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Aase-Smith Syndrome (Diamond-Blackfan Anemia)	Aase Syndrome
Acrocallosal Syndrome	Acrodysostosis
Acro-Fronto-Facio-Nasal Dysostosis	Adrenoleukodystrophy
Agenesis of the Corpus Callosum	Agyria
Aicardi Syndrome	Alexander's Disease
Alper's Syndrome	Amelia
Angelman Syndrome	Aniridia
Anophthalmia/Microphthalmia	Antley-Bixler Syndrome
Apert Syndrome	Arachnoid cyst with neuro-developmental delay
Arhinencephaly	Arthogryposis
Ataxia	Atelosteogenesis
Autism	Baller-Gerold Syndrome
Bannayan-Riley-Ruvalcaba Syndrome	Bardet-Biedl Syndrome
Bartoscas-Papas Syndrome	Beals Syndrome (congenital contractual arachnodactyly)
Bixler Syndrome	Blackfan-Diamond Syndrome
Bobble Head Doll Syndrome	Borjeson-Forssman-Lehmann Syndrome
Brachial Plexopathy	Brancio-Oto-Renal (BOR) Syndrome
Campomelic Dysplasia	Canavan Disease
Carbohydrate Deficient Glycoprotein Syndrome	Cardio-Facio-Cutaneous Syndrome
Carpenter Syndrome	Cataracts-Congenital
Caudal Dysplasia	Cerebro-Costo-Mandibular Syndrome
Cerebellar Aplasis/Hypoplasia/Degeneration	Cerebral Atrophy
Cerebral Palsy	Cerebro-oculo-facial-skeletal syndrome
CHARGE Association	Chediak Higashi Syndrome
Chondrodysplasia Punctata	Christian Syndrome
Chromosome Abnormality	CNS Aneurysm with Neuro-Developmental Delay
a. Unbalanced numerical (autosomal)	
b. Numerical trisomy (chromosomes 1-22)	
c. Sex chromosomes XXX; XXXX; XXXXX;	
XXXY; XXXXY	
CNS Tumor with Neuro-Developmental Delay	Cockayne Syndrome
Coffin Lowry Syndrome	Coffin Siris Syndrome
Cohen Syndrome	Cone Dystrophy
Congenital Cytomegalovirus	Congenital Herpes
Congenital Rubella	Congenital Syphilis
Congenital Toxoplasmosis	Cortical Blindness
Costello Syndrome	Cri Du Chat Syndrome
Cryotophthalmos	Cutis Laxa
Cytochrome-c Oxidase Deficiency	Dandy Walker Syndrome
DeBarsy Syndrome	DeBoquois Syndrome
Dejerine-Sottas Syndrome	DeLange Syndrome
DeSanctis Cacchione Syndrome	Diastrophic Dysplasia
DiGeorge Syndrome	Distal Arthrogryrosis
Donohue Syndrome	Down Syndrome
Dubowitz Syndrome	Dyggve Melchor-Calusen Syndrome
Dyssegmental Dysplasia	Dystonia
EEC (Ectrodactyly-ectodermal dysplasia-clefting)	Encephalocele
Syndrome	·
Encephalo-Cranio-Cutaneous Syndrome	





Familial Dysautonomia (Riley-Day Syndrome) Farbor Syndrome Fetal Alcohol Syndrome/Effects Fetal Pydantoin Syndrome Fetal Alcohol Syndrome Fetal Varione Fetal Varione Fetal Varione Filozohondregenesis Ficating I Syndrome Filozohondregenesis Fragile X Syndrome Fryns Syndrome Galactosemia Glutanc Anduria Type I and II Glycogen Storage Disease Goluberg-Shprintzen Syndrome Head Trauma with Neurological Sequelae/Developmental Delay Gerbe Syndrome Head Trauma with Neurological Hemimegalencephaly Hemimegalencephaly Hemorrhage-Intraventricular Grade III and IV Hereditary Sensory Motor Neuropathy (Charcot Marie Tooth Disease) Heterotopias Holt-Oram Syndrome Hunter Syndrome (MPS II) Hurter Syndrome (MPS II) Hydrosephalus Hypomicalnosis of ITO Hypoxic Ischemic Encephalopathy Hypoxic Ischemic Encephalopathy Ininencephaly Jensory Jenso	Facio-Auriculo-Radial Dysplasia	Facio-Cardio Renal (Eastman-Bixler) Syndrome
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		MERRF
	Metachromatic Leukodystrophy	Metatropic Dysplasia





Methylmalonic Acidemia	Microcephaly
Microtia-Bilateral	Midas Syndrome
Miller (postaxial acrofacial-dysostosis) Syndrome	Miller-Dieker Syndrome
Mitochondrial Disorder	Mobius Syndrome
Morquio Syndrome	Moya-Moya Disease
Mucolipidosis II and III	Multiple congenital anomalies (major organ birth defects)
Multiple Pterygium Syndrome	Muscular Dystrophy
Myasthenia Gravis-Congenital	Myelocystocele
Myopathy –Congenital	Myotonic Dystrophy
Nager (Acrofacial Dysostosis) Syndrome	Nance Horan Syndrome
NARP	Neonatal Meningitis/Encephalitis
Neuronal Ceroid Lipofuscinoses	Neuronal Migration Disorder
Nonketotic Hyperglycinemia	Noonan Syndrome
Ocular Albinism	Oculocerebrocutaneous Syndrome
Oculo-Cutaneous Albinism	Optic Atrophy
Optic Nerve Hypoplasia	Oral-Facial digital Syndrome, Types I-VII
Osteogenesis Imperfecta, Types III and IV	Osteopetrosis (Autosomal Recessive)
Oto-Palato-Digital Syndrome, Types I and II	Pachygyria
Pallister Mosaic Syndrome	Pallister-Hall Syndrome
Pelizaeus-Merzbacher Disease	Pendred's Syndrome
Periventricular Leukomalacia	Pervasive Developmental Disorder
Peters Anomaly	Phocomelia
Poland Sequence	Polymicrogyria
Popliteal Pterygium Syndrome	Porencephaly
Prader-Willi Syndrome	Progeria
Propionic Acidema	Proteus Syndrome
Pyruvate Carboxylase Deficiency	Pyruvate Dehydrogenase Deficiency
Radial Aplasia/Hypoplasia	Refsum Disease
Retinoblastoma	Retinoic Acid Embryopathy
Retinoplational Retinoplation of Prematurity, Stages III and IV	Rett Syndrome
Rickets	Rieger Syndrome
Roberts SC Phocomelia	Robinow Syndrome
	Sanfilippo Syndrome (MPS III)
Rubinstein-Taybin Syndrome	
Schinzel-Giedion Syndrome	Schimmelpenning Syndrome (Epidermal Nevus Syndrome)
Schizencephaly	Schwartz-Jampel Syndrome
Seckel Syndrome	Septo-Optic Dysplasis
Severe Attachment Disorder	Shaken Baby Syndrome
Short Syndrome	Sialidosis
Simpson-Golabi-Behmel Syndrome	Sly Syndrome (MPS IV)
Smith-Fineman-Myers Syndrome	Smith_Limitz-Opitz Syndrome
Smith-Magenis Syndrome	Sotos Syndrome
Spina Bifida (Meningomyelocele)	Spinal Muscular Atrophy
Spondyloepiphyseal Dysplasia Congenita	Spondylometaphyseal Dysplasia
Stroke	Sturge-Weber Syndrome
TAR (Thrombocytopenia-Absent Radii Syndrome)	Thanatophoric Dysplasia
Tibial Aplasis (Hypoplasia)	Toriello-Carey Syndrome
Townes-Brocks Syndrome	Trecher-Collins Syndrome
Trisomy 13	Trisomy 18
Tuberous Sclerosis	Urea Cycle Defect





Valocardiofacial Syndrome	Wildervanck Syndrome
Walker-Warburg Syndrome	Weaver Syndrome
Wiedemann-Rautenstrauch Syndrome	Williams Syndrome
Winchester Syndrome	Wolf Hirschhorn Syndrome
Yunis-Varon Syndrome	Zellweger Syndrome