The Community Health Clinic

Disease List

AUTOSOMAL DOMINANT

- Apert syndrome (FGFR2)
- Autosomal dominant polycystic kidney disease
- Beckwith-Wiedemann syndrome
- 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 (CACNA1A)
- Familial thoracic aortic aneurysms and dissections (FTAAD, MYLK)
- FOXG1 syndrome
- Hereditary breast & ovarian cancer (BRCA1)
- Hereditary hemorrhagic telangiectasia (ENG)
- Hereditary melanoma-pancreatic cancer syndrome (CDKN2A)
- Hereditary pancreatitis (PRSS1)
- HNF4A-related hyperinsulinism and maturity-onset diabetes of the young 1 (MODY1)
- Hypertrophic cardiomyopathy 4 (MYBPC3)
- Hypotrichosis 1
- KIF2A-related disorder, complex cortical dysplasia with other brain malformations (CDCBM3)
- MAP3K7 related disorder
- Marfan syndrome (FBN1)
- Mental retardation 5 (SYNGAP1)
- Mental retardation 7 (DYRK1A)
- Mental retardation 26 (AUTS2)
- Mental retardation 31 (PURA syndrome)
- Moebius syndrome
- Myotonic dystrophy 1 (DMPK)
- Neurofibromatosis 1
- Osteogenesis imperfecta, different types (COL1A1, COL1A2)
- Rubinstein-Taybi syndrome (CREBBP)
- SCN2A-related disorder
- Septo-optic dysplasia
- Stickler syndrome (COL2A1)
- Syndactyly type 1
- White-Sutton syndrome (POGZ)

AUTOSOMAL RECESSIVE

- 2-methylbutyryl CoA dehydrogenase deficiency (2MBCD)
- 3-methylcrotonyl CoA carboxylase deficiency (3MCC)
- 21 hydroxylase deficiency (congenital adrenal hyperplasia (CAH))
- Alpha-1 antitrypsin deficiency

AUTOSOMAL RECESSIVE (continued)

- Alpha thalassemia
- Amish brittle hair syndrome
- Beta ketothialase deficiency (BKT)
- Biotinidase deficiency
- Cartilage hair hypoplasia (CHH)
- CCDC47-related disorder (new syndrome)
- 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)
- Cortical dysplasia and focal epilepsy syndrome
- Crigler-Najjar syndrome type 1
- Cystic fibrosis
- Galactosemia (classic and Duarte variant)
- Gitelman syndrome
- Glutaric aciduria 1 (GA 1)
- GM3 synthase deficiency
- Friedreich ataxia
- Hemochromatosis
- Hereditary spastic paraplegia 20 (SPG20)
- Hereditary spastic paraplegia 45 (NT5C2)
- Hypomyelinating leukodystrophy
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric acidemia
- Juvenile Parkinson's disease (ARPD-PARK2)
- 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 (MMA)
- Microcephalic osteodysplastic primordial dwarfism type I (MOPD1)
- Mitochondrial depletion syndrome 12 (SLC25A4, ANT1)
- Non-ketotic hyperglycinemia (NKH)
- Oculocutaneous albinism type 1B (Amish albinism)
- Peroxisomal biogenesis disorders – Zellweger syndrome spectrum (infantile Refsum disease and neonatal adrenoleukodystrophy) (PEX26)
- Phenylalanine hydroxylase (PAH) deficiency (PKU)
- Psychomotor retardation, epilepsy, and craniofacial dysmorphism (PMRED) (SNIP1)
- Pontocerebellar hypoplasia type 1 (EXOSC3)
- Primary autosome recessive microcephaly-6 (MCPH)/Seckel syndrome 4
- Primary diliary dyskinesia (HYDIN)
- Propionic acidemia (PA)
- RSRC1-related disorder (new syndrome)
- Short chain acyl CoA dehydrogenase deficiency (SCADD)
- Sitosterolemia (ABCG8)
- 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
- Very long chain acyl CoA dehydrogenase deficiency (VLCADD)
- Wilson disease
- Wolfram syndrome

MITOCHONDRIAL

- Leber hereditary optic neuropathy
- Myoclonic epilepsy with ragged red fibers (MERRF)

X-LINKED

- Creatine transporter deficiency
- Duchenne muscular dystrophy
- Hemophilia B
- MICPCH syndrome (CASK related disorder)
- Neurodegeneration with brain iron accumulation 5 (NBIA6 or 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)