PAN-ETHNIC PANEL

Cystic fibrosis (CFTR) ACOG ACMG	Spinal muscular atrophy (SMA) ACOG ACMG
Fragile X syndrome ACOG ACMG	

BROAD PAN-ETHNIC PANEL

Alpha-thalassemia (HBA1/HBA2) ACOG	Bloom syndrome (BLM) ACMG
Canavan disease (ASPA) ACOG ACMG	Citrullinemia type 1 (ASS1)
Congenital disorders of glycosylation (PMM2-related)	Cystic Fibrosis (CFTR) ACOG ACMG
Dihydrolipoamide dehydrogenase deficiency (DLD)	DMD-related dystrophinopathy (DMD)
Familial dysautonomia (IKBKAP) ACOG ACMG	Familial hyperinsulinism (ABCC8-related)
Fanconi anemia type C (FANCC) ACMG	Fragile X syndrome (FMR1) ACOG ACMG
Galactosemia (GALT)	Gaucher disease (GBA) ACMG
GJB2-related DFNB1 nonsyndromic hearing loss and deafness (GJB2)	Glycogen storage disease type Ia (G6PC)
Glycogen storage disease type II (Pompe disease) (GAA)	HBB-related hemoglobinopathies (including Beta-thalassemia and Sickle cell disease) (HBB) ACOG
Joubert syndrome 2/ TMEM216-related disorders (TMEM216)	Krabbe disease (GALC)
Maple syrup urine disease (MSUD) type 1A (BCKDHA)	Maple syrup urine disease (MSUD) type 1B (BCKDHB)
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (ACADM)	Mucolipidosis type IV (MCOLN1) ACMG
Mucopolysaccharidosis type I (includes Hurler, Hurler-Scheie, and Scheie syndromes) (IDUA)	Nemaline myopathy 2 (NEB)
Neuronal ceroid-lipofuscinosis (CLN3-related)	Niemann-Pick disease type A/B (SMPD1) ACMG
Ornithine transcarbamylase (OTC) deficiency	Pendred syndrome (SLC26A4)
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU)) (PAH)	Polycystic kidney disease (PKHD1-related) (PKHD1)
Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related) (PEX7)	Smith-Lemli-Opitz syndrome (DHCR7)
Spinal muscular atrophy (SMN1) ACMG	Tay-Sachs disease/ Hexosaminidase A deficiency (HEXA) ACOG ACMG
Tyrosinemia type I (FAH)	Usher syndrome type IF/ PCDH15-related disorders (PCDH15)
Usher syndrome type IIA/ USH2A-related disorders (USH2A)	Usher syndrome type IIIA (CLRN1)
Walker-Warburg syndrome/ FKTN-related disorders (FKTN)	X-linked adrenoleukodystrophy (ABCD1)
X-linked severe combined immunodeficiency (X-SCID) (IL2RG)	Zellweger spectrum disorder (PEX1-related)

Zellweger spectrum disorder (PEX6-related)	

COMPREHENSIVE PANEL

11-beta-hydroxylase-deficient congenital adrenal hyperplasia (CYP11B1)	17-alpha-hydroxylase-deficient congenital adrenal hyperplasia (CYP17A1)
3-beta-hydroxysteroid dehydrogenase type II deficiency (Congenital adrenal hyperplasia) (HSD3B2)	3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency (HMGCL)
3-methylglutaconic aciduria type III (Costeff optic atrophy) (OPA3)	Abetalipoproteinemia (MTTP)
ACAD9 deficiency (ACAD9)	Achromatopsia (CNGB3-related)
Acrodermatitis enteropathica (SLC39A4)	Adenosine deaminase deficiency (ADA)
Aicardi-Goutieres syndrome (SAMHD1-related)	Alpha-mannosidosis (MAN2B1)
Alpha-thalassemia (HBA1/HBA2) ACOG	Alpha-thalassemia X-linked intellectual disability syndrome (ATRX)
Alport Syndrome (COL4A3-related)	Alport Syndrome (COL4A4-related)
Alport Syndrome, X-linked (COL4A5-related)	Alström syndrome (ALMS1)
Andermann syndrome (SLC12A6)	Arginase deficiency (ARG1)
Argininosuccinic aciduria (ASL)	Aromatase deficiency (CYP19A1)
Asparagine synthetase deficiency (ASNS)	Aspartylglucosaminuria (AGA)
Ataxia with vitamin E deficiency (TTPA)	Ataxia-telangiectasia (ATM)
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AIRE)	Autosomal recessive deafness 77 (DFNB77) (LOXHD1)
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) (SACS)	Bardet-Biedl syndrome (BBS1-related)
Bardet-Biedl syndrome (BBS10-related)	Bardet-Biedl syndrome (BBS12-related)
Bardet-Biedl syndrome (BBS2-related)	Bartter syndrome type IV (BSND)
Beta-ketothiolase deficiency (ACAT1)	Bloom syndrome (BLM) ACMG
Canavan disease (ASPA) ACOG ACMG	Carbamoylphosphate synthetase I deficiency (CPS1)
Carnitine palmitoyltransferase I deficiency (CPT1A)	Carnitine palmitoyltransferase II deficiency (CPT2)
Carpenter Syndrome (RAB23)	Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders (RMRP)
Cerebrotendinous xanthomatosis (CYP27A1)	Charcot-Marie-Tooth disease (NDRG1-related)
Charcot-Marie-Tooth disease, X-linked (GJB1-related)	Chorea-acanthocytosis (VPS13A)
Choroideremia (CHM)	Chronic granulomatous disease (CYBA-related)
Chronic granulomatous disease (CYBB-related)	Citrin deficiency (SLC25A13)
Citrullinemia type 1 (ASS1)	Cockayne syndrome type A (ERCC8)
Cockayne syndrome type B (ERCC6)	Cohen syndrome (VPS13B)

Combined malonic and methylmalonic aciduria (ACSF3-related)	Combined oxidative phosphorylation deficiency (GFM1-related)
Combined oxidative phosphorylation deficiency (TSFM-related)	Combined pituitary hormone deficiency (LHX3-related)
Combined pituitary hormone deficiency (PROP1-related)	Combined SAP Deficiency (PSAP)
Congenital amegakaryocytic thrombocytopenia (MPL)	Congenital disorder of glycosylation (ALG6-related)
Congenital disorder of glycosylation (MPI-related)	Congenital disorders of glycosylation (PMM2-related)
Congenital ichthyosis (TGM1-related)	Congenital insensitivity to pain with anhidrosis (NTRK1)
Congenital myasthenic syndrome (CHRNE-related)	Congenital myasthenic syndrome (RAPSN-related)
Congenital neutropenia (HAX1-related)	Corneal dystrophy and perceptive deafness (SLC4A11)
Corticosterone methyloxidase deficiency (CYP11B2)	Cystic fibrosis (CFTR) ACOG ACMG
Cystinosis (CTNS)	D-bifunctional protein deficiency (HSD17B4)
DHDDS-related disorders (including Congenital disorder of glycoslylation/ Retinitis pigmentosa 59)	Dihydrolipoamide dehydrogenase deficiency (DLD)
DMD-related dystrophinopathy (Including Duchenne/ Becker muscular dystrophy and Dilated cardiomyopathy) (DMD)	Dysferlinopathy (including Limb-girdle muscular dystrophy type 2B) (DYSF)
Dystrophic epidermolysis bullosa (COL7A1-related)	Ehlers-Danlos syndrome type VIIC (ADAMTS2)
Ellis-van Creveld syndrome (EVC-related)	Ellis-van Creveld syndrome (EVC2-related)
Emery-Dreifuss muscular dystrophy (EMD-related)	Enhanced S-cone syndrome/ Retinitis pigmentosa 37 (NR2E3)
Ethylmalonic encephalopathy (ETHE1)	Fabry disease (GLA)
Factor IX deficiency (Hemophilia B) (F9)	Familial dysautonomia (IKBKAP) ACOG ACMG
Familial hypercholesterolemia (LDLR-related)	Familial hypercholesterolemia (LDLRAP1-related)
Familial hyperinsulinism (ABCC8-related)	Familial hyperinsulinism (KCNJ11-related)
Fanconi anemia type A (FANCA)	Fanconi anemia type C (FANCC) ACMG
Fanconi anemia type G (FANCG)	Fragile X syndrome (FMR1) ACOG ACMG
Fumarate hydratase deficiency (FH)	Galactokinase deficiency galactosemia (GALK1)
Galactosemia (GALT)	Gaucher disease (GBA) ACMG
Gitelman syndrome (SLC12A3)	GJB2-related DFNB1 nonsyndromic hearing loss and deafness (GJB2)
Glutaric acidemia type I (GCDH)	Glutaric acidemia type II (ETFA-related)
Glutaric acidemia type II (ETFDH-related)	Glycine encephalopathy (AMT-related)
Glycine encephalopathy (GLDC-related)	Glycogen storage disease type Ia (G6PC)

Glycogen storage disease type Ib (SLC37A4)	Glycogen storage disease type II (Pompe disease) (GAA)
Glycogen storage disease type III (AGL)	Glycogen storage disease type IV/ Adult polyglucosan body disease (GBE1)
Glycogen storage disease type V (PYGM)	Glycogen storage disease type VII (PFKM)
GRACILE syndrome/ BCS1L-related disorders (including Mitochondrial complex III deficiency, Bjornstad syndrome, Leigh syndrome) ()	Guanidinoacetate methyltransferase deficiency (GAMT)
HBB-related hemoglobinopathies (including Beta-thalassemia and Sickle cell disease) ACOG	Hereditary fructose intolerance (ALDOB)
Hereditary hemochromatosis (TFR2-related)	Hermansky-Pudlak syndrome (HPS1-related)
Hermansky-Pudlak syndrome (HPS3-related)	Holocarboxylase synthetase deficiency (HLCS)
Homocystinuria (CBS-related)	Homocystinuria due to MTHFR deficiency (MTHFR)
Homocystinuria, cobalamin E type (MTRR)	Hydrolethalus syndrome type 1 (HYLS1)
Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome (SLC25A15)	Hypohidrotic ectodermal dysplasia (EDA-related)
Hypophosphatasia (ALPL)	Inclusion body myopathy 2 (GNE)
Isovaleric acidemia (IVD)	Joubert syndrome 2/ TMEM216-related disorders
Junctional epidermolysis bullosa (LAMA3-related)	Junctional epidermolysis bullosa (LAMB3-related)
Junctional epidermolysis bullosa (LAMC2-related)	Krabbe disease (GALC)
LAMA2-related muscular dystrophy (LAMA2)	Leber congenital amaurosis 10/ CEP290-related disorders
Leber congenital amaurosis 13 (RDH12)	Leber congenital amaurosis 2 (RPE65)
Leber congenital amaurosis 5 (LCA5)	Leber congenital amaurosis 8/ CRB1-related disorders (CRB1)
Leigh syndrome, French Canadian type (LRPPRC)	Lethal congenital contracture syndrome 1 / Lethal arthrogryposis with anterior horn cell disease (GLE1)
Leukoencephalopathy with vanishing white matter (EIF2B5-related)	Limb-girdle muscular dystrophy type 2A/ Calpainopathy (CAPN3)
Limb-girdle muscular dystrophy type 2C (SGCG)	Limb-girdle muscular dystrophy type 2D (SGCA)
Limb-girdle muscular dystrophy type 2E (SGCB)	Lipoid congenital adrenal hyperplasia (STAR)
Lipoprotein lipase deficiency (LPL)	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency (HADHA)
Lysinuric protein intolerance (SLC7A7)	Lysosomal acid lipase deficiency (includes Wolman disease and Cholesterol ester storage disease) (LIPA)
Major histocompatibility complex class II deficiency (CIITA)	Maple syrup urine disease (MSUD) type 1A (BCKDHA)
Maple syrup urine disease (MSUD) type 1B (BCKDHB)	Maple syrup urine disease (MSUD) type 2 (DBT)
Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (ACADM)	Megalencephalic leukoencephalopathy with subcortical cysts type 1 (MLC1)

Menkes disease/ ATP7A-related disorders (including Occipital horn syndrome and Distal hereditary motor neuropathy) (ATP7A)	Metachromatic leukodystrophy (ARSA)
Methylmalonic acidemia (MMAA-related)	Methylmalonic acidemia (MMAB-related)
Methylmalonic acidemia (MUT-related)	Methylmalonic acidemia with homocystinuria, cobalamin C type (MMACHC)
Methylmalonic acidemia with homocystinuria, cobalamin D type (MMADHC)	Microphthalmia / clinical anophthalmia (VSX2)
Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)	Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6- related)
Mitochondrial DNA depletion syndrome (MPV17)	Mitochondrial myopathy and sideroblastic anemia 1 (PUS1)
Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease (TYMP)	MKS1-related disorders (MKS1)
Mucolipidosis type II/III (GNPTAB-related)	Mucolipidosis type III (GNPTG-related)
Mucolipidosis type IV (MCOLN1) ACMG	Mucopolysaccharidosis type I (includes Hurler, Hurler-Scheie, and Scheie syndromes) (IDUA)
Mucopolysaccharidosis type II (Hunter syndrome) (IDS)	Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome) (SGSH)
Mucopolysaccharidosis type IIIB (NAGLU)	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome)/ Retinitis pigmentosa 73 (HGSNAT)
Mucopolysaccharidosis type IIID (Sanfilippo syndrome) (GNS)	Mucopolysaccharidosis type IVB (Morquio B syndrome)/ GM1 gangliosidosis (GLB1)
Mucopolysaccharidosis type IX (HYAL1)	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) (ARSB)
Multiple sulfatase deficiency (SUMF1)	N-Acetylglutamate synthase deficiency (NAGS)
Nemaline myopathy 2 (NEB)	Nephrogenic diabetes insipidus (AQP2-related)
Nephrotic syndrome/ Congenital Finnish nephrosis (NPHS1-related)	Nephrotic syndrome/Steroid-resistant nephrotic syndrome (NPHS2-related)
Neuronal ceroid lipofuscinosis (TPP1-related)	Neuronal ceroid-lipofuscinosis (CLN3-related)
Neuronal ceroid-lipofuscinosis (CLN5-related)	Neuronal ceroid-lipofuscinosis (CLN6-related)
Neuronal ceroid-lipofuscinosis (MFSD8-related)	Neuronal ceroid-lipofuscinosis (PPT1-related)
Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)	Niemann-Pick disease type A/B (SMPD1) ACMG
Niemann-Pick disease type C (NPC1-related)	Niemann-Pick disease type C (NPC2-related)
Nijmegen breakage syndrome (NBN)	Ornithine aminotransferase deficiency (OAT)
Ornithine transcarbamylase (OTC) deficiency	Osteopetrosis (TCIRG1-related)
Pendred syndrome (SLC26A4)	Peroxisomal acyl-CoA oxidase deficiency (ACOX1)
Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU)) (PAH)	Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome (PHGDH)
Polycystic kidney disease (PKHD1-related)	Polymicrogyria (ADGRG1-related)

POMGNT1-related disorders (including Muscle eye brain disease) (POMGNT1)	Pontocerebellar hypoplasia (RARS2-related)
Pontocerebellar hypoplasia (SEPSECS-related)	Pontocerebellar hypoplasia (VRK1-related)
Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)	Primary carnitine deficiency (SLC22A5)
Primary Ciliary Dyskinesia (DNAH5-related)	Primary Ciliary Dyskinesia (DNAI1-related)
Primary Ciliary Dyskinesia (DNAI2-related)	Primary hyperoxaluria type 1 (AGXT)
Primary hyperoxaluria type 2 (GRHPR)	Primary hyperoxaluria type 3 (HOGA1)
Progressive familial intrahepatic cholestasis type 2 (ABCB11)	Propionic acidemia (PCCA-related)
Propionic acidemia (PCCB-related)	PRPS1-related disorders (including Charcot-Marie-Tooth disease type 5 and Arts syndrome) (PRPS1)
Pycnodysostosis (CTSK)	Pyruvate carboxylase deficiency (PC)
Pyruvate dehydrogenase deficiency (PDHA1-related)	Pyruvate dehydrogenase deficiency (PDHB-related)
Renal tubular acidosis with deafness (ATP6V1B1-related)	Retinitis pigmentosa 25 (EYS)
Retinitis pigmentosa 26 (CERKL)	Retinitis Pigmentosa 28 (FAM161A)
Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)	Rhizomelic chondrodysplasia punctata type 3 (AGPS)
Roberts syndrome (ESCO2)	RPGRIP1L-related disorders (including Joubert syndrome 7, COACH syndrome and Meckel syndrome 5) (RPGRIP1L)
RTEL-1-related disorders (including Dyskeratosis congenita) (RTEL1)	Sandhoff disease (HEXB)
Schimke immuno-osseous dysplasia (SMARCAL1)	Severe combined immune deficiency (DCLRE1C-related)
Severe combined immunodeficiency/ Omenn syndrome (RAG2-related)	Severe congenital neutropenia (VPS45-related)
Sialic acid storage disorders (SLC17A5)	Sjögren-Larsson syndrome (ALDH3A2)
SLC26A2-related disorders (including Diatrophic dysplasia, Atelosteogenesis type 2, Achondrogenesis type 1B/ Multiple metaphyseal dysplasia) (SLC26A2)	SLC35A3-related disorder (SLC35A3)
Smith-Lemli-Opitz syndrome (DHCR7)	Spastic paraplegia type 15 (ZFYVE26)
Spastic paraplegia type 49 (TECPR2)	Spinal muscular atrophy (SMN1) ACMG
Spondylothoracic dysostosis (MESP2)	Steel Syndrome (COL27A1)
Stüve-Wiedemann syndrome (LIFR)	Tay-Sachs disease/ Hexosaminidase A deficiency (HEXA) ACOG ACMG
Tetrahydrobiopterin deficiency (PTS-related)	Transient infantile liver failure (TRMU-related)
Tyrosine hydroxylase deficiency (TH)	Tyrosinemia type I (FAH)

Tyrosinemia type II (TAT)	Usher syndrome type IB/ MYO7A-related disorders
Usher syndrome type IC/ USH1C-related disorders	Usher syndrome type ID (CDH23)
Usher syndrome type IF/ PCDH15-related disorders	Usher syndrome type IIA/ USH2A-related disorders
Usher syndrome type IIIA (CLRN1)	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (ACADVL)
Walker-Warburg syndrome/ FKRP-related disorders	Walker-Warburg syndrome/ FKTN-related disorders
Wilson disease (ATP7B)	WNT10A-related disorders (including Odonto-onycho-dermal dysplasia and Schopf-Schulz-Passarge syndrome) (WNT10A)
X-linked adrenoleukodystrophy (ABCD1)	X-linked creatine transporter deficiency (SLC6A8)
X-linked juvenile retinoschisis (RS1)	X-linked myotubular myopathy (MTM1)
X-linked severe combined immunodeficiency (X-SCID) (IL2RG)	Xeroderma pigmentosum complemetation group A (XPA)
Xeroderma pigmentosum complemetation group C (XPC)	Zellweger spectrum disorder (PEX10-related)
Zellweger spectrum disorder (PEX12-related)	Zellweger spectrum disorder (PEX2-related)
Zellweger spectrum disorder (PEX6-related)	