

COMPREHENSIVE PANEL



3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency, <i>HMGCL</i>	3-Methylcrotonyl-CoA Carboxylase Deficiency 1, <i>MCCC1</i>	3-Methylcrotonyl-CoA Carboxylase Deficiency 2, <i>MCCC2</i>
3-Methylglutaconic Aciduria, Type 3 [Costeff Syndrome], <i>OPA3</i>	3-Phosphoglycerate Dehydrogenase Deficiency, <i>PHGDH</i>	6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency, <i>PTS</i>
Abetalipoproteinemia, <i>MTPP</i>	Achondrogenesis, Type 1B, <i>SLC26A2</i>	Achromatopsia (CNGB3-related), <i>CNGB3</i>
Acute Infantile Liver Failure (TRMU-related), <i>TRMU</i>	Acyl-CoA Oxidase I Deficiency, <i>ACOX1</i>	Adrenoleukodystrophy, X-Linked, <i>ABCD1</i>
Aicardi-Goutières Syndrome, <i>SAMHD1</i>	Alpha Thalassemia, <i>HBA1, HBA2</i> ●	Alport Syndrome (COL4A3-related), <i>COL4A3</i>
Alport Syndrome, X-Linked, <i>COL4A5</i>	Alstrom Syndrome, <i>ALMS1</i>	Andermann Syndrome, <i>SLC12A6</i>
Argininosuccinate Lyase Deficiency, <i>ASL</i>	Aromatase Deficiency, <i>CYP19A1</i>	Arthrogryposis Mental Retardation Seizures, <i>SLC35A3</i>
Asparagine Synthetase Deficiency, <i>ASNS</i>	Aspartylglycosaminuria, <i>AGA</i>	Ataxia with Vitamin E Deficiency, <i>TTPA</i>
Ataxia-Telangiectasia, <i>ATM</i>	Autoimmune Polyglandular Syndrome, Type 1, <i>AIRE</i>	Autosomal Recessive Polycystic Kidney Disease, <i>PKHD1</i>
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay, <i>SACS</i>	Bardet-Biedl Syndrome (BBS1-related), <i>BBS1</i>	Bardet Biedl syndrome (BBS12-related), <i>BBS12</i>
Bare Lymphocyte Syndrome (CIITA-related), <i>CIITA</i>	Bartter Syndrome (BSND-related), <i>BSND</i>	Batten Disease (CLN3-related), <i>CLN3</i>
Beta Thalassemia, <i>HBB</i> ●	Biotinidase Deficiency, <i>BTD</i>	Bloom Syndrome, <i>BLM</i> ◆
Canavan Disease, <i>ASPA</i> ◆	Carnitine Palmitoyltransferase IA Deficiency, <i>CPT1A</i>	Carnitine Palmitoyltransferase II Deficiency, <i>CPT2</i>
Carpenter Syndrome, <i>RAB23</i>	Cartilage-Hair Hypoplasia, <i>RMRP</i>	Cerebrotendinous Xanthomatosis, <i>CYP27A1</i>
Choreacanthocytosis, <i>VPS13A</i>	Choroideremia, X-Linked, <i>CHM</i>	Chronic Granulomatous Disease, X-Linked, <i>CYBB</i>
Citrin Deficiency, <i>SLC25A13</i>	Citrullinemia, Type 1, <i>ASS1</i>	Combined Malonic and Methylmalonic Aciduria, <i>ACSF3</i>
Combined Oxidative Phosphorylation Deficiency 1, <i>GFM1</i>	Combined Oxidative Phosphorylation Deficiency 3, <i>TSFM</i>	Combined Pituitary Hormone Deficiency 2, <i>PROPI</i>
Congenital Disorder of Glycosylation, Type 1A (PMM2- related), <i>PMM2</i>	Congenital Disorder of Glycosylation, Type 1B, <i>MPI</i>	Congenital Disorder of Glycosylation Type 1C, <i>ALG6</i>
Congenital Finnish Nephrosis, <i>NPHS1</i>	Congenital Insensitivity to Pain with Anhidrosis, <i>NTRK1</i>	Congenital Myasthenic Syndrome (CHRNE-related), <i>CHRNE</i>
Congenital Myasthenic Syndrome (RAPSN-related), <i>RAPSN</i>	Congenital Neutropenia (HAX1-related), <i>HAX1</i>	Congenital Neutropenia (VPS45-related), <i>VPS45</i>
Corneal Dystrophy and Perceptive Deafness, <i>SLC4A11</i>	Corticosterone Methyloxidase Deficiency, <i>CYP11B2</i>	CRB1-related Retinal Dystrophies, <i>CRB1</i>
Creatine Transporter Defect [Cerebral Creatine Deficiency Syndrome 1] X-Linked, <i>SLC6A8</i>	Crigler Najjar syndrome, Type I, <i>UGT1A1</i>	Cystic Fibrosis, <i>CFTR</i> ●
Cystinosis, <i>CTNS</i>	D-Bifunctional Protein Deficiency, <i>HSD17B4</i>	Deafness, Autosomal Recessive 77, <i>LOXHD1</i>
Duchenne Muscular Dystrophy, X-linked, <i>DMD</i> ●	Dystrophic Epidermolysis Bullosa (COL7A1-related), <i>COL7A1</i>	Ehlers-Danlos Syndrome, Type VIIC, <i>ADAMTS</i>
Emery-Dreifuss Muscular Dystrophy 1, X-Linked, <i>EMD</i>	Enhanced S-Cone Syndrome, <i>NR2E3</i>	Ethylmalonic Encephalopathy, <i>ETHE1</i>
Fabry Disease, X-Linked, <i>GLA</i>	Factor IX Deficiency, X-Linked, <i>F9</i>	Factor V Leiden Thrombophilia, <i>F5</i>
Factor XI Deficiency, <i>F11</i>	Familial Dysautonomia, <i>IKBKAP</i> ◆	Familial Hypercholesterolemia (LDLR-related), <i>LDLR</i>
Familial Mediterranean Fever, <i>MEFV</i>	Familial Nephrogenic Diabetes Insipidus (AQP2- related), <i>AQP2</i>	Fanconi Anemia, Type G, <i>FANCG</i>
Fanconi Anemia, Type C, <i>FANCC</i> ◆	Fragile X Syndrome, X-Linked, <i>FMR1</i> ●	Galactokinase Deficiency [Galactosemia, Type II], <i>GALK1</i>
Galactosemia, <i>GALT</i> ◆	Gaucher Disease, <i>GBA</i> ◆	Glutaric Acidemia, Type 1, <i>GCDH</i>
Glutaric Acidemia, Type 2A, <i>ETFA</i>	Glycine Encephalopathy (GLDC-related), <i>GLDC</i>	Glycine Encephalopathy (AMT-related), <i>AMT</i>
Glycogen Storage Disease, Type 1A, <i>G6PC</i>	Glycogen Storage Disease, Type 1B, <i>SLC37A4</i>	Glycogen Storage Disease, Type 2 [Pompe Disease], <i>GAA</i>
Glycogen Storage Disease, Type 3, <i>AGL</i>	Glycogen Storage Disease, Type 4, <i>GBE1</i>	Glycogen Storage Disease, Type 5 [McArdle Disease], <i>PYGM</i>
Glycogen Storage Disease, Type 7, <i>PFKM</i>	GRACILE Syndrome, <i>BCS1L</i>	Hemochromatosis, Type 2A, <i>HFE2</i>
Hemochromatosis, Type 3 (TFR2-related), <i>TFR2</i>	Hereditary Fructose Intolerance, <i>ALDOB</i>	Hermansky-Pudlak Syndrome (HPS1-related), <i>HPS1</i>
Hermansky-Pudlak Syndrome (HPS3-related), <i>HPS3</i>	Holocarboxylase Synthetase Deficiency, <i>HLCS</i>	Homocystinuria (CBS-related), <i>CBS</i>
Homocystinuria, Type c1E, <i>MTRR</i>	Hydrolethalus Syndrome, <i>HYLS1</i>	Hypohidrotic Ectodermal Dysplasia, X-Linked, <i>EDA</i>
Hypophosphatasia (ALPL-related), <i>ALPL</i>	Inclusion Body Myopathy Type 2, <i>GNE</i>	Isovaleric Acidemia, <i>IVD</i>

Joubert Syndrome, Type 2, <i>TMEM216</i>	Junctional Epidermolysis Bullosa, Herlitz type, <i>LAMC2</i>	Juvenile Retinoschisis, <i>X-Linked</i> , <i>RS1</i>
Krabbe Disease, <i>GALC</i>	Lamellar Ichthyosis, Type 1, <i>TGM1</i>	<i>Leber Congenital Amaurosis (LCA5-related)</i> , <i>LCA5</i>
Leber Congenital Amaurosis, Type CEP290, <i>CEP290</i>	Leigh Syndrome, French-Canadian Type, <i>LRPPRC</i>	Leukoencephalopathy with Vanishing White Matter, <i>EIF2B5</i>
Leydig Cell Hypoplasia [Luteinizing Hormone Resistance], <i>LHCGR</i>	Limb-Girdle Muscular Dystrophy, Type 2A, <i>CAPN3</i>	Limb-Girdle Muscular Dystrophy, Type 2C, <i>SGCG</i>
Limb-Girdle Muscular Dystrophy, Type 2B, <i>DYSF</i>	Limb-Girdle Muscular Dystrophy, Type 2C, <i>SGCG</i>	Limb-Girdle Muscular Dystrophy, Type 2D, <i>SGCA</i>
Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3], <i>DLSD</i>	Lipoid Adrenal Hyperplasia, <i>STAR</i>	Lipoprotein Lipase Deficiency, <i>LPL</i>
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency, <i>HADHA</i>	Lysinuric Protein Intolerance, <i>SLC7A7</i>	Maple Syrup Urine Disease, Type 1B, <i>BCKDHB</i>
Meckel-Gruber Syndrome, Type 1, <i>MKS1</i>	Medium Chain Acyl-CoA Dehydrogenase Deficiency, <i>ACADM</i> ◆	Megalencephalic Leukoencephalopathy with Subcortical Cysts, <i>MLC1</i>
Metachromatic Leukodystrophy (ARSA-related), <i>ARSA</i>	Metachromatic Leukodystrophy (PSAP-related) <i>PSAP</i>	Methylmalonic Aciduria (MMAA-related), <i>MMAA</i>
Methylmalonic Aciduria (MMAB-related), <i>MMAB</i>	Methylmalonic Aciduria and Homocystinuria, Type cblC, <i>MMACHC</i>	Methylmalonic Aciduria and Homocystinuria, Type cblD, <i>MMADHC</i>
Methylmalonic Aciduria, Type mut(O), <i>MUT</i>	Microphthalmia/Anophthalmia (VSX2-related), <i>VSX2</i>	Mitochondrial Complex 1 Deficiency (ACAD9-related), <i>ACAD9</i>
Mitochondrial Complex 1 Deficiency (NDUFAF5-related), <i>NDUFAF5</i>	Mitochondrial Complex 1 Deficiency (NDUFS6-related), <i>NDUFS6</i>	Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1), <i>PUS1</i>
Mucopolipidosis II/III, <i>GNPTAB</i>	Mucopolipidosis III Gamma, <i>GNPTG</i>	Mucopolipidosis, Type IV, <i>MCOLN1</i> ◆
Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked, <i>IDS</i>	Mucopolysaccharidosis, Type IIIB [Sanfilippo B], <i>NAGLU</i>	Mucopolysaccharidosis, Type IIIC [Sanfilippo C], <i>HGSNAT</i>
Mucopolysaccharidosis IIID [Sanfilippo D], <i>GNS</i>	Mucopolysaccharidosis, Type IX, <i>HYAL1</i>	Multiple Sulfatase Deficiency, <i>SUMF1</i>
Myoneurogastrointestinal Encephalopathy (MNGIE), <i>TYMP</i>	Myotubular Myopathy, X-Linked, <i>MTM1</i>	N-acetylglutamate Synthase Deficiency, <i>NAGS</i>
Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome], <i>MPV17</i>	Neurological Ceroid Lipofuscinosis, TPP1-related, <i>TPP1</i>	Neuronal Ceroid Lipofuscinosis (MFSD8-related), <i>MFSD8</i>
Neuronal Ceroid Lipofuscinosis (CLN5-related), <i>CLN5</i>	Neuronal Ceroid Lipofuscinosis (CLN6-related), <i>CLN6</i>	Neuronal Ceroid Lipofuscinosis (CLN8-related), <i>CLN8</i>
Neuronal Ceroid Lipofuscinosis (PPT1-related), <i>PPT1</i>	Niemann-Pick Disease, Types A/B, <i>SMPD1</i> ◆	Niemann-Pick Disease, Type C1/D, <i>NPC1</i>
Niemann-Pick Disease, Type C2, <i>NPC2</i>	Nijmegen Breakage Syndrome, <i>NBN</i>	Non-Syndromic Hearing Loss (GJB2-related), <i>GJB2</i>
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome, <i>WNT10A</i>	Omenn Syndrome, RAG2-related, <i>RAG2</i>	Ornithine Aminotransferase Deficiency, <i>OAT</i>
Ornithine Transcarbamylase Deficiency, <i>OTC</i>	Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome], <i>SLC25A15</i>	Pendred Syndrome, <i>SLC26A4</i>
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related), <i>PEX1</i>	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related), <i>PEX2</i>	Phenylketonurea, <i>PAH</i> ◆
Pituitary Hormone Deficiency, Combined 3, <i>LHX3</i>	Pontocerebellar Hypoplasia, RARS2-related, <i>RARS2</i>	Pontocerebellar Hypoplasia, Type 1A, <i>VRK1</i>
Pontocerebellar Hypoplasia, Type 2D, <i>SEPSECS</i>	Pontocerebellar Hypoplasia, Type 2E, <i>VPS53</i>	Primary Ciliary Dyskinesia (DNAH5-related), <i>DNAH5</i>
Primary Ciliary Dyskinesia, DNAI1-related, <i>DNAI1</i>	Primary Ciliary Dyskinesia, DNAI2-related, <i>DNAI2</i>	Primary Hyperoxaluria, Type 1, <i>AGXT</i>
Primary Hyperoxaluria, Type 2, <i>GRHPR</i>	Primary Hyperoxaluria, Type 3, <i>HOGA1</i>	Pycnodysostosis, <i>CTSK</i>
Pyruvate Dehydrogenase Deficiency (PDHB-related), <i>PDHB</i>	Pyruvate Dehydrogenase Deficiency, X-Linked, <i>PDHA1</i>	Renal Tubular Acidosis and Deafness (ATP6V1B1-related), <i>ATP6V1B1</i>
Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy], <i>RLBP</i>	Retinitis Pigmentosa 59 (DHDDS-related), <i>DHDDS</i>	Retinitis Pigmentosa 25 (EYS-related), <i>EYS</i>
Retinitis Pigmentosa 26, <i>CERKL</i>	Retinitis Pigmentosa 28, <i>FAM161A</i>	Retinitis Pigmentosa, X-linked, <i>RPGR</i>
Rhizomelic Chondrodysplasia Punctata, Type 1, <i>PEX7</i>	Rhizomelic Chondrodysplasia Punctata, Type 3, <i>AGPS</i>	Roberts Syndrome, <i>ESCO2</i>
Salla Disease, <i>SLC17A5</i>	Sandhoff Disease, <i>HEXB</i>	Schimke Immunoosseous Dysplasia, <i>SMARCA1</i>
Segawa Syndrome, TH-related, <i>TH</i>	Severe Combined Immunodeficiency, Type Athabaskan, <i>DCLRE1C</i>	Severe Combined Immunodeficiency, X-Linked, <i>IL2RG</i>
Sickle-Cell Disease, <i>HBB</i> ●	Sjögren-Larsson Syndrome, <i>ALDH3A2</i>	Smith-Lemli-Opitz Syndrome, <i>DHCR7</i> ◆
Spinal Muscular Atrophy, <i>SMN1</i> , <i>SMN2</i> ●	Steroid-Resistant Nephrotic Syndrome, <i>NPHS2</i>	Stuve-Wiedemann Syndrome, <i>LIFR</i>
Tay-Sachs Disease, <i>HEXA</i> ◆	Tyrosinemia, Type 1, <i>FAH</i>	Usher Syndrome, Type 1C, <i>USH1C</i>
Usher Syndrome, Type 1F, <i>PCDH15</i>	Usher Syndrome, Type 2A, <i>USH2A</i>	Usher Syndrome, Type 3, <i>CLRN1</i>
Wilson Disease, <i>ATP7B</i>	Wolman Disease, <i>LIPA</i>	Zellweger Spectrum Disorders, (PEX6-related), <i>PEX6</i>
Zellweger Spectrum Disorders (PEX10-related), <i>PEX10</i>		

● AVAILABLE AS FOCUS PANELS OR PART OF GUIDELINES-BASED PANEL

◆ INCLUDED IN GUIDELINES-BASED PANEL