Condition List: Early Advantage Panel

Screens 68 Conditions

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

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

ADA-Related Conditions: *ADA*

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

Argininosuccinic Aciduria: *ASL*

Beta-Ketothiolase Deficiency: *ACAT1*

Beta-Thalassemia: *HBB*

Biotinidase Deficiency: *BTD*

Bloom Syndrome: *BLM*

Canavan Disease: *ASPA*

Carnitine Palmitoyltransferase II Deficiency: *CPT2*

Citrin Deficiency: *SLC25A13*

Citrullinemia Type 1: *ASS1*

Congenital Disorder of Glycosylation, PMM2-Related: *PMM2*

Cystic Fibrosis and CFTR-Related Disorders: *CFTR*

Dihydrolipoamide Dehydrogenase Deficiency: *DLD*

Familial Dysautonomia: *ELP1*

Familial Hyperinsulinism, ABCC8-Related: *ABCC8*

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 I: *GCDH*

Glycogen Storage Disease Type IA: *G6PC*

Glycogen Storage Disease Type II (Pompe Disease): *GAA*

Glycogen Storage Disease Type IV/Adult Polyglucosan Body Disease: *GBE1*

Holocarboxylase Synthetase Deficiency: *HLCS*

Homocystinuria, Cobalamin E Type: *MTRR*

Homocystinuria due to Cystathionine Beta-Synthase Deficiency: CBS

Isovaleric Acidemia: *IVD*

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

Krabbe Disease: *GALC*

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

Maple Syrup Urine Disease Type 1A: *BCKDHA*

Maple Syrup Urine Disease Type 1B: *BCKDHB*

Medium Chain Acyl-CoA Dehydrogenase Deficiency: *ACADM*

Metachromatic Leukodystrophy, ARSA-Related: *ARSA*

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

Methylmalonic Acidemia with Homocystinuria, Cobalamin C Type: M*MACHC*

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

Mucolipidosis IV: *MCOLN1*

Mucopolysaccharidosis Type I (Hurler Syndrome): *IDUA*

Nemaline Myopathy 2: *NEB*

Niemann-Pick Disease Type A/B: *SMPD1*

Nonsyndromic Hearing Loss: *GJB2/GJB3/GJB6*

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

Phenylalanine Hydroxylase Deficiency: *PAH*

Polycystic Kidney Disease, Autosomal Recessive: *PKHD1*

Primary Carnitine Deficiency: *SLC22A5*

Propionic Acidemia, PCCA, PCCB-Related: *PCCA/PCCB*

SLC26A2-Related Disorders: *SLC26A2*

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

Smith-Lemli-Opitz Syndrome: *DHCR7*

Spinal Muscular Atrophy: *SMN1/SMN2*

Tay-Sachs Disease: *HEXA*

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 conditions*

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