Condition List: Super Panel

Screens 145/147 Conditions

3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) Lyase Deficiency: *HMGCL*

3-Methylcrotonyl-CoA Carboxylase (3-MCC) Deficiency, MCCC1, MCCC2-Related: *MCCC1/MCCC2*

3-Methylglutaconic Aciduria Type III (Costeff Optic Atrophy): *OPA3*

Abetalipoproteinemia: *MTTP*

Achromatopsia: *CNGB3*

ADA-Related Conditions: *ADA*

Alkaptonuria: *HGD*

Alpha-Thalassemia/Alpha-Globin Triplication: *HBA1/HBA2*

Alpha-Mannosidosis: *MAN2B1*

Alport Syndrome, COL4A4-Related: *COL4A4*

Andermann Syndrome: *SLC12A6*

Argininosuccinic Aciduria: *ASL*

Arthrogryposis, Mental Retardation, and Seizures (AMRS): *SLC35A3*

Aspartylglucosaminuria: *AGA*

Ataxia-Telangiectasia: *ATM*

Ataxia With Vitamin E Deficiency: *TTPA*

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS): *SACS*

Bardet-Biedl Syndrome, BBS1-Related: *BBS1*

Bardet-Biedl Syndrome, BBS10-Related: *BBS10*

Bardet-Biedl Syndrome, MKKS-Related: *MKKS*

Beta-Ketothiolase Deficiency: *ACAT1*

Beta-Thalassemia: HBB Biotinidase Deficiency: *BTD*

Bloom Syndrome: *BLM*

Canavan Disease: *ASPA*

Carnitine Palmitoyltransferase I Deficiency: *CPT1A*

Carnitine Palmitoyltransferase II Deficiency: *CPT2*

Cartilage-Hair Hypoplasia, Anauxetic Dysplasia Spectrum Disorders: *RMRP*

Choroideremia: *CHM**

Citrin Deficiency: *SLC25A13*

Citrullinemia Type 1: *ASS1*

Cohen Syndrome: *VPS13B*

Combined Pituitary Hormone Deficiency: *PROP1*

Congenital Amegakaryocytic Thrombocytopenia: *MPL*

Congenital Disorder of Glycosylation Type Ia: *PMM2*

Congenital Disorder of Glycosylation Type Ib: *MPI*

Cystic Fibrosis and Other CFTR-Related Disorders: *CFTR*

Cystinosis: *CTNS*

D-Bifunctional Protein Deficiency: *HSD17B4*

DHDDS-Related Disorders (including Retinitis Pigmentosa 59): *DHDDS*

Dihydrolipoamide Dehydrogenase Deficiency: *DLD*

Dyskeratosis Congenita: *NOLA3*

Ehlers-Danlos Syndrome, Dermatosparaxis Type: *ADAMTS2*

Familial Dysautonomia: *ELP1*

Familial Hyperinsulinism ABCC8-Related: *ABCC8*

Familial Mediterranean Fever: *MEFV*

Fanconi Anemia Type C: *FANCC*

FKTN-Related Disorders (including Walker-Warburg Syndrome): *FKTN*

Fragile X Syndrome and FMR1-Related Disorders: *FMR1**

Galactokinase Deficiency: *GALK1*

Galactosemia, GALT-Related: *GALT*

Gaucher Disease: *GBA*

Glucose-6-Phosphate Dehydrogenase Deficiency: *G6PD**

Glutaric Acidemia Type 1: *GCDH*

Glycine Encephalopathy, AMT-Related: *AMT*

Glycine Encephalopathy, GLDC-Related: *GLDC*

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*

GRACILE Syndrome/BCS1L-Related Disorders: *BCS1L*

Hemophilia C/Factor XI Deficiency: *F11*

Hereditary Fructose Intolerance: *ALDOB*

Hereditary Thymine-Uraciluria: *DPYD*

Holocarboxylase Synthetase Deficiency: *HLCS*

Homocystinuria Due to Cystathionine Beta-Synthase Deficiency: *CBS*

Homocystinuria, Cobalamin E Type: *MTRR*

Hypophosphatasia: *ALPL*

Inclusion Body Myopathy 2: *GNE*

Isovaleric Acidemia: *IVD*

**X-linked conditions*

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Joubert Syndrome 2/TMEM216-Related Disorders: *TMEM216*

Junctional Epidermolysis Bullosa, LAMA3-Related: *LAMA3*

Junctional Epidermolysis Bullosa, LAMB3-Related: *LAMB3*

Junctional Epidermolysis Bullosa, LAMC2-Related: *LAMC2*

Krabbe Disease: *GALC*

Limb-Girdle Muscular Dystrophy Type 2A: *CAPN3*

Limb-Girdle Muscular Dystrophy Type 2D: *SGCA*

Limb-Girdle Muscular Dystrophy Type 2E: *SGCB*

Lipoid Congenital Adrenal Hyperplasia, STAR-Related: *STAR*

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency: *HADHA*

Lysosomal Acid Lipase Deficiency: *LIPA*

Maple Syrup Urine Disease Type 1A: *BCKDHA*

Maple Syrup Urine Disease Type 1B: *BCKDHB*

Medium Chain Acyl-CoA Dehydrogenase Deficiency: *ACADM*

Megalencephalic Leukoencephalopathy with Subcortical Cysts Type 1: *MLC1*

Metachromatic Leukodystrophy, ARSA-Related: *ARSA*

Methylmalonic Acidemia with Homocystinuria, Cobalamin C Type: *MMACHC*

Methylmalonic Acidemia with Homocystinuria, Cobalamin D Type: *MMADHC*

Methylmalonic Acidemia, MMAA, MMAB-Related: *MMAA/MMAB*

Mucolipidosis Type IV: *MCOLN1*

Mucopolysaccharidosis Type I (Hurler Syndrome): *IDUA*

Multiple Sulfatase Deficiency: *SUMF1*

Nemaline Myopathy 2: *NEB*

Nephrotic Syndrome/Congenital Finnish Nephrosis, NPHS1-Related: *NPHS1*

Nephrotic Syndrome/Steroid-Resistant Nephrotic Syndrome, NPHS2-Related: *NPHS2*

Neuronal Ceroid Lipofuscinosis, CLN3-Related: *CLN3*

Neuronal Ceroid Lipofuscinosis, CLN5-Related: *CLN5*

Neuronal Ceroid Lipofuscinosis/Northern Epilepsy, CLN8-Related: *CLN8*

Neuronal Ceroid Lipofuscinosis, PPT1-Related: *PPT1*

Neuronal Ceroid Lipofuscinosis, TPP1-Related: *TPP1*

Niemann-Pick Type A/B: *SMPD1*

Niemann-Pick Type C, NPC1-Related: *NPC1*

Nijmegen Breakage Syndrome: *NBN*

Nonsyndromic Hearing Loss: *GJB2/GJB3/GJB6*

Pendred Syndrome: *SLC26A4/FOXI1/KCNJ10*
(also associated with SeSAME syndrome)

Phenylalanine Hydroxylase Deficiency: *PAH*

Phosphoglycerate Dehydrogenase Deficiency/ Neu-Laxova Syndrome: *PHGDH*

Polycystic Kidney Disease, Autosomal Recessive: *PKHD1*

Polyglandular Autoimmune Syndrome Type 1: *AIRE*

POMGNT1-Related Disorders: *POMGNT1*

Primary Carnitine Deficiency: *SLC22A5*

Primary Hyperoxaluria Type 1, AGXT-Related: *AGXT*

Primary Hyperoxaluria Type 2, GRHPR-Related: *GRHPR*

Propionic Acidemia, PCCA-Related: *PCCA*

Propionic Acidemia, PCCB-Related: *PCCB*

Pseudocholinesterase Deficiency: *BCHE*

Pycnodysostosis: *CTSK*

Rhizomelic Chondrodysplasia Punctata Type 1/Refsum Disease: *PEX7*

Salla Disease: *SLC17A5*

Short Chain Acyl-CoA Dehydrogenase Deficiency: *ACADS*

Sickle Cell (HbS) and HbC Disease: *HBB*

Sjögren-Larsson Syndrome: *ALDH3A2*

SLC26A2-Related Disorders: *SLC26A2*

Smith-Lemli-Opitz Syndrome: *DHCR7*

Spinal Muscular Atrophy: *SMN1/SMN2*

Tay-Sachs Disease: *HEXA*

Tyrosine Hydroxylase Deficiency: *TH*

Tyrosinemia Type I: *FAH*

Tyrosinemia Type II: *TAT*

Usher Syndrome Type IB/MYO7A-Related Disorders: *MYO7A*

Usher Syndrome Type ID/CDH23-Related Disorders: *CDH23*

Usher Syndrome Type IF/PCDH15-Related Disorders: *PCDH15*

Usher Syndrome Type IIA/USH2A-Related Disorders: *USH2A*

Usher Syndrome Type IIIA: *CLRN1*

Very Long Chain Acyl-CoA Dehydrogenase Deficiency: *ACADVL*

Wilson Disease: *ATP7B*

X-Linked Adrenoleukodystrophy: *ABCD1**

X-Linked Congenital Adrenal Hypoplasia: *NR0B1**

X-Linked Juvenile Retinoschisis: *RS1**

Zellweger Syndrome Spectrum, PEX1-Related: *PEX1*

Available Upon Request (Super Panel 147)

Congenital Adrenal Hyperplasia: *HSD3B2*, *CYP11B1*, *CYP17A1*, *CYP21A2*

DMD-Related Dystrophinopathy (Duchenne Muscular Dystrophy and Becker Muscular Dystrophy): *DMD**

**X-linked conditions*

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