JScreen Test Panel

The following genetic diseases are included on the test panel. **Conditions in bold are common in those with Jewish ancestry.**

- **21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia**
- **ABCC8-Related Hyperinsulinism**
- Achromatopsia
- **Alpha Thalassemia**
- Alpha-Mannosidosis
- Andermann Syndrome
- ARSACS
- Aspartylglycosaminuria
- Ataxia With Vitamin E Deficiency
- **Ataxia-Telangiectasia**
- **Autosomal Recessive Polycystic Kidney Disease**
- Bardet-Biedl Syndrome, BBS10-Related
- Bardet-Biedl Syndrome, BBS1-Related
- **Beta Thalassemia**
- Biotinidase Deficiency
- **Bloom Syndrome**
- **Canavan Disease**
- **Carnitine Palmitoyltransferase IA Deficiency**
- Carnitine Palmitoyltransferase II Deficiency
- Cartilage-Hair Hypoplasia
- Choroideremia
- Citrullinemia Type 1
- CLN3-Related Neuronal Ceroid Lipofuscinosis
- CLN5-Related Neuronal Ceroid Lipofuscinosis
- Cohen Syndrome
- **Congenital Disorder of Glycosylation Type Ia**
- Congenital Disorder of Glycosylation Type Ib
- Congenital Finnish Nephrosis
- **Costeff Optic Atrophy Syndrome**
- **Cystic Fibrosis**
- Cystinosis
- D-Bifunctional Protein Deficiency
- **Factor XI Deficiency**
- **Familial Dysautonomia**
- **Familial Mediterranean Fever**
- **Fanconi Anemia Type C**
- **Fragile X Syndrome**
- **Galactosemia**
- **Gaucher Disease**
- GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness
- **Glucose-6-Phosphate Dehydrogenase Deficiency**
- Glutaric Acidemia Type 1
- **Glycogen Storage Disease Type Ia**
- Glycogen Storage Disease Type Ib
- **Glycogen Storage Disease Type III**
- Glycogen Storage Disease Type V
- GRACILE Syndrome
- Hereditary Fructose Intolerance
- Hereditary Thymine-Uraciluria
- Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related
- Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related
- Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related
- Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency
- Hurler Syndrome
- Hypophosphatasia, Autosomal Recessive
- **Inclusion Body Myopathy 2**
- Isovaleric Acidemia
- **Joubert Syndrome 2**
- Krabbe Disease
- Limb-Girdle Muscular Dystrophy Type 2D
- Limb-Girdle Muscular Dystrophy Type 2E
- **Lipoamide Dehydrogenase Deficiency**
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
- **Maple Syrup Urine Disease Type 1B**
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- **Megalencephalic Leukoencephalopathy With Subcortical Cysts**
- **Metachromatic Leukodystrophy**
- **Mucolipidosis IV**
- Muscle-Eye-Brain Disease
- **NEB-Related Nemaline Myopathy**
- Niemann-Pick Disease Type C
- **Niemann-Pick Disease, SMPD1-Associated**
- Nijmegen Breakage Syndrome
- Northern Epilepsy
- Pendred Syndrome
- PEX1-Related Zellweger Syndrome Spectrum
- **Phenylalanine Hydroxylase Deficiency**
- **Polyglandular Autoimmune Syndrome Type 1**
- Pompe Disease
- PPT1-Related Neuronal Ceroid Lipofuscinosis
- Primary Carnitine Deficiency
- Primary Hyperoxaluria Type 1
- Primary Hyperoxaluria Type 2
- PROP1-Related Combined Pituitary Hormone Deficiency
- **Pseudocholinesterase Deficiency**
- Pycnodysostosis
- Rhizomelic Chondrodysplasia Punctata Type 1
- Salla Disease
- Segawa Syndrome
- Short Chain Acyl-CoA Dehydrogenase Deficiency
- **Sickle Cell Disease**
- Sjogren-Larsson Syndrome
- **Smith-Lemli-Opitz Syndrome**
- **Spinal Muscular Atrophy**
- Steroid-Resistant Nephrotic Syndrome
- Sulfate Transporter-Related Osteochondrodysplasia
- **Tay-Sachs Disease**
- TPP1-Related Neuronal Ceroid Lipofuscinosis
- **Tyrosinemia Type I**
- **Usher Syndrome Type 1F**
- **Usher Syndrome Type 3**
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency
- **Walker Warburg syndrome**
- **Wilson Disease**
- X-Linked Juvenile Retinoschisis