Carrier Screening
for Inherited Genetic Disorders

Certain genetic disorders have a similar incidence across all ethnicities; others occur more frequently in a select ethnic group. Because these disorders are inherited in an autosomal recessive or X-linked manner, your patient may be at risk for being a carrier for a genetic disorder without even knowing it. We offer carrier screening for a large number of diseases.

What kind of disorders can Mount Sinai Genetic Testing Laboratory screen for?

Each disease we offer screening for can severely impact an individual's life in one way or another:

- Some diseases shorten lifespan
- Others have no treatment or curative options
- Certain diseases cause physical impairment
- While others cause mental impairment
- Most diseases require lifelong treatment and management
- Oftentimes, multiple organ systems are affected
- Some disorders are lethal *in utero*

What diseases should my patients be tested for?

Talk to your account manager about developing one or several customized panels to suit your patients' needs.

Suggested panel for individuals of Ashkenazi Jewish background:

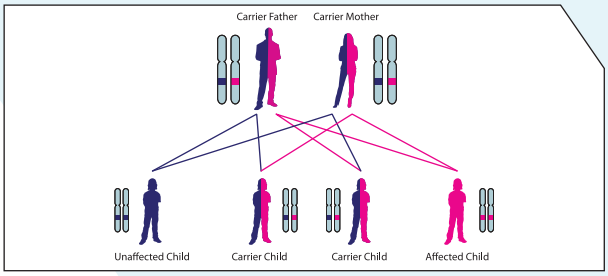
- *Abetalipoproteinemia*
- *Alport Syndrome, Autosomal Recessive*
- *Arthrogryposis, Mental Retardation and Seizures*
- *Bardet-Biedl Syndrome, BBS2-Related*
- *Bloom Syndrome*
- *Canavan Disease*
- *Carnitine Palmitoyltransferase II Deficiency*
- *Congenital Amegakaryocytic Thrombocytopenia*
- *Congenital Disorder of Glycosylation Ia*
- *Cystic Fibrosis*
- *Dyskeratosis Congenita, Autosomal Recessive*
- *Ehlers-Danlos VIIC*
- *Familial Dysautonomia*
- *Familial Hyperinsulinism, ABCC8-Related*
- *Fanconi Anemia C*
- *Fragile X Syndrome*
- *Galactosemia*
- *Gaucher Disease*
- *Glycogen Storage Disease Ia*
- *Joubert Syndrome 2*
- *Lipoamide Dehydrogenase Deficiency (E3)*
- *Maple Syrup Urine Disease Ib*
- *Mucolipidosis IV*
- *Multiple Sulphatase Deficiency*
- *Nemaline Myopathy, NEB-Related*
- *Niemann-Pick Disease A and B*
- *3-Phosphoglycerate Dehydrogenase Deficiency*
- *Polycystic Kidney Disease, Autosomal Recessive*
- *Retinitis Pigmentosa 59*
- *Smith-Lemli-Opitz Syndrome*
- *Spinal Muscular Atrophy*
- *Tay-Sachs disease (by DNA and enzyme)*
- *Tyrosinemia I*
- *Usher Syndrome IF*
- *Usher Syndrome III*
- *Walker-Warburg Syndrome, FKTN-Related*
- *Wilson Disease*
- *Zellweger Syndrome, PEX2-Related*

Suggested standard pan-ethnic panel for individuals of non-Jewish background*:

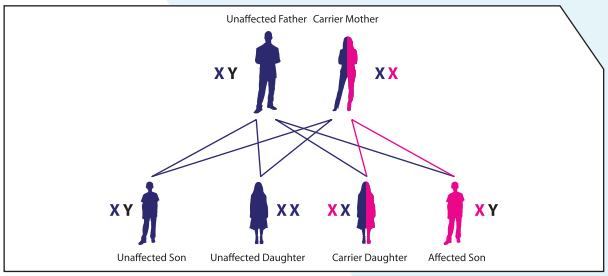
- *Cystic Fibrosis*
- *Fragile X Syndrome*
- *Spinal Muscular Atrophy (including enhanced SMA testing for more accurate residual risk estimates)*
- *Smith-Lemli-Opitz Syndrome*

** Screening for hemoglobinopathies by CBC and Hemoglobin Electrophoresis is recommended for individuals of African, Asian, Hispanic, and Mediterranean ethnicity. Follow up DNA testing is available through Mount Sinai Genetic Testing Laboratory.*

How are these disorders inherited?



Autosomal Recessive: *(Screening of both members of a couple can occur concurrently or sequentially).*



X-Linked: *(Screening for these conditions usually takes place on the female member of the couple).*

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Mount Sinai Genetic Testing Laboratory Expanded Pan-Ethnic Panel

The following is a list of all the diseases for which carrier screening may be ordered:

Abetalipoproteinemia*
Achromatopsia
Alkaptonuria
Alpha-Mannosidosis
Alport Syndrome, Autosomal Recessive*
Andermann Syndrome
ARSACS
Aspartylglycosaminuria
Ataxia with Vitamin E Deficiency
Ataxia-Telangiectasia
Arthrogryposis, Mental Retardation and Seizures*
Bardet-Biedl Syndrome, *BBS1*-Related
Bardet-Biedl Syndrome, *BBS2*-Related*
Bardet-Biedl Syndrome, *BBS10*-Related
Beta Thalassemia
Biotinidase Deficiency
Bloom Syndrome*
Canavan Disease*
Carnitine Palmitoyltransferase IA Deficiency
Carnitine Palmitoyltransferase II Deficiency*
Cartilage-Hair Hypoplasia
Choroideremia
Citrullinemia 1
CLN3-Related Neuronal Ceroid Lipofuscinosis
CLN5-Related Neuronal Ceroid Lipofuscinosis
Cohen Syndrome
Congenital Amegakaryocytic Thrombocytopenia*
Congenital Disorder of Glycosylation Ia*
Congenital Disorder of Glycosylation Ib
Congenital Finnish Nephrosis
Costeff Optic Atrophy Syndrome
Cystic Fibrosis*
Cystinosis
D-Bifunctional Protein Deficiency
Dyskeratosis Congenita, Autosomal Recessive *
Ehlers-Danlos VIIC*
Familial Dysautonomia*
Familial Hyperinsulinism, *ABCC8*-Related *
Familial Mediterranean Fever
Fanconi Anemia C*
Fragile X Syndrome*
Galactosemia*
Gaucher Disease*
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness (Connexin 26)
Glutaric Acidemia 1
Glycogen Storage Disease Ia*
Glycogen Storage Disease Ib
Glycogen Storage Disease III
Glycogen Storage Disease 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 2D
Limb-Girdle Muscular Dystrophy 2E
Lipoamide Dehydrogenase Deficiency (E3)*
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
Maple Syrup Urine Disease Ib*
Medium Chain Acyl-CoA Dehydrogenase deficiency
Megalencephalic Leukoencephalopathy with Subcortical Cysts
Metachromatic Leukodystrophy
Mucolipidosis IV*
Multiple Sulphatase Deficiency*
Muscle-Eye-Brain Disease
Nemaline Myopathy, *NEB*-Related*
Niemann-Pick Disease C
Niemann-Pick Disease A and B*
Nijmegen Breakage Syndrome
Northern Epilepsy
Pendred Syndrome
Phenylalanine Hydroxylase Deficiency
3-Phosphoglycerate Dehydrogenase Deficiency*
Polycystic Kidney Disease, Autosomal Recessive*
Polyglandular Autoimmune Syndrome 1
Pompe Disease
PPT1-Related Neuronal Ceroid Lipofuscinosis
Primary Carnitine Deficiency
Primary Hyperoxaluria 1
Primary Hyperoxaluria 2
PROP1-Related Combined Pituitary Hormone Deficiency
Pycnodysostosis
Retinitis Pigmentosa 59*
Rhizomelic Chondrodysplasia Punctata 1
Salla Disease
Sandhoff Disease (by enzyme)
Segawa Syndrome
Sickle Cell Disease
Sjogren-Larsson Syndrome
Smith-Lemli-Opitz Syndrome*
Spinal Muscular Atrophy*
Steroid-Resistant Nephrotic Syndrome
Sulfate Transporter-Related Osteochondrodysplasia
Tay-Sachs disease (by DNA and enzyme)*
TPP1-Related Neuronal Ceroid Lipofuscinosis
Tyrosinemia I*
Usher Syndrome IF*
Usher Syndrome III*
Very Long Chain Acyl-CoA Dehydrogenase Deficiency
Walker-Warburg Syndrome, *FKTN*-Related*
Wilson Disease*
X-Linked Juvenile Retinoschisis
Zellweger Syndrome, *PEX1*-Related
Zellweger Syndrome, *PEX2*-Related*

**Diseases included in our Askhkenzai Jewish Panel*

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