthritis
post poliomyelitic syndrome
postaxial polydactyly mental retardation
posterior valve urethra
potassium aggravated myotonia
potter disease type 1
potter disease type 3
potter sequence cleft cardiopathy
potter syndrome dominant type
powell buist stenzel syndrome
powell chandra saal syndrome
powell venencie gordon syndrome
prader willi syndrome
prata liberal goncalves syndrome
preauricular pits renal disease
preaxial deficiency postaxial polydactyly hypospadias
preaxial polydactyly colobomata mental retardation

precocious epileptic encephalopathy
precocious myoclonic encephalopathy
precocious puberty gonadotropin-dependant
precocious puberty idiopathic or tumoral
precocious puberty male limited
preeyasombat viravithya syndrome
prekallikrein deficiency
premature aging, okamoto type
premature atherosclerosis photomyoclonic epilepsy diabetes mellitus nephropathy degenerative neurologic disease
premature menopause, familial
prieto badia mulas syndrome
prieur griscelli syndrome
primary biliary cirrhosis
primary ciliary dyskinesia
primary lymphedema
primary malignant lymphoma
primary pulmonar fibrosis
primary syringomyelia
primary tubular proximal acidosis
primerose syndrome
primitive neuroectodermal tumor
primordial microcephalic dwarfism crachami type
progeria
progeria short stature pigmented nevi
progeria variant syndrome ruvalcaba type
progeroid syndrome de barsy type
progeroid syndrome neonatal
progeroid syndrome petty type
progeroid syndrome, penttinen type
prognathism dominant
progressive acromelanosis
progressive black carbon hyperpigmentation of infancy
progressive diaphyseal dysplasia
progressive external ophtalmoplegia
progressive hearing loss stapes fixation
progressive supranuclear palsy atypical
progressive symmetrical erythrokeratoderma
prolactinoma
prolerating trichilemmal cyst
prolidase deficiency
proline oxydase deficiency
prominent glabella microcephaly hypogenitalism
properdin deficiency
propionic acidemia
propionic acidemia type 1
propionic acidemia type 2
propionyl coa carboxylase deficiency
propionyl coa carboxylase deficiency type 1
propionyl coa carboxylase deficiency type 2
propping zerres syndrome
proptosis robin association overlapping fingers hypospadias
prosencephaly cerebellar dysgenesis
prostaglandin antenatal infection
prostate cancer familial
protein c deficiency
protein r deficiency
protein s acquired deficiency
protein s deficiency
proteus like syndrome mental retardation eye defects
proteus syndrome
prothrombin deficiency
protoporphyria erythropoietic
proud levine carpenter syndrome
proximal myotonic dystrophy
proximal myotonic myopathy
proximal spinal muscular atrophy
proximal tubulopathy diabetes mellitus cerebellar ataxia
prune belly syndrome

pseudo hurler polydystrophy
pseudo pelade of Brocq
pseudo trisomy 13 syndrome
pseudo turner syndrome
pseudo zellweger syndrome
pseudoachondroplasia
pseudoachondroplastic dysplasia
pseudoachondroplastic dysplasia 1
pseudoadrenoleukodystrophy
pseudoaminopterin syndrome
pseudohermaphrodism anorectal anomalies
pseudohermaphroditism female skeletal anomalies
pseudohermaphroditism male with gynecomastia
pseudohermaphroditism mental retardation
Pseudohyperaldosteronism
pseudohypoaldosteronism type 1
pseudohypoaldosteronism type 2
pseudohypoparathyroidism
pseudomarfanism
pseudomongolism
pseudoobstruction idiopathic intestinal
pseudopapilledema blepharophimosis hand anomalies
pseudoprogeria syndrome
pseudotoxoplasmosis syndrome
pseudovaginal perineoscrotal hypospadias
pseudoxanthoma elasticum
pseudoxanthoma elasticum dominant form
pseudoxanthoma elasticum recessive form
pterygia mental retardation facial dysmorphism
pterygium colli mental retardation digital anomalies
pterygium of the conjunctiva
pterygium syndrome antecubital
pterygium syndrome multiple dominant type
pterygium syndrome x linked
ptosis coloboma mental retardation
ptosis coloboma trigonocephaly
ptosis strabismus diastasis
ptosis strabismus ectopic pupils
ptosis vocal cord paralysis
pulmonar arterioveinous aneurysm
pulmonary agenesis
pulmonary alveolar proteinosis congenital
pulmonary aortic stenosis obstructive uropathy
pulmonary arterio-veinous fistula
pulmonary artery agenesis
pulmonary artery coming from the aorta
pulmonary artery familial dilatation
pulmonary atresia with ventricular septal defect
pulmonary branches stenosis
pulmonary cystic lymphangiectasis
pulmonary hypertension primary
pulmonary hypoplasia familial primary
pulmonary sequestration
pulmonary stenosis cafe au lait spots
pulmonary supravalvular stenosis
pulmonary surfactant protein b deficiency of
pulmonary valve stenosis
pulmonary valves agenesis
pulmonary veins stenosis
pulmonary venous return anomaly
pulmonaryatresia intact ventricular septum
punctate acrokeratoderma freckle like pigmentation
pure macular dystrophy
Puretic syndrome
puretic syndrome
purine nucleoside phosphorylase deficiency
purtilo syndrome
pycnodysostosis
pyknoachondrogenesis

pyle disease
Pyomyositis
pyramidal molar glaucoma upper abnormal lip
pyridoxine deficit
pyrimidinemia familial
pyroglutamicaciduria
pyropoikilocytosis
pyruvate carboxylase deficiency
pyruvate decarboxylase deficiency
pyruvate dehydrogenase deficiency
pyruvate kinase deficiency
pyruvate kinase deficiency liver type
pyruvate kinase deficiency muscle type
qazi markouizos syndrome
quattrin mcpherson syndrome
Quinquaud's decalvans folliculitis
Quinquaud's decalvans folliculitis
rabies
rabson mendenhall syndrome
radial defect robin sequence
radial deficiency tibial hypoplasia
radial hypoplasia triphalangeal thumbs hypospadias maxillary diastema
radial ray agenesis
radial ray hypoplasia choanal atresia
radiculomegaly of canine teeth congenital cataracts
radio digito facial dysplasia
radio renal syndrome
radio-ulnar synostosis
radioulnar synostosis mental retardation hypotonia
radioulnar synostosis retinal pigment abnormalities
radius absent anogenital anomalies
raine syndrome
rambam hasharon syndrome
rambaud galian syndrome
ramer ladda syndrome
ramon syndrome
ramos arroyo clark syndrome
ramsay hunt syndrome
rapadilino syndrome
rapp hodgkin syndrome
rasmussen johnsen thomsen syndrome
rasmussen subacute encephalitis
ray peterson scott syndrome
rayner lampert rennert syndrome
reardon hall slaney syndrome
reardon wilson cavanagh syndrome
recurrent peripheral facial palsy
red fever
reductional transverse limb defects
refetoff syndrome
Refetoff syndrome
refsum disease
refsum disease infantile form
reginato schiapachasse syndrome
reifenstein syndrome
reinhardt pfeiffer syndrome
relapsing polychondritis
renal adysplasia dominant type
renal agenesis
renal agenesis meningomyelocele mullerian defect
renal caliceal diverticuli deafness
renal carcinoma familial
renal dysplasia diffuse autosomal recessive
renal dysplasia diffuse cystic
renal dysplasia hepatic fibrosis dandy walker
renal dysplasia limb defects
renal dysplasia megalocystis sirenomelia
renal dysplasia mesomelia radiohumeral fusion
renal dysplasia retinal aplasia

renal dysplasia retinal aplasia
renal genital middle ear anomalies
renal hepatic pancreatic dysplasia dandy walker cyst
renal tubular acidosis distal
renal tubular acidosis distal autosomal dominant
renal tubular acidosis distal autosomal recessive
renal tubular acidosis distal type 3
renal tubular acidosis distal type 4
renal tubular acidosis progressive nerve deafness
rendu osler weber disease
renier gabreels jasper syndrome
renoanogenital syndrome
renotubular dysgenesis
resistance to LH
resistance to thyroid stimulating hormone
respiratory chain deficiency malformations
reticulosis familial histiocytic
retinal degeneration nanophthalmos glaucoma
retinal detachment occipital encephalocele
retinal dysplasia x linked
retinal telangiectasia hypogammaglobulinemia
retinis pigmentosa deafness hypogenitalism
retinis pigmentosa-deafness
retinitis pigmentosa
retinitis pigmentosa mental retardation deafness
retinoblastoma
retinohepatoendocrinologic syndrome
retinopathy anemia cns anomalies
retinopathy aplastic anaemia neurological abnormalities
retinopathy pigmentary mental retardation
retinopathy pigmentosa
retinoschisis juvenile
retinoschisis x linked
retraction syndrome
rett like syndrome
rett syndrome
revesz debuse syndrome
reye syndrome
reynolds neri hermann syndrome
Reynolds syndrome
rhabdomyomatous dysplasia cardiopathy genital anomalies
rhabdomyosarcoma
rhabdomyosarcoma 1
rhabdomyosarcoma 2
rhabdomyosarcoma alveolar
rhabdomyosarcoma embryonal
rhizomelic dysplasia type patterson lowry
rhizomelic pseudopolyarthritis
rhizomelic syndrome
rhumatoid purpura
richards rundle syndrome
richieri costa colletto otto syndrome
richieri costa da silva syndrome
richieri costa gorlin syndrome
richieri costa guion almeida acrofacial dysostosis
richieri costa guion almeida cohen syndrome
richieri costa guion almeida dwarfism
richieri costa guion almeida rodini syndrome
richieri costa guion almeida syndrome
richieri costa montagnoli syndrome
richieri costa orquizas syndrome
richieri costa silveira pereira syndrome
rickettsiosis
rieger syndrome
right atrium familial dilatation
right ventricule hypoplasia
rigid mask like face deafness polydactyly
rigid spine syndrome
riley day syndrome

riley smith syndrome
ring chromosome 17
ringed hair disease
rippberger aase syndrome
ritscher schinzel syndrome
rivera perez salas syndrome
roberts syndrome
robin sequence oligodactyly
robinow like syndrome
robinow sorauf syndrome
robinow syndrome
robinow syndrome recessive form
robinson miller bensimon syndrome
Roch-Leri mesosomatous lipomatosis
rod myopathy
rodini richieri costa syndrome
rokitansky kuster hauser syndrome
rokitansky sequence
romano ward syndrome
rombo syndrome
rommen mueller sybert syndrome
rosenberg chutorian syndrome
rosenberg lohr syndrome
rotor syndrome
roussy levy hereditary areflexic dystasia
roy maroteaux kremp syndrome
rozin hertz goodman syndrome
rubella virus antenatal infection
rubinstein taybi like syndrome
rubinstein taybi syndrome
rudd klimek syndrome
rudiger syndrome
ruiz rivas ramirez syndrome
russell weaver bull syndrome
rutherfurd syndrome
rutledge friedman harrod syndrome
ruvalcaba churesigaew myhre syndrome
ruvalcaba myhre smith syndrome
ruvalcaba myhre syndrome
ruvalcaba syndrome
ruzicka goerz anton syndrome
saal bulas syndrome
saal greenstein syndrome
sabinas brittle hair syndrome
saccharopine dehydrogenase deficiency
saccharopinuria
sackey sakati aur syndrome
sacral agenesis
sacral defect anterior sacral meningocele
sacral hemangiomas multiple congenital abnormalities
sacral meningocele conotrancal heart defects
sacrococcygeal dysgenesis association
saethre chotzen syndrome
saito kuba tsuruta syndrome
sakati nyhan syndrome
salcedo syndrome
saldino mainzer syndrome
saldino noonan maccreanor syndrome
salla disease
sallis beighton syndrome
salmonellosis
salti salem syndrome
sammartino decreccio syndrome
samson gardner syndrome
samson viljoen syndrome
sanderson fraser syndrome
sandhaus ben ami syndrome
sandhoff disease
sandrow sullivan steel syndrome

sanfilippo disease
sanfilippo syndrome type a
sanfilippo syndrome type b
sanfilippo syndrome type c
sanfilippo syndrome type d
santavuori disease
santavuori haltia disease
santos mateus leal syndrome
sapho syndrome
sarcoidosis
sarcosin dehydrogenase complex deficiency
sarcosinemia
satoyoshi syndrome
saul wilkes stevenson syndrome
say barber hobbs syndrome
say barber miller syndrome
say carpenter syndrome
say field coldwell syndrome
say meyer syndrome
sc phocomelia syndrome
scad deficiency
scalp defects postaxial polydactyly
scalp ear nipple syndrome
scapuloiliac dysostosis
scarf syndrome
schaap taylor baraitser syndrome
schaefer stein oshman syndrome
scheie syndrome
scheuermann juvenile kyphosis dominant form
schiel stengel rutkowski syndrome
schimke syndrome
schindler disease
schinzel acrocallosal syndrome
schinzel giedion midface retraction syndrome
schinzel syndrome
schisis association
schistosomiasis
schizencephaly
schizophrenia
schizophrenia mental retardation deafness retinitis
schlegelberger grote syndrome
schmidt syndrome
schmitt gillenwater kelly syndrome
schneckenbecken dysplasia
schofer beetz bohl syndrome
scholte begeer van essen syndrome
schrander stumpel theunissen hulsmans syndrome
schroer hammer mauldin syndrome
schwannomatosis
schwartz jampel syndrome
schwartz newark syndrome
schweitzer kemink malcolm syndrome
scimitar syndrome
scleroatonic myopathy
sclerocornea syndactyly ambiguous genitalia
scleroderma
sclerosing bone dysplasia mental retardation
sclerosing cholangitis
sclerosteosis
scoditti geminiani colonna syndrome
scoliosis with unilateral unsegmented bar
scot deficiency
scott bryant graham syndrome
scott syndrome
seaver cassidy syndrome
sebocystomatosis
seckel like syndrome majoor krakauer type
seckel like syndrome type buebel
seckel syndrome

sedaghatian chondrodysplasia
seemanova lesny syndrome
seemanova syndrome type 2
segawa syndrome
seghers syndrome
segmental vertebral anomalies
seizures benign familial neonatal recessive form
seizures mental retardation hair dysplasia
selig benacerraf greene syndrome
sellars beighton syndrome
seminoma
semmerkrot haraldsson weenaes syndrome
sengers hamel otten syndrome
senior syndrome
sensenbrenner syndrome
sensory and autonomic neuropathy type 4 hereditary
sensory neuropathy deafness dementia
sensory neuropathy type 1
sensory radicular neuropathy recessive form
senter syndrome
seow najjar syndrome
septooptic dysplasia
septooptic dysplasia digital anomalies
sequeiros sack syndrome
seres santamaria arimany muniz syndrome
serious digitalic intoxication
serpentine fibula polycystic kidneys
serpentine fibula syndrome
setleis syndrome
severe combined immunodeficiency alymphocytotic type
severe combined immunodeficiency due to adenosine deaminase deficiency
severe combined immunodeficiency hla class 2-negative
sezary's lymphoma
sezary's syndrome
sharma kapoor ramji syndrome
sharp syndrome
shigellosis
shith filkins syndrome
shokeir syndrome
short broad great toe macrocranium
short limb dwarf lethal colavita kozlowski type
short limb dwarf lethal mcalister crane type
short limb dwarf mental retardation myopia
short limb dwarf oedema iris coloboma
short limb dwarfism al gazali type
short limbs abnormal face congenital heart disease
short limbs subluxed knees cleft palate
short rib polydactyly syndrome
short rib polydactyly syndrome majewski type
short rib polydactyly syndrome saldino noonan type
short rib polydactyly syndrome verma naumoff type
short rib syndrome beemer type
short ribs craniosynostosis polysyndactyly
short stature abnormal skin pigmentation mental retardation
short stature brussels type
short stature contractures hypotonia
short stature cranial hyperostosis hepatomegaly diabetes
short stature deafness neutrophil dysfunction dysmorphism
short stature dysmorphic face pelvic scapula dysplasia
short stature heart defect craniofacial anomalies
short stature hyperkaliemia acidosis
short stature locking fingers
short stature mental retardation eye anomalies cleft lip palate
short stature mental retardation eye defects absent patella
short stature microcephaly heart defect
short stature microcephaly seizures deafness
short stature monodactylous ectrodactyly cleft palate
short stature prognathism short femoral necks
short stature robin sequence cleft madible hand anomalies clubfoot

short stature talipes natal teeth
short stature valvular heart disease characteristic facies
short stature webbed neck heart disease
short stature wormian bones dextrocardia
short syndrome
short tarsus absence of lower eyelashes
shoulder and thorax deformity congenital heart disease
shoulder and thorax deformity congenital heart disease
shoulder girdle defect mental retardation familial type
shprintzen golberg craniosynostosis
shprintzen omphalocele syndrome
shulman syndrome
shwachman-diamond syndrome
shy drager syndrome
sialidosis type 1 and 2
sialuria french type
sickle cell anemia
sickle cell anemia
sideroblastic anemia
sidransky feinstein goodman syndrome
siegler brewer carey syndrome
silengo lerone pelizzo syndrome
sillence syndrome
silver russell dwarfism
silvery hair syndrome
simosa penchaszadeh bustos syndrome
simpson golabi behmel syndrome
singh chhaparwal dhanda syndrome
single upper central incisor
single ventricle
single ventricule heart
sino-auricular heart block
sinus node disease and myopia
sipple syndrome
sirenomelia
sitosterolemia
situs inversus viscerum cardiopathy
situs inversus x linked
sjogren larsson like syndrome
sjogren larsson syndrome
sjogren syndrome
skeletal dysplasia brachydactyly
skeletal dysplasia epilepsy short stature
skeletal dysplasia orofacial anomalies
skeletal dysplasia san diego type
skeleto cardiac syndrome with thrombocytopenia
sketetal dysplasia coarse facies mental retardation
Skin peeling syndrome
slavotinek hurst syndrome
sly disease
small non cleaved cell lymphoma
small patella syndrome
smith fineman myers syndrome
smith lemli opitz syndrome
smith lemli opitz syndrome type 1
smith lemli opitz syndrome type 2
smith magenis syndrome
smith martin dodd syndrome
sneddon syndrome
sohval soffer syndrome
somatostatinoma
sommer hines syndrome
sommer rathbun battles syndrome
sommer young wee frye syndrome
sondheimer syndrome
sonoda syndrome
sosby syndrome
sotos syndrome
sparse hair ptosis mental retardation

spastic angina with healthy coronary artery
spastic ataxia charlevoix-saguenay type
spastic diplegia infantile type
spastic paraparesis
spastic paraparesis deafness
spastic paraparesis, infantile
spastic paraplegia epilepsy mental retardation
spastic paraplegia facial cutaneous lesions
spastic paraplegia familial
spastic paraplegia familial autosomal recessive form
spastic paraplegia glaucoma precocious puberty
spastic paraplegia mental retardation corpus callosum thin
spastic paraplegia nephritis deafness
spastic paraplegia neuropathy poikiloderma
spastic paraplegia type 1 x linked
spastic paraplegia type 2 x linked
spastic paraplegia type 3 dominant
spastic paraplegia type 4 dominant
spastic paraplegia type 5a recessive
spastic paraplegia type 5b recessive
spastic paraplegia type 6 dominant
spastic paraplegia-pigmentary abnormalities
spastic paresis glaucoma mental retardation
spastic quadriplegia retinitis pigmentosa mental retardation
spasticity mental retardation
spasticity multiple exostoses
spatic paraparesis vitiligo premature graying
spellacy gibbs watts syndrome
spherocytosis hereditary
spherophakia brachymorphia syndrome
sphingomyelinase deficiency
spielmeyer vogt disease
spina bifida
spina bifida hypospadias
spinal and bulbar muscular atrophy
spinal atrophy ophthalmoplegia pyramidal syndrome
spinal dysostosis type anhalt
spinal muscular atrophy type 1
spinal muscular atrophy type 2
spinal muscular atrophy type 3
spinal muscular atrophy type i with congenital bone fractures
spine rigid cardiomyopathy
spinocerebellar ataxia 1
spinocerebellar ataxia 2
spinocerebellar ataxia 3
spinocerebellar ataxia 4
spinocerebellar ataxia 5
spinocerebellar ataxia 6
spinocerebellar ataxia 7
spinocerebellar ataxia 8
spinocerebellar ataxia amyotrophy deafness
spinocerebellar ataxia dysmorphism
spinocerebellar atrophy type 3
spinocerebellar degeneration corneal dystrophy
spinocerebellar degenerescence book type
splenic agenesis syndrome
splenogonadal fusion limb defects micrognatia
split hand deformity
split hand deformity mandibulofacial dysostosis
split hand split foot malformation autosomal recessive form
split hand split foot mandibular hypoplasia
split hand split foot nystagmus
split hand split foot x linked
split hand urinary anomalies spina bifida
sponastrime dysplasia
spondylo camptodactyly syndrome
spondylo costal dysostosis dandy walker
spondylo metaphyseal dysplasia algerian type
spondylo peripheral epiphyseal dysplasia

spondylocarpotarsal synostosis
spondylocostal dysostosis
spondylocostal dysplasia dominant
spondylodysplasia brachyolmia
spondyloenchondrodysplasia
spondyloendochromatosis
spondyloepimetaphysal dysplasia shoat lachman type
spondyloepimetaphyseal dysplasia
spondyloepimetaphyseal dysplasia joint laxity
spondyloepimetaphyseal dysplasia leonard type
spondyloepimetaphyseal dysplasia rao type
spondyloepimetaphyseal dysplasia type camera
spondyloepiphyseal dysplasia
spondyloepiphyseal dysplasia congenital dominant type
spondyloepiphyseal dysplasia macdermot type
spondyloepiphyseal dysplasia nephrotic syndrome
spondyloepiphyseal dysplasia reardon type
spondyloepiphyseal dysplasia stanescu type
spondyloepiphyseal dysplasia tarda dysmorphic facies
spondyloepiphyseal dysplasia tarda mental retardation
spondyloepiphyseal dysplasia tarda progressive arthropathy
spondyloepiphyseal dysplasia tarda toledo type
spondyloepiphyseal dysplasia with atlantoaxial instability
spondyloepiphyseal dysplasia with instability
spondylohumerofemoral hypoplasia
spondylohypoplasia arthrogryposis popliteal pterygium
spondylometa epiphyseal dysplasia borochowitz type
spondylometaepiphyseal dysplasia congenita strudwick type
spondylometaphyseal dysplasia
spondylometaphyseal dysplasia absent distal ulna
spondylometaphyseal dysplasia dentinogenesis imperfecta
spondylometaphyseal dysplasia hunter type
spondylometaphyseal dysplasia kozlowski type
spondylometaphyseal dysplasia lethal advanced bone
spondylometaphyseal dysplasia recessive form
spondylometaphyseal dysplasia with abnormal dentition
spondylometaphyseal dysplasia with endochromatous changes
spondylometaphyseal dysplasia with hypotrichosis
spondyloperipheral dysplasia short ulna
spongy degeneration of central nervous system
spontaneous pneumothorax familial type
Sporotrichosis
spranger schinzel yers syndrome
sprengel deformity
stalker chitayat syndrome
stampe sorensen syndrome
stanescu maroteaux syndrome
STAR protein deficiency
stargardt disease
steatocystoma multiplex
steatocystoma multiplex natal teeth
steele richardson olszewski syndrome atypical
stein-leventhal syndrome
Steinert disease
steinert myotonic dystrophy
steinfeld syndrome
sterility due to immotile flagella
stern lubinsky durrie syndrome
sternal cleft
sternal cyst vascular anomalies
sternal malformation vascular dysplasia association
steroid dehydrogenase deficiency dental anomalies
steroid sulfatase deficiency
stickler syndrome
stickler syndrome type 1
stickler syndrome type 2
stiff baby syndrome
stiff man syndrome
stiff skin syndrome

Still disease
still juvenile type disease
stimmler syndrome
stoelinga de koomen davis syndrome
stoll alembik dott syndrome
stoll alembik finck syndrome
stoll geraudel chauvin syndrome
stoll kieny dott syndrome
stoll levy francfort syndrome
storage pool platelet disease
stormorken sjaastad langslet syndrome
stratton garcia young syndrome
stratton parker syndrome
striatal degeneration familial
striatonigral degeneration infantile
strumpell lorrain disease
stuart factor deficiency
stuccokeratosis
sturge weber syndrome
stuve wiedemann dysplasia
subacute sclerosing panencephalitis
subaortic stenosis short stature syndrome
subcortical laminar heterotopia
subependymal nodular heterotopia
subpulmonary stenosis
subvalvular aortic stenosis
succinate coenzyme q reductase deficiency of
succinic acidemia
succinic acidemia lactic acidosis congenital
succinic semialdehyde dehydrogenase deficiency
succinic semialdehyde deshydrogenase deficiency
succinyl-coa acetoacetate transferase deficiency
sugarman syndrome
sujansky leonard syndrome
sulfatidosis juvenile austin type
sulfite and xanthine oxydase deficiency
sulfite oxidase deficiency
summitt syndrome
suprabulbar paresis congenital
supranuclear palsy progressive
Susac syndrome
sussman kelly rosenbaum syndrome
sutherland haan syndrome
sweet syndrome
swyer syndrome
sybert smith syndrome
symmetrical thalamic calcifications
symphalangism brachydactyly
symphalangism brachydactyly
symphalangism brachydactyly craniosynostosis
symphalangism cushing type
symphalangism distal
symphalangism familial proximal
symphalangism short stature accessory testis
symphalangism with multiple anomalies of hands and feet
syncamptodactyly scoliosis
syndactyly
syndactyly between 4 and 5
syndactyly cataract mental retardation
syndactyly cenani lenz type
syndactyly ectodermal dysplasia cleft lip palate hand foot
syndactyly type 1 microcephaly mental retardation
syndactyly type 2
syndactyly type 3
syndactyly type 5
syndactyly-polydactyly-ear lobe syndrome
syngnathia cleft palate
syngnathia multiple anomalies
synostosis microcephaly scoliosis

synostosis of talus and calcaneus short stature
synovialosarcoma
synovitis acne pustulosis hyperostosis osteitis syndrome
synovitis granulomatous uveitis cranial neuropathies
synpolydactyly
synspondylism
syphilis embryopathy
Syringocystadenoma papilliferum
syringomas natal teeth oligodontia
syringomelia hyperkeratosis
systemic arterio-veinous fistula
systemic lupus erythematosus
systemic mastocytosis
systemic polyarthritis
t cell immunodeficiency primary
t cell lymphoma
tabatznik syndrome
tachycardia hypertension microphthalmos hyperglycinuria
taiga encephalitis
tajara pinheiro syndrome
takayasu arteritis
takayasu's disease
tamari goodman syndrome
tang hsi ryu syndrome
tangier disease
tap 2 deficiency
tar syndrome
tardive dyskinesia
Tarui disease
tau syndrome
taurodontia absent teeth sparse hair
taurodontism
tay sachs disease
tay syndrome
taybi linder syndrome
taybi syndrome
teebi al saleh hassoon syndrome
teebi kaurah syndrome
teebi naguib alawadi syndrome
teebi shaltout syndrome
teebi syndrome
teeth noneruption of with maxillary hypoplasia and genu valgum
tel hashomer camptodactyly syndrome
telangiectasia hemorrhagic familial
telecanthus associated abnormalities
telecanthus hypertelorism pes cavus
telfer sugar jaeger syndrome
temporal arteritis
temporomandibular ankylosis
temtamy shalash syndrome
ter haar hamel hendricks syndrome
ter haar syndrome
teratoma
terminal transverse defects of arm
testicular feminization syndrome
testicular regression syndrome
testicular regression syndrome
tetanus
tetraamelia ectodermal dysplasia
tetraamelia multiple malformations
tetraamelia pulmonary hypoplasia
tetraamelia-syrinx
tetralogy of fallot
tetraploidy
tetrasomy 12p
tetrasomy 15q
tetrasomy 18p
tetrasomy 21q
tetrasomy 5p

tetrasomy 9p
tetrasomy x
thakker donnai syndrome
thalamic degeneration symmetrical infantile
thalamic degenerescence infantile
thanatophoric dwarfism
thanatophoric dysplasia
thanatophoric dysplasia cloverleaf skull
thanatophoric dysplasia glasgow variant
thanos stewart zonana syndrome
theodor hertz goodman syndrome
thiele syndrome
thiemann epiphyseal disease
thies reis syndrome
thin ribs tubular bones dysmorphism
thiolase deficiency
thiopurine s methyltranferase deficiency
thomas jewett raines syndrome
thomas syndrome
thombocytopenia X linked
thompson baraitser syndrome
thomsen disease
thong douglas ferrante syndrome
thoracic celosomia
thoracic dysplasia hydrocephalus syndrome
thoracic pelvic phalangeal dystrophy
thoraco abdominal enteric duplication
thoraco limb dysplasia rivera type
thoracoabdominal syndrome
thoracolaryngopelvic dysplasia
thoracopelvic dysostosis
Thost-Unna palmoplantar keratoderma
thrombocythemia essential
thrombocytopathy asplenia miosis
thrombocytopenia absent radius syndrome
thrombocytopenia absent ulnar
thrombocytopenia cerebellar hypoplasia short stature
thrombocytopenia chromosome breakage
thrombocytopenia multiple congenital anomaly
thrombocytopenia robin sequence
thrombocytopenic purpura autoimmune
thrombomodulin anomalies familial
thrombotic microangiopathy, familial
thumb absence hypoplastic halluces
thumb absent short stature immune deficiency
thumb deformity alopecia pigmentation anomaly
thumb stiff brachydactyly mental retardation
thymic carcinoma
thymic renal anal lung dysplasia
thymoma
thymus epithelial tumor
thymus malignant tumor
thyrocerebrorenal syndrome
thyroglossal tract cyst
thyroid carcinoma follicular
thyroid carcinoma papillary
thyroid hormone uesponsiveness
thyroid hormonogenesis genetic defect in iib
thyroid renal digital anomalies
tibia absent polydactyly
tibia absent polydactyly arachnoid cyst
tibiae bowed radial anomalies osteopennia fractures
tibial aplasia ectrodactyly
tibial aplasia ectrodactyly hydrocephalus
tibial hemimelia cleft lip palate
tibial muscular dystrophy tardive
tick-borne encephalitis
tollner horst manzke syndrome
toluene antenatal infection

toluene embryopathy
tomaculous neuropathy
tome brune fardeau syndrome
toni debre fanconi maladie
tonoki ohura niikawa syndrome
tooth and nail syndrome
toriello carey syndrome
toriello higgins miller syndrome
toriello lacassie droste syndrome
toriello syndrome
torres ayber syndrome
torticollis keloids cryptorchidism renal dysplasia
tosti misciali barbareschi syndrome
tourette syndrome
townes brocks syndrome
toxocariasis
toxopachyoteose diaphysaire tibio peroniere
toxoplasma fetal syndrome
toxoplasmosis
TPA, familial defective release of
tracheal agenesis
tracheobronchomegaly
tracheobronchopathia osteoplastica
tracheoesophageal fistula symphalangism
tracheophageal fistula hypospadias
tranebjaerg svejgaard syndrome
transcobalamin 2 deficiency
transient neonatal arthrogryposis
transverse limb deficiency hemangioma
treacher collins syndrome
treft sanborn carey syndrome
tremor hereditary essential
tremor nystagmus duodenal ulcer
trevor disease
triatrial heart
trichinellosis
Trichinosis
tricho dento osseous syndrome type 1
tricho odonto onycho dermal syndrome
tricho odonto onychodysplasia syndactyly dominant
tricho onychic dysplasia
tricho onycho hypohidrotic dysplasia
tricho retino dento digital syndrome
tricho-hepato-enterique syndrome
trichodental syndrome
trichodermal syndrome mental retardation
trichodermodysplasia dental alterations
trichodysplasia xeroderma
trichoepithelioma multiple familial
Trichofolliculloma
Trichomalacia
trichomegaly cataract hereditary spherocytosis
trichomegaly retina pigmentary degeneration dwarfism
trichoodontoonychial dysplasia
trichorhinophalangeal syndrome
trichorhinophalangeal syndrome type 2
Trichostasis spinulosa
trichothiodystrophie
trichothiodystrophy sun sensitivity
trichothiodystrophy with congenital ichtyosis
trico oculo dermo vertebral syndrome
tricuspid atresia
tricuspid dysplasia
trigonocephaly
trigonocephaly bifid nose acral anomalies
trigonocephaly broad thumbs
trigonocephaly ptosis coloboma
trigonocephaly ptosis mental retardation
trigonocephaly short stature developmental delay

trigonomacrocephaly tibial defect polydactyly
trihydroxycholestanoylcoa oxidase isolated deficiency
trimethadione antenatal infection
triopia
triose phosphate isomerase deficiency
triphalangeal thumb non opposable
triphalangeal thumb polysyndactyly syndrome
triphalangeal thumbs brachyectrodactyly
triphalangeal thumbs dislocation of patella
triphalangeal thumbs thrombocytopathy deafness
triple a syndrome
triploidy
trismus pseudocamptodactyly syndrome
trisomy 1 mosaicism
trisomy 10p
trisomy 10pter p13
trisomy 10q partial
trisomy 11q
trisomy 11q23
trisomy 12 mosaicism
trisomy 12p
trisomy 12q
trisomy 13
trisomy 13p
trisomy 13q
trisomy 14 mosaicism
trisomy 14qprox
trisomy 14qter
trisomy 15 mosaicism
trisomy 15q
trisomy 16 mosaicism
trisomy 16p
trisomy 16q
trisomy 17 mosaicism
trisomy 17p
trisomy 17p11 2
trisomy 17q22
trisomy 18
trisomy 18 mosaicism
trisomy 18p
trisomy 18q
trisomy 19q
trisomy 1p21 p32
trisomy 1q32 qter
trisomy 1q42 11 q42 12
trisomy 1q42 qter
trisomy 2 mosaicism
trisomy 20 mosaicism
trisomy 20p
trisomy 21
trisomy 22
trisomy 22q11 q13
trisomy 2p
trisomy 2p13 p21
trisomy 2pter p24
trisomy 2q
trisomy 2q37
trisomy 3 mosaicism
trisomy 3p
trisomy 3p25
trisomy 3q
trisomy 3q13 2 q25
trisomy 4p
trisomy 4q
trisomy 4q21
trisomy 4q25 qter
trisomy 5p
trisomy 5pter p13 3
trisomy 5q

trisomy 6p
trisomy 6q
trisomy 7 mosaicism
trisomy 7p
trisomy 7p13 p12 2
trisomy 7q
trisomy 8
trisomy 8p
trisomy 8q
trisomy 9 mosaicism
trisomy 9p partial
trisomy 9q32
trisomy x
trisomy xp3
trisomy xpter xq13
trisomy xq
trisomy xq25
trochlear dysplasia
troncus arteriosus malformation
troyer syndrome
true hermaphrodism xx
trueb burg bottani syndrome
tsao ellingson syndrome
tsukahara azuno kajii syndrome
tsukahara kajii syndrome
tsukuhara syndrome
tuberculosis
tuberous sclerosis
tuberous sclerosis type 1
tuberous sclerosis type 2
tucker syndrome
tuffli laxova syndrome
tufted angioma
tularaemia
Tungiasis
tunglang savage bellman syndrome
turcot syndrome
turner kieser syndrome
turner syndrome
tutuncuoglu syndrome
typhoid
tyrosine transaminase deficiency
tyrosine-oxidase temporary deficiency
tyrosinemia
tyrosinemia type 1
tyrosinemia type 2
Udd tibial myopathy
udp galactose-4-epimerase deficiency
uhl anomaly
ulbright hodes syndrome
ulerythema ophryogenesis
Ullrich disease
ulna and fibula absence of severe limb deficit
ulna hypoplasia
ulna hypoplasia mental retardation
ulna metaphyseal dysplasia syndrome
ulnar hypoplasia lobster claw deformity of feet
ulnar mammary syndrome
ulnar mammary syndrome of pallister
umbilical cord ulceration intestinal atresia
uncompable hair syndrome
uniparental disomy
uniparental disomy of 10
uniparental disomy of 11
uniparental disomy of 13
uniparental disomy of 14
uniparental disomy of 15
uniparental disomy of 16
uniparental disomy of 2

uniparental disomy of 21
uniparental disomy of 22
uniparental disomy of 5
uniparental disomy of 6
uniparental disomy of 7
uniparental disomy of 8
uniparental disomy of 9
unusual facies pectus carinatum joint laxity
upington disease
upper limb defect eye and ear abnormalities
upper limb mesomelic dysplasia
upton young syndrome
urachal cyst
urban rogers meyer syndrome
urban schosser spohn syndrome
urethral obstruction sequence
uridine monophosphate synthetase deficiency
urioste martinez frias syndrome
urioste martinez frias syndrome
urofacial syndrome
urogenital adysplasia
urophathy distal obstructive polydactyly
urticaria deafness amyloidosis
urticaria pigmentosa
ly
vagina absence of
vaginal atresia
vagneur triolle ripert syndrome
valproate syndrome
valproic acid antenatal infection
valvular dysplasia of the child
van allen myhre syndrome
van bervliet syndrome
van biervliet hendrickx van ertbruggen syndrome
van de berghe dequeker syndrome
van den bosch syndrome
van den ende brunner syndrome
van der woude syndrome
van goethem syndrome
van maldergem wetzburger verloes syndrome
van regemorter pierquin vamos syndrome
varadi papp syndrome
varicella virus antenatal infection
vas deferens absence
vas deferens congenital bilateral aplasia of
vascular disruption sequence
vascular facial pain
vascular malposition
vasquez hurst sotos syndrome
vater association
vein of galen aneurysm
velo cardio facial syndrome
velofacioskeletal syndrome
velopharyngeal incompetence
venencie powell winkelmann syndrome
ventricular extrasystoles perodactyly robin sequence
ventricular familial preexcitation syndrome
ventricular septal defect
ventriculo-arterial discordance, isolated
ventruto digirolamo festa syndrome
verloes bourguignon syndrome
verloes david syndrome
verloes gillerot fryns syndrome
verloes van maldergem marneffe syndrome
verloove vanhorick brubakk syndrome
Verneuil disease
verrucous nevus
verrucous nevus acanthokeratolytic
vertebral body fusion overgrowth

vertebral fusion posterior lumbosacral blepharoptosis
vestibulocochlear dysfunction progressive familial type
viljoen kallis voges syndrome
viljoen smart syndrome
viljoen winship syndrome
vipoma
viral hemorragic fever
virilizing ovarian tumor
visceral myopathy familiail external ophthalmoplegia
viscero-atrial heterotaxia
vitamin a embryopathy
vitamin b12 responsive methylmalonic acidemia
vitamin b12 responsive methylmalonic acidemia (cbl a)
vitamin b12 responsive methylmalonic acidemia (cbl b)
vitamin b12 responsive methylmalonic acidemia mutvitamin
b12 responsive methylmalonicaciduria
vitamin b12 responsive methylmalonicaciduria (cbl a)
vitamin b12 responsive methylmalonicaciduria (cbl b)
vitamin b12 responsive methylmalonicaciduria mutvitamin
d resistant rickets x linked
vitamin e familial isolated deficiency of
vitiligo
vitiligo mental retardation facial dysmorphism urethral duplication
vitiligo psychomotor retardation cleft palate facial dysmorphism
vitreoretinal degeneration
vitreoretinochoroidopathy dominant
vlcad deficiency
vocal cord dysfunction familial
vogt koyanagi harada disease
vohwinkel syndrome
volcke soekarman syndrome
von gierke disease
von hippel lindau disease
von recklinghausen disease
von voss cherstvoy syndrome
von willebrand disease dominant form
von willebrand disease, recessive form
vsr syndrome
w syndrome
waaler aarskog syndrome
waardenburg shah syndrome
waardenburg syndrome type 1
waardenburg syndrome type 2
waardenburg syndrome type 2a
waardenburg syndrome type 2b
waardenburg syndrome type 3
waardenburg syndrome type 4
waardenburg type pierpont
wagner disease
wagr syndrome
walbaum titran durieux crepin syndrome
walker dyson syndrome
walker-warburg syndrome
wallis cremin beighton syndrome
wallis zieff goldblatt syndrome
walt disney dwarfism
warburg sjo fledelius syndrome
warburg thomsen syndrome
warburton anyane yeboa syndrome
warfarin antenatal infection
warman mulliken hayward syndrome
warman mulliken syndrome
watson syndrome
weaver johnson syndrome
weaver like syndrome
weaver syndrome
weaver williams syndrome
weber cockayne type epidermolysis bullosa simplex
webster deming syndrome

wegener's granulomatosis
wegmann jones smith syndrome
weill marchesani syndrome
weinstein kliman scully syndrome
weismann netter syndrome
weissenbacher zweymuller syndrome
weleber hecht bigley syndrome
wellesley carmen french syndrome
wells jankovic syndrome
Wells syndrome
werdnig hoffman disease
werdnig hoffman type 1 bone fractures
wermer syndrome
werner syndrome
west syndrome
west syndrome x linked
westerhof beemer cormane syndrome
westphall disease
whipple disease
whitaker syndrome
white forelock with malformations
white matter hypoplasia corpus callosum agenesia mental retardation
whooping cough
whyte murphy syndrome
wieacker syndrome
wieacker wolff syndrome
wiedemann beckwith syndrome
wiedemann grosse dibbern syndrome
wiedemann oldigs oppermann syndrome
wiedemann opitz syndrome
wiedemann rautenstrauch syndrome
wildervanck syndrome
wilkes stevenson syndrome
wilkie taylor scambler syndrome
willebrand disease
willems de vries syndrome
willi prader syndrome
williams syndrome
wilms tumor
wilms tumor and pseudohermaphroditism
wilms tumor aniridia
wilms tumour radial bilateral aplasia
wilson disease
wilson turner syndrome
winchester disease
winkelman bethge pfeiffer syndrome
winship ocular albinism (type 1b)
winship viljoen leary syndrome
winter harding hyde syndrome
winter shortland temple syndrome
wisconsin syndrome
wiskott aldrich syndrome
witkop ocular albinism (type 2)
witkop syndrome
wl syndrome
wolcott rallison syndrome
wolf hirschhorn syndrome
wolff parkinson white syndrome
wolff zimmermann syndrome
wolfram syndrome
wolman disease
woodhouse sakati syndrome
woods black norbury syndrome
woods leversha rogers syndrome
woolly hair hypotrichosis everted lower lip outstanding ears
woolly hair palmoplantar keratoderma cardiac anomaly
Wooly hair naevus
wooly hair syndrome
worster drought syndrome

worth syndrome
wright dick syndrome
wrinkly skin syndrome
writer's cramp
wt limb blood syndrome
x linked hypogonadism gynecomastia mental retardation
x linked lymphoproliferative disease
x linked mental retardation craniofacial abn microcephaly club
x linked mental retardation de silva type
x linked mental retardation hamel type
x linked mental retardation hypotonia
x linked mental retardation short stature obesity
x linked mental retardation type brooks
x linked mental retardation type gu
x linked mental retardation type martinez
x linked mental retardation type raynaud
x linked mental retardation type schutz
x linked mental retardation type snyder
x linked mental retardation type wittner
x linked severe combined immunodeficiency disease
xanthic urolithiasis
xanthine oxydase deficiency
xanthinuria
xanthomatosis cerebrotendinous
xeroderma pigmentosum
xeroderma pigmentosum type 1
xeroderma pigmentosum type 2
xeroderma pigmentosum type 3
xeroderma pigmentosum type 5
xeroderma pigmentosum type 6
xeroderma pigmentosum type 7
xeroderma pigmentosum variant type
xeroderma talipes enamel defects
xk aprosencephaly
xy gonadal agenesis syndrome
xylitol dehydrogenase deficiency
yellow nail syndrome
yim ebbin syndrome
yolk sac tumor
yorifuji okuno syndrome
yoshimura-takeshita syndrome de
young harper syndrome
young hugues syndrome
young maders syndrome
young mc keever squier syndrome
young simpson syndrome
young syndrome
yunis varon syndrome
zadik barak levin syndrome
zap 70 deficiency
zazam sheriff phillips syndrome
zellweger syndrome
zerres rietschel majewski syndrome
zeta-associated-protein 70 deficiency
zimmer phocomelia
zimmer taub sova syndrome
zimmerman laband syndrome
zlotogora syndrome
zollinger ellison syndrome
zori stalker williams syndrome
zunich-kaye syndrome