CHC Disease List

Autosomal Dominant

- ADCY5-related Dyskinesia
- Apert Syndrome (*FGFR2*).
- Autosomal Dominant Polycystic Kidney Disease
- Benign familial infantile seizures 2 (BFIS2) (*PRRT2*)
- Catecholaminergic polymorphic ventricular tachycardia-1 (CPVT1)
- Central Core Disease (RYR1 related disorder)
- Charcot-Marie-Tooth type 1A
- Childhood-onset epileptic encephalopathy (EEOC) (*CHD2*)
- Congenital myasthenic syndrome (CMS-*CHRNE*)
- Dravet Syndrome (SCN1A)
- Dystonia 6
- Ehlers Danlos syndrome, hypermobility type
- Ehlers Danlos syndrome, vascular type
- Familial hemiplegic migraines 1 (C*ACNA1A*)
- Familial Partial Lipodystrophy (LMNA)
- Familial thoracic aortic aneurysms and dissections (FTAAD, *MYLK*)
- FOXG1 syndrome
- Gorlin Syndrome (PTCH1)
- Hereditary breast & ovarian cancer (*BRCA1)*
- Hereditary hemorrhagic telangiectasia (*ENG*)
- Hereditary melanoma-pancreatic cancer syndrome (*CDKN2A*)
- Hereditary pancreatitis (*PRSS1*)
- Hereditary paraganglioma pheochromocytoma type 1 (*SDHA* and *SDHD*)
- HNF4A-related hyperinsulinism and Maturity-onset diabetes of the young 1 (MODY1)
- Hypertrophic cardiomyopathy 4 (*MYBPC3*)
- Hypotrichosis 1
- Kabuki Syndrome (*KMT2D*)
- KBG Syndrome (ANKRD11)
- KIF2A-related disorder, complex cortical dysplasia with other brain malformations, CDCBM3
- Kleefstra Syndrome (EHMT1)
- Leri-Weill dyschondrosteosis (LWD(
- Long QT syndrome (*KCNQ1*)
- Lymphedema-Distichiasis Syndrome (LDS, associated with FOXC2)
- Lynch Syndrome (*MSH2*)
- MAP3K7 related disorder
- Marfan syndrome (*FBN1*)
- Mental retardation 5 (*SYNGAP1*)
- Mental retardation 7 (*DYRK1A*)
- Mental retardation 23 (*SETD5*)
- Mental retardation 26 (*AUTS2*)
- Mental retardation 31 (*PURA* syndrome)
- Mental retardation 46 (*KCNQ5)*
- Moebius syndrome
- Mowat Wilson Syndrome (*ZEB2)*
- Myotonic dystrophy 1 (*DMPK*)
- Neurofibromatosis 1
- Osteogenesis Imperfecta, different types (*COL1A1, COL1A2*)
- Retinitis pigmentosa (*NR2E3*)
- *RET*-related disorder (Hirschsprung disease)
- Rubinstein-Taybi syndrome (*CREBBP and EP300*)
- *SCN2A*-related disorder
- Septooptic dysplasia
- Sotos Syndrome 2
- Stickler syndrome (*COL2A1*)
- Syndactyly type 1
- Tuberous Sclerosis (*TSC2*)
- White-Sutton Syndrome (*POGZ* gene)

Autosomal Recessive

- 2-methylbutyryl CoA dehydrogenase deficiency (2MBCD)
- 3-methylcrotonyl CoA carboxylase deficiency (3MCC)
- 21 hydroxylase deficiency (congenital adrenal hyperplasia (CAH)
- Adenosine Deaminase (ADA SICD) deficiency
- Adenosine Deaminase 2 (ADA 2) deficiency
- Alpha-1 antitrypsin deficiency
- Alpha thalassemia
- Amish brittle hair syndrome
- Argininosucciate lyase (ASL) deficiency
- Beta ketothialase deficiency (BKT)
- Biotinidase deficiency
- Cartilage hair hypoplasia (CHH)
- Cerebro-facio-thoracic Dysplasia (TMCO1)
- Charcot-Marie-Tooth neuropathy type 4A
- CLN 6 disease; Late Infantile Neuronal Ceroid Lipofuscinosis (rare form of Batten disease)
- Cobalamin C defect (MMA + Homocystinuria)
- Cockayne Syndrome (type B)
- Combined oxidative phosphorylation deficiency-13 (COXPD13) (*PNPT1*)
- Cortical dysplasia and focal epilepsy syndrome (*CNTNAP2*) Crigler-Najjar syndrome type 1
- Cystic Fibrosis
- Dubin Johnson Syndrome
- Galactosemia (Classical and Duarte variant)
- Galloway Mowat Syndrome (Yoder Dystonia)
- Gitelman syndrome
- Glutaric aciduria 1 (GA 1)
- Glutaric aciduria 2 (GA 2)
- GM3 synthase deficiency
- Friedreich Ataxia
- Hemochromatosis
- Hereditary spastic paraplegia 20 (SPG20), Troyer Syndrome
- Hereditary spastic paraplegia 21 (SPG21), Mast Syndrome
- Hereditary spastic paraplegia 45 (NT5C2)
- Hypomyelinating leukodystrophy (*GJC2*)
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric Acidemia
- Juvenile Parkinson's Disease (ARPD-PARKN)
- Limb Girdle Muscular Dystrophy type 2A
- Limb Girdle Muscular Dystrophy type 2E
- Maple syrup urine disease (MSUD)
- Meckel Syndrome (Meckel-Gruber)
- Medium chain acyl CoA dehydrogenase deficiency (MCADD)
- Mental retardation 44 (*METTL23*)
- Methylmalonic acidemia, isolated (MMA)
- Microcephalic osteodysplastic primordial dwarfism type I (MOPD1)
- Mitochondrial depletion syndrome 12 (*SLC25A4*, ANT1)
- MYH-associated polyposis. (*MUTYH)*
- Non-ketotic hyperglycinemia (NKH)
- Oculocutaneous albinism type 1B (Amish albinism)
- Phenylalanine Hydroxylase (PAH) deficiency (PKU)
- PMRED-SNIP1
- Pompe Disease (GAA)
- Pontocerebellar hypoplasia type 1 (*EXOSC3*)
- Primary autosome recessive microcephaly-6 (MCPH)/Seckel syndrome 4
- Primary Ciliary Dyskinesia (*HYDIN*)
- Propionic acidemia (*PCCB*)
- *RAG-1* Severe Combined Immunodeficiency (SCID)
- Retinitis. Pigmentosa 49 (*CNGA1*)
- *RSRC1*-related disorder
- Sanfilippo syndrome type B (MPS III B – *NAGLU*)
- Short chain acyl CoA dehydrogenase deficiency (SCADD)
- Short Rib Thoracic Dysplasia type 4 *(TTC21B*)
- Sitosterolemia (*ABCG8*)
- SMC5-related disorder (microcephalic dwarfism)
- Spastic Ataxia 4 (*MTPAP*)
- Spinal muscular atrophy 1 (SMA)
- Systemic primary carnitine deficiency
- TANGO2-related recurrent metabolic encephalomyopathic crises associated with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration (MECRCN).
- TMCO1 defect
- Trichohepatoneurodevelopmental syndrome (THNS) (*CCDC47*)
- Tripeptidyl peptidase II (*TPP2*) deficiency
- Thyroid Dyshormonogenesis 2A (*TPO*)
- Ullrich Congenital Muscular Dystrophy (COL6A1)
- Very long chain acyl CoA dehydrogenase deficiency (VLCADD)
- Wilson disease
- Wolfram syndrome (*WFS1*)
- Zellweger Spectrum Disorder - Peroxisomal Biogenesis (*PEX26*)

Mitochondrial

- Leber Hereditary Optic Neuropathy
- Myoclonic Epilepsy with Ragged Red Fibers (MERRF)

X-linked

- Creatine Transporter Deficiency (CRTR)
- Duchenne Muscular Dystrophy
- Hemophilia B
- MICPCH syndrome *(CASK* related disorder)
- Ohdo Syndrome (*MED12*)
- Beta-propeller protein-associated neurodegeneration (BPAN) (WDR45)
- Oral-facial-digital syndrome
- Partial Androgen Insensitivity syndrome *(AR*)
- Steroid Sulfatase Deficiency (X-linked Ichthyosis)
- X-linked syndromic mental retardation, Turner type (*HUWE1*)

Chromosomal abnormalities

- 1q terminal deletion syndrome
- 1p36 deletion syndrome (mosaic)
- 1q21.1 deletion syndrome
- 1q43q44 deletion syndrome
- 1q43 terminal duplication syndrome
- 4q partial trisomy syndrome
- 4q12q23 duplication
- 5p, 9q, 16p duplication syndromes (triple duplication)
- 5p minus syndrome (Cri du chat)
- 6q27 microdeletion syndrome
- 7p22.3 duplication
- 7q11.23 microduplication syndrome
- 9p terminal deletion syndrome
- 10p 12.1 microdeletion syndrome
- 10q22.3 duplication syndrome
- 10q24 deletion syndrome
- 10q partial trisomy syndrome (mosaic)
- 14q11.2 microdeletion syndrome
- 15q11.2 microdeletion syndrome
- 15q11.2 triplication/duplication (isodicentric Chr 15)
- 16p11.2 microdeletion syndrome
- 16p13.11 microdeletion syndrome
- 16q22 deletion
- 17q12 microdeletion
- 18q21.32 deletion syndrome
- 20q13.33 duplication syndrome
- 22q11.2 deletion syndrome
- Distal Chromosome 18q deletion
- Klinefelter syndrome (47, XXY)
- Tetrasomy 12p, mosaic (PKS)
- Trisomy 9 (mosaic)
- Trisomy 13 (Patau syndrome)
- Trisomy 21 (Down syndrome)
- Turner Syndrome (45, X)
- Xp11.22 duplication
- Xp22.32 terminal deletion (Turner-like syndrome)
- Xp22.3 deletion
- Xp22.33 deletion
- XYY syndrome

Imprinting Disorders:

- Beckwith-Wiedemann syndrome (BWS) (*IC2* methylation defect)
- Russell-Silver Syndrome
- UPD 6
- Prader-Willi Syndrome

Other (central nervous system malformation, clinical diagnosis, cancer, etc.)

- Acute disseminated encephalomyelitis (ADEM)
- Acute myeloid leukemia
- Acute tic disorder
- Alcohol Related Neurodevelopmental Disorder (ARND)
- Addison's disease
- Autism Spectrum Disorder
- Breast cancer
- Breath holding spells
- Carnitine deficiency
- CHARGE syndrome
- Chronic inflammatory Demyelinating Polyneuropathy
- Cleft lip and/or palate
- Cold urticaria/periodic fever
- Congenital CMV infection
- Congenital Hypothyroidism
- Recurrent Colon cancer
- Endometriosis
- Fetal Alcohol Syndrome (FAS)
- Hemimegalencephaly
- Hirschsprung disease
- Hypoxic Ischemic Encephalopathy
- In utero stroke
- Jeavons Syndrome
- Klippel-Trenaunay syndrome
- Lissencephaly
- Microcephaly + Deafness
- Pancreatic insufficiency
- Paraganglioma
- PHACE syndrome
- Polycystic kidney disease – suspected recessive inheritance
- Prenatal Tobacco Exposure (PTE)
- Primary selective IgM deficiency
- Renal oncocytosis
- Retinitis pigmentosa
- Sagittal craniosynostosis (isolated)
- Schizencephaly & porencephaly
- Severe prematurity
- Spastic paraplegia (unknown type/gene)
- VATER syndrome
- Vein of Galen arteriovenous malformation