The Community Health Clinic

Disease List

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AUTOSOMAL DOMINANT

- Autosomal Dominant Polycystic Kidney Disease
- Beckwith-Wiedemann syndrome
- Catecholaminergic polymorphic ventricular tachycardia-1 (CPVT1)
- Central Core Disease (RYR1 related disorder)
- Charcot-Marie-Tooth type 1A
- Congenital myasthenic syndrome (CMS-CHRNE)
- Dystonia 6
- Ehlers Danlos syndrome, hypermobility type
- Ehlers Danlos syndrome, vascular type
- Familial hemiplegic migraines 1 (CACNA1A)
- Hereditary breast & ovarian cancer (BRCA1)
- Hereditary hemorrhagic telangiectasia (ENG)
- Hereditary pancreatitis (PRSS1)
- Hypertrophic cardiomyopathy 4 (MYBPC3)
- Hypotrichosis 1
- MAP3K7 related disorder
- Marfan syndrome
- Melanoma-Pancreatic Cancer syndrome
- Mental retardation 5 (SYNGAP1)
- Mental retardation 7 (DYRK1A)
- Mental retardation 26 (AUTS2)
- Moebius syndrome
- Myotonic dystrophy 1 (DMPK)
- Neurofibromatosis 1
- Osteogenesis Imperfecta type 1 (COL1A1 related disorder)
- Rubinstein-Taybi syndrome (CREBBP)
- Septooptic dysplasia
- Stickler syndrome (COL2A1)
- Syndactyly type 1

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
- Alpha thalassemia
- Amish brittle hair syndrome
- Beta ketothialase deficiency (BKT)
- Biotinidase deficiency
- Cartilage hair hypoplasia (CHH)
- 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
- Duarte galactosemia
- 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)
- Maple syrup urine disease (MSUD)
- Meckel Syndrome (Meckel-Gruber)
- Medium chain acyl CoA dehydrogenase deficiency (MCADD)
- Methylmalonic acidemia (MMA)
- 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)
- Phenylalanine Hydroxylase(PAH) deficiency (PKU)
- PMRED-SNIP1

AUTOSOMAL RECESSIVE (continued)

- Primary autosome recessive microcephaly-6 (MCPH)/Seckel syndrome 4
- Propionic acidemia (PA)
- RSRC1 and MLF1 deletion syndrome (new syndrome)
- Short chain acyl CoA dehydrogenase deficiency (SCADD)
- Spinal muscular atrophy 1 (SMA)
- Systemic primary carnitine deficiency
- 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

- Duchenne Muscular Dystrophy
- Hemophilia B
- MICPCH syndrome (CASK related disorder)
- Partial Androgen Insensitivity syndrome
- Steroid Sulfatase Deficiency (X-linked Ichthyosis)
- X-linked syndromic mental retardation, Turner type (HUWE1)

CHROMOSOMAL ABNORMALITIES

- 1p36 deletion syndrome (mosaic)
- 1q43q44 deletion syndrome
- 4q partial trisomy syndrome
- 5p, 9q, 16p duplication syndromes (triple duplication)
- 6q27 microdeletion syndrome
- 7p22.3 duplication
- 7q11.23 microduplication syndrome
- 10q22.3 duplication syndrome
- 10q partial trisomy syndrome (mosaic)
- 15q11.2 microdeletion sydrome
- 16p11.2 microdeletion syndrome
- 16p13.11 microdeletion syndrome
- 20q13.33 dupulication syndrome
- 22q11.2 microdeletion syndrome (not DiGeorge)
- Klinefelter syndrome (47, XXY)
- Trisomy 13 (Patau syndrome)
- Trisomy 21 (Down syndrome)
- Turner Syndrome (45, X)
- Xp11.22 duplication
- Xp22.32 terminal deletion (Turner-like syndrome)

OTHER

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

- Acute myeloid leukemia
- Acute tic disorder
- Alcohol Related Neurodevelopmental Disorder (ARND)
- Addison's disease
- Breast cancer
- CHARGE syndrome
- Colon cancer
- Endometriosis
- Hemimegalencephaly
- In utero stroke
- Klippel-Trenaunay syndrome
- Lissencephaly
- 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