Conditions associated with fatigue, some more relevant than others Pathological Conditions, Signs and Symptoms

- Morphological and Microscopic Findings
 - Adenocarcinoma in Situ
 - Atypical Squamous Cells of the Cervix
 - Squamous Intraepithelial Lesions of the Cervix
- Pathological Conditions, Anatomical
 - Accessory Atrioventricular Bundle
 - Airway Remodeling
 - Agenesis of Corpus Callosum
 - Absent corpus callosum cataract immunodeficiency
 - Acrocallosal Syndrome
 - Acrocallosal syndrome, Schinzel type
 - Aicardi Syndrome
 - X-linked infantile spasm syndrome
 - Ben Ari Shuper Mimouni syndrome
 - Calloso-genital dvsplasia
 - CAMFAK syndrome
 - Chudley-Mccullough syndrome
 - Combined Oxidative Phosphorylation Deficiency 2
 - Corpus callosum agenesis neuronopathy
 - Corpus Callosum, Agenesis of, with Facial Anomalies and Robin Sequence
 - Corpus Callosum, Agenesis of, with Mental Retardation, Ocular Coloboma, and Micrognathia
 - Corpus Callosum, Partial Agenesis of, X-Linked
 - Curatolo Cilio Pessagno syndrome
 - Donnai-Barrow syndrome
 - Duker Weiss Siber syndrome

- <u>Ectodermal Dysplasia</u>, <u>Hypohidrotic</u>, <u>with Hypothyroidism</u> and <u>Agenesis of the Corpus Callosum</u>
- Faye-Petersen Ward Carey syndrome
- Cyclopia
 - Dysgnathia complex
 - Genoa syndrome
 - Holoprosencephaly 10
 - Holoprosencephaly 2
 - Holoprosencephaly 3
 - Holoprosencephaly 4
 - Holoprosencephaly 5
 - Holoprosencephaly 6
 - Holoprosencephaly 7
 - Holoprosencephaly 8
 - Holoprosencephaly 9
 - Holoprosencephaly, Ectrodactyly, and Bilateral Cleft Lip-Palate
 - Holoprosencephaly, recurrent infections, and monocytosis
 - Holoprosencephaly with Fetal Akinesia-Hypokinesia Sequence
 - Lambotte syndrome
 - Microgastria limb reduction defect
 - Nonsyndromic Holoprosencephaly
 - Pseudotrisomy 13 syndrome
 - Steinfeld Syndrome
- Kozlowski Ouvrier syndrome
- Lissencephaly and agenesis of corpus callosum
- Median cleft lip, corpus callosum, lipoma, and skin polyps
- Microcephaly, corpus callosum dysgenesis and cleft lippalate

- Nakamura Osame syndrome
- Opitz-Kaveggia syndrome
- Osteochondrodysplasia, Rhizomelic, with Callosal Agenesis, Thrombocytopenia, Hydrocephalus, and Hypertension
- Partial agenesis of corpus callosum
- Proud Syndrome
- Recurrent spontaneous hypothermia with hypoplasia of the corpus callosum
- Saal Bulas syndrome
- Sakoda Complex
- Shapiro syndrome
- Short Stature, Mental Retardation, Callosal Agenesis, Heminasal Hypoplasia, Microphthalmia, And Atypical Clefting
- Stargardt Macular Degeneration Absent or Hypoplastic Corpus Callosum Mental Retardation and Dysmorphic Features
- Temtamy syndrome
- Thrombocytopenia Robin sequence
- Androgenetic alopecia
 - Alopecia, Androgenetic, 2
 - Alopecia, Androgenetic, 3
 - Alopecia areata
 - Alopecia Areata 1
 - Alopecia Areata 2
 - Diffuse alopecia
 - Alopecia congenita keratosis palmoplantaris
 - Alopecia contractures dwarfism mental retardation
 - Alopecia epilepsy oligophrenia syndrome of Moynahan
 - Alopecia, epilepsy, pyorrhea, mental subnormality

- Alopecia, Familial Focal
- Alopecia hypogonadism extrapyramidal disorder
- Alopecia-Mental Retardation Syndrome 1
- Alopecia-Mental Retardation Syndrome 2
- ALOPECIA-MENTAL RETARDATION SYNDROME 3
- Alopecia-Mental Retardation Syndrome with Convulsions and Hypergonadotropic Hypogonadism
- Alopecia, Neurologic Defects, and Endocrinopathy Syndrome
- Alopecia universalis
- Alopecia Universalis Congenita, XY Gonadal Dysgenesis, and Laryngomalacia
- Alopecia universalis onychodystrophy vitiligo
- Atrichia with Papular Lesions
- Bullous Dystrophy, Hereditary Macular Type
- Cataract, alopecia, sclerodactyly
- <u>Cerebral Autosomal Recessive Arteriopathy with</u>
 Subcortical Infarcts and Leukoencephalopathy
- Congenital alopecia X-linked
- Dermatopathia pigmentosa reticularis
- Ectodermal dysplasia alopecia preaxial polydactyly
- Ectodermal Dysplasia Syndrome with Distinctive Facial Appearance and Preaxial Polydactyly of Feet
- Follicular hamartoma alopecia cystic fibrosis
- FRONTONASAL DYSPLASIA 2
- Garret Tripp syndrome
- Glomerulonephritis sparse hair telangiectases
- Gomez Lopez Hernandez syndrome
- Growth retardation, Alopecia, Pseudoanodontia and Optic atrophy
- Hypergonadotropic Hypogonadism And Partial Alopecia

- <u>Ichthyosis follicularis atrichia photophobia syndrome</u>
- <u>Ichthyosis, Leukocyte Vacuoles, Alopecia, And Sclerosing</u>
 Cholangitis
- Jagell Holmgren Hofer syndrome
- Johnson neuroectodermal syndrome
- Keratosis Follicularis Spinulosa Decalvans, Autosomal Dominant
- Kuster Majewski Hammerstein syndrome
- Loose Anagen Hair Syndrome
 - Noonan-Like Syndrome With Loose Anagen Hair
 - NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR
- Macrocephaly, Alopecia, Cutis Laxa, and Scoliosis
- Microcephaly sparse hair mental retardation seizures
- Moloney syndrome
- Mucinosis, Follicular
- PARC syndrome
- Patel Bixler syndrome
- Perniola Krajewska Carnevale syndrome
- Satoyoshi syndrome
- Scholte syndrome
- Slti Salem syndrome
- <u>T-cell immunodeficiency, congenital alopecia and nail dystrophy</u>
- Thumb deformity, alopecia, pigmentation anomaly
- Thumb Deformity And Alopecia
- Total Hypotrichosis, Mari type
- Urban Schosser Spohn syndrome
- Woolly hair, hypotrichosis, everted lower lip and outstanding ears
- Woodhouse Sakati syndrome

- Atrial Remodeling
- Atrophy
 - MICROCEPHALY, POSTNATAL PROGRESSIVE, WITH SEIZURES AND BRAIN ATROPHY
 - Muscular Atrophy
 - Allan-Herndon-Dudley syndrome
 - Arnold Stickler Bourne syndrome
 - Facial Dysmorphism with Multiple Malformations
 - Furukawa Takagi Nakao syndrome
 - Lethal Congenital Contractural Syndrome 3
 - MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE
 - Miyoshi Muscular Dystrophy 2
 - Miyoshi Muscular Dystrophy 3
 - Miyoshi myopathy
 - Muscular Atrophy, Ataxia, Retinitis Pigmentosa, and Diabetes Mellitus
 - Muscular Atrophy, Malignant Neurogenic
 - Nathalie syndrome
 - Primrose syndrome
 - Sarcopenia
 - Thakker Donnai syndrome
 - Wieacker syndrome
- Blister
 - Hypotrichosis And Recurrent Skin Vesicles
 - Poikiloderma of Kindler
 - Pulmonary Bullae Causing Pneumothorax
- Calculi
 - Dental Calculus
 - Gallstones

- Cholestasis with Gallstone, Ataxia, and Visual Disturbance
- GALLBLADDER DISEASE 1
- Salivary Calculi
 - Salivary Duct Calculi
 - Salivary Gland Calculi
- Testicular Microlithiasis
- Urinary Calculi
 - Kidney stone
 - Gorlin Bushkell Jensen syndrome
 - <u>NEPHROLITHIASIS</u>, <u>URIC ACID</u>, SUSCEPTIBILITY TO
 - Renal hypouricemia
 - Ureteral Calculi
 - Urinary Bladder Calculi
- Choristoma
 - Choroidal Osteoma, Bilateral
 - Dermoids of cornea
- Stricture
 - Calabro syndrome
 - Celiac Artery Stenosis from Compression by Median Arcuate Ligament of Diaphragm
 - Infundibulopelvic dysgenesis
 - <u>Laryngotracheal Stenosis</u>, <u>Progressive</u>, <u>with Short Stature</u> <u>and Arthropathy</u>
 - Lumbar Stenosis, Familial
 - Pseudoainhum
 - Tracheal agenesis
 - Tracheobronchial Stenosis, Congenital
 - Urethral obstruction sequence

- Cysts
 - Aplasia Cutis Congenita, Congenital Heart Defect, And Frontonasal Cysts
 - Arachnoid Cysts
 - Bobble-head doll syndrome
 - Chudley-Mccullough syndrome
 - PACHYGYRIA WITH MENTAL RETARDATION, SEIZURES, AND ARACHNOID CYSTS
 - Spinal intradural arachnoid cysts
 - Tibia, Absence or Hypoplasia of, with Polydactyly, Retrocerebellar Arachnoid Cyst, and Other Anomalies
 - Tibia absent polydactyly arachnoid cyst
 - Bone Cysts
 - Bone Cysts, Aneurysmal
 - Jaw Cysts
 - Nonodontogenic Cysts
 - Odontogenic Cysts
 - Basal Cell Nevus Syndrome
 - Aloi Tomasini Isaia syndrome
 - Dentigerous Cyst
 - Odontogenic Cyst, Calcifying
 - Periodontal Cyst
 - Radicular Cyst
 - Polycystic bone disease
 - Branchioma
 - Breast Cyst
 - Galactocele
 - Bronchogenic Cyst
 - Chalazion

- Choledochal Cyst
 - Caroli Disease
 - Caroli disease isolated
- Colloid Cysts
 - Colloid cysts of third ventricle
- Cystic medial necrosis of aorta
- Dermoid Cyst
 - 46,XX Gonadal dysgenesis epibulbar dermoid
 - Aplasia Cutis Congenita with Epibulbar Dermoids
 - Dermoid Cysts, Familial Frontonasal
 - Ring dermoid of cornea
 - Zadik Barak Levin syndrome
- Ectodermal dysplasia adrenal cyst
- Epidermal Cyst
 - FLOTCH syndrome
 - Follicular Atrophoderma, Perioral Pigmented, with Milia and Epidermoid Cysts
 - Trichilemmal Cyst 1
- Esophageal Cyst
- Follicular Cyst
 - Trichofolliculoma
- Ganglion Cysts
- Leukoencephalopathy, Cystic, Without Megalencephaly
- Lymphocele
 - Campomelia Cumming type
- Mediastinal Cyst
- Megalencephalic leukoencephalopathy with subcortical cysts
- Mesenteric Cyst
- Microphthalmia associated with colobomatous cyst

- Mucocele
- Ovarian Cysts
 - Fraser-Like Syndrome
 - Neonatal ovarian cyst
 - Polycystic ovary syndrome
 - Fowler Christmas Chapple syndrome
 - Polycystic Ovarian Disease due to 17-Ketosteroid Reductase Deficiency
- Pancreatic Cyst
 - Pancreatic Pseudocyst
 - Renal hepatic pancreatic dysplasia Dandy Walker cyst
- Parovarian Cyst
- Pilonidal Sinus
- Polycystic liver disease
- Ranula
- Sener syndrome
- Spermatocele
- Synovial Cyst
 - Popliteal Cyst
- Tarlov Cysts
- Thyroglossal Cyst
 - Thyroglossal tract cyst
- Urachal Cyst
 - Benign non-infected urachal cyst
- Van der Woude syndrome
 - VAN DER WOUDE SYNDROME 1, MODIFIER OF
- Dilatation, Pathologic
- Diverticulum
 - Bladder Diverticulum

- Diverticulosis, Small Intestinal
- Diverticulum, Colon
- Diverticulum, Esophageal
 - Zenker Diverticulum
- Diverticulum, Stomach
- Meckel Diverticulum
- Facial Asymmetry
 - Facial Hemihypertrophy
 - Fronto-facio-nasal dysplasia
 - Hemifacial Hyperplasia With Strabismus
 - Hemifacial myohyperplasia
 - Mehes syndrome
 - Oculodentoosseous dysplasia recessive
 - Opitz Reynolds Fitzgerald syndrome
- Fistula
 - Cutaneous Fistula
 - Digestive System Fistula
 - Biliary Fistula
 - Esophageal Fistula
 - Tracheoesophageal Fistula
 - Esophageal atresia with or without tracheoesophageal fistula
 - Martinez-Frias Syndrome
 - Oculodigitoesophagoduodenal syndrome
 - Vater Association With Hydrocephalus
 - Gastric Fistula
 - Intestinal Fistula
 - Rectal Fistula
 - Rectovaginal Fistula

- Pancreatic Fistula
- Lachiewicz Sibley syndrome
- Oral Fistula
 - Dental Fistula
 - Oroantral Fistula
 - Salivary Gland Fistula
- Respiratory Tract Fistula
 - Bronchial Fistula
 - Tracheoesophageal Fistula
 - Esophageal atresia with or without tracheoesophageal fistula
 - Martinez-Frias Syndrome
 - Oculodigitoesophagoduodenal syndrome
 - Vater Association With Hydrocephalus
- Urinary Fistula
 - Urinary Bladder Fistula
 - Vesicovaginal Fistula
- Vaginal Fistula
 - Rectovaginal Fistula
 - Vesicovaginal Fistula
- Vascular Fistula
 - Arterio-Arterial Fistula
 - Aorta-pulmonary artery fistula
 - Bland White Garland Syndrome
 - Arteriovenous Fistula
 - Angiokeratoma Corporis Diffusum with Arteriovenous Fistulas
 - Carotid-Cavernous Sinus Fistula
 - Pulmonary Arteriovenous Fistulas
 - Wyburn Mason's syndrome

- Zuska's Disease
- Hernia
 - Acromegaloid features, overgrowth, cleft palate, and hernia
 - Encephalocele
 - Cerebellar Vermis Aplasia with Associated Features suggesting Smith-Lemli-Opitz Syndrome and Meckel Syndrome
 - Cerebrorenodigital Syndrome with Limb Malformations and Triradiate Acetabula
 - <u>Dandy-Walker Malformation With Occipital</u>
 <u>Cephalocele</u>, Autosomal Dominant
 - DK Phocomelia Syndrome
 - Fronto-facio-nasal dysplasia
 - Knobloch syndrome
 - Knobloch Syndrome Type II
 - Knobloch Syndrome Type III
 - <u>Laryngeal Atresia</u>, <u>Encephalocele</u>, <u>and Limb</u>
 Deformities
 - Meckel-Like Cerebrorenodigital Syndrome
 - Meckel syndrome type 1
 - Meckel syndrome type 2
 - Meckel syndrome type 3
 - Meckel Syndrome, Type 4
 - Meckel Syndrome, Type 5
 - Meckel Syndrome, Type 6
 - Parietal Foramina
 - Parietal Foramina 1
 - Parietal Foramina 2
 - Parietal Foramina 3
 - Parietal Foramina With Cleidocranial Dysplasia

- Podder-Tolmie syndrome
- Sakoda Complex
- Zechi-Ceide Syndrome
- Hernia, Abdominal
 - <u>Diverticulosis of Bowel, Hernia, and Retinal</u>
 <u>Detachment</u>
 - Gastroschisis
 - Paraomphalocele
 - Hernia, Femoral
 - Hernia, Inguinal
 - Arthrogryposis multiplex with deafness, inguinal hernias, and early death
 - Hernia, Double Inguinal
 - Lambert syndrome
 - Microspherophakia with hernia
 - Hernia, Ventral
 - Hernia, Umbilical
 - AXENFELD-RIEGER SYNDROME, TYPE 1
 - Omphalocele cleft palate syndrome lethal
 - Omphalocele, Diaphragmatic Hernia,
 And Radial Ray Defects
 - Omphalocele exstrophy imperforate anus
 - Pseudodiastrophic dysplasia
 - Shprintzen omphalocele syndrome
- Hernia, Diaphragmatic
 - Diaphragmatic Hernia 1
 - Diaphragmatic Hernia 2
 - Diaphragmatic Hernia 3

- Diaphragm, Complete Agenesis Of
- Fryns syndrome
- Hernia, Diaphragmatic, Traumatic
- Hernia, Hiatal
 - Galloway Mowat syndrome
 - Gastric Volvulus, Intrathoracic
 - Sucrosuria, Hiatus Hernia and Mental Retardation
- Hernias, Diaphragmatic, Congenital
 - Donnai-Barrow syndrome
 - Epidermolysis Bullosa With Diaphragmatic Hernia
 - Hernia, Anterior Diaphragmatic
 - Kennerknecht Sorgo Oberhoffer syndrome
 - Nasopharyngeal teratoma with Dandy Walker diaphragmatic hernia
 - Omphalocele, Diaphragmatic Hernia, And Radial Ray Defects
 - Saal Bulas syndrome
- Hernia, Obturator
- Incisional Hernia
- Intervertebral Disc Displacement
 - Intervertebral disc disease
 - Spastic paraplegia 25, autosomal recessive
- Megarbane syndrome
- Meningocele
 - <u>Cerebellar Vermis Aplasia with Associated Features</u> suggesting Smith-Lemli-Opitz Syndrome and Meckel Syndrome
 - Lateral meningocele syndrome
 - Sacral Agenesis Syndrome

- Sacral defect and anterior sacral meningocele
- Sakoda Complex
- Rectocele
- Hypertrophy
 - Cardiac hypertrophy
 - Cantu syndrome
 - Dilated cardiomyopathy
 - 3-Methylglutaconic Aciduria, Type V
 - Cardiomyopathy, Dilated, 1z
 - Cardiomyopathy, Dilated, 2a
 - Cardiomyopathy, Dilated, 1CC
 - Cardiomyopathy, Dilated, 1AA
 - Cardiomyopathy, Dilated, 1BB
 - Cardiomyopathy, Dilated, 1C
 - Cardiomyopathy, Dilated, 1D
 - Cardiomyopathy, Dilated, 1DD
 - Cardiomyopathy, Dilated, 1E
 - Cardiomyopathy, Dilated, 1EE
 - Cardiomyopathy, Dilated, 1FF
 - Cardiomyopathy, Dilated, 1g
 - Cardiomyopathy, Dilated, 1i
 - Cardiomyopathy, Dilated, 1J
 - Cardiomyopathy, Dilated, 1K
 - Cardiomyopathy, Dilated, 11
 - Cardiomyopathy, Dilated, 1M
 - Cardiomyopathy, Dilated, 1N
 - Cardiomyopathy, Dilated, 1o
 - Cardiomyopathy, Dilated, 1p
 - Cardiomyopathy, Dilated, 1q

- Cardiomyopathy, Dilated, 1s
- Cardiomyopathy, Dilated, 1t
- Cardiomyopathy, Dilated, 1u
- Cardiomyopathy, Dilated, 1V
- Cardiomyopathy, Dilated, 1w
- Cardiomyopathy, Dilated, 1x
- Cardiomyopathy, Dilated, 1y
- Cardiomyopathy, Dilated, 3A
- Cardiomyopathy, Dilated, with Left Ventricular Noncompaction
- Cardiomyopathy, Right Ventricular Dilated
- Dmd-Associated Dilated Cardiomyopathy
- Familial dilated cardiomyopathy
- Idiopathic dilation cardiomyopathy
- Krasnow Qazi syndrome
- Malouf syndrome
- Uhl anomaly
- Winship Viljoen Leary syndrome
- Ethanolaminosis
- GELEOPHYSIC DYSPLASIA 1
- Left ventricular hypertrophy
- Hypertrophy, Right Ventricular
- Gigantomastia
- Hepatomegaly
 - Cryohydrocytosis, Stomatin-Deficient, with Mental Retardation, Seizures, Cataracts, and Massive Hepatosplenomegaly
 - Dykes Markes Harper syndrome
 - Refsum disease with increased pipecolic acidemia
 - Tang Hsi Ryu syndrome

- Hypertrophia Musculorum Vera
- Kocher-Debre-Semelaigne syndrome
- Masticatory Muscles, Hypertrophy of
- Myostatin-related muscle hypertrophy
- Splenomegaly
 - Banti's syndrome
 - Cryohydrocytosis, Stomatin-Deficient, with Mental Retardation, Seizures, Cataracts, and Massive Hepatosplenomegaly
 - Dykes Markes Harper syndrome
 - Myeloid splenomegaly
 - Splenomegaly Syndrome With Splenic Germinal Center Hypoplasia And Reduced Circulating T-Helper Cells
 - Tang Hsi Ryu syndrome
- Prolapse
 - Pelvic Organ Prolapse
 - Cystocele
 - Rectal Prolapse
 - Uterine Prolapse
 - Visceral Prolapse
- Leg Length Inequality
- Leukoplakia
 - Leukoplakia, Oral
 - Leukoplakia, Hairy
- Nails, Malformed
 - Alopecia universalis onychodystrophy vitiligo
 - Ameloonychohypohidrotic syndrome
 - Anonychia congenita
 - Anonychia-Ectrodactyly

- Anonychia onychodystrophy
- Anonychia-onychodystrophy with brachydactyly type B and ectrodactyly
- Anonychia, Total, with Microcephaly
- Anonychia with Flexural Pigmentation
- Basan syndrome
- Brachymorphism-onychodysplasia-dysphalangism syndrome
- Cartwright Nelson Fryns syndrome
- Curly hair-acral keratoderma-caries syndrome
- Curly hair-ankyloblepharon-nail dysplasia syndrome
- Deafness, Congenital, and Onychodystrophy, Autosomal Dominant
- Deafness enamel hypoplasia nail defects
- Dermoodontodysplasia
- Digitorenocerebral Syndrome
- Double Nail for Fifth Toe
- Gorlin Bushkell Jensen syndrome
- Hereditary koilonychia
- Hypospadias-Mental Retardation Syndrome
- MAMMARY-DIGITAL-NAIL SYNDROME
- Otoonychoperoneal Syndrome
- NAIL DISORDER, NONSYNDROMIC CONGENITAL, 9
- Oculotrichodysplasia
- Onycholysis, Partial, with Scleronychia
- Onychotrichodysplasia and neutropenia
- Pili torti onychodysplasia
- Pinheiro Freire-Maia Miranda syndrome
- Propping Zerres syndrome
- Santos Syndrome

- Schinzel-Giedion syndrome
- Steatocystoma multiplex with natal teeth
- Teebi Kaurah syndrome
- Temple-Baraitser Syndrome
- Toenail Dystrophy, Isolated
- Tonoki syndrome
- Twenty-Nail Dystrophy
- Ulna hypoplasia with mental retardation
- Ulnar Hypoplasia with Mental Retardation
- Witkop syndrome
- Yellow Nail Syndrome
 - Lymphedema of the lower extremities, recurrent pneumonia, bronchiectasis, and yellowed nails
- Zori Stalker Williams syndrome
- Plaque, Amyloid
 - <u>Dementia-Parkinsonism With Non-Alzheimer Amyloid</u>
 <u>Plaques</u>
- Plaque, Atherosclerotic
- Polyps
 - Intestinal Polyps
 - Colonic Polyps
 - Nasal Polyps
 - Asthma and Nasal Polyps
 - Asthma, Nasal Polyps, And Aspirin Intolerance
 - Median cleft lip, corpus callosum, lipoma, and skin polyps
 - Polyposis Of Gastric Fundus Without Polyposis Coli
- Rupture, Spontaneous
 - Bone Fragility with Contractures, Arterial Rupture, and Deafness

- Torsion Abnormality
 - Bone Anteversion
 - Coxa Vara
 - Jacobs syndrome
 - Patella aplasia, coxa vara, tarsal synostosis
 - Bone Retroversion
 - Coxa Valga
 - Dystonia 13, Torsion
 - Intestinal Volvulus
 - McPherson Clemens syndrome
 - Volvulus Of Midgut
 - Tibial Torsion, Bilateral Medial
 - Uterine Retroversion
- Vascular Remodeling
- Ventricular Remodeling
- Pathologic Processes
 - Acantholysis
 - Grover's disease
 - Cardiac arrhythmia
 - Arrhythmia, Sinus
 - Sick Sinus Syndrome
 - Sick Sinus Syndrome 1, Autosomal Recessive
 - Sick Sinus Syndrome 2, Autosomal Dominant
 - Sinus Node Disease and Myopia
 - Sinus Arrest, Cardiac
 - Atrial fibrillation
 - Atrial Fibrillation, Familial, 8
 - Atrial fibrillation, familial 1

- Atrial Fibrillation, Familial, 2
- Atrial Fibrillation, Familial, 3
- Atrial Fibrillation, Familial, 4
- Atrial Fibrillation, Familial, 5
- Atrial Fibrillation, Familial, 6
- Atrial Fibrillation, Familial, 7
- Atrial Flutter
- Atrial Septal Defect, Secundum, with Various Cardiac and Noncardiac Defects
- Bradycardia
- Brugada syndrome
 - Brugada Syndrome 6
 - Brugada Syndrome 7
 - Brugada Syndrome 2
 - Brugada Syndrome 3
 - Brugada Syndrome 4
 - Brugada Syndrome 5
 - Brugada Syndrome 8
 - Cardiac Conduction Defect
 - Cardiac Conduction Defect, Nonspecific
 - Sudden unexpected nocturnal death syndrome
- Cardiac Arrhythmia, Ankyrin-B-Related
- Cardiac Complexes, Premature
 - Atrial Premature Complexes
 - <u>Extrasystoles</u>, <u>Multiform Ventricular</u>, <u>with Short</u>
 Stature, Hyperpigmentation and Microcephaly
 - Ventricular Premature Complexes
 - Stoll Alembik Dott syndrome
- Cardiac Conduction Defect, Nonprogressive
- Commotio Cordis

- Heart Block
 - Adams-Stokes Syndrome
 - Atrial Septal Defect with Atrioventricular Conduction Defects
 - ATRIAL SEPTAL DEFECT 7 WITH OR WITHOUT ATRIOVENTRICULAR CONDUCTION DEFECTS
 - Atrial Standstill
 - Atrioventricular Block
 - ATRIAL SEPTAL DEFECT 7 WITH OR WITHOUT ATRIOVENTRICULAR CONDUCTION DEFECTS
 - Bundle-Branch Block
 - Bundle Branch Block, Familial Isolated Complete Right
 - Congenital heart block
 - PROGRESSIVE FAMILIAL HEART BLOCK, TYPE IA
 - Heart Block, Nonprogressive
 - Hereditary bundle branch system defect
 - Progressive Familial Heart Block, Type Ib
 - Progressive Familial Heart Block, Type II
 - Sick Sinus Syndrome
 - Sick Sinus Syndrome 1, Autosomal Recessive
 - Sick Sinus Syndrome 2, Autosomal Dominant
 - Sinus Node Disease and Myopia
 - Sinoatrial Block
 - Mental retardation, keratoconus, febrile seizures, and sinoatrial block
- Long QT Syndrome

- Andersen Syndrome
- Jervell-Lange Nielsen Syndrome
 - Jervell And Lange-Nielsen Syndrome 2
- Long QT syndrome type 3
- Long Qt Syndrome 10
- Long Qt Syndrome 11
- Long Qt Syndrome 12
- LONG QT SYNDROME 13
- Long Qt Syndrome 2
- Long Qt Syndrome 2-3
- Long Qt Syndrome 2-5
- Long Qt Syndrome 3
- Long Qt Syndrome 3-6
- Long Qt Syndrome 4
- Long Qt Syndrome 5
- Long Qt Syndrome 6
- Long Qt Syndrome 9
- Romano-Ward Syndrome
 - Long Qt Syndrome 1-2
- Timothy syndrome
- Parasystole
- Pre-Excitation Syndromes
 - Lown-Ganong-Levine Syndrome
 - Pre-Excitation, Mahaim-Type
 - Wolff-Parkinson-White Syndrome
- QT INTERVAL, VARIATION IN
- Short Qt Syndrome
- Short QT Syndrome 1
- Short QT Syndrome 2

- Short QT Syndrome 3
- Simpson-Golabi-Behmel syndrome
- Tachycardia
 - Atrial Tachyarrhythmia with Short PR Interval
 - Bidirectional tachycardia
 - <u>Tachycardia</u>, <u>Hypertension</u>, <u>Microphthalmia</u>, <u>And</u> <u>Hyperglycinuria</u>
 - Tachycardia, Paroxysmal
 - Adams Nance syndrome
 - Tachycardia, Reciprocating
 - Tachycardia, Atrioventricular Nodal Reentry
 - Tachycardia, Sinoatrial Nodal Reentry
 - Tachycardia, Supraventricular
 - Tachycardia, Ectopic Atrial
 - Tachycardia, Ectopic Junctional
 - Tachycardia, Sinus
 - Tachycardia, Ventricular
 - Accelerated Idioventricular Rhythm
 - Polymorphic catecholergic ventricular tachycardia
 - VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC
 POLYMORPHIC, 1, WITH OR
 WITHOUT ATRIAL DYSFUNCTION
 AND/OR DILATED
 CARDIOMYOPATHY
 - VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC, 2
 - VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC, 3
 - Torsades de pointes

VENTRICULAR TACHYCARDIA, FAMILIAL

- Ventricular Fibrillation
 - Paroxysmal ventricular fibrillation
 - Ventricular Fibrillation, Paroxysmal Familial, 1
 - Ventricular Fibrillation, Paroxysmal Familial, 2
- Ventricular Flutter
- Woodhouse Sakati syndrome
- Ascites
- Atrial Remodeling
- Azotemia
- Cardiotoxicity
- Channelopathies
 - Indifference to Pain, Congenital, Autosomal Recessive
- Chromosome Aberrations
 - Abnormal Karyotype
 - XYY Karyotype
 - 47, XYY syndrome
 - Aneuploidy
 - 49,XXXXX syndrome
 - Chromosome 15q, tetrasomy
 - Chromosome 18, tetrasomy 18p
 - Chromosome 21, tetrasomy 21q
 - Chromosome 9, duplication 9g21
 - Chromosome 9, tetrasomy 9p
 - Monosomy
 - Chromosome 18 mosaic monosomy
 - Chromosome 21 monosomy
 - Chromosome 8 deletion

- CHROMOSOME 8q21.11 DELETION SYNDROME
- Chromosome Deletion
 - 10p Deletion Syndrome (Partial)
 - 13q deletion syndrome
 - CHROMOSOME 13q14 DELETION SYNDROME
 - 15q24 Microdeletion
 - 16p11.2 Deletion Syndrome
 - 7p2 monosomy syndrome
 - Chromosome 1, monosomy 1p32
 - Chromosome 1, monosomy 1p34 p32
 - Chromosome 1, monosomy 1q25 q32
 - Chromosome 1, monosomy 1q32 q42
 - Chromosome 6, monosomy 6q2
 - Chromosome 1, monosomy 1q4
 - Chromosome 3, monosomy 3q21 23
 - Chromosome 3, monosomy 3q27
 - Chromosome 17, deletion 17q23 q24
 - Chromosome 6, deletion 6q13 q15
 - Chromosome 1p36 Deletion Syndrome
 - Chromosome 1, monosomy 1p22 p13
 - Chromosome 1, monosomy 1p31 p22
 - Chromosome 6, monosomy 6p23
 - Chromosome 6, monosomy 6q
 - Chromosome 10, monosomy 10q
 - Chromosome 10q23 Deletion Syndrome
 - Chromosome 10q26 Deletion Syndrome

- Chromosome 11, deletion 11p
- Chromosome 11p Deletion Syndrome
- Chromosome 11p, partial deletion
- Chromosome 11q partial deletion
- Chromosome 12p deletion
- Chromosome 12p partial deletion
- Chromosome 14q, partial deletions
- Chromosome 14q, terminal deletion
- Chromosome 15q13.3 Microdeletion Syndrome
- Chromosome 15q, partial deletion
- Chromosome 17 deletion
 - CHROMOSOME 17p13.1 DELETION SYNDROME
- Chromosome 17q21.31 Deletion Syndrome
- Chromosome 18, deletion 18q23
- Chromosome 18 deletion syndrome
- Chromosome 18p deletion syndrome
- Chromosome 19q13.11 Deletion Syndrome
- Chromosome 1, deletion g21 g25
- Chromosome 1, monosomy 1p
- Chromosome 1q21.1 Deletion Syndrome, 1.35-Mb
- Chromosome 1q21.1 Duplication Syndrome
- Chromosome 1q43-Q44 Deletion Syndrome
- Chromosome 20, deletion 20p
- Chromosome 21, monosomy 21q22

- Chromosome 22, microdeletion 22 q11
- Chromosome 22q11.2 Deletion Syndrome, Distal
- Chromosome 6, monosomy 6q1
- Chromosome 2, monosomy 2p22
- Chromosome 2, monosomy 2pter p24
- Chromosome 2, monosomy 2q
- Chromosome 2, monosomy 2q24
- Chromosome 2p16.1-P15 Deletion Syndrome
- Chromosome 2q31.2 Deletion Syndrome
- Chromosome 2q32-Q33 Deletion Syndrome
- Chromosome 2g37 deletion syndrome
- Chromosome 3, monosomy 3p
- Chromosome 3, monosomy 3p14 p11
- Chromosome 3, monosomy 3p2
- Chromosome 3, monosomy 3p25
- Chromosome 3, monosomy 3q13
- CHROMOSOME 3pter-p25 DELETION SYNDROME
- Chromosome 3q29 Deletion Syndrome
- Chromosome 4, 4q Terminal Deletion Syndrome
- Chromosome 4, monosomy 4p14 p16
- Chromosome 4, monosomy 4q32
- Chromosome 4q- Syndrome
- Chromosome 4 short arm deletion
- Chromosome 5g Deletion Syndrome

- Chromosome 6pter-P24 Deletion Syndrome
- Chromosome 7, monosomy
- Chromosome 7, monosomy 7q2
- Chromosome 7, monosomy 7q21
- Chromosome 7, monosomy 7q3
- CHROMOSOME 7q11.23 DELETION SYNDROME, DISTAL, 1.2-MB
- Chromosome 8, monosomy 8p
- Chromosome 8, monosomy 8p23 1
- Chromosome 8, monosomy 8q
- Chromosome 8p deletion syndrome (partial)
- CHROMOSOME 8q21.11 DELETION SYNDROME
- Chromosome 9, partial monosomy 9p
- Chromosome 9p Deletion Syndrome
- <u>Deafness, Sensorineural, And Male</u> <u>Infertility</u>
- Deletion 13q syndrome, partial
- Deletion 6q16 q21
- Fragile Site 16p12
- Holoprosencephaly 10
- Homozygous 11p15-p14 Deletion Syndrome
- Hypertrichosis Terminalis,
 Generalized, with or without Gingival
 Hyperplasia
- Hypotonia-Cystinuria Syndrome
- Kleefstra Syndrome
- Male sterility due to Y-chromosome deletions

- Monosomy 7 of Bone Marrow
- NF1 Microdeletion Syndrome
- Otodental Dysplasia
- EXOSTOSES, MULTIPLE, TYPE II
- Prader-Willi-Like Syndrome
 Associated With Chromosome 6
- <u>Telomeric 22q13 Monosomy</u>
 <u>Syndrome</u>
- X chromosome, monosomy Xp22 pter
- X chromosome, monosomy Xq28
- Schmid-Fraccaro syndrome
- Tetrasomy
- Trisomy
 - 6q+ Syndrome, Partial
 - Chromosome 12, trisomy 12q
 - Chromosome 13p duplication
 - Chromosome 8, trisomy
 - Chromosome 10q duplication syndrome
 - Chromosome 10, trisomy 10p
 - Chromosome 10, trisomy 10pter p13
 - Chromosome 10, uniparental disomy of
 - Chromosome 11, partial trisomy 11q
 - Chromosome 11g trisomy
 - Chromosome 12, 12p trisomy
 - Chromosome 13q trisomy
 - Chromosome 14q, proximal duplication
 - Chromosome 14q, terminal duplication
 - Chromosome 14 trisomy
 - Chromosome 14, trisomy mosaic
 - Chromosome 15q, trisomy

- Chromosome 15, trisomy mosaicism
- Chromosome 16, trisomy
- Chromosome 16, trisomy 16q
- Chromosome 17 trisomy
- Chromosome 17, trisomy 17p
- Chromosome 17, trisomy 17p11 2
- Chromosome 17, trisomy 17q22
- Chromosome 18, trisomy 18p
- Chromosome 18, trisomy 18q
- Chromosome 19, trisomy 19q
- Chromosome 1, duplication 1p21 p32
- Chromosome 1, q42 11 q42 12 duplication
- Chromosome 1q, duplication 1q12 q21
- Chromosome 1, trisomy 1q32 qter
- Chromosome 1, trisomy 1q42 qter
- Chromosome 1, uniparental disomy 1q12 q21
- Chromosome 20, trisomy
- Chromosome 21, uniparental disomy of
- Chromosome 5, uniparental disomy
- Chromosome 22, trisomy
- Chromosome 22, trisomy q11 q13
- Chromosome 2, trisomy 2p
- Chromosome 2, trisomy 2p13 p21
- Chromosome 2, trisomy 2pter p24
- Chromosome 2, trisomy 2q
- Chromosome 2, trisomy 2q37
- Chromosome 3 duplication syndrome
- Chromosome 3, trisomy 3p

- Chromosome 3, trisomy 3p25
- Chromosome 3, trisomy 3q
- Chromosome 3, trisomy 3q13 2 q25
- Chromosome 4, partial trisomy distal 4q
- Chromosome 4, trisomy 4q
- Chromosome 4, trisomy 4q21
- Chromosome 4, trisomy 4q25 qter
- Chromosome 5, monosomy 5q35
- Chromosome 5, trisomy 5pter p13 3
- Chromosome 5, trisomy 5q
- Chromosome 6, trisomy 6p
- Chromosome 6, trisomy 6q
- Chromosome 7, trisomy 7p
- Chromosome 7, trisomy 7p13 p12 2
- Chromosome 7, trisomy 7q
- Chromosome 7, trisomy mosaic
- Chromosome 8, mosaic trisomy
- Chromosome 8, partial trisomy
- Chromosome 8, trisomy 8p
- Chromosome 8, trisomy 8q
- Chromosome 9, partial trisomy 9p
- Chromosome 9, trisomy
- Chromosome 9, trisomy 9p
- Chromosome 9, trisomy 9q
- Chromosome 9, trisomy 9q32
- Chromosome 9, trisomy mosaic
- Chromosome Xq duplication syndrome
- Distal Trisomy 10g Syndrome
- Duplication 4p Syndrome

- Partial Duplication 15q Syndrome
- Partial Trisomy 3q Syndrome
- Pseudotrisomy 13 syndrome
- Triple X syndrome
- Trisomy 13 syndrome
- Trisomy 18
- Trisomy 18-Like Syndrome
- Trisomy 20p
- Trisomy 22 mosaicism syndrome
- Warburton Anyane Yeboa syndrome
- X chromosome, duplication Xq13 1 q21 1
- X chromosome, trisomy Xp3
- X chromosome, trisomy Xpter Xq13
- X chromosome, trisomy Xq25

Chromosomal Instability

- Chromosomal Instability with Tissue-Specific Radiosensitivity
- Chromosome Fragility
- Radiation Sensitivity Chromosome Instability
 Syndrome, Autosomal Dominant

Chromosome Breakage

- Craniosynostosis Microcephaly with Chromosomal Breakage and Other Abnormalities
- THROMBOCYTOPENIA 2
- WARSAW BREAKAGE SYNDROME
- Chromosome Duplication
 - Chromosome 17p13.3 Duplication Syndrome
 - Chromosome 22q11.2 Microduplication Syndrome
 - Chromosome 3q29 Duplication Syndrome
 - Chromosome 5p13 Duplication Syndrome

- Chromosome Xp11.23-P11.22 Duplication Syndrome
- Chromosome Xq28 Duplication Syndrome
- NF1 Microduplication Syndrome
- Potocki-Lupski syndrome
- Tetrasomy
- Trisomy
 - 6q+ Syndrome, Partial
 - Chromosome 12, trisomy 12q
 - Chromosome 13p duplication
 - Chromosome 8, trisomy
 - Chromosome 10q duplication syndrome
 - Chromosome 10, trisomy 10p
 - Chromosome 10, trisomy 10pter p13
 - Chromosome 10, uniparental disomy of
 - Chromosome 11, partial trisomy 11q
 - Chromosome 11g trisomy
 - Chromosome 12, 12p trisomy
 - Chromosome 13q trisomy
 - Chromosome 14q, proximal duplication
 - Chromosome 14q, terminal duplication
 - Chromosome 14 trisomy
 - Chromosome 14, trisomy mosaic
 - Chromosome 15q, trisomy
 - Chromosome 15, trisomy mosaicism
 - Chromosome 16, trisomy
 - Chromosome 16, trisomy 16q
 - Chromosome 17 trisomy
 - Chromosome 17, trisomy 17p

- Chromosome 17, trisomy 17p11 2
- Chromosome 17, trisomy 17q22
- Chromosome 18, trisomy 18p
- Chromosome 18, trisomy 18q
- Chromosome 19, trisomy 19q
- Chromosome 1, duplication 1p21 p32
- Chromosome 1, q42 11 q42 12 duplication
- Chromosome 1q, duplication 1q12 q21
- Chromosome 1, trisomy 1q32 qter
- Chromosome 1, trisomy 1q42 qter
- Chromosome 1, uniparental disomy 1q12 q21
- Chromosome 20, trisomy
- Chromosome 21, uniparental disomy of
- Chromosome 5, uniparental disomy
- Chromosome 22, trisomy
- Chromosome 22, trisomy g11 g13
- Chromosome 2, trisomy 2p
- Chromosome 2, trisomy 2p13 p21
- Chromosome 2, trisomy 2pter p24
- Chromosome 2, trisomy 2q
- Chromosome 2, trisomy 2q37
- Chromosome 3 duplication syndrome
- Chromosome 3, trisomy 3p
- Chromosome 3, trisomy 3p25
- Chromosome 3, trisomy 3q
- Chromosome 3, trisomy 3q13 2 q25
- Chromosome 4, partial trisomy distal 4q
- Chromosome 4, trisomy 4q

- Chromosome 4, trisomy 4q21
- Chromosome 4, trisomy 4q25 qter
- Chromosome 5, monosomy 5q35
- Chromosome 5, trisomy 5pter p13 3
- Chromosome 5, trisomy 5q
- Chromosome 6, trisomy 6p
- Chromosome 6, trisomy 6q
- Chromosome 7, trisomy 7p
- Chromosome 7, trisomy 7p13 p12 2
- Chromosome 7, trisomy 7q
- Chromosome 7, trisomy mosaic
- Chromosome 8, mosaic trisomy
- Chromosome 8, partial trisomy
- Chromosome 8, trisomy 8p
- Chromosome 8, trisomy 8q
- Chromosome 9, partial trisomy 9p
- Chromosome 9, trisomy
- Chromosome 9, trisomy 9p
- Chromosome 9, trisomy 9q
- Chromosome 9, trisomy 9q32
- Chromosome 9, trisomy mosaic
- Chromosome Xq duplication syndrome
- Distal Trisomy 10q Syndrome
- Duplication 4p Syndrome
- Partial Duplication 15g Syndrome
- Partial Trisomy 3q Syndrome
- Pseudotrisomy 13 syndrome
- Triple X syndrome
- Trisomy 13 syndrome

- Trisomy 18
- Trisomy 18-Like Syndrome
- Trisomy 20p
- Trisomy 22 mosaicism syndrome
- Warburton Anyane Yeboa syndrome
- X chromosome, duplication Xq13 1 q21 1
- X chromosome, trisomy Xp3
- X chromosome, trisomy Xpter Xq13
- X chromosome, trisomy Xq25
- Chromosome Inversion
- Duplication 15q11-q13 Syndrome
- Edinburgh Malformation Syndrome
- IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES SYNDROME 1
- Isochromosomes
- Micronuclei, Chromosome-Defective
- Nondisjunction, Genetic
 - Uniparental Disomy
 - Chromosome 10. uniparental disomy of
 - Chromosome 15, trisomy mosaicism
 - Chromosome 16, uniparental disomy
 - Chromosome 1, uniparental disomy 1q12 q21
 - Chromosome 21, uniparental disomy of
 - Chromosome 5, uniparental disomy
 - Chromosome 7, trisomy mosaic
 - Chromosome 8, mosaic trisomy
 - Chromosome 9, trisomy mosaic
 - Trisomy 22 mosaicism syndrome
 - Uniparental disomy of 11

- Uniparental disomy of 13
- Uniparental disomy of chromosome 2
- <u>Uniparental disomy, paternal, chromosome</u>
 14
- Polyploidy
 - Diploid-Triploid Mosaicism
 - Tetraploidy
 - Triploidy
- PREMATURE CHROMATID SEPARATION TRAIT
- Ring Chromosomes
 - Chromosome 10 ring
 - Chromosome 12 ring
 - Chromosome 13 ring
 - Chromosome 15 ring
 - Chromosome 17 ring
 - Chromosome 18 ring
 - Chromosome 19 ring
 - Chromosome 1 ring
 - Chromosome 20 ring
 - Chromosome 21 ring
 - Chromosome 22 ring
 - Chromosome 6 ring syndrome
 - Chromosome 7 ring syndrome
 - Chromosome 8 ring
 - Chromosome 9 Ring
 - Ring Chromosome 14 Syndrome
 - Ring Chromosome 20 Syndrome
 - Ring chromosome 4 syndrome
- Sex Chromosome Aberrations

- 49,XXXXX syndrome
- Chromosome Xq duplication syndrome
- Male sterility due to Y-chromosome deletions
- Tetrasomy X
- Triple X syndrome
- X chromosome, duplication Xq13 1 q21 1
- X chromosome, monosomy Xp22 pter
- X chromosome, monosomy Xq28
- X chromosome, trisomy Xp3
- X chromosome, trisomy Xpter Xq13
- X chromosome, trisomy Xq25
- XYY Karyotype
 - 47, XYY syndrome
- Young Hughes syndrome
- Translocation, Genetic
 - Chromosome 9 inversion or duplication
 - Chromosomes 1 and 2, monosomy 2q duplication
 1p
 - Philadelphia Chromosome
 - SUPERNUMERARY DER(22)t(8
- Death
- Dehydration
- Delayed Graft Function
- Disease
 - Syndrome
- Disease Attributes
 - Acute Disease
 - Asymptomatic Diseases
 - Asymptomatic Infections

- Catastrophic Illness
- Chronic Disease
- Convalescence
- Critical Illness
- Disease Progression
 - Remission, Spontaneous
- Disease Resistance
- Diseases in Twins
- Disease Susceptibility
 - C6 Deficiency, Subtotal
 - Echo Virus 11 Sensitivity
 - Genetic Predisposition to Disease
 - Anticipation, Genetic
 - FASTING INSULIN LEVEL QUANTITATIVE TRAIT LOCUS 1
 - Mycobacterium tuberculosis, susceptibility to infection by
 - MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TO
 - MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TO, 1
 - MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TO, 2
 - MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TO, 3
 - MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TO, X-LINKED
- Emergencies
- Facies
 - Al Gazali Aziz Salem syndrome
 - Al Gazali Hirschsprung syndrome

- Anonychia-onychodystrophy with hypoplasia or absence of distal phalanges
- Aortic arch anomaly with peculiar facies and mental retardation
- Arthrogryposis, Distal, with Mental Retardation and Characteristic Facies
- Axenfeld-Rieger Anomaly with Partially Absent Eye Muscles, Distinctive Face, Hydrocephaly, and Skeletal Abnormalities
- AXENFELD-RIEGER SYNDROME, TYPE 1
- BEAULIEU-BOYCOTT-INNES SYNDROME
- Beemer Ertbruggen syndrome
- Boomerang dysplasia
- Bowen syndrome
- Brachytelephalangy characteristic facies Kallmann
- Brooks-Wisniewski-Brown Syndrome
- Brunoni syndrome
- Burn-Mckeown syndrome
- Cardiofaciocutaneous syndrome
- Cataracts, Congenital, with Sensorineural Deafness, Down Syndrome-Like Facial Appearance, Short Stature, and Mental Retardation
- Charcot-Marie-Tooth Disease, Guadalajara Neuronal Type
- Chondrodysplasia, Megarbane-Dagher-Melki Type
- Chromosome 10g26 Deletion Syndrome
- Chromosome 5p13 Duplication Syndrome
- Chromosome Xq28 Duplication Syndrome
- Clark-Baraitser syndrome
- Cleft palate, midfacial hypoplasia, triangular facies, and sensorineural hearing loss

- Cortical Blindness, Retardation, and Postaxial Polydactyly
- <u>Craniosynostosis, Calcification of Basal Ganglia, and Facial Dysmorphism</u>
- Creases, Infra-Auricular Cutaneous, with Tall Stature and Advanced Bone Age
- Cree Mental Retardation Syndrome
- Cold-Induced Sweating Syndrome 1
- Crumpled helices and small mouth
- C SYNDROME
- Cubitus Valgus with Mental Retardation and Unusual Facies
- Cyprus facial neuromusculoskeletal syndrome
- Davis Lafer syndrome
- Deafness-Craniofacial Syndrome
- Der Kaloustian Mcintosh Silver syndrome
- <u>Diabetes Mellitus, Permanent Neonatal, with Cerebellar Agenesis</u>
- <u>Dislocation of Hip, Congenital, with</u>
 <u>Hyperextensibility of Fingers and Facial</u>
 Dysmorphism
- Disproportionate Short Stature with Ptosis and Valvular Heart Lesions
- Dubowitz syndrome
- Ectodermal dysplasia alopecia preaxial polydactyly
- Ectodermal dysplasia, sensorineural hearing loss, and distinctive facial features
- <u>Ectodermal Dysplasia Syndrome with Distinctive</u>
 Facial Appearance and Preaxial Polydactyly of Feet
- Ectrodactyly cardiopathy dysmorphism
- Edinburgh Malformation Syndrome
- FACES syndrome

- Facial Dysmorphism, Selective Tooth Agenesis, and Choroid Calcification
- Faciocardiomelic Syndrome
- Feingold Trainer syndrome
- Fibrochondrogenesis
- Fibromatosis, Gingival, with Distinctive Facies
- Filippi syndrome
- Fryns-Aftimos Syndrome
- Fryns Macrocephaly
- Fryns syndrome
- GELEOPHYSIC DYSPLASIA 1
- Giacheti Syndrome
- Granddad Syndrome
- Growth Retardation, Developmental Delay, Coarse Facies, And Early Death
- Growth Deficiency and Mental Retardation with Facial Dysmorphism
- Growth mental deficiency syndrome of Myhre
- Hadziselimovic Syndrome
- Haspeslagh Fryns Muelenaere syndrome
- Holoprosencephaly 10
- Hydronephrosis, Congenital, with Cleft Palate, Characteristic Facies, Hypotonia, and Mental Retardation
- Hydrops Fetalis, Nonimmune, With Gracile Bones
 And Dysmorphic Features
- Rudiger Syndrome
- Hypertrichosis, hyperkeratosis, mental retardation, and distinctive facial features
- Hypotonia, Seizures, And Precocious Puberty
- Iris dysplasia hypertelorism deafness

- Irons Bhan syndrome
- Kahrizi Syndrome
- Kaufman oculocerebrofacial syndrome
- KBG syndrome
- Kozlowski-Krajewska syndrome
- Kozlowski Rafinski Klicharska syndrome
- Larsen-Like Syndrome
- Leri pleonosteosis
- Lichtenstein syndrome
- Lymphedema, Cardiac Septal Defects, And Characteristic Facies
- Lymphedema, microcephaly and chorioretinopathy syndrome
- Macrocephaly Autism Syndrome
- Macrocephaly with Multiple Epiphyseal Dysplasia and Distinctive Facies
- Malocclusion and Short Stature
- Marfanoid Mental Retardation Syndrome, Autosomal
- McDonough syndrome
- McPherson Clemens syndrome
- Mental Retardation, Buenos Aires Type
- Mental Retardation, Microcephaly, Epilepsy, And Coarse Face
- Mental Retardation, Short Stature, Facial Anomalies, and Joint Dislocations
- Otoonychoperoneal Syndrome
- Metaphyseal Dysplasia with Maxillary Hypoplasia and Brachydactyly
- Microcephaly cervical spine fusion anomalies
- Microcephaly deafness syndrome

- Microcephaly, Growth Retardation, Cataract, Hearing Loss, And Unusual Appearance
- Microcephaly, Severe, with Skeletal Anomalies including Posterior Rib-Gap Defects
- Microcephaly with Chemotactic Defect and <u>Transient Hypogammaglobulinemia</u>
- Mowat-Wilson syndrome
- Multiple Pterygium Syndrome, X-Linked
- NF1 Microduplication Syndrome
- Nicolaides Baraitser syndrome
- Night blindness skeletal anomalies unusual facies
- Omodysplasia 2
- Oroacral Syndrome, Verloes-Koulischer Type
- Osteolysis syndrome recessive
- Palant cleft palate syndrome
- Partington Anderson syndrome
- Pfeiffer Palm Teller syndrome
- Pierre Robin Sequence with Facial and Digital Anomalies
- Pitt-Hopkins syndrome
- Pituitary Hormone Deficiency, Combined, 1
- Plantar Lipomatosis, Unusual Facies, and Developmental Delay
- Progeroid Facial Appearance with Hand Anomalies
- Radioulnar synostosis retinal pigment abnormalities
- Ramos Arroyo Clark syndrome
- Renal and Mullerian Duct Hypoplasia
- Renal dysplasia limb defects syndrome
- Renal Tubular Acidosis, Distal, With Nephrocalcinosis, Short Stature, Mental Retardation, And Distinctive Facies

- Robin Sequence with Distinctive Facial Appearance and Brachydactyly
- Roifman-Chitayat Syndrome
- Ruvalcaba Syndrome
- Schrander-Stumpel Theunissen Hulsmans syndrome
- Seckel syndrome 1
- Seckel Syndrome 3
- Short Stature, Facial Dysmorphism, Severe Brachydactyly, and Syndactyly
- Shprintzen omphalocele syndrome
- Spastic paraplegia 23
- Speech Development, Delayed, With Facial Asymmetry, Strabismus, And Transverse Earlobe Crease
- Spinocerebellar Ataxia with Dysmorphism
- Spondyloepiphyseal Dysplasia Tarda with Characteristic Facies
- Thakker Donnai syndrome
- Thomas Jewett Raines syndrome
- Thrombocytopenia Robin sequence
- Trichohepatoenteric Syndrome
- Trichorhinophalangeal Syndrome, Type III
- Urofacial syndrome
- Uruguay Faciocardiomusculoskeletal Syndrome
- White forelock with malformations
- Winter Harding Hyde syndrome
- Young Simpson syndrome
- Zechi-Ceide Syndrome
- latrogenic Disease
 - Cross Infection

- Pneumonia, Ventilator-Associated
- Late Onset Disorders
- Neglected Diseases
- Rare Diseases
- Recurrence
 - Symptom Flare Up
- Dysbiosis
- Emphysema
 - Emphysema, Congenital, With Deafness, Penoscrotal Web, And Mental Retardation
 - Hemolytic Anemia, Congenital, with Emphysema and Cutis Laxa
 - Mediastinal Emphysema
 - Subcutaneous Emphysema
 - Alpha 1-Antitrypsin Deficiency
 - Alpha-1-Antitrypsin Deficiency, Autosomal Recessive
- Extravasation of Diagnostic and Therapeutic Materials
- Femoracetabular Impingement
- Fibrosis
 - Cicatrix
 - Cicatrix, Hypertrophic
 - Keloid
 - Acne Keloid
 - Torticollis keloids cryptorchidism renal dysplasia
 - Tissue Adhesions
 - Fibrosis Of Extraocular Muscles, Congenital, 2
 - Fibrosis Of Extraocular Muscles, Congenital, 3A, with or without Extraocular Involvement
 - Fibrosis of Extraocular Muscles, Congenital, 3B

- Fibrosis of Extraocular Muscles, Congenital, 3C
- <u>Fibrosis of Extraocular Muscles, Congenital, with</u> Synergistic Divergence
- Nephrogenic Fibrosing Dermopathy
- Peritoneal Fibrosis
- Retroperitoneal Fibrosis
 - Multifocal fibrosclerosis
- Tukel syndrome
- Genomic Instability
 - Chromosomal Instability
 - Chromosomal Instability with Tissue-Specific Radiosensitivity
 - Chromosome Fragility
 - Radiation Sensitivity Chromosome Instability
 Syndrome, Autosomal Dominant
 - Microsatellite Instability
- Gliosis
- Granuloma
 - Angiolymphoid Hyperplasia with Eosinophilia
 - Churg-Strauss Syndrome
 - Eosinophilic Granuloma
 - Granuloma, Pyogenic
 - Granuloma, Respiratory Tract
 - Granuloma, Laryngeal
 - Granuloma Annulare
 - Granuloma, Foreign-Body
 - Granuloma, Giant Cell
 - Noonan like syndrome
 - NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR

NOONAN SYNDROME-LIKE DISORDER WITH OR WITHOUT JUVENILE MYELOMONOCYTIC LEUKEMIA

- Granuloma, Plasma Cell
- Granulomas, congenital cerebral
- Necrobiotic Xanthogranuloma
- Xanthogranulomatous cholecystitis
- Xanthogranulomatous sialadenitis
- Granulomatosis, Orofacial
- Growth Disorders
 - Acrocapitofemoral Dysplasia
 - Acrocephalopolydactylous Dysplasia
 - Acromegaloid features, overgrowth, cleft palate, and hernia
 - Agonadism, XY, with Mental Retardation, Short Stature, Retarded Bone Age, and Multiple Extragenital Malformations
 - Al Gazali Khidr Prem Chandran syndrome
 - Aphalangia syndactyly microcephaly
 - Auriculoosteodysplasia
 - Bellini Chiumello Rimoldi syndrome
 - Berk-Tabatznik syndrome
 - Bhaskar Jagannathan syndrome
 - Blepharophimosis with ptosis, syndactyly, and short stature
 - Borieson-Forssman-Lehmann syndrome
 - Boudhina Yedes Khiari syndrome
 - BRACHYDACTYLY, TYPE E2
 - Brooks-Wisniewski-Brown Syndrome
 - Cantalamessa Baldini Ambrosi syndrome
 - Cantu Sanchez-Corona Fragoso syndrome

- Cataracts, ataxia, short stature, and mental retardation
- <u>Cataracts, Congenital, with Sensorineural Deafness,</u>
 <u>Down Syndrome-Like Facial Appearance, Short Stature,</u>
 and Mental Retardation
- Chitty Hall Baraitser syndrome
- Chromosome 15g26-Qter Deletion Syndrome
- Chromosome 18 Pericentric Inversion
- CHROMOSOME 3pter-p25 DELETION SYNDROME
- Clark-Baraitser syndrome
- CODAS syndrome
- Coffin syndrome 1
- Contractures ectodermal dysplasia cleft lip palate
- Cote Katsantoni syndrome
- Coxoauricular Syndrome
- Craniofacial Abnormalities, Cataracts, Congenital Heart Disease, Sacral Neural Tube Defects, and Growth and Developmental Retardation
- Creases, Infra-Auricular Cutaneous, with Tall Stature and Advanced Bone Age
- Crumpled helices and small mouth
- Curatolo Cilio Pessagno syndrome
- Daish Hardman Lamont syndrome
- Dermoids of cornea
- Devriendt syndrome
- <u>Dubowitz syndrome</u>
- Dyschondrosteosis and Nephritis
- Epithelial Squamous Dysplasia, Keratinizing Desquamative, of Urinary Tract
- Extrasystoles, Multiform Ventricular, with Short Stature,
 Hyperpigmentation and Microcephaly
- Fallot complex with severe mental and growth retardation

- Fetal Growth Retardation
 - Bowen-Conradi syndrome
 - Camptodactyly syndrome Guadalajara type 2
 - Cartwright Nelson Fryns syndrome
 - Craniomicromelic Syndrome
 - <u>Diabetes Mellitus, Permanent Neonatal, with</u>
 Cerebellar Agenesis
 - Fetal akinesia syndrome, X-linked
 - Finnish lethal neonatal metabolic syndrome
 - Granddad Syndrome
 - Holoprosencephaly with Fetal Akinesia-Hypokinesia Sequence
 - Hoyeraal Hreidarsson syndrome
 - Intrauterine Growth Retardation, Metaphyseal <u>Dysplasia</u>, Adrenal Hypoplasia Congenita, And Genital Anomalies
 - Lambotte syndrome
 - <u>Microcephalic osteodysplastic primordial dwarfism</u>, type 1
 - Microcephalic osteodysplastic primordial dwarfism, type 3
 - Microcephalic Osteodysplastic Primordial Dwarfism, Type II
 - Microcephaly, Severe, with Skeletal Anomalies including Posterior Rib-Gap Defects
 - Neu Laxova syndrome
 - Progeroid syndrome, neonatal
 - Ray Peterson Scott syndrome
 - SECKEL SYNDROME 4
 - Sharma Kapoor Ramji syndrome
 - Short stature and locking fingers
 - Thymic-Renal-Anal-Lung dysplasia

- Trichohepatoenteric Syndrome
- Woods Leversha Rogers syndrome
- Filippi syndrome
- Floating-harbor syndrome
- FORSYTHE-WAKELING SYNDROME
- Frias syndrome
- Game Friedman Paradice syndrome
- Gay Feinmesser Cohen syndrome
- GELEOPHYSIC DYSPLASIA 1
- GEMSS syndrome
- GOMBO syndrome
- Gomez Lopez Hernandez syndrome
- Gonadal Dysgenesis, Hypergonadotropic, XX Type, Short Stature, and Recurrent Metabolic Acidosis
- Goniodysgenesis-Mental Retardation-Short Stature Syndrome
- Growth and Developmental Retardation, Ocular Ptosis,
 Cardiac Defect, and Anal Atresia
- Growth Deficiency and Mental Retardation with Facial <u>Dysmorphism</u>
- Growth Failure, Microcephaly, Mental Retardation,
 Cataracts, Large Joint Contractures, Osteoporosis,
 Cortical Dysplasia, and Cerebellar Atrophy
- Growth mental deficiency syndrome of Myhre
- Growth retardation, Alopecia, Pseudoanodontia and Optic atrophy
- Growth Retardation, Small and Puffy Hands and Feet, and Eczema
- Hairy elbows
- Heme Oxygenase 1 Deficiency
- Hersh Podruch Weisskopk syndrome
- Hooft disease

- Hunter-McAlpine syndrome
- Hutterite cerebroosteonephrodysplasia syndrome
- Hypoparathyroidism-retardation-dysmorphism syndrome
- Insulin-Like Growth Factor I Deficiency
- Insulin-Like Growth Factor I, Resistance To
- Insulin-Like Growth Factor I, Resistance to, due to Increased Binding Protein
- Johanson Blizzard syndrome
- Jorgenson Lenz syndrome
- Krause-Kivlin syndrome
- Kozlowski Rafinski Klicharska syndrome
- Kuster Majewski Hammerstein syndrome
- Ladda Zonana Ramer syndrome
- Langer mesomelic dysplasia
- <u>Laryngotracheal Stenosis</u>, <u>Progressive</u>, <u>with Short Stature</u> <u>and Arthropathy</u>
- Leri-Weil syndrome
- <u>Leukomelanoderma, Infantilism, Mental Retardation,</u>
 <u>Hypodontia, Hypotrichosis</u>
- LIG4 Syndrome
- <u>Lipodystrophy, Generalized, with Mental Retardation,</u>
 <u>Deafness, Short Stature, and Slender Bones</u>
- Lowry Maclean syndrome
- Lowry Wood syndrome
- Madelung Deformity
- Macrosomia Adiposa Congenita
- Malocclusion and Short Stature
- Megarbane syndrome
- Meier-Gorlin syndrome
- Mental and Growth Retardation with Amblyopia

- Mental retardation-hypotonic facies syndrome, x-linked, 1
- Mental Retardation, Microcephaly, Growth Retardation, Joint Contractures, and Facial Dysmorphism
- Mental retardation Mietens Weber type
- Mental Retardation with Optic Atrophy, Facial Dysmorphism, Microcephaly, and Short Stature
- MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE
- Mental Retardation, X-Linked, with Short Stature
- Microcephaly cervical spine fusion anomalies
- Microdontia hypodontia short stature
- Milner Khallouf Gibson syndrome
- Mitochondrial myopathy with lactic acidosis
- Mollica Pavone Antener syndrome
- Morillo-Cucci Passarge syndrome
- Myelodysplasia, Immunodeficiency, Facial Dysmorphism, Short Stature, and Psychomotor Delay
- Nathalie syndrome
- Neurofaciodigitorenal syndrome
- Nijmegen Breakage Syndrome-Like Disorder
- Micromelic dysplasia, congenital, with dislocation of radius
- Onat syndrome
- OSTEOCHONDRITIS DISSECANS, SHORT STATURE, AND EARLY-ONSET OSTEOARTHRITIS
- Osteolysis syndrome recessive
- Partington Anderson syndrome
- <u>Pectus Excavatum, Macrocephaly, Short Stature,</u>
 Dysplastic Nails
- Polydysspondyly
- Petty Laxova Wiedemann syndrome
- Pfeiffer Kapferer syndrome

- Pfeiffer Mayer syndrome
- <u>Pfeiffer Palm Teller syndrome</u>
- Pili torti developmental delay neurological abnormalities
- Pilotto syndrome
- Premature aging, Okamoto type
- Progeria short stature pigmented nevi
- Progeroid Syndrome, Congenital, Petty Type
- Qazi Markouizos syndrome
- Radioulnar synostosis retinal pigment abnormalities
- RAJAB SYNDROME
- Ramon Syndrome
- Reardon Wilson Cavanagh syndrome
- Renal Tubular Acidosis, Distal, With Nephrocalcinosis,
 Short Stature, Mental Retardation, And Distinctive Facies
- Rhizomelic dysplasia, scoliosis, and retinitis pigmentosa
- Rodrigues blindness
- Rommen Mueller Sybert syndrome
- Rowley-Rosenberg syndrome
- Say Meyer syndrome
- Say syndrome
- Schaap Taylor Baraitser syndrome
- Schimke X-linked mental retardation syndrome
- Seemanova Lesny syndrome
- Severe Combined Immunodeficiency with Microcephaly,
 Growth Retardation, and Sensitivity to Ionizing Radiation
- Short Stature, Facial Dysmorphism, Severe Brachydactyly, and Syndactyly
- Short Stature, Idiopathic, X-Linked
- Short stature syndrome, Brussels type
- SHORT syndrome

- Slavotinek Pike Mills Hurst syndrome
- Sonoda syndrome
- Spastic paraplegia 9, autosomal dominant
- Spondylodysplasia And Premature Pubarche
- Spondylometaphyseal dysplasia, 'corner fracture' type
- Stern Lubinsky Durrie syndrome
- Synostosis of Talus and Calcaneus with Short Stature
- Theodor Hertz Goodman syndrome
- Thrombocytopenia Robin sequence
- Thumb Agenesis, Short Stature, And Immunodeficiency
- Thumb, Hypoplastic, with Choroid Coloboma, Poorly Developed Antihelix, and Deafness
- Tonoki syndrome
- Tsukahara Syndrome
- Vater-Like Defects with Pulmonary Hypertension, Laryngeal Webs, and Growth Deficiency
- Vertebral body fusion overgrowth
- Viljoen Kallis Voges syndrome
- Volcke Soekarman syndrome
- Wellesley Carmen French syndrome
- Wiedemann Grosse Dibbern syndrome
- Winchester syndrome
- Young Hughes syndrome
- Zerres Rietschel Majewski syndrome
- Zinc Deficiency, Neonatal, due to Low Breast Milk Zinc

Hemolysis

- Red cell phospholipid defect with hemolysis
- <u>Thrombocytopenia</u>, <u>Platelet Dysfunction</u>, <u>Hemolysis</u>, <u>and</u> <u>Imbalanced Globin Synthesis</u>
- Hemorrhage

- BLEEDING DISORDER, PLATELET-TYPE, 8
- Bleeding Disorder Due To P2rx1 Defect
- Blood Loss, Surgical
- Circumvallate Placenta Syndrome
- Ecchymosis
- Epistaxis
 - Epistaxis, Hereditary
- Exsanguination
- Eye Hemorrhage
 - Choroid Hemorrhage
 - Hyphema
 - Retinal Hemorrhage
 - Brain Small Vessel Disease with Hemorrhage
 - Frenkel Russe syndrome
 - Vitreous Hemorrhage
- Gastrointestinal Hemorrhage
 - Hematemesis
 - Melena
 - Peptic Ulcer Hemorrhage
- Hemarthrosis
- Hematocele
- Hematoma
 - Hematoma, Epidural, Cranial
 - Hematoma, Epidural, Spinal
 - Hematoma, Subdural
 - Hematoma, Subdural, Acute
 - Hematoma, Subdural, Chronic
 - Hematoma, Subdural, Intracranial

- Hematoma, Subdural, Spinal
- Hematuria
 - Coloboma, cleft lip-palate and mental retardation syndrome
 - Hematuria, Benign Familial
- Hemobilia
- Hemoperitoneum
- Hemoptysis
- Hemothorax
 - Hemopneumothorax
- Intracranial Hemorrhages
 - Intracerebral hemorrhage
 - Basal Ganglia Hemorrhage
 - Putaminal Hemorrhage
 - Cerebral Hemorrhage, Traumatic
 - Cerebral hemorrhage with amyloidosis, hereditary, Dutch type
 - Hematoma, Epidural, Cranial
 - Hematoma, Subdural
 - Hematoma, Subdural, Acute
 - Hematoma, Subdural, Chronic
 - Hematoma, Subdural, Intracranial
 - Hematoma, Subdural, Spinal
 - Intracranial Hemorrhage, Hypertensive
 - Intracranial Hemorrhage, Traumatic
 - Brain Hemorrhage, Traumatic
 - Brain Stem Hemorrhage, Traumatic
 - Cerebral Hemorrhage, Traumatic
 - Hematoma, Epidural, Cranial
 - Hematoma, Subdural

- Hematoma, Subdural, Acute
- Hematoma, Subdural, Chronic
- Hematoma, Subdural, Intracranial
- Hematoma, Subdural, Spinal
- Subarachnoid Hemorrhage, Traumatic
- Pituitary Apoplexy
- Subarachnoid Hemorrhage
 - Aneurysm, Intracranial Berry, 1
 - Aneurysm, Intracranial Berry, 4
 - Aneurysm, Intracranial Berry, 6
 - Aneurysm, Intracranial Berry, 7
 - Aneurysm, Intracranial Berry, 8
 - Aneurysm, Intracranial Berry, 9
 - Subarachnoid Hemorrhage, Traumatic
- Oral Hemorrhage
 - Gingival Hemorrhage
 - Bazopoulou Kyrkanidou syndrome
- Postoperative Hemorrhage
 - Endoleak
- Purpura
 - Ethylmalonic encephalopathy
 - Pigmented purpuric eruption
 - Purpura Fulminans
 - Purpura, Hyperglobulinemic
 - Purpura, Schoenlein-Henoch
 - Vascular purpura
 - Purpura simplex
 - Purpura, Thrombocytopenic
 - Acquired amegakaryocytic thrombocytopenia

- Posttransfusion Purpura
- Purpura, Thrombocytopenic, Idiopathic
- Purpura, Thrombotic Thrombocytopenic
 - <u>Thrombotic thrombocytopenic purpura, acquired</u>
- Waterhouse-Friderichsen Syndrome
 - <u>Fatal pneumococcal Waterhouse-</u>
 <u>Friderichsen syndrome</u>
- Rapidly progressive glomerulonephritis with pulmonary hemorrhage
- Retrobulbar Hemorrhage
- Shock, Hemorrhagic
- Uterine Hemorrhage
 - Menorrhagia
 - Metrorrhagia
 - Postpartum Hemorrhage
- Hyperamylasemia
- Hyperammonemia
 - HHH syndrome
 - ORNITHINE TRANSCARBAMYLASE DEFICIENCY, HYPERAMMONEMIA DUE TO
 - Systemic carnitine deficiency
- Hyperbilirubinemia
 - Hyperbilirubinemia, Neonatal
 - Hyperbilirubinemia, Transient Familial Neonatal
 - Jaundice, Neonatal
 - Breastfeeding Jaundice
 - Jaundice, Chronic Idiopathic
 - Jaundice
 - Deal Barratt Dillon syndrome

- Dysmyelination With Jaundice
- Jaundice, Obstructive
 - Anemia, hereditary spherocytic hemolytic
 - Jaundice, Familial Obstructive, of Infancy
- Lutz Richner Landolt syndrome
- Kernicterus
- Hyperplasia
 - Facial Hemihypertrophy
 - Hemihyperplasia, Isolated
 - Pseudoangiomatous stromal hyperplasia
 - Sebaceous gland hyperplasia, familial presenile
- Hyperuricemia
 - Glomerulocystic Kidney Disease with Hyperuricemia and Isosthenuria
 - Hyperuricemia, Infantile, with Abnormal Behavior and Normal Hypoxanthine Guanine Phosphoribosyltransferase
 - HYPERURICEMIC NEPHROPATHY, FAMILIAL JUVENILE, 1
 - Hyperuricemic Nephropathy, Familial Juvenile 2
 - HYPERURICEMIC NEPHROPATHY, FAMILIAL JUVENILE, 3
 - Juvenile gout
 - URIC ACID CONCENTRATION, SERUM, QUANTITATIVE TRAIT LOCUS 1
 - URIC ACID CONCENTRATION, SERUM, QUANTITATIVE TRAIT LOCUS 4
- Hypovolemia
- Inflammation
 - Acute-Phase Reaction
 - Foreign-Body Reaction
 - Granuloma, Foreign-Body

- Implant Capsular Contracture
- Neurogenic Inflammation
- Rasmussen subacute encephalitis
- Seroma
- Serositis
- Suppuration
 - Abscess
 - Abdominal Abscess
 - Liver Abscess
 - Liver Abscess, Amebic
 - Liver Abscess, Pyogenic
 - Subphrenic Abscess
 - Brain Abscess
 - Toxoplasmosis, Cerebral
 - Epidural Abscess
 - Lung Abscess
 - Periapical Abscess
 - Periodontal Abscess
 - Peritonsillar Abscess
 - Psoas Abscess
 - Retropharyngeal Abscess
 - Zuska's Disease
 - Cellulitis
 - Orbital Cellulitis
 - <u>Perifolliculitis Capitis Abscedens Et Suffodiens, Familial</u>
 - Wells syndrome
 - Empyema
 - Empyema, Pleural

- Empyema, Tuberculous
- Empyema, Subdural
- Hidradenitis Suppurativa
 - Hidradenitis suppurativa, familial
- Otitis Media, Suppurative
- Pyomyositis
- Thyroiditis, Suppurative
- Uveitis, Suppurative
 - Panophthalmitis
- Systemic Inflammatory Response Syndrome
 - Sepsis
 - Bacteremia
 - Endotoxemia
 - Flavimonas oryzihabitans Bacteremia
 - Hemorrhagic Septicemia
 - Fungemia
 - Candidemia
 - Parasitemia
 - Septic shock
 - Viremia
 - Hemorrhagic Septicemia, Viral
 - Septic shock
- Intraoperative Complications
 - Blood Loss, Surgical
 - Intraoperative Awareness
 - Malignant Hyperthermia
 - <u>Contractures, Congenital, Torticollis, and Malignant</u> Hyperthermia
 - King Denborough syndrome

- Malignant fever
- 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
- Multiple pterygium syndrome
 - MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE
- Native American myopathy
- Ischemia
 - Infarction
 - No-Reflow Phenomenon
 - Rambaud Galian syndrome
- Leukoaraiosis
- Leukocytosis
 - Holoprosencephaly, recurrent infections, and monocytosis
 - Leukemoid Reaction
 - Myeloproliferative Syndrome, Transient
 - Lymphocytosis
 - Persistent Polyclonal B-Cell Lymphocytosis
 - Sclerosing lymphocytic lobulitis
- Lithiasis
- Long Term Adverse Effects
- Malacoplakia
 - Prostatic malacoplakia associated with prostatic abscess
- Menstruation Disturbances
 - Amenorrhea

- Al Awadi syndrome
- Calloso-genital dysplasia
- Nonpuerperal galactorrhea
- Dysmenorrhea
- Hymen, Imperforate
- Menorrhagia
- Oligomenorrhea
- Premenstrual Syndrome
 - Premenstrual Dysphoric Disorder
- Metaplasia
 - Neovascularization, Pathologic
 - Choroidal Neovascularization
 - Eales disease
 - Retinal Neovascularization
- Muscle Weakness
 - Acyl-CoA Dehydrogenase Family, Member 9, Deficiency of
 - Camera Marugo Cohen syndrome
 - Coenzyme Q10 Deficiency
 - COENZYME Q10 DEFICIENCY, PRIMARY, 1
 - Episodic Muscle Weakness, X-Linked
- Necrosis
 - Dental Pulp Necrosis
 - DNA Degradation, Necrotic
 - Fat Necrosis
 - Gangrene
 - Infarction
 - Osteonecrosis
 - Bilateral Kienbock's disease

- Bisphosphonate-Associated Osteonecrosis of the Jaw
- Dieterich's disease
- Femur Head Necrosis
 - Legg-Calve-Perthes Disease
 - Garret Tripp syndrome
- Neointima
- Neoplastic Processes
 - Anaplasia
 - Carcinogenesis
 - Cell Transformation, Neoplastic
 - Blast Crisis
 - Cell Transformation, Viral
 - Progressive Transformation of Germinal Centers
 - Cocarcinogenesis
 - Neoplasm Invasiveness
 - Leukemic Infiltration
 - Neoplasm Metastasis
 - Lymphatic Metastasis
 - Neoplasm Micrometastasis
 - Neoplasm Seeding
 - Neoplasms, Unknown Primary
 - Neoplastic Cells, Circulating
 - Neoplasm Recurrence, Local
 - Neoplasm Regression, Spontaneous
 - Neoplasm, Residual
- Nerve Degeneration
 - Cerebellar degeneration, subacute
 - Retrograde Degeneration

- Skeletal Dysplasia And Progressive Central Nervous System Degeneration, Lethal
- Striatal Degeneration, Autosomal Dominant
- Subacute Combined Degeneration
- Wallerian Degeneration
 - Wallerian degeneration of the pyramidal tract
- Ochronosis
 - Alkaptonuric ochronosis
 - Ochronosis, hereditary
- Ossification, Heterotopic
 - Desbuquois syndrome
 - Eagle syndrome
 - Leri pleonosteosis
 - Osseous Heteroplasia, Progressive
 - Ossification of Posterior Longitudinal Ligament
 - Ossification of the posterior longitudinal ligament of the spine
 - Superior Transverse Scapular Ligament, Calcification Of, Familial
- Polydipsia
 - Polydipsia, Psychogenic
- Postoperative Complications
 - Afferent Loop Syndrome
 - Anastomotic Leak
 - Corneal Endothelial Cell Loss
 - Coronary-Subclavian Steal Syndrome
 - Delayed Emergence from Anesthesia
 - Failed Back Surgery Syndrome
 - Gas bloat syndrome
 - Graft Occlusion, Vascular

- Incisional Hernia
- Malignant Hyperthermia
 - <u>Contractures, Congenital, Torticollis, and Malignant</u> <u>Hyperthermia</u>
 - King Denborough syndrome
 - Malignant fever
 - 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
 - Multiple pterygium syndrome
 - MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE
 - Native American myopathy
- Pain, Postoperative
 - Phantom Limb
- Postoperative Hemorrhage
 - Endoleak
- Postcholecystectomy Syndrome
- Postgastrectomy Syndromes
 - Dumping Syndrome
- Postoperative Nausea and Vomiting
- Postpericardiotomy Syndrome
- Prosthesis Failure
 - Implant Capsular Contracture
- Prosthesis-Related Infections
- Reperfusion Injury
 - Myocardial Reperfusion Injury

- Primary Graft Dysfunction
- Shock, Surgical
- Short Bowel Syndrome
- Slit Ventricle Syndrome
- Surgical Wound Infection
- Surgical Wound Dehiscence
- Vasoplegia
- Protein Aggregation, Pathological
- Respiratory Aspiration
 - Respiratory Aspiration of Gastric Contents
- Retropneumoperitoneum
- Sclerosis
 - Mediastinal Fibrosis
 - Mesangial sclerosis, diffuse
 - Mesangial Sclerosis, Diffuse Renal, with Ocular Abnormalities
 - Osteosclerosis with ichthyosis and premature ovarian failure
 - Poikiloderma, Hereditary Sclerosing
 - Sclerosing lymphocytic lobulitis
 - Scleroatonic muscular dystrophy
 - Sclerosing bone dysplasia mental retardation
- Shock
 - Multiple Organ Failure
 - Shock, Cardiogenic
 - Shock, Hemorrhagic
 - Shock, Surgical
 - Shock, Traumatic
 - Crush Syndrome
 - Systemic Inflammatory Response Syndrome

- Sepsis
 - Bacteremia
 - Endotoxemia
 - Flavimonas oryzihabitans Bacteremia
 - Hemorrhagic Septicemia
 - Fungemia
 - Candidemia
 - Parasitemia
 - Septic shock
 - Viremia
 - Hemorrhagic Septicemia, Viral
- Septic shock
- Teratogenesis
- Ulcer
 - Umbilical cord ulceration and intestinal atresia
- Vascular Remodeling
- Yang deficiency
- Yin Deficiency
- Signs and Symptoms
 - Aging, Premature
 - Wolfram Syndrome 2
 - Asthenia
 - Body Temperature Changes
 - Fever
 - Cold-Induced Sweating Syndrome 1
 - Fever of Unknown Origin
 - Fever, Familial Lifelong Persistent
 - Hyperthermia, Cutaneous, With Headaches And Nausea

- Pelger-Huet-Like Anomaly and Episodic Fever with Abdominal Pain
- Periodic fever, familial, autosomal dominant
- Sweating Sickness
- Hypothermia
 - Recurrent spontaneous hypothermia with hypoplasia of the corpus callosum
 - Shapiro syndrome
- Body weight
 - Birth weight
 - Fetal Macrosomia
 - Macrosomia obesity macrocephaly ocular abnormalities
 - Macrosomia with lethal microphthalmia
 - Nephroblastomatosis, fetal ascites, macrosomia and Wilms tumor
 - Pseudotrisomy 13 syndrome
 - Body Weight Changes
 - Weight Gain
 - Weight Loss
 - Emaciation
 - Cachexia
 - FACES syndrome
 - Groll Hirschowitz syndrome
 - Fetal Weight
 - Overweight
 - Obesity
 - ADIPONECTIN, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS 2
 - ADIPONECTIN, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS 3

- Ayazi syndrome
- Biemond Syndrome II
- Biemond syndrome type 2
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 12
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 10
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 11
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 13
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 14
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 4
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 7
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 8
- BODY MASS INDEX QUANTITATIVE TRAIT LOCUS 9
- Borjeson-Forssman-Lehmann syndrome
- Camera Marugo Cohen syndrome
- Clark-Baraitser syndrome
- Cohen syndrome
- Coloboma-Obesity-Hypogenitalism-Mental Retardation Syndrome
- FASTING INSULIN LEVEL QUANTITATIVE TRAIT LOCUS 1
- <u>Macrosomia obesity macrocephaly ocular</u> abnormalities
- MEHMO syndrome

- Midface Hypoplasia, Obesity, Developmental Delay, and Neonatal Hypotonia
- MOMES Syndrome
- MORM syndrome
- Obesity, Abdominal
- Obesity, Hyperphagia, and Developmental Delay
- Obesity Hypoventilation Syndrome
- Obesity, Metabolically Benign
- Obesity, Morbid
- Pediatric Obesity
- Prader-Willi Syndrome
 - Prader-Willi habitus, osteopenia, and camptodactyly
 - Prader-Willi-Like Syndrome
 Associated With Chromosome 6
- Prolactin Deficiency with Obesity and Enlarged Testes
- Proopiomelanocortin Deficiency
- Proprotein Convertase 1 3 Deficiency
- Short Stature-Obesity Syndrome
- Vasquez Hurst Sotos syndrome
- Wilms Tumor, Aniridia, Genitourinary
 Anomalies, Mental Retardation, and Obesity
 Syndrome
- Wilson-Turner X-linked mental retardation syndrome
- Young Hughes syndrome
- Thinness
 - CK SYNDROME
- Cardiac Output, High
- Cardiac Output, Low

- Chills
- Cyanosis
 - Cyanosis and Hepatic Disease
 - Infantile Apparent Life-Threatening Event
 - Rombo syndrome
- Eye Manifestations
 - Eye Pain
 - Eye Hemorrhage
 - Choroid Hemorrhage
 - Hyphema
 - Retinal Hemorrhage
 - Vitreous Hemorrhage
 - Susac Syndrome
- Edema
 - Edema, Familial Idiopathic, Prepubertal
 - Edema, Cardiac
 - Enteropathy, Familial, with Villous Edema and Immunoglobulin G2 Deficiency
 - Hydrops Fetalis
 - <u>Dehydrated Hereditary Stomatocytosis</u>,
 <u>Pseudohyperkalemia</u>, and <u>Perinatal Edema</u>
 - Hydrops Fetalis, Nonimmune, With Gracile Bones
 And Dysmorphic Features
 - Nuchal bleb, familial
 - Xerocytosis, hereditary
 - Ketoadipicaciduria
 - Landy Donnai syndrome
- Failure to Thrive
 - Cardiofaciocutaneous syndrome

- Delayed Cranial Ossification due to CBFB Haploinsufficiency
- Edinburgh Malformation Syndrome
- Growth Retardation, Developmental Delay, Coarse Facies, And Early Death
- Secretory Diarrhea, Myopathy, and Deafness
- Fatigue
 - Corticosteroid-Binding Globulin Deficiency
 - Mental Fatigue
 - Compassion Fatigue
- Feminization
- Fetal Distress
- Flushing
 - Chlorpropamide-Alcohol Flushing
 - Harlequin syndrome
- Heart Murmurs
 - Systolic Murmurs
- Hot Flashes
- Hypergammaglobulinemia
 - Capillary leak syndrome with monoclonal gammopathy
 - Hyperimmunoglobulin G1(A1) Syndrome
 - Lymphopenic Hypergammaglobulinemia, Antibody Deficiency, Autoimmune Hemolytic Anemia, and Glomerulonephritis
 - Mevalonate Kinase Deficiency
 - Monoclonal Gammopathy of Undetermined Significance
 - Schnitzler Syndrome
- Hyperlactatemia
- Hypertriglyceridemic Waist
- Intermittent Claudication

- Mobility Limitation
- Motion sickness
 - Mal de debarquement
 - Space Motion Sickness
- Myocardial Stunning
- Neurologic Manifestations
 - Cerebrospinal Fluid Leak
 - Cerebrospinal Fluid Otorrhea
 - Cerebrospinal Fluid Rhinorrhea
 - Decerebrate State
 - <u>Diabetes Mellitus, Permanent Neonatal, With Neurologic Features</u>
 - Dyskinesias
 - Ataxia
 - Abetalipoproteinemia neuropathy
 - Arts syndrome
 - Ataxia and Polyneuropathy, Adult-Onset
 - Ataxia, Deafness, and Cardiomyopathy
 - Ataxia-Microcephaly-Cataract Syndrome
 - ATAXIA, SENSORY, 1, AUTOSOMAL DOMINANT
 - Ataxia, Sensory, Autosomal Dominant
 - Ataxia, Spastic, with Congenital Miosis
 - Ataxia with Fasciculations
 - Ataxia with Myoclonic Epilepsy and Presenile Dementia
 - Ataxia with vitamin E deficiency
 - Atonic-Astatic Syndrome of Foerster
 - Bangstad syndrome
 - Behr syndrome

- Bhaskar Jagannathan syndrome
- CANOMAD syndrome
- Carnitine Acetyltransferase Deficiency
- Cataract ataxia deafness
- <u>Cataracts, ataxia, short stature, and mental</u> <u>retardation</u>
- Cerebellar Ataxia
 - 3-Methylglutaconic Aciduria, Type V
 - Aniridia cerebellar ataxia mental deficiency
 - Ataxia, Spastic, 1, Autosomal Dominant
 - Ataxia, Spastic, 2, Autosomal Recessive
 - Ataxia, Spastic, 3, Autosomal Recessive
 - Autosomal Recessive Cerebellar Ataxia Type 1
 - Brachydactyly-Nystagmus-Cerebellar Ataxia
 - Branchial Myoclonus with Spastic Paraparesis and Cerebellar Ataxia
 - CAPOS syndrome
 - Cerebellar Ataxia and Hypergonadotropic Hypogonadism
 - Cerebellar Ataxia and Hypogonadotropic Hypogonadism
 - Cerebellar Ataxia and Neurosensory Deafness
 - Cerebellar Ataxia, Benign, with Thermoanalgesia
 - Cerebellar Ataxia, Cayman Type

- Cerebellar Ataxia, Deafness, and Narcolepsy
- Cerebellar ataxia ectodermal dysplasia
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 2
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 3
- Cerebellar hypoplasia with endosteal sclerosis
- Cerebelloparenchymal Disorder II
- Dementia, familial Danish
- Dysequilibrium syndrome
- Early-onset ataxia with oculomotor apraxia and hypoalbuminemia
- Episodic Ataxia, Type 6
- Episodic Ataxia, Type 5
- Furukawa Takagi Nakao syndrome
- Harding ataxia
- Hemiplegic migraine, familial type 1
- Herrmann syndrome
- <u>Laryngeal Abductor Paralysis with</u>
 <u>Cerebellar Ataxia and Motor</u>
 <u>Neuropathy</u>
- Mainzer-Saldino Disease
- Marinesco-Sjogren-like syndrome (MSLS)
- Myelocerebellar Disorder
- Myoclonus, Cerebellar Ataxia, and Deafness
- Neuhauser Eichner Opitz syndrome
- Renal Tubulopathy, Diabetes Mellitus, and Cerebellar Ataxia due to Duplication of Mitochondrial DNA

- Spinocerebellar Ataxias
 - Anemia, sideroblastic spinocerebellar ataxia
 - Ataxia Telangiectasia
 - Ataxia Telangiectasia Like Disorder
 - TELANGIECTASIA-LIKE
 DISORDER 1
 - Ataxia-Telangiectasia
 Variant
 - Ataxia-Telangiectasia Variant V2
 - Ataxia-Telangiectasia with Generalized Skin Pigmentation and Early Death
 - Chorioretinal Dystrophy,
 Spinocerebellar Ataxia, and
 Hypogonadotropic
 Hypogonadism
 - Gemignani syndrome
 - Machado-Joseph Disease
 - Spinocerebellar ataxia, Xlinked, 3
 - MITOCHONDRIAL DNA DEPLETION SYNDROME 7 (HEPATOCEREBRAL TYPE)
 - Spastic Ataxia
 - Spastic ataxia Charlevoix-Saguenay type
 - Spinocerebellar Ataxia 10
 - Spinocerebellar Ataxia 11
 - Spinocerebellar Ataxia 12
 - Spinocerebellar ataxia 13

- Spinocerebellar ataxia 14
- Spinocerebellar Ataxia 15
- Spinocerebellar Ataxia 17
- Spinocerebellar ataxia 20
- Spinocerebellar ataxia 25
- Spinocerebellar ataxia 26
- Spinocerebellar ataxia 28
- Spinocerebellar ataxia 30
- Spinocerebellar Ataxia 31
- Spinocerebellar Ataxia And Plaque-Like Deposits
- Spinocerebellar ataxia, autosomal recessive 1
- Spinocerebellar Ataxia, Autosomal Recessive 7
- Spinocerebellar Ataxia, Autosomal Recessive 8
- Spinocerebellar Ataxia, Autosomal Recessive 9
- Spinocerebellar Ataxia with Dysmorphism
- Spinocerebellar Ataxia with Epilepsy
- Spinocerebellar Ataxia With Rigidity And Peripheral Neuropathy
- Spinocerebellar Ataxia, X-Linked 1
- Spinocerebellar Ataxia, X-Linked 5
- Cerebral Palsy, Ataxic, Autosomal Recessive
- Cerebroretinal Microangiopathy with Calcifications and Cysts

- COACH syndrome
- Coenzyme Q10 Deficiency
- COENZYME Q10 DEFICIENCY, PRIMARY,
 1
- Deafness hyperuricemia neurologic ataxia
- Diaminopentanuria
- Episodic Ataxia, Type 7
- Episodic Ataxia
- Episodic Ataxia, Type 1
- Episodic Ataxia, Type 2
- Episodic Ataxia, Type 3
- Episodic Ataxia, Type 4
- Erythrokeratodermia with ataxia
- Fragile X Tremor Ataxia Syndrome
- Gait Ataxia
- Hypotonia, congenital nystagmus, ataxia and abnormal auditory brainstem response
- Joubert Syndrome 7
- <u>Leukodystrophy</u>, <u>Hypomyelinating</u>, <u>with</u> <u>Hypodontia and Hypogonadotropic</u> Hypogonadism
- Mental Retardation, X-Linked, Syndromic, Christianson Type
- <u>Muscular Atrophy, Ataxia, Retinitis</u>
 <u>Pigmentosa, and Diabetes Mellitus</u>
- Myokymia 1
- Paroxysmal Tonic Upgaze, Benign Childhood, With Ataxia
- <u>Partington X-linked mental retardation</u> syndrome

- Peripheral Neuropathy, Ataxia, Focal Necrotizing Encephalopathy, and Spongy Degeneration of Brain
- Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, And Cataract
- Posterior column ataxia with retinitis pigmentosa
- Reardon Wilson Cavanagh syndrome
- Richards-Rundle syndrome
- SPASTIC ATAXIA 4, AUTOSOMAL RECESSIVE
- Spastic Paraplegia, Ataxia, And Mental Retardation
- Spondyloepimetaphyseal dysplasia, Genevieve type
- Tapetoretinal Degeneration with Ataxia
- Treft Sanborn Carey syndrome
- Tremor of Intention, Ataxia, and Lipofuscinosis
- Tryptophanuria With Dwarfism
- Athetosis
 - Choreoathetosis, Familial Inverted
 - Choreoathetosis, Hypothyroidism, And Neonatal Respiratory Distress
 - Hhhh Syndrome
- Bobble-head doll syndrome
- Catalepsy
- Chorea
 - Chorea, Benign Familial
 - Chorea Gravidarum
 - Chorea, remitting with nystagmus and cataracts

- Choreoathetosis, Familial Inverted
- Choreoathetosis, Hypothyroidism, And Neonatal Respiratory Distress
- Costeff optic atrophy syndrome
- Generalized Epilepsy and Paroxysmal Dyskinesia
- Huntington Disease
 - Huntington Disease-Like 1
 - Huntington Disease-Like 3
 - Westphal disease
- Huntington Disease-Like 2
- Neuroacanthocytosis
 - Neuroacanthocytosis, Mcleod Type
- Paroxysmal nonkinesigenic dyskinesia
- Paroxysmal Nonkinesigenic Dyskinesia 2
- Choreoathetosis-Spasticity, Episodic
- Dyskinesia, Drug-Induced
- Dyskinesia, Familial, with Facial Myokymia
- Limb dystonia
 - Amyotrophic Dystonic Paraplegia
 - Cervical Dystonia, Primary
 - Dystonia 13, Torsion
 - <u>Dystonia, Dopa-Responsive, due to</u>
 <u>Sepiapterin Reductase Deficiency</u>
 - Dystonia with Cerebellar Atrophy
 - Epilepsy, rolandic with paroxysmal exerciseinduced dystonia and writer's cramp
 - Familial paroxysmal dystonia
 - Dystonia 18
 - <u>LEUKOENCEPHALOPATHY WITH</u>
 <u>DYSTONIA AND MOTOR NEUROPATHY</u>

- <u>Leukoencephalopathy with Dystonia and Motor Neuropathy, SCPx-Deficient</u>
- Marsden syndrome
- Jensen syndrome
- Torsion dystonia with onset in infancy
- Torticollis
 - Congenital torticollis
 - Contractures, Congenital, Torticollis, and Malignant Hyperthermia
 - Sandifer syndrome
 - Torticollis keloids cryptorchidism renal dysplasia
- Episodic Kinesigenic Dyskinesia 2
- Hyperkinesis
- Hypokinesia
 - German Syndrome
- Infantile convulsions and paroxysmal choreoathetosis, familial
- Mental Retardation, X-Linked, Syndromic 10
- Microcephaly pontocerebellar hypoplasia dyskinesia
- MIRROR MOVEMENTS 1
- Myoclonus
 - Branchial Myoclonus with Spastic
 Paraparesis and Cerebellar Ataxia
 - Convulsive Disorder, Familial, with Prenatal or Early Onset
 - EPILEPSY, PROGRESSIVE MYOCLONIC,
 4, WITH OR WITHOUT RENAL FAILURE
 - Feigenbaum Bergeron Richardson syndrome
 - Herrmann syndrome
 - Jankovic Rivera syndrome

- Myoclonus, Cerebellar Ataxia, and Deafness
- Nystagmus, Myoclonic
- Opsoclonus-Myoclonus Syndrome
 - Infantile polymyoclonus
- Paroxysmal Exertion-Induced Dyskinesia And Hemolytic Anemia
- Psychomotor Agitation
 - Akathisia, Drug-Induced
- Synkinesis
 - Kallmann Syndrome 2 with Bimanual Synkinesia
 - Levator-Medial Rectus Synkinesis
- Tics
- Tremor
 - Fragile X Tremor Ataxia Syndrome
 - Geniospasm
 - MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE
 - Primary orthostatic tremor
 - Tremor hereditary essential, 2
 - Tremor of Intention, Ataxia, and Lipofuscinosis
- Gait Disorders, Neurologic
 - Gait Apraxia
 - Gait Ataxia
- Meningism
- Neurobehavioral Manifestations
 - Anhedonia
 - Catatonia
 - Presenile dementia, Kraepelin type

- Communication Disorders
 - Childhood-Onset Fluency Disorder
 - Language Disorders
 - Agraphia
 - Anomia
 - Dyslexia
 - Dyslexia, Acquired
 - Alexia, Pure
 - Stormorken Syndrome
 - Giacheti Syndrome
 - Hyperlexia
 - Language Development Disorders
 - Guanidinoacetate methyltransferase deficiency
 - Mehes syndrome
 - MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND WITH OR WITHOUT AUTISTIC FEATURES
 - Speech Development, Delayed,
 With Facial Asymmetry,
 Strabismus, And Transverse
 Earlobe Crease
 - SPECIFIC LANGUAGE IMPAIRMENT 1
 - SPECIFIC LANGUAGE IMPAIRMENT 2
 - SPECIFIC LANGUAGE IMPAIRMENT 3
 - SPEECH-LANGUAGE DISORDER 1
 - Speech Disorders
 - Aphasia

- Aphasia, Broca
- Aphasia, Conduction
- Aphasia, Primary Progressive
 - Primary Progressive Nonfluent Aphasia
- Aphasia, Wernicke
- <u>Dysphasia</u>, <u>Familial</u>
 Developmental
- Arginine:Glycine
 Amidinotransferase Deficiency
- Articulation Disorders
 - Dysarthria
 - Auditory perceptual impairment
 - <u>Bahemuka Brown</u> syndrome
 - <u>Fitzsimmons-</u> Guilbert syndrome
 - Foix Chavany
 Marie syndrome
 - Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis
 - Worster Drought syndrome
- Atonic-Astatic Syndrome of Foerster
- CHROMOSOME 17p13.1 DELETION SYNDROME
- Echolalia
- Maxillofacial Dysostosis

- MENTAL RETARDATION, AUTOSOMAL DOMINANT 20
- Mutism
- Non-lissencephalic cortical dysplasia
- Opticocochleodentate
 Degeneration
- Rolandic Epilepsy, Mental Retardation, and Speech Dyspraxia, X-Linked
- Speech disturbance use of faulty phrasing and unrelated words
- Stuttering
 - STUTTERING, FAMILIAL PERSISTENT, 1
 - Stuttering, Familial Persistent 2
- Learning Disorders
 - Chromosome 10q26 Deletion Syndrome
 - CHROMOSOME 7q11.23 DELETION SYNDROME, DISTAL, 1.2-MB
 - Dyscalculia
 - Dyslexia
 - Dyslexia, Acquired
 - Alexia, Pure
 - Stormorken Syndrome
 - Epilepsy, X-Linked, with Variable Learning Disabilities and Behavior Disorders
 - Giacheti Syndrome
 - NF1 Microdeletion Syndrome

- Riddle Syndrome
- Slavotinek Pike Mills Hurst syndrome
- Specific Learning Disorder
- Social Communication Disorder
- Speech Sound Disorder
 - Speech-Sound Disorder, hereditary
- Confusion
 - Carnitine Acetyltransferase Deficiency
 - Delirium
- Consciousness Disorders
 - Unconsciousness
 - Coma
 - Persistent Vegetative State
 - Stupor
 - Syncope
 - Stoll Alembik Dott syndrome
 - Syncope, Vasovagal
 - Familial neurocardiogenic syncope
- Intellectual Disability
 - 15q24 Microdeletion
 - 16p11.2 Deletion Syndrome
 - Absent Eyebrows and Eyelashes with Mental Retardation
 - Acrodysostosis
 - Agonadism, XY, with Mental Retardation, Short Stature, Retarded Bone Age, and Multiple Extragenital Malformations
 - AICAR Transformylase Inosine Monophosphate Cyclohydrolase Deficiency
 - Akesson syndrome

- Alaninuria with Microcephaly, Dwarfism,
 Enamel Hypoplasia, and Diabetes Mellitus
- Al Gazali Aziz Salem syndrome
- Alopecia contractures dwarfism mental retardation
- Alopecia epilepsy oligophrenia syndrome of Moynahan
- Alopecia, epilepsy, pyorrhea, mental subnormality
- Alopecia-Mental Retardation Syndrome 1
- Alopecia-Mental Retardation Syndrome 2
- ALOPECIA-MENTAL RETARDATION SYNDROME 3
- Alopecia-Mental Retardation Syndrome with Convulsions and Hypergonadotropic Hypogonadism
- Alopecia, Neurologic Defects, and Endocrinopathy Syndrome
- Alpha-Thalassemia Mental Retardation Syndrome, Deletion-Type
- Alport Syndrome, Mental Retardation, Midface Hypoplasia, and Elliptocytosis
- Amino Aciduria with Mental Deficiency,
 Dwarfism, Muscular Dystrophy,
 Osteoporosis, and Acidosis
- Amyloidosis of Gingiva and Conjunctiva, with Mental Retardation
- Amyotrophic Dystonic Paraplegia
- Anemia, Congenital Hypoplastic, with <u>Multiple Congenital Anomalies-Mental</u> <u>Retardation Syndrome</u>
- Aniridia cerebellar ataxia mental deficiency
- Ansell Bywaters Elderking syndrome
- Aortic arch anomaly with peculiar facies and mental retardation

- Aphalangia, Partial, with Syndactyly and Duplication of Metatarsal IV
- Arachnodactyly ataxia cataract aminoaciduria mental retardation
- Arginine:Glycine Amidinotransferase Deficiency
- Arthrogryposis, distal, with hypopituitarism, mental retardation, and facial anomalies
- Arthrogryposis, Distal, with Mental Retardation and Characteristic Facies
- Aughton syndrome
- Aural Atresia, Multiple Congenital Anomalies, and Mental Retardation
- Baraitser Rodeck Garner syndrome
- Battaglia Neri syndrome
- BEAULIEU-BOYCOTT-INNES SYNDROME
- Behr syndrome
- Bellini Chiumello Rimoldi syndrome
- Biemond Syndrome II
- Biemond syndrome type 2
- Birk-Barel Mental Retardation Dysmorphism Syndrome
- Blepharophimosis syndrome Ohdo type
- Blepharophimosis with Facial and Genital Anomalies and Mental Retardation
- Bohring syndrome
- Boudhina Yedes Khiari syndrome
- Brain Anomalies, Retardation, Ectodermal Dysplasia, Skeletal Malformations, Hirschsprung Disease, Ear-Eye Anomalies, Cleft Palate-Cryptorchidism, And Kidney Dysplasia-Hypoplasia
- Brunner Syndrome

- Bullous Dystrophy, Hereditary Macular Type
- Camera Marugo Cohen syndrome
- CAHMR syndrome
- Cantalamessa Baldini Ambrosi syndrome
- Cantu Sanchez-Corona Fragoso syndrome
- Cartwright Nelson Fryns syndrome
- <u>Cataract, Congenital, with Mental Impairment</u> and Dentate Gyrus Atrophy
- <u>Cataracts, ataxia, short stature, and mental</u> retardation
- Cataracts, Congenital, with Sensorineural Deafness, Down Syndrome-Like Facial Appearance, Short Stature, and Mental Retardation
- Cephalin Lipidosis
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 2
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 3
- Cerebral Cavernous Malformations 2
- Cerebral Cavernous Malformations 3
- Cerebrocostomandibular Syndrome
- Cerebrofaciothoracic Dysplasia
- Cerebrooculofacioskeletal Syndrome 2
- Cerebrooculofacioskeletal Syndrome 4
- Cerebrooculonasal Syndrome
- Choroid plexus calcification with mental retardation
- CHROMOSOME 13q14 DELETION SYNDROME
- Chromosome 15q13.3 Microdeletion Syndrome

- Chromosome 15q26-Qter Deletion Syndrome
- CHROMOSOME 17p13.1 DELETION SYNDROME
- Chromosome 17q21.31 Deletion Syndrome
- Chromosome 18 Pericentric Inversion
- Chromosome 1q21.1 Duplication Syndrome
- Chromosome 1q43-Q44 Deletion Syndrome
- Chromosome 2q31.2 Deletion Syndrome
- Chromosome 2q32-Q33 Deletion Syndrome
- Chromosome 3q29 Deletion Syndrome
- CHROMOSOME 7q11.23 DELETION SYNDROME, DISTAL, 1.2-MB
- CHROMOSOME 8q21.11 DELETION SYNDROME
- Chromosome Xq28 Duplication Syndrome
- Chudley-Rozdilsky syndrome
- Cleft palate
- Coffin-Siris syndrome
- Coffin syndrome 1
- Cohen syndrome
- Coloboma, cleft lip-palate and mental retardation syndrome
- Coloboma-Obesity-Hypogenitalism-Mental Retardation Syndrome
- Coloboma, Uveal, with Cleft Lip and Palate and Mental Retardation
- Convulsive Disorder, Familial, with Prenatal or Early Onset
- Corpus Callosum, Agenesis of, with Mental Retardation, Ocular Coloboma, and Micrognathia

- Cortical Blindness, Retardation, and Postaxial Polydactyly
- Craniofaciofrontodigital Syndrome
- <u>Craniosynostosis Mental Retardation Clefting</u>
 Syndrome
- <u>Craniosynostosis-Mental Retardation</u>
 Syndrome of Lin and Gettig
- Cree Mental Retardation Syndrome
- Cri-du-Chat Syndrome
 - Chromosome 5, uniparental disomy
 - Chromosome 5, monosomy 5q35
 - Chromosome 5, trisomy 5p
 - Chromosome 5, trisomy 5pter p13 3
 - Chromosome 5, trisomy 5q
 - Monosomy 5p
- Cryohydrocytosis, Stomatin-Deficient, with Mental Retardation, Seizures, Cataracts, and Massive Hepatosplenomegaly
- Cubitus Valgus with Mental Retardation and Unusual Facies
- Curatolo Cilio Pessagno syndrome
- Cutis Verticis Gyrata and Mental Deficiency
- Cystic Fibrosis with Helicobacter Pylori Gastritis, Megaloblastic Anemia, and Subnormal Mentality
- Davis Lafer syndrome
- Deafness, Cochlear, with Myopia and Intellectual Impairment
- <u>Deafness</u>, <u>congenital onychodystrophy</u>, recessive form
- De Barsy syndrome
- De Lange Syndrome

- Brachmann-De Lange-Like Facial
 Changes With Microcephaly,
 Metatarsus Adductus, And
 Developmental Delay
- De Sanctis-Cacchione syndrome
- Devriendt syndrome
- <u>Diabetes Insipidus, Nephrogenic, with Mental</u>
 Retardation and Intracerebral Calcification
- Dicarboxylicaminoaciduria
- Digitorenocerebral Syndrome
- <u>Dislocated Elbows, Bowed Tibias, Scoliosis,</u>
 <u>Deafness, Cataract, Microcephaly, And</u>
 Mental Retardation
- Down syndrome
- Dubowitz syndrome
- Duker Weiss Siber syndrome
- Duplication 15q11-q13 Syndrome
- Dwarfism, Low-Birth-Weight Type, with Unresponsiveness to Growth Hormone
- Dyggve-Melchior-Clausen syndrome
- Dysequilibrium syndrome
- Dysmyelination With Jaundice
- <u>Ectodermal Dysplasia</u>, <u>Hypohidrotic</u>, <u>with</u>
 <u>Hypothyroidism and Agenesis of the Corpus</u>
 Callosum
- <u>Ectodermal dysplasia mental retardation</u> <u>syndactyly</u>
- Elliott Ludman Teebi syndrome
- Emanuel syndrome
- Emphysema, Congenital, With Deafness,
 Penoscrotal Web, And Mental Retardation

- Encephalopathy with Intracranial
 Calcification, Growth Hormone Deficiency,
 Microcephaly, and Retinal Degeneration
- Epidermolysis bullosa, late-onset localized junctional, with mental retardation
- Epilepsy, Female-Restricted, with Mental Retardation
- Epilepsy, Photogenic, with Spastic Diplegia and Mental Retardation
- Epilepsy telangiectasia
- Facial Abnormalities, Kyphoscoliosis, and Mental Retardation
- Faciocardiomelic Syndrome
- Fallot complex with severe mental and growth retardation
- Feingold Trainer syndrome
- Fg Syndrome 5
- <u>Fibromatosis</u>, <u>Gingival</u>, <u>with Hypertrichosis</u> and Mental Retardation
- Filippi syndrome
- Fine-Lubinsky syndrome
- Fitzsimmons-McLachlan-Gilbert syndrome
- Fitzsimmons Walson Mellor syndrome
- Fountain syndrome
- FRONTONASAL DYSPLASIA 3
- Fryns-Aftimos Syndrome
- Garret Tripp syndrome
- Genitopatellar Syndrome
- Goniodysgenesis-Mental Retardation-Short Stature Syndrome
- Growth and Developmental Retardation, Ocular Ptosis, Cardiac Defect, and Anal Atresia

- Growth and mental retardation, mandibulofacial dysostosis, microcephaly, and cleft palate
- Growth Deficiency and Mental Retardation with Facial Dysmorphism
- Growth Failure, Microcephaly, Mental Retardation, Cataracts, Large Joint Contractures, Osteoporosis, Cortical Dysplasia, and Cerebellar Atrophy
- Growth mental deficiency syndrome of Myhre
- Gurrieri Sammito Bellussi syndrome
- Hair defect with photosensitivity and mental retardation
- Hall Riggs mental retardation syndrome
- Harrod Doman Keele syndrome
- Haspeslagh Fryns Muelenaere syndrome
- Histidinemia
- Hittner Hirsch Kreh syndrome
- Holoprosencephaly, Ectrodactyly, and Bilateral Cleft Lip-Palate
- Hooft disease
- Hordnes Engebretsen Knudtson syndrome
- Hoyeraal Hreidarsson syndrome
- Hunter-McAlpine syndrome
- Hydronephrosis, Congenital, with Cleft Palate, Characteristic Facies, Hypotonia, and Mental Retardation
- Hydroxylysinuria
- Hyperleucine-Isoleucinemia
- Hyperlysinemia Due To Defect In Lysine Transport Into Mitochondria
- Hyperphosphatasia with Mental Retardation

- HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 1
- HYPERPHOSPHATASIA WITH MENTAL RETARDATION SYNDROME 3
- Hypertelorism, Severe, With Midface Prominence, Myopia, Mental Retardation, And Bone Fragility
- Hypertrichosis, hyperkeratosis, mental retardation, and distinctive facial features
- Hyperuricemia, Infantile, with Abnormal Behavior and Normal Hypoxanthine Guanine Phosphoribosyltransferase
- Hypogonadism, Male, With Mental Retardation And Skeletal Anomalies
- Hypogonadism with Low-Grade Mental Deficiency and Microcephaly
- Hypoparathyroidism-retardationdysmorphism syndrome
- Hypospadias-Mental Retardation Syndrome
- Hypotonia-Cystinuria Syndrome
- Ichthyosis and male hypogonadism
- <u>Ichthyosis, mental retardation, dwarfism, and renal impairment</u>
- Ichthyosis-Mental Retardation Syndrome with Large Keratohyalin Granules in the Skin
- Indolylacroyl Glycinuria with Mental Retardation
- Iris Coloboma with Ptosis, Hypertelorism, and Mental Retardation
- Jagell Holmgren Hofer syndrome
- Johanson Blizzard syndrome
- Joubert Syndrome 7
- Joubert Syndrome 9

- Kahrizi Syndrome
- Kaler Garrity Stern syndrome
- Kapur Toriello syndrome
- Karandikar Maria Kamble syndrome
- Katsantoni Papadakou Lagoyanni syndrome
- Kaufman oculocerebrofacial syndrome
- KBG syndrome
- Kleefstra Syndrome
- Koone Rizzo Elias syndrome
- Kosztolanyi syndrome
- Kozlowski-Krajewska syndrome
- Kozlowski Ouvrier syndrome
- Kozlowski Rafinski Klicharska syndrome
- Kuzniecky syndrome
- Lambert syndrome
- Lenz Majewski hyperostotic dwarfism
- <u>Leukomelanoderma</u>, <u>Infantilism</u>, <u>Mental</u>
 <u>Retardation</u>, <u>Hypodontia</u>, <u>Hypotrichosis</u>
- <u>Light Fixation Seizure Syndrome</u>
- <u>Limb Defects</u>, <u>Distal Transverse</u>, <u>with Mental</u>
 Retardation and Spasticity
- <u>Lipodystrophy, Generalized, with Mental</u> <u>Retardation, Deafness, Short Stature, and</u> <u>Slender Bones</u>
- Lissencephaly 3
- Lowry Maclean syndrome
- Lowry Wood syndrome
- Lubani Al Saleh Teebi syndrome
- Lynch Lee Murday syndrome
- Macrogyria, pseudobulbar palsy and mental retardation

- Macrosomia obesity macrocephaly ocular abnormalities
- Male pseudohermaphroditism-mental retardation syndrome, Verloes type
- Mandibulofacial Dysostosis with Mental Deficiency
- Marfanoid Mental Retardation Syndrome, Autosomal
- Marinesco-Sjogren-like syndrome (MSLS)
- <u>Martin-Probst Deafness-Mental Retardation</u>
 Syndrome
- Martsolf syndrome
- MASA (Mental Retardation, Aphasia, Shuffling Gait, Adducted Thumbs) Syndrome
- McDonough syndrome
- Mental and Growth Retardation with Amblyopia
- MENTAL RETARDATION, ANTERIOR
 MAXILLARY PROTRUSION, AND
 STRABISMUS
- Mental Retardation associated with Psoriasis
- Mental Retardation, Autosomal Dominant 1
- MENTAL RETARDATION, AUTOSOMAL DOMINANT 20
- Mental Retardation, Autosomal Dominant 3
- Mental Retardation, Autosomal Dominant 4
- Mental Retardation, Autosomal Dominant 5
- Mental Retardation, Autosomal Recessive 1
- Mental Retardation, Autosomal Recessive 10
- Mental Retardation, Autosomal Recessive 11
- Mental Retardation, Autosomal Recessive 12
- Mental Retardation, Autosomal Recessive 13

- MENTAL RETARDATION, AUTOSOMAL RECESSIVE 16
- Mental Retardation, Autosomal Recessive 2
- Mental Retardation, Autosomal Recessive 3
- Mental Retardation, Autosomal Recessive 4
- Mental Retardation, Autosomal Recessive 5
- Mental Retardation, Autosomal Recessive 6
- Mental Retardation, Autosomal Recessive 7
- Mental Retardation, Autosomal Recessive 8
- Mental Retardation, Autosomal Recessive 9
- Mental Retardation, Buenos Aires Type
- Mental Retardation, Fra12a Type
- Mental Retardation, Joint Hypermobility, And Skin Laxity, With Or Without Metabolic Abnormalities
- Mental retardation, keratoconus, febrile seizures, and sinoatrial block
- Mental retardation, macrocephaly, short stature and craniofacial dysmorphism
- Mental Retardation, Microcephaly, Epilepsy, And Coarse Face
- Mental Retardation, Microcephaly, Growth Retardation, Joint Contractures, and Facial Dysmorphism
- Mental retardation Mietens Weber type
- Mental Retardation, Severe, With Spasticity And Pigmentary Tapetoretinal Degeneration
- Mental Retardation, Short Stature, Facial Anomalies, and Joint Dislocations
- Mental Retardation, Skeletal Dysplasia, and Abducens Palsy
- Mental retardation Smith Fineman Myers type

- Mental retardation spasticity ectrodactyly
- Mental retardation syndrome, Belgian type
- MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND WITH OR WITHOUT AUTISTIC FEATURES
- Mental Retardation with Optic Atrophy, Facial <u>Dysmorphism</u>, Microcephaly, and Short <u>Stature</u>
- Mental Retardation with Spastic Paraplegia
- Mental retardation Wolff type
- Mental Retardation, X-Linked
 - Abidi X-linked mental retardation syndrome
 - Adrenoleukodystrophy
 - Peroxisomal ACYL-COA oxidase deficiency
 - PEROXISOME BIOGENESIS DISORDER 2B
 - Wells Jankovic syndrome
 - Aldred syndrome
 - Allan-Herndon-Dudley syndrome
 - Armfield X-Linked Mental Retardation Syndrome
 - Atkin syndrome
 - Arena syndrome
 - ATR-X syndrome
 - Borjeson-Forssman-Lehmann syndrome
 - Brooks-Wisniewski-Brown Syndrome
 - Chromosome Xp11.3 Deletion Syndrome
 - Coffin-Lowry Syndrome

- CK SYNDROME
- Clark-Baraitser syndrome
- Classical Lissencephalies and Subcortical Band Heterotopias
 - Band Heterotopia of Brain
 - Double cortex
 - <u>Lissencephaly and agenesis of corpus callosum</u>
 - Lissencephaly, X-Linked, 2
 - Subcortical Band Heterotopia, X-Linked
- Cowchock syndrome
- Creatine deficiency, X-linked
- Encephalopathy, Neonatal Severe,
 Due To Mecp2 Mutations
- <u>Faciogenital Dysplasia with Attention</u>
 <u>Deficit-Hyperactivity Disorder</u>
- Fragile X Syndrome
 - Fragile X Tremor Ataxia
 Syndrome
 - Primary Ovarian Insufficiency, Fragile X-Associated
 - Saul Wilkes Stevenson syndrome
- Glycogen Storage Disease Type IIb
- Lesch-Nyhan Syndrome
 - Lesch-Nyhan Syndrome, Neurologic Variant
- <u>Lubs X-linked mental retardation</u> syndrome
- Lujan Fryns syndrome
- Mental Retardation, X-Linked 45

- Mental Retardation, X-Linked 46
- MEHMO syndrome
- Menkes Kinky Hair Syndrome
 - Hair defect with photosensitivity and mental retardation
- Mental Retardation And Microcephaly
 With Pontine And Cerebellar
 Hypoplasia
- Mental retardation-hypotonic facies syndrome, x-linked, 1
- Mental retardation, X-linked 14
- Mental Retardation, X-Linked 1
- Mental Retardation, X-Linked 17
- Mental Retardation, X-Linked 19
- Mental Retardation, X-Linked 2
- Mental Retardation, X-Linked 20
- MENTAL RETARDATION, X-LINKED
 21
- Mental Retardation, X-Linked 23
- Mental Retardation, X-Linked 3
- Mental Retardation, X-Linked 30
- Mental Retardation, X-Linked 31
- Mental Retardation, X-Linked 34
- Mental Retardation, X-Linked 42
- Mental Retardation, X-Linked 47
- MENTAL RETARDATION, X-LINKED 49
- Mental Retardation, X-Linked 50
- Mental Retardation, X-Linked 52
- Mental Retardation, X-Linked 53
- Mental Retardation, X-Linked 58

- Mental Retardation, X-Linked 63
- Mental Retardation, X-Linked 72
- Mental Retardation, X-Linked 73
- Mental Retardation, X-Linked 77
- Mental Retardation, X-Linked 78
- Mental Retardation, X-Linked 81
- Mental Retardation, X-Linked 82
- Mental Retardation, X-Linked 84
- Mental Retardation, X-Linked 89
- Mental Retardation, X-Linked 9
- Mental Retardation, X-Linked 91
- Mental Retardation, X-Linked 92
- Mental Retardation, X-Linked 93
- Mental Retardation, X-Linked 94
- Mental Retardation, X-Linked 95
- MENTAL RETARDATION, X-LINKED 96
- Mental Retardation, X-Linked Nonsyndromic
- Mental retardation X-linked, South African type
- Mental Retardation, X-Linked, Syndromic 10
- Mental Retardation with Psychosis,
 Pyramidal Signs, and Macroorchidism
- Mental Retardation, X-Linked 16
- Mental Retardation, X-Linked 79
- Mental Retardation, X-Linked, Syndromic 14
- Mental Retardation, X-Linked 59

- Mental retardation X-linked syndromic
 <u>7</u>
- Mental Retardation, X-Linked, Syndromic 9
- Mental Retardation, X-Linked, Syndromic, Jarid1c-Related
- MENTAL RETARDATION, X-LINKED, SYNDROMIC, RAYMOND TYPE
- Mental Retardation, X-Linked, Syndromic, Ube2a-Related
- Mental Retardation, X-Linked, Syp-Related
- Mental Retardation, X-Linked, With Brachydactyly And Macroglossia
 - MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE
- Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance
- Mental Retardation, X-Linked, with Epilepsy
- Mental Retardation, X-Linked, with Isolated Growth Hormone Deficiency
- Mental Retardation, X-Linked, With Or Without Seizures, Arx-Related
- Mental Retardation, X-Linked, with Short Stature
- Mental Retardation, X-Linked, With Spasticity
- Microphthalmia, Syndromic 4
- Miles-Carpenter x-linked mental retardation syndrome
- Mucopolysaccharidosis II
- Opitz-Kaveggia syndrome

- Orofaciodigital syndrome, Shashi type
- Partington X-linked mental retardation syndrome
- Plagiocephaly and X-linked mental retardation
- Ppm-X Syndrome
- Prieto X-linked mental retardation syndrome
- <u>Pyruvate Dehydrogenase Complex</u>
 Deficiency Disease
 - <u>Phosphoenolpyruvate</u> carboxykinase 2 deficiency
 - Pyruvate Dehydrogenase E1-Beta Deficiency
 - Pyruvate Dehydrogenase E2 Deficiency
 - Pyruvate Dehydrogenase E3-Binding Protein Deficiency
- Renpenning syndrome 1
- Rett Syndrome
 - Epileptic Encephalopathy, Early Infantile, 2
 - Rett Syndrome, Atypical
 - <u>RETT SYNDROME</u>, CONGENITAL VARIANT
 - Rett Syndrome, Preserved Speech Variant
 - Rett Syndrome, Zappella Variant
- Roifman syndrome
- Schimke X-linked mental retardation syndrome
- Siderius X-linked mental retardation syndrome

- Snyder Robinson syndrome
- Stocco dos Santos syndrome
- Tranebjaerg Svejgaard syndrome
- Wilson-Turner X-linked mental retardation syndrome
- X-linked mental retardation Gustavson type
- X-linked mental retardation type Wittwer
- Mental Retardation, X-Linked, Syndromic 12
- Mental Retardation, X-Linked, Syndromic, Christianson Type
- Mental Retardation, X-Linked, Syndromic, Turner Type
- Mental Retardation, X-Linked, Syndromic,
 Zdhhc9-Related
- Mental Retardation, X-Linked, With Panhypopituitarism
- Mental Retardation, X-Linked, Znf711-Related
- Metaphyseal Dysostosis, Mental Retardation, and Conductive Deafness
- Methionine Malabsorption Syndrome
- Microcephalic primordial dwarfism Toriello type
- Microcephaly cervical spine fusion anomalies
- Microcephaly, corpus callosum dysgenesis and cleft lip-palate
- Microcephaly deafness syndrome
- Microcephaly, Facial Abnormalities, Micromelia, and Mental Retardation
- Microcephaly, Macrotia, And Mental Retardation

- Microcephaly seizures mental retardation heart disorders
- Microcephaly sparse hair mental retardation seizures
- Microcephaly with Mental Retardation and Digital Anomalies
- Microphthalmia and mental deficiency
- Mirhosseini-Holmes-Walton syndrome
- Jensen syndrome
- Mollica Pavone Antener syndrome
- MOMES Syndrome
- Morillo-Cucci Passarge syndrome
- MORM syndrome
- Mowat-Wilson syndrome
- Muscular Dystrophy, Congenital, associated with Calf Hypertrophy, Microcephaly, and Severe Mental Retardation
- Muscular Dystrophy, Congenital, plus Mental Retardation
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 1
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 2
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 3
- Muscular Dystrophy, Congenital, Type 1D
- Myotonia with Skeletal Abnormalities and Mental Retardation
- Nakamura Osame syndrome
- Neuhauser syndrome

- Neurofaciodigitorenal syndrome
- Neurologic Disease, Infantile Multisystem, with Osseous Fragility
- NF1 Microdeletion Syndrome
- NF1 Microduplication Syndrome
- Nicolaides Baraitser syndrome
- N syndrome
- Oculodigitoesophagoduodenal syndrome
- Oliver-McFarlane syndrome
- Oliver Syndrome
- Onychotrichodysplasia and neutropenia
- Ophthalmoplegia, Progressive, with Scrotal Tongue and Mental Deficiency
- Opitz trigonocephaly syndrome
- Osteolysis syndrome recessive
- PACHYGYRIA WITH MENTAL RETARDATION, SEIZURES, AND ARACHNOID CYSTS
- Palant cleft palate syndrome
- Pallister W syndrome
- Parastremmatic dwarfism
- Parkinsonism, early onset with mental retardation
- Pashayan syndrome
- Patella hypoplasia mental retardation
- Pavone Fiumara Rizzo syndrome
- Perisylvian syndrome
- Perniola Krajewska Carnevale syndrome
- Pfeiffer Kapferer syndrome
- Pfeiffer Mayer syndrome
- Pfeiffer Tietze Welte syndrome

- Pilotto syndrome
- Pitt-Hopkins syndrome
- Piussan Lenaerts Mathieu syndrome
- Prader-Willi Syndrome
 - Prader-Willi habitus, osteopenia, and camptodactyly
 - Prader-Willi-Like Syndrome
 Associated With Chromosome 6
- Primrose syndrome
- Prolonged Bleeding Time, Brachydactyly, and Mental Retardation
- Proud Syndrome
- Prune Belly Syndrome with Pulmonic
 Stenosis, Mental Retardation, and Deafness
- Pseudoaminopterin syndrome
- Pseudouridinuria and Mental Defect
- Pterygium colli mental retardation digital anomalies
- Qazi Markouizos syndrome
- Radioulnar synostosis retinal pigment abnormalities
- Ramon Syndrome
- Ramos Arroyo Clark syndrome
- Reardon Wilson Cavanagh syndrome
- Renal Tubular Acidosis, Proximal, With Ocular Abnormalities And Mental Retardation
- Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism
- Richards-Rundle syndrome
- Robin Sequence with Distinctive Facial Appearance and Brachydactyly

- Rolandic Epilepsy, Mental Retardation, And Speech Dyspraxia, Autosomal Dominant
- Rolandic Epilepsy, Mental Retardation, and Speech Dyspraxia, X-Linked
- Rubinstein-Taybi Syndrome
 - Chromosome 16p13.3 Deletion Syndrome
 - Rubinstein Taybi like syndrome
- Rud Syndrome
- Ruzicka Goerz Anton syndrome
- Sammartino De Crecchio Syndrome
- Sao Paulo MCA-MR Syndrome
- Scaphocephaly, Maxillary Retrusion, And Mental Retardation
- SCARF syndrome
- Schinzel-Giedion syndrome
- Schofer Beetz Bohl syndrome
- Scholte syndrome
- Schrander-Stumpel Theunissen Hulsmans syndrome
- Sclerosing bone dysplasia mental retardation
- Scott Bryant Graham syndrome
- Seckel Syndrome 3
- SECKEL SYNDROME 4
- Seemanova Lesny syndrome
- SeSAME syndrome
- Short Stature, Mental Retardation, Callosal Agenesis, Heminasal Hypoplasia, Microphthalmia, And Atypical Clefting
- Simpson-Golabi-Behmel syndrome
- Singh Chhaparwal Dhanda syndrome

- Skeletal Defects, Genital Hypoplasia, And Mental Retardation
- Sketetal dysplasia coarse facies mental retardation
- Spastic Ataxia
- Spastic diplegia infantile type
- Spastic paraplegia 14, autosomal recessive
- Spastic Paraplegia 18, Autosomal Recessive
- Spastic Paraplegia 32, Autosomal Recessive
- Spastic Paraplegia, Ataxia, And Mental Retardation
- Spastic paraplegia epilepsy mental retardation
- Spastic Paraplegia, Sensorineural Deafness, Mental Retardation, And Progressive Nephropathy
- Spastic Paresis, Glaucoma, and Mental Retardation
- Spastic Quadriplegia, Retinitis Pigmentosa, and Mental Retardation
- Spinal Muscular Atrophy with Mental Retardation
- Spinal Muscular Atrophy with Microcephaly and Mental Subnormality
- Spondyloepimetaphyseal dysplasia, Genevieve type
- Spondyloepiphyseal Dysplasia Tarda with Mental Retardation
- Spondyloepiphyseal Dysplasia With Coronal Craniosynostosis, Cataracts, Cleft Palate, And Mental Retardation
- Stevenson-Carey Syndrome
- Sucrosuria, Hiatus Hernia and Mental Retardation

- SUPERNUMERARY DER(22)t(8)
- Tamari Goodman syndrome
- Temple-Baraitser Syndrome
- Temtamy preaxial brachydactyly syndrome
- Tetrasomy X
- Tonoki syndrome
- Trichodental syndrome
- TRICHOTHIODYSTROPHY 1, PHOTOSENSITIVE
- Tryptophanuria With Dwarfism
- Tsukahara Syndrome
- Ulna hypoplasia with mental retardation
- Ulnar Hypoplasia with Mental Retardation
- Upton Young syndrome
- Van Bogaert-Hozay syndrome
- Van Den Bosch Syndrome
- Van Maldergem Wetzburger Verloes syndrome
- Vasquez Hurst Sotos syndrome
- Verloes Gillerot Fryns syndrome
- Viljoen Kallis Voges syndrome
- <u>Vitiligo, Progressive, with Mental Retardation</u> and <u>Urethral Duplication</u>
- Volcke Soekarman syndrome
- WAGR Syndrome
 - Chromosome 11p Deletion Syndrome
 - Wilms Tumor, Aniridia, Genitourinary Anomalies, Mental Retardation, and Obesity Syndrome
- Walker Dyson syndrome
- Warburg Sio Fledelius syndrome

- Warburton Anyane Yeboa syndrome
- Wiedemann Grosse Dibbern syndrome
- Wiedemann Oldigs Oppermann syndrome
- Williams Syndrome
 - Williams-Beuren Region Duplication Syndrome
- Winship Viljoen Leary syndrome
- Worster Drought syndrome
- Woodhouse Sakati syndrome
- Yorifuji Okuno syndrome
- Young Hughes syndrome
- Young Simpson syndrome
- Zazam Sheriff Phillips syndrome
- Zechi-Ceide Syndrome
- Zerres Rietschel Majewski syndrome
- Zlotogora-Ogur syndrome
- Zunich neuroectodermal syndrome
- 15g24 Microdeletion
- 16p11.2 Deletion Syndrome
- Absent Eyebrows and Eyelashes with Mental Retardation
- Acrodysostosis
- Agonadism, XY, with Mental Retardation, Short Stature, Retarded Bone Age, and Multiple Extragenital Malformations
- AICAR Transformylase Inosine Monophosphate Cyclohydrolase Deficiency
- Akesson syndrome
- Alaninuria with Microcephaly, Dwarfism,
 Enamel Hypoplasia, and Diabetes Mellitus
- Al Gazali Aziz Salem syndrome

- Alopecia contractures dwarfism mental retardation
- Alopecia epilepsy oligophrenia syndrome of Moynahan
- Alopecia, epilepsy, pyorrhea, mental subnormality
- Alopecia-Mental Retardation Syndrome 1
- Alopecia-Mental Retardation Syndrome 2
- ALOPECIA-MENTAL RETARDATION SYNDROME 3
- Alopecia-Mental Retardation Syndrome with Convulsions and Hypergonadotropic Hypogonadism
- Alopecia, Neurologic Defects, and Endocrinopathy Syndrome
- Alpha-Thalassemia Mental Retardation Syndrome, Deletion-Type
- Alport Syndrome, Mental Retardation, Midface Hypoplasia, and Elliptocytosis
- Amino Aciduria with Mental Deficiency, Dwarfism, Muscular Dystrophy, Osteoporosis, and Acidosis
- Amyloidosis of Gingiva and Conjunctiva, with Mental Retardation
- Amyotrophic Dystonic Paraplegia
- Anemia, Congenital Hypoplastic, with <u>Multiple Congenital Anomalies-Mental</u> Retardation Syndrome
- Aniridia cerebellar ataxia mental deficiency
- Ansell Bywaters Elderking syndrome
- Aortic arch anomaly with peculiar facies and mental retardation
- Aphalangia, Partial, with Syndactyly and Duplication of Metatarsal IV

- Arachnodactyly ataxia cataract aminoaciduria mental retardation
- Arginine:Glycine Amidinotransferase Deficiency
- Arthrogryposis, distal, with hypopituitarism, mental retardation, and facial anomalies
- Arthrogryposis, Distal, with Mental Retardation and Characteristic Facies
- Aughton syndrome
- Aural Atresia, Multiple Congenital Anomalies, and Mental Retardation
- Baraitser Rodeck Garner syndrome
- Battaglia Neri syndrome
- BEAULIEU-BOYCOTT-INNES SYNDROME
- Behr syndrome
- Bellini Chiumello Rimoldi syndrome
- Biemond Syndrome II
- Biemond syndrome type 2
- Birk-Barel Mental Retardation Dysmorphism Syndrome
- Blepharophimosis syndrome Ohdo type
- Blepharophimosis with Facial and Genital Anomalies and Mental Retardation
- Bohring syndrome
- Boudhina Yedes Khiari syndrome
- Brain Anomalies, Retardation, Ectodermal Dysplasia, Skeletal Malformations,
 Hirschsprung Disease, Ear-Eye Anomalies,
 Cleft Palate-Cryptorchidism, And Kidney
 Dysplasia-Hypoplasia
- Brunner Syndrome
- Bullous Dystrophy, Hereditary Macular Type
- Camera Marugo Cohen syndrome

- CAHMR syndrome
- Cantalamessa Baldini Ambrosi syndrome
- Cantu Sanchez-Corona Fragoso syndrome
- Cartwright Nelson Fryns syndrome
- Cataract, Congenital, with Mental Impairment and Dentate Gyrus Atrophy
- <u>Cataracts, ataxia, short stature, and mental</u> retardation
- Cataracts, Congenital, with Sensorineural Deafness, Down Syndrome-Like Facial Appearance, Short Stature, and Mental Retardation
- Cephalin Lipidosis
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 2
- Cerebellar Ataxia, Mental Retardation, And Dysequilibrium Syndrome 3
- Cerebral Cavernous Malformations 2
- Cerebral Cavernous Malformations 3
- Cerebrocostomandibular Syndrome
- Cerebrofaciothoracic Dysplasia
- Cerebrooculofacioskeletal Syndrome 2
- Cerebrooculofacioskeletal Syndrome 4
- Cerebrooculonasal Syndrome
- Choroid plexus calcification with mental retardation
- CHROMOSOME 13q14 DELETION SYNDROME
- Chromosome 15q13.3 Microdeletion Syndrome
- Chromosome 15q26-Qter Deletion Syndrome
- CHROMOSOME 17p13.1 DELETION SYNDROME

- Chromosome 17q21.31 Deletion Syndrome
- Chromosome 18 Pericentric Inversion
- Chromosome 1q21.1 Duplication Syndrome
- Chromosome 1q43-Q44 Deletion Syndrome
- Chromosome 2q31.2 Deletion Syndrome
- Chromosome 2g32-Q33 Deletion Syndrome
- Chromosome 3g29 Deletion Syndrome
- CHROMOSOME 7q11.23 DELETION SYNDROME, DISTAL, 1.2-MB
- CHROMOSOME 8q21.11 DELETION SYNDROME
- Chromosome Xq28 Duplication Syndrome
- Chudley-Rozdilsky syndrome
- Cleft palate
- Coffin-Siris syndrome
- Coffin syndrome 1
- Cohen syndrome
- Coloboma, cleft lip-palate and mental retardation syndrome
- Coloboma-Obesity-Hypogenitalism-Mental Retardation Syndrome
- Coloboma, Uveal, with Cleft Lip and Palate and Mental Retardation
- Convulsive Disorder, Familial, with Prenatal or Early Onset
- Corpus Callosum, Agenesis of, with Mental Retardation, Ocular Coloboma, and Micrognathia
- Cortical Blindness, Retardation, and Postaxial Polydactyly
- Craniofaciofrontodigital Syndrome

- <u>Craniosynostosis Mental Retardation Clefting</u>
 <u>Syndrome</u>
- Craniosynostosis-Mental Retardation
 Syndrome of Lin and Gettig
- Cree Mental Retardation Syndrome
- Cri-du-Chat Syndrome
- Cryohydrocytosis, Stomatin-Deficient, with Mental Retardation, Seizures, Cataracts, and Massive Hepatosplenomegaly
- Cubitus Valgus with Mental Retardation and Unusual Facies
- Curatolo Cilio Pessagno syndrome
- Cutis Verticis Gyrata and Mental Deficiency
- Cystic Fibrosis with Helicobacter Pylori Gastritis, Megaloblastic Anemia, and Subnormal Mentality
- Davis Lafer syndrome
- Deafness, Cochlear, with Myopia and Intellectual Impairment
- <u>Deafness</u>, <u>congenital onychodystrophy</u>, <u>recessive form</u>
- De Barsy syndrome
- De Lange Syndrome
- De Sanctis-Cacchione syndrome
- Devriendt syndrome
- <u>Diabetes Insipidus, Nephrogenic, with Mental</u>
 <u>Retardation and Intracerebral Calcification</u>
- Dicarboxylicaminoaciduria
- Digitorenocerebral Syndrome
- Dislocated Elbows, Bowed Tibias, Scoliosis, Deafness, Cataract, Microcephaly, And Mental Retardation
- Down syndrome

- Dubowitz syndrome
- Duker Weiss Siber syndrome
- Duplication 15q11-q13 Syndrome
- Dwarfism, Low-Birth-Weight Type, with Unresponsiveness to Growth Hormone
- Dyggve-Melchior-Clausen syndrome
- Dysequilibrium syndrome
- Dysmyelination With Jaundice
- Ectodermal Dysplasia, Hypohidrotic, with Hypothyroidism and Agenesis of the Corpus Callosum
- <u>Ectodermal dysplasia mental retardation</u> syndactyly
- Elliott Ludman Teebi syndrome
- Emanuel syndrome
- Emphysema, Congenital, With Deafness,
 Penoscrotal Web, And Mental Retardation
- Encephalopathy with Intracranial
 Calcification, Growth Hormone Deficiency,
 Microcephaly, and Retinal Degeneration
- Epidermolysis bullosa, late-onset localized iunctional, with mental retardation
- Epilepsy, Female-Restricted, with Mental Retardation
- Epilepsy, Photogenic, with Spastic Diplegia and Mental Retardation
- Epilepsy telangiectasia
- <u>Facial Abnormalities, Kyphoscoliosis, and</u>
 Mental Retardation
- Faciocardiomelic Syndrome
- Fallot complex with severe mental and growth retardation
- Feingold Trainer syndrome

- Fg Syndrome 5
- <u>Fibromatosis</u>, <u>Gingival</u>, <u>with Hypertrichosis</u> and Mental Retardation
- Filippi syndrome
- Fine-Lubinsky syndrome
- Fitzsimmons-McLachlan-Gilbert syndrome
- Fitzsimmons Walson Mellor syndrome
- Fountain syndrome
- FRONTONASAL DYSPLASIA 3
- Fryns-Aftimos Syndrome
- Garret Tripp syndrome
- Genitopatellar Syndrome
- Goniodysgenesis-Mental Retardation-Short Stature Syndrome
- Growth and Developmental Retardation, Ocular Ptosis, Cardiac Defect, and Anal Atresia
- Growth and mental retardation, mandibulofacial dysostosis, microcephaly, and cleft palate
- Growth Deficiency and Mental Retardation with Facial Dysmorphism
- Growth Failure, Microcephaly, Mental Retardation, Cataracts, Large Joint Contractures, Osteoporosis, Cortical Dysplasia, and Cerebellar Atrophy
- Growth mental deficiency syndrome of Myhre
- Gurrieri Sammito Bellussi syndrome
- Hair defect with photosensitivity and mental retardation
- Hall Riggs mental retardation syndrome
- Harrod Doman Keele syndrome
- Haspeslagh Fryns Muelenaere syndrome

- Histidinemia
- Hittner Hirsch Kreh syndrome
- Holoprosencephaly, Ectrodactyly, and Bilateral Cleft Lip-Palate
- Hooft disease
- Hordnes Engebretsen Knudtson syndrome
- Hoyeraal Hreidarsson syndrome
- Hunter-McAlpine syndrome
- Hydronephrosis, Congenital, with Cleft
 Palate, Characteristic Facies, Hypotonia, and
 Mental Retardation
- Hydroxylysinuria
- Hyperleucine-Isoleucinemia
- Hyperlysinemia Due To Defect In Lysine Transport Into Mitochondria
- Hyperphosphatasia with Mental Retardation
- Hypertelorism, Severe, With Midface Prominence, Myopia, Mental Retardation, And Bone Fragility
- Hypertrichosis, hyperkeratosis, mental retardation, and distinctive facial features
- Hyperuricemia, Infantile, with Abnormal Behavior and Normal Hypoxanthine Guanine Phosphoribosyltransferase
- Hypogonadism, Male, With Mental Retardation And Skeletal Anomalies
- Hypogonadism with Low-Grade Mental Deficiency and Microcephaly
- Hypoparathyroidism-retardationdysmorphism syndrome
- Hypospadias-Mental Retardation Syndrome
- Hypotonia-Cystinuria Syndrome
- Ichthyosis and male hypogonadism

- Ichthyosis, mental retardation, dwarfism, and renal impairment
- Ichthyosis-Mental Retardation Syndrome with Large Keratohyalin Granules in the Skin
- Indolylacroyl Glycinuria with Mental Retardation
- Iris Coloboma with Ptosis, Hypertelorism, and Mental Retardation
- Jagell Holmgren Hofer syndrome
- Johanson Blizzard syndrome
- Joubert Syndrome 7
- Joubert Syndrome 9
- Kahrizi Syndrome
- Kaler Garrity Stern syndrome
- Kapur Toriello syndrome
- Karandikar Maria Kamble syndrome
- Katsantoni Papadakou Lagoyanni syndrome
- Kaufman oculocerebrofacial syndrome
- KBG syndrome
- Kleefstra Syndrome
- Koone Rizzo Elias syndrome
- Kosztolanyi syndrome
- Kozlowski-Krajewska syndrome
- Kozlowski Ouvrier syndrome
- Kozlowski Rafinski Klicharska syndrome
- Kuzniecky syndrome
- Lambert syndrome
- Lenz Majewski hyperostotic dwarfism
- <u>Leukomelanoderma, Infantilism, Mental</u> <u>Retardation, Hypodontia, Hypotrichosis</u>
- Light Fixation Seizure Syndrome

- <u>Limb Defects</u>, <u>Distal Transverse</u>, <u>with Mental Retardation and Spasticity</u>
- <u>Lipodystrophy, Generalized, with Mental</u> <u>Retardation, Deafness, Short Stature, and</u> Slender Bones
- Lissencephaly 3
- Lowry Maclean syndrome
- Lowry Wood syndrome
- Lubani Al Saleh Teebi syndrome
- Lynch Lee Murday syndrome
- <u>Macrogyria</u>, <u>pseudobulbar palsy and mental</u> <u>retardation</u>
- Macrosomia obesity macrocephaly ocular abnormalities
- Male pseudohermaphroditism-mental retardation syndrome, Verloes type
- Mandibulofacial Dysostosis with Mental Deficiency
- Marfanoid Mental Retardation Syndrome, Autosomal
- Marinesco-Sjogren-like syndrome (MSLS)
- <u>Martin-Probst Deafness-Mental Retardation</u>
 Syndrome
- Martsolf syndrome
- MASA (Mental Retardation, Aphasia, Shuffling Gait, Adducted Thumbs) Syndrome
- McDonough syndrome
- Mental and Growth Retardation with Amblyopia
- MENTAL RETARDATION, ANTERIOR MAXILLARY PROTRUSION, AND STRABISMUS
- Mental Retardation associated with Psoriasis

- Mental Retardation, Autosomal Dominant 1
- MENTAL RETARDATION, AUTOSOMAL DOMINANT 20
- Mental Retardation, Autosomal Dominant 3
- Mental Retardation, Autosomal Dominant 4
- Mental Retardation, Autosomal Dominant 5
- Mental Retardation, Autosomal Recessive 1
- Mental Retardation, Autosomal Recessive 10
- Mental Retardation, Autosomal Recessive 11
- Mental Retardation, Autosomal Recessive 12
- Mental Retardation, Autosomal Recessive 13
- MENTAL RETARDATION, AUTOSOMAL RECESSIVE 16
- Mental Retardation, Autosomal Recessive 2
- Mental Retardation, Autosomal Recessive 3
- Mental Retardation, Autosomal Recessive 4
- Mental Retardation, Autosomal Recessive 5
- Mental Retardation, Autosomal Recessive 6
- Mental Retardation, Autosomal Recessive 7
- Mental Retardation, Autosomal Recessive 8
- Mental Retardation, Autosomal Recessive 9
- Mental Retardation, Buenos Aires Type
- Mental Retardation, Fra12a Type
- Mental Retardation, Joint Hypermobility, And Skin Laxity, With Or Without Metabolic Abnormalities
- Mental retardation, keratoconus, febrile seizures, and sinoatrial block
- Mental retardation, macrocephaly, short stature and craniofacial dysmorphism

- Mental Retardation, Microcephaly, Epilepsy, And Coarse Face
- Mental Retardation, Microcephaly, Growth Retardation, Joint Contractures, and Facial Dysmorphism
- Mental retardation Mietens Weber type
- Mental Retardation, Severe, With Spasticity And Pigmentary Tapetoretinal Degeneration
- Mental Retardation, Short Stature, Facial Anomalies, and Joint Dislocations
- Mental Retardation, Skeletal Dysplasia, and Abducens Palsy
- Mental retardation Smith Fineman Myers type
- Mental retardation spasticity ectrodactyly
- Mental retardation syndrome, Belgian type
- MENTAL RETARDATION WITH LANGUAGE IMPAIRMENT AND WITH OR WITHOUT AUTISTIC FEATURES
- Mental Retardation with Optic Atrophy, Facial Dysmorphism, Microcephaly, and Short Stature
- Mental Retardation with Spastic Paraplegia
- Mental retardation Wolff type
- Mental Retardation, X-Linked
- Mental Retardation, X-Linked, Syndromic 12
- Mental Retardation, X-Linked, Syndromic, Christianson Type
- Mental Retardation, X-Linked, Syndromic, Turner Type
- Mental Retardation, X-Linked, Syndromic, Zdhhc9-Related
- Mental Retardation, X-Linked, With Panhypopituitarism

- Mental Retardation, X-Linked, Znf711-Related
- Metaphyseal Dysostosis, Mental Retardation, and Conductive Deafness
- Methionine Malabsorption Syndrome
- Microcephalic primordial dwarfism Toriello type
- Microcephaly cervical spine fusion anomalies
- Microcephaly, corpus callosum dysgenesis and cleft lip-palate
- Microcephaly deafness syndrome
- Microcephaly, Facial Abnormalities, Micromelia, and Mental Retardation
- Microcephaly, Macrotia, And Mental Retardation
- <u>Microcephaly seizures mental retardation</u> heart disorders
- Microcephaly sparse hair mental retardation seizures
- Microcephaly with Mental Retardation and Digital Anomalies
- Microphthalmia and mental deficiency
- Mirhosseini-Holmes-Walton syndrome
- Jensen syndrome
- Mollica Pavone Antener syndrome
- MOMES Syndrome
- Morillo-Cucci Passarge syndrome
- MORM syndrome
- Mowat-Wilson syndrome
- Muscular Dystrophy, Congenital, associated with Calf Hypertrophy, Microcephaly, and Severe Mental Retardation

- Muscular Dystrophy, Congenital, plus Mental Retardation
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 1
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 2
- MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (CONGENITAL WITH MENTAL RETARDATION), TYPE B, 3
- Muscular Dystrophy, Congenital, Type 1D
- Myotonia with Skeletal Abnormalities and Mental Retardation
- Nakamura Osame syndrome
- Neuhauser syndrome
- Neurofaciodigitorenal syndrome
- Neurologic Disease, Infantile Multisystem, with Osseous Fragility
- NF1 Microdeletion Syndrome
- NF1 Microduplication Syndrome
- Nicolaides Baraitser syndrome
- N syndrome
- Oculodigitoesophagoduodenal syndrome
- Oliver-McFarlane syndrome
- Oliver Syndrome
- Onychotrichodysplasia and neutropenia
- Ophthalmoplegia, Progressive, with Scrotal Tongue and Mental Deficiency
- Opitz trigonocephaly syndrome
- Osteolysis syndrome recessive

- PACHYGYRIA WITH MENTAL RETARDATION, SEIZURES, AND ARACHNOID CYSTS
- Palant cleft palate syndrome
- Pallister W syndrome
- Parastremmatic dwarfism
- Parkinsonism, early onset with mental retardation
- Pashayan syndrome
- Patella hypoplasia mental retardation
- Pavone Fiumara Rizzo syndrome
- Perisylvian syndrome
- Perniola Krajewska Carnevale syndrome
- Pfeiffer Kapferer syndrome
- Pfeiffer Mayer syndrome
- Pfeiffer Tietze Welte syndrome
- Pilotto syndrome
- Pitt-Hopkins syndrome
- Piussan Lenaerts Mathieu syndrome
- Prader-Willi Syndrome
- Primrose syndrome
- Prolonged Bleeding Time, Brachydactyly, and Mental Retardation
- Proud Syndrome
- Prune Belly Syndrome with Pulmonic
 Stenosis, Mental Retardation, and Deafness
- Pseudoaminopterin syndrome
- Pseudouridinuria and Mental Defect
- <u>Pterygium colli mental retardation digital anomalies</u>
- Qazi Markouizos syndrome

- Radioulnar synostosis retinal pigment abnormalities
- Ramon Syndrome
- Ramos Arroyo Clark syndrome
- Reardon Wilson Cavanagh syndrome
- Renal Tubular Acidosis, Proximal, With Ocular Abnormalities And Mental Retardation
- Retinitis Pigmentosa, Deafness, Mental Retardation, and Hypogonadism
- Richards-Rundle syndrome
- Robin Sequence with Distinctive Facial Appearance and Brachydactyly
- Rolandic Epilepsy, Mental Retardation, And Speech Dyspraxia, Autosomal Dominant
- Rolandic Epilepsy, Mental Retardation, and Speech Dyspraxia, X-Linked
- Rubinstein-Taybi Syndrome
- Rud Syndrome
- Ruzicka Goerz Anton syndrome
- Sammartino De Crecchio Syndrome
- Sao Paulo MCA-MR Syndrome
- Scaphocephaly, Maxillary Retrusion, And Mental Retardation
- SCARF syndrome
- Schinzel-Giedion syndrome
- Schofer Beetz Bohl syndrome
- Scholte syndrome
- Schrander-Stumpel Theunissen Hulsmans syndrome
- Sclerosing bone dysplasia mental retardation
- Scott Bryant Graham syndrome
- Seckel Syndrome 3

- SECKEL SYNDROME 4
- Seemanova Lesny syndrome
- SeSAME syndrome
- Short Stature, Mental Retardation, Callosal Agenesis, Heminasal Hypoplasia, Microphthalmia, And Atypical Clefting
- Simpson-Golabi-Behmel syndrome
- Singh Chhaparwal Dhanda syndrome
- Skeletal Defects, Genital Hypoplasia, And Mental Retardation
- Sketetal dysplasia coarse facies mental retardation
- Spastic Ataxia
- Spastic diplegia infantile type
- Spastic paraplegia 14, autosomal recessive
- Spastic Paraplegia 18, Autosomal Recessive
- Spastic Paraplegia 32, Autosomal Recessive
- Spastic Paraplegia, Ataxia, And Mental Retardation
- Spastic paraplegia epilepsy mental retardation
- Spastic Paraplegia, Sensorineural Deafness, Mental Retardation, And Progressive Nephropathy
- Spastic Paresis, Glaucoma, and Mental Retardation
- Spastic Quadriplegia, Retinitis Pigmentosa, and Mental Retardation
- Spinal Muscular Atrophy with Mental Retardation
- Spinal Muscular Atrophy with Microcephaly and Mental Subnormality

- Spondyloepimetaphyseal dysplasia, Genevieve type
- Spondyloepiphyseal Dysplasia Tarda with Mental Retardation
- Spondyloepiphyseal Dysplasia With Coronal Craniosynostosis, Cataracts, Cleft Palate, And Mental Retardation
- Stevenson-Carey Syndrome
- Sucrosuria, Hiatus Hernia and Mental Retardation
- SUPERNUMERARY DER(22)t(8
- Tamari Goodman syndrome
- Temple-Baraitser Syndrome
- Temtamy preaxial brachydactyly syndrome
- Tetrasomy X
- Tonoki syndrome
- Trichodental syndrome
- TRICHOTHIODYSTROPHY 1, PHOTOSENSITIVE
- Tryptophanuria With Dwarfism
- Tsukahara Syndrome
- Ulna hypoplasia with mental retardation
- Ulnar Hypoplasia with Mental Retardation
- Upton Young syndrome
- Van Bogaert-Hozay syndrome
- Van Den Bosch Syndrome
- Van Maldergem Wetzburger Verloes syndrome
- Vasquez Hurst Sotos syndrome
- Verloes Gillerot Frvns syndrome
- Viljoen Kallis Voges syndrome

- Vitiligo, Progressive, with Mental Retardation and Urethral Duplication
- Volcke Soekarman syndrome
- WAGR Syndrome
- Walker Dyson syndrome
- Warburg Sjo Fledelius syndrome
- Warburton Anyane Yeboa syndrome
- Wiedemann Grosse Dibbern syndrome
- Wiedemann Oldigs Oppermann syndrome
- Williams Syndrome
- Winship Viljoen Leary syndrome
- Worster Drought syndrome
- Woodhouse Sakati syndrome
- Yorifuji Okuno syndrome
- Young Hughes syndrome
- Young Simpson syndrome
- Zazam Sheriff Phillips syndrome
- Zechi-Ceide Syndrome
- Zerres Rietschel Majewski syndrome
- Zlotogora-Ogur syndrome
- Zunich neuroectodermal syndrome
- Lethargy
- Memory Disorders
 - Amnesia
 - Alcohol Amnestic Disorder
 - Alcohol induced encephalopathy
 - Korsakoff Syndrome
 - Amnesia, Anterograde
 - Amnesia, Retrograde

- Amnesia, Transient Global
- Korsakoff Syndrome
- Perceptual Disorders
 - Agnosia
 - Gerstmann Syndrome
 - Primary visual agnosia
 - Prosopagnosia
 - Prosopagnosia, hereditary
 - Alice in Wonderland Syndrome
 - Allesthesia
 - Auditory Perceptual Disorders
 - Tune Deafness
 - Hallucinations
 - Illusions
 - Phantom Limb
 - Synesthesia
- Psychomotor Disorders
 - Aniridia, Partial, with Unilateral Renal Agenesis and Psychomotor Retardation
 - Apraxias
 - Alien Hand Syndrome
 - Alzheimer Disease, Familial, 3, with Spastic Paraparesis and Apraxia
 - Apraxia, Ideomotor
 - Apraxia, oculomotor, Cogan type
 - Early-onset ataxia with oculomotor apraxia and hypoalbuminemia
 - Gait Apraxia
 - Rolandic Epilepsy, Mental Retardation, And Speech Dyspraxia, Autosomal Dominant

- Rolandic Epilepsy, Mental Retardation, and Speech Dyspraxia, X-Linked
- Specific Language Impairment 4
- SPEECH-LANGUAGE DISORDER 1
- Wieacker syndrome
- Bowen-Conradi syndrome
- CHROMOSOME 3pter-p25 DELETION SYNDROME
- C SYNDROME
- De Hauwere Leroy Adriaenssens syndrome
- <u>Developmental Delay, Epilepsy, and</u>
 Neonatal Diabetes
- Diaminopentanuria
- <u>Dystonia, Dopa-Responsive, due to</u>
 <u>Sepiapterin Reductase Deficiency</u>
- Edinburgh Malformation Syndrome
- Fumaric aciduria
- Genitopatellar Syndrome
- Growth Retardation, Small and Puffy Hands and Feet, and Eczema
- Hypomyelination, Global Cerebral
- Kozlowski Rafinski Klicharska syndrome
- MacDermot Winter syndrome
- Megarbane syndrome
- Myelodysplasia, Immunodeficiency, Facial Dysmorphism, Short Stature, and Psychomotor Delay
- Phosphoglycerate Dehydrogenase Deficiency
- Phosphoserine Aminotransferase Deficiency
- Polyhydramnios, Megalencephaly, And Symptomatic Epilepsy

- Psychomotor Agitation
 - Akathisia, Drug-Induced
- Trigonobrachycephaly, Bulbous Bifid Nose, Micrognathia, and Abnormalities of the Hands and Feet
- Neurogenic Inflammation
- Neuromuscular Manifestations
 - Fasciculation
 - Muscle Cramp
 - Muscle Hypertonia
 - Muscle Hypotonia
 - Muscle Weakness
 - Muscular Atrophy
 - Myokymia
 - Myotonia
 - Spasm
 - Tetany
- Irritable heart
 - Orthostatic hypotension
 - Idiopathic orthostatic hypotension
 - Orthostatic Hypotensive Disorder, Streeten
 Type
 - Post-Exercise Hypotension
 - Postural Orthostatic Tachycardia Syndrome
 - Syncope, Vasovagal
 - Familial neurocardiogenic syncope
- Neurocirculatory Asthenia
 - Orthostatic hypotension
 - Idiopathic orthostatic hypotension

- Orthostatic Hypotensive Disorder, Streeten
 Type
- Post-Exercise Hypotension
- Postural Orthostatic Tachycardia Syndrome
- Syncope, Vasovagal
 - Familial neurocardiogenic syncope
- Pain
 - Abdominal Pain
 - Abdomen, Acute
 - Pelger-Huet-Like Anomaly and Episodic Fever with Abdominal Pain
 - Acute Pain
 - Arthralgia
 - Shoulder Pain
 - Back Pain
 - Failed Back Surgery Syndrome
 - Low Back Pain
 - Breakthrough Pain
 - Chest Pain
 - Angina Pectoris
 - Angina, Stable
 - Angina, Unstable
 - Angina Pectoris, Variant
 - Prinzmetal's variant angina
 - Microvascular Angina
 - Chronic Pain
 - Earache
 - Eye Pain
 - Facial Pain

- Flank Pain
- Glossalgia
- Headache
 - Hyperthermia, Cutaneous, With Headaches And Nausea
 - Slit Ventricle Syndrome
- Labor Pain
- Levator syndrome
- Mastodynia
- Metatarsalgia
- Musculoskeletal Pain
 - Myalgia
 - Pelvic Girdle Pain
- Neck Pain
- Paroxysmal Extreme Pain Disorder
- Neuropathic pain
 - Causalgia
 - Neuralgia, Postherpetic
 - Piriformis Muscle Syndrome
 - Hip socket neuropathy
 - Pudendal Neuralgia
 - Sciatica
 - Hip socket neuropathy
- Neuropathy, Painful
- Nociceptive Pain
 - Visceral Pain
- Pain, Intractable
- Pain, Postoperative
 - Phantom Limb

- Pain, Referred
- Pelvic Pain
 - Dysmenorrhea
 - Pelvic Girdle Pain
 - Piriformis Muscle Syndrome
 - Hip socket neuropathy
- Renal Colic
- Paralysis
- Paresis
 - Encephalopathy, Spastic Tetraparesis, and Hypogonadism
 - Hhhh Syndrome
 - Paraparesis
 - Paraparesis, Spastic
 - Alzheimer Disease, Familial, 3, with Spastic Paraparesis and Apraxia
 - Alzheimer Disease, Familial, 3, with Spastic Paraparesis and Unusual Plaques
 - Branchial Myoclonus with Spastic Paraparesis and Cerebellar Ataxia
 - Gemignani syndrome
 - Multiple Exostoses with Spastic Tetraparesis
 - Spastic Paresis, Glaucoma, and Mental Retardation
- Psychophysiologic Disorders
- Pupil Disorders
 - Anisocoria
 - Congenital Corneal Opacities, Cornea Guttata, and Corectopia
 - Ectopia Lentis with Ectopia of Pupil

- Ectopia pupillae
- McPherson Robertson Cammarano syndrome
- Microcoria, congenital
- Microphthalmia, Isolated, With Corectopia
- Miosis
- Mydriasis
- Pierson syndrome
- Ptosis, Strabismus, And Ectopic Pupils
- Tonic Pupil
- Reflex, Abnormal
 - Bahemuka Brown syndrome
 - CAPOS syndrome
 - External Ophthalmoplegia, Synergistic Divergence,
 Jaw Winking, and Oculocutaneous
 Hypopigmentation
 - Hyperekplexia and Epilepsy
 - Marcus Gunn phenomenon

Seizures

- Adams Nance syndrome
- Alcohol Withdrawal Seizures
- Alopecia-Mental Retardation Syndrome with Convulsions and Hypergonadotropic Hypogonadism
- Baraitser Rodeck Garner syndrome
- Cerebroretinal Microangiopathy with Calcifications and Cysts
- Chromosome 15q13.3 Microdeletion Syndrome
- Convulsive Disorder, Familial, with Prenatal or Early Onset
- Copper deficiency, familial benign

- Cryohydrocytosis, Stomatin-Deficient, with Mental Retardation, Seizures, Cataracts, and Massive Hepatosplenomegaly
- Dysmyelination With Jaundice
- EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 11
- EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 12
- EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 5
- EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, 7
- Hyper-Beta-Alaninemia
- Hyperleucine-Isoleucinemia
- HYPERMETHIONINEMIA DUE TO ADENOSINE KINASE DEFICIENCY
- Hyperphosphatemia, Polyuria, and Seizures
- Hypoparathyroidism-retardation-dysmorphism syndrome
- Hypotonia, Seizures, And Precocious Puberty
- Infantile convulsions and paroxysmal choreoathetosis, familial
- Mental retardation, keratoconus, febrile seizures, and sinoatrial block
- Mental Retardation, X-Linked 59
- Methionine Malabsorption Syndrome
- MICROCEPHALY, POSTNATAL PROGRESSIVE, WITH SEIZURES AND BRAIN ATROPHY
- MICROCEPHALY, SEIZURES, AND DEVELOPMENTAL DELAY
- Microcephaly seizures genital hypoplasia
- <u>Microcephaly seizures mental retardation heart</u> disorders

- Microcephaly sparse hair mental retardation seizures
- Muller Barth Menger syndrome
- PACHYGYRIA WITH MENTAL RETARDATION, SEIZURES, AND ARACHNOID CYSTS
- Partington X-linked mental retardation syndrome
- Perniola Krajewska Carnevale syndrome
- Phosphoglycerate Dehydrogenase Deficiency
- Phosphoserine Aminotransferase Deficiency
- Pyridoxamine 5-Prime-Phosphate Oxidase Deficiency
- Qazi Markouizos syndrome
- SeSAME syndrome
- Tranebjaerg Svejgaard syndrome
- Warman Mulliken Hayward syndrome
- X-linked mental retardation Gustavson type
- Sensation Disorders
 - Dizziness
 - Primary orthostatic tremor
 - Hearing Disorders
 - Hearing loss
 - Abidi X-linked mental retardation syndrome
 - Behr syndrome
 - Branchial arch syndrome X-linked
 - CATSHL syndrome
 - Chromosome 6pter-P24 Deletion Syndrome
 - Deafness
 - Albinism deafness syndrome

- Arthrogryposis multiplex with deafness, inguinal hernias, and early death
- Ayazi syndrome
- Branchiogenic-Deafness
 Syndrome
- Burn-Mckeown syndrome
- Cardioauditory syndrome of Sanchez Cascos
- Charcot-Marie-Tooth disease and deafness
- Corneal Degeneration,
 Ribbonlike, with Deafness
- Coxoauricular Syndrome
- Davenport Donlan syndrome
- Deaf-Blind Disorders
- DEAFNESS, AUTOSOMAL DOMINANT 22; DFNA22
 DEAFNESS, AUTOSOMAL DOMINANT 22, WITH HYPERTROPHIC CARDIOMYOPATHY,
- DEAFNESS, AUTOSOMAL DOMINANT 27
- DEAFNESS, AUTOSOMAL DOMINANT 33
- <u>Deafness, Autosomal Dominant</u>
 <u>4</u>
- DEAFNESS, AUTOSOMAL DOMINANT 50
- DEAFNESS, AUTOSOMAL DOMINANT 51
- <u>Deafness, Autosomal Recessive</u>
 18

- DEAFNESS, AUTOSOMAL RECESSIVE 25
- DEAFNESS, AUTOSOMAL RECESSIVE 36, WITH OR WITHOUT VESTIBULAR INVOLVEMENT
- DEAFNESS, AUTOSOMAL RECESSIVE 45
- DEAFNESS, AUTOSOMAL RECESSIVE 4, WITH ENLARGED VESTIBULAR AQUEDUCT
- DEAFNESS, AUTOSOMAL RECESSIVE 74
- DEAFNESS, AUTOSOMAL RECESSIVE 83
- DEAFNESS, AUTOSOMAL RECESSIVE 84A
- DEAFNESS, AUTOSOMAL RECESSIVE 85
- DEAFNESS, AUTOSOMAL RECESSIVE 89
- DEAFNESS, AUTOSOMAL RECESSIVE 91
- <u>Deafness, Cataract, Retinitis</u>
 <u>Pigmentosa, And Sperm</u>
 <u>Abnormalities</u>
- <u>Deafness, Congenital, and</u>
 Familial Myoclonic Epilepsy
- Deafness, Congenital, and Onychodystrophy, Autosomal Dominant
- <u>Deafness, congenital</u> <u>onychodystrophy, recessive</u> form
- <u>Deafness, Congenital, with</u>
 Vitiligo and Achalasia

- Deafness hyperuricemia neurologic ataxia
- DEAFNESS,
 NONSYNDROMIC, MODIFIER
 1
- DEAFNESS, Y-LINKED 1
- Dementia, familial Danish
- <u>Dislocated Elbows, Bowed</u>
 <u>Tibias, Scoliosis, Deafness,</u>
 <u>Cataract, Microcephaly, And</u>
 <u>Mental Retardation</u>
- Emphysema, Congenital, With Deafness, Penoscrotal Web, And Mental Retardation
- Epiphyseal Dysplasia of Femoral Head, Myopia, and Deafness
- Fine-Lubinsky syndrome
- Fountain syndrome
- Herrmann syndrome
- Hirschsprung Disease with Polydactyly, Renal Agenesis, and Deafness
- Hyperlipoproteinemia, Type II, and Deafness
- HYPERTELORISM,
 PREAURICULAR SINUS,
 PUNCTAL PITS, AND
 DEAFNESS
- Ichthyosiform erythroderma, corneal involvement, deafness
- Johnson neuroectodermal syndrome
- Jones syndrome
- Keratitis, Ichthyosis, and Deafness (KID) Syndrome

- Konigsmark Knox Hussels syndrome
- Lynch Lee Murday syndrome
- Mental retardation-hypotonic facies syndrome, x-linked, 1
- Meyenburg-Altherr-Uehlinger syndrome
- Microcephaly deafness syndrome
- MUCKLE-WELLS SYNDROME
- Myoclonic Epilepsy, Congenital Deafness, Macular Dystrophy, and Psychiatric Disorders
- Myoclonus, Cerebellar Ataxia, and Deafness
- Nasodigitoacoustic syndrome
- Nathalie syndrome
- Nephrosis deafness urinary tract digital malformation
- Noninsulin-dependent diabetes mellitus with deafness
- Nonsyndromic Deafness
- Opticocochleodentate
 Degeneration
- PERRAULT SYNDROME 3
- Ramos Arroyo Clark syndrome
- Retinitis Pigmentosa, X-Linked, And Sinorespiratory Infections, With Or Without Deafness
- Richards-Rundle syndrome
- Schimke X-linked mental retardation syndrome
- Schlegelberger Grote syndrome

- Secretory Diarrhea, Myopathy, and Deafness
- Spastic paraplegia 24
- Temtamy preaxial brachydactyly syndrome
- <u>Tibia, Absence of, with</u>
 Congenital Deafness
- Tietz syndrome
- Wells Jankovic syndrome
- Wright Dyck syndrome
- X-linked mental retardation Gustavson type
- Yemenite deaf-blind hypopigmentation syndrome
- <u>Deafness, Autosomal Dominant, Due</u>
 <u>To Mutation In Myo1a</u>
- <u>Deafness, Autosomal Recessive 36,</u>
 Without Vestibular Involvement
- <u>Deafness, Congenital Heart Defects,</u> and Posterior Embryotoxon
- Deafness-Craniofacial Syndrome
- <u>Deafness, Unilateral, With Delayed</u>
 Endolymphatic Hydrops
- <u>Deafness with Anhidrotic Ectodermal</u>
 <u>Dysplasia</u>
- Hearing Loss, Bilateral
- HEARING LOSS, CISPLATIN-INDUCED, SUSCEPTIBILITY TO
- Hearing Loss, Conductive
- Hearing Loss, Functional
- Hearing Loss, High-Frequency
- Hearing Loss, Mixed Conductive-Sensorineural

- Hearing Loss, Sensorineural
- Hearing Loss, Sudden
- Hearing Loss, Unilateral
- Iris dysplasia hypertelorism deafness
- <u>Lacrimoauriculodentodigital syndrome</u>
- Microcephaly, Growth Retardation, Cataract, Hearing Loss, And Unusual Appearance
- <u>Microtia, Hearing Impairment, And</u>
 Cleft Palate
- Myopathy, Mitochondrial Progressive, With Congenital Cataract, Hearing Loss, And Developmental Delay
- Optic Atrophy, Hearing Loss, and Peripheral Neuropathy, Autosomal Dominant
- Reardon Wilson Cavanagh syndrome
- Spondylomegaepiphyseal Dysplasia
 With Upper Limb Mesomelia, Punctate
 Calcifications, And Deafness
- Hyperacusis
- Ossicular Malformations, familial
- Tinnitus
 - Episodic Ataxia, Type 3
- Olfaction Disorders
 - Congenital anosmia
 - Johnson neuroectodermal syndrome
 - Musk, Inability to Smell
 - Neuropathy, Hereditary Sensory and Autonomic, Adult-Onset, with Anosmia
- Posterior column ataxia
- Somatosensory Disorders

- Taste Disorders
- Vision Disorders
- Sleep Wake Disorders
 - Autosomal Dominant Lateral Temporal Lobe
 Epilepsy
 - Dyssomnias
 - Sleep Deprivation
 - Sleep Disorders, Circadian Rhythm
 - Advanced Sleep-Phase Syndrome, Familial
 - Jet Lag Syndrome
 - Sleep Disorders, Intrinsic
 - Hypersomnia
 - Hyper-Beta-Alaninemia
 - Hypersomnolence, Idiopathic
 - Kleine-Levin Syndrome
 - Narcolepsy
 - Cataplexy
 - Cerebellar Ataxia,
 Deafness, and
 Narcolepsy
 - <u>Disseminated Sclerosis</u>
 <u>with Narcolepsy</u>
 - Irresistible sleepiness, cataplexy and onset of sleep in desynchronized phase
 - Narcolepsy 1
 - Nocturnal Myoclonus Syndrome
 - Restless legs syndrome
 - Restless legs syndrome 1
 - Restless legs syndrome 2

- Sleep Apnea Syndromes
 - Glaucoma and Sleep Apnea
 - Sleep Apnea, Central
 - Congenital central hypoventilation syndrome
 - Obstructive sleep apnea
 - Obesity Hypoventilation Syndrome
- Insomnia
 - Insomnia, Fatal Familial
- Parasomnias
 - Nocturnal Myoclonus Syndrome
 - Nocturnal Paroxysmal Dystonia
 - REM Sleep Parasomnias
 - REM Sleep Behavior Disorder
 - Sleep Paralysis
 - Restless legs syndrome
 - Restless legs syndrome 1
 - Restless legs syndrome 2
 - Sleep Arousal Disorders
 - Night Terrors
 - Somnambulism
 - Epilepsy, Nocturnal Frontal Lobe, Type 4
 - Sleep Bruxism
 - Faciomandibular myoclonus, nocturnal
 - Sleep-Wake Transition Disorders
- Sleep Deprivation
- Susac Syndrome
- Urinary Bladder, Neurogenic

- Vertigo
 - Benign Paroxysmal Positional Vertigo
 - Episodic Ataxia, Type 3
 - Vertigo, Benign Recurrent
 - Vertigo, Benign Recurrent, 1
 - Vertigo, Benign Recurrent, 2
- Voice Disorders
 - Aphonia
 - Dysphonia
 - <u>Laryngeal Abductor Paralysis with Cerebellar</u>
 <u>Ataxia and Motor Neuropathy</u>
 - Hoarseness
 - Asrar Facharzt Haque syndrome
 - Lipoid Proteinosis of Urbach and Wiethe
 - Dystonia musculorum deformans 4
- Oral Manifestations
 - Oral Hemorrhage
 - Gingival Hemorrhage
 - Bazopoulou Kyrkanidou syndrome
- Polydipsia
 - Polydipsia, Psychogenic
- Prodromal Symptoms
- Pseudophakia
- Renal Colic
- Reticulocytosis
- Signs and Symptoms, Digestive
 - Abdominal Pain
 - Abdomen, Acute
 - Pelger-Huet-Like Anomaly and Episodic Fever with Abdominal Pain

- Aerophagy
- Anorexia
 - FACES syndrome
- Dyspepsia
- Constipation
 - Anal sphincter dysplasia
 - Fg Syndrome 5
 - Opitz-Kaveggia syndrome
- Diarrhea
 - Bile Acid Malabsorption, Primary
 - Congenital chloride diarrhea
 - Diarrhea 3, Secretory Sodium, Congenital
 - <u>Diarrhea 3, Secretory Sodium, Congenital,</u>
 Syndromic
 - Diarrhea 4, Malabsorptive, Congenital
 - Diarrhea, Chronic, with Villous Atrophy
 - <u>Diarrhea, Glucose-Stimulated Secretory, with</u>
 Common Variable Immunodeficiency
 - Diarrhea, Infantile
 - Immune Dysregulation, Polyendocrinopathy, Enteropathy, X-Linked Syndrome
 - Satoyoshi syndrome
 - Secretory Diarrhea, Myopathy, and Deafness
 - Trehalase Deficiency
 - Vascular Hyalinosis
- Encopresis
 - Anal sphincter dysplasia
- Vomiting
 - Cyclic Vomiting Syndrome-Plus

- Cyclic Vomiting Syndrome with Neuromuscular <u>Disease</u>
- Familial cyclic vomiting syndrome
- Hematemesis
- Vomiting, Anticipatory
- Morning Sickness
- Postoperative Nausea and Vomiting
- Eructation
- Flatulence
- Gagging
- Halitosis
- Heartburn
- Hiccup
- Hyperphagia
- Nausea
- Signs and Symptoms, Respiratory
 - Anoxia
 - Fetal Hypoxia
 - Apnea
 - Butyrylcholinesterase deficiency
 - Congenital myasthenic syndrome with episodic apnea
 - Sleep Apnea Syndromes
 - Glaucoma and Sleep Apnea
 - Sleep Apnea, Central
 - Congenital central hypoventilation syndrome
 - Obstructive sleep apnea
 - Obesity Hypoventilation Syndrome
 - Cheyne-Stokes Respiration

- Cough
 - Neuropathy, Hereditary Sensory And Autonomic,
 Type I, With Cough And Gastroesophageal Reflux
- Dyspnea
 - Dyspnea, Paroxysmal
- Hemoptysis
- Hoarseness
 - Asrar Facharzt Haque syndrome
 - Lipoid Proteinosis of Urbach and Wiethe
- Hypercapnia
- Hyperoxia
- Hyperventilation
 - Alkalosis, Respiratory
 - Pitt-Hopkins syndrome
- Hypocapnia
- Hypoventilation
 - Congenital central hypoventilation syndrome
 - Obesity Hypoventilation Syndrome
 - Perry Syndrome
- Infantile Apparent Life-Threatening Event
- Mouth Breathing
- Respiratory Sounds
 - Snoring
 - Stridor, Congenital
- Sneezing
 - Autosomal dominant compelling helio ophthalmic outburst syndrome
 - Gastric Sneezing
- Tachypnea
 - Transient Tachypnea of the Newborn

- Skin Manifestations
 - Cafe-au-Lait Spots
 - Cafe au lait spots, multiple
 - Gastrocutaneous syndrome
 - Legius syndrome
 - WATSON SYNDROME
 - Ecchymosis
 - Jaundice
 - Deal Barratt Dillon syndrome
 - Dysmyelination With Jaundice
 - Jaundice, Obstructive
 - Anemia, hereditary spherocytic hemolytic
 - Jaundice, Familial Obstructive, of Infancy
 - Lutz Richner Landolt syndrome
 - Livedo Reticularis
 - Aortic Aneurysm, Familial Thoracic 6
 - Necrolytic Migratory Erythema
 - Pallor
 - Pruritus
 - Purpura
 - Striae Distensae
- Urological Manifestations
 - Hypercalciuria
 - Hypercalciuric Hypocalcemia, Familial
 - Hypercalciuria, Absorptive, 1
 - Hypercalciuria, Absorptive, 2
 - Hypercalciuria, childhood idiopathic
 - Hypomagnesemia primary

- Hypophosphatemic Rickets with Hypercalciuria, Hereditary
- Lower Urinary Tract Symptoms
 - Dysuria
 - Prostatism
 - Nocturia
 - Urinary Bladder, Overactive
 - Urinary Incontinence
 - Urinary Incontinence, Stress
 - Urinary Incontinence, Urge
- Oliguria
- Polyuria
 - Hyperphosphatemia, Polyuria, and Seizures
- Proteinuria
 - Albuminuria
 - Donnai-Barrow syndrome
 - Hemoglobinuria
 - CD59 Deficiency
- Urinoma
- Virilism
 - Achard-Thiers syndrome
 - Hirsutism
 - Acanthosis nigricans muscle cramps acral enlargement
 - Barber Say syndrome
 - Cortisone reductase deficiency
 - Spondyloepimetaphyseal dysplasia, Genevieve type
 - Tel Hashomer camptodactyly syndrome
 - Wiedemann Oldigs Oppermann syndrome