Universal Genetic Test

The Universal Genetic Test uses targeted DNA mutation analysis to simultaneously determine the carrier status of an individual or couple for 100+ Mendelian diseases. Counsyl reports which mutations, if any, were detected for each disease. The risk of conceiving a child affected with a disease is presented below and calculated using the test results as well as published data on each disease. The child risk summary is provided as an aid to genetic counseling.

Partner

DNA test shows that he is a carrier of HFE-associated hereditary hemochromatosis.

Child Risk Summary

Your Universal Genetic Test indicates that your future children have a reduced risk for the diseases tested, including those listed below which are common in your ethnicity. Note that child risks are not calculated for mild diseases, including HFE-associated hereditary hemochromatosis, which are described in the next section.

- Autosomal Recessive Polycystic Kidney Disease
- Cystic Fibrosis
- Phenylalanine Hydroxylase Deficiency
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Spinal Muscular Atrophy

*Limitations:* Interpretation is given as a probability due to the inheritance pattern of these diseases and because only targeted mutations are detected. Other nearby genetic variants may interfere with this detection. Inaccurate reporting of ethnicity or clinical information may cause errors in risk calculation.
Mild Disease Summary

The chart below shows carrier status for 3 mild diseases. The conditions on this list have been highlighted because they are extremely common in the general population and usually do not cause major health problems. In many cases, individuals with these mild conditions remain asymptomatic. For this reason, the results in this section of the report are unlikely to influence reproductive choices. However for those who do show symptoms, knowledge of one’s genetic status for these conditions can be helpful to recognize the disease and direct treatment.

<table>
<thead>
<tr>
<th>Mild Disease</th>
<th>Status</th>
</tr>
</thead>
<tbody>
<tr>
<td>Factor V Leiden Thrombophilia</td>
<td>No disease-causing mutations detected.</td>
</tr>
<tr>
<td>Glucose-6-Phosphate Dehydrogenase Deficiency</td>
<td>No disease-causing mutations detected.</td>
</tr>
</tbody>
</table>

For details on HFE-associated hereditary hemochromatosis, see page 3.
Mild Disease Positive Report: HFE-Associated Hereditary Hemochromatosis

This disease report is included due to positive result for

Patient Results

<table>
<thead>
<tr>
<th>Result</th>
<th>No partner tested</th>
</tr>
</thead>
<tbody>
<tr>
<td>HFE:p.Cys282Tyr (C282Y) heterozygote.</td>
<td>N/A</td>
</tr>
</tbody>
</table>

Interpretation:
This individual is a carrier of HFE-associated hereditary hemochromatosis. Carriers generally do not experience symptoms. Clinical symptoms are also uncommon in C282Y homozygotes.

Variants on the Counsyl panel

What is HFE-Associated Hereditary Hemochromatosis?

HFE-associated hereditary hemochromatosis (HFE-HHC) is a common and treatable inherited disease in which the body absorbs and stores too much iron, potentially damaging organs such as the liver, heart, and pancreas. If the disease is diagnosed and treated before symptoms develop, people with HFE-HHC typically have a normal lifespan. If the disease is untreated, however, it can lead to fatal liver and heart failure.

For reasons not well understood, the majority of people with the genetic mutations that cause HFE-HHC do not develop symptoms of the disease at any point in their lives. For these people, simple blood tests can determine whether or not the body is storing too much iron. If it is, beginning treatment early can leave a person virtually symptom-free for life.

Depending on the specific mutation(s) a person has, he or she can be more or less likely to develop the iron overload symptoms of HFE-HHC.

People who have two copies of the C282Y mutation are most likely to have dangerously elevated levels of iron in their blood. Studies have found that among those with the C282Y/C282Y combination, men are more likely to develop symptoms of iron overload than women, perhaps because women's menstrual cycles lower their iron levels on a regular basis. Do keep in mind, however, that the majority of people with two copies of the C282Y mutation do not develop any symptoms of HFE-HHC.

Those who have C282Y in combination with another HFE-HHC mutation are much less likely to develop symptoms of the disease. Only 0.5% to 2% of people with C282Y in combination with another mutation are thought to have clinical signs of the disease. People with this genetic combination who have another disease of the liver may be more likely to develop HFE-HHC symptoms.

Among people who have two copies of any other HFE-HHC mutation, including a very common mutation known as H63D, the likelihood of developing symptoms is extremely low. In the absence of another liver disease, two copies of any HFE-HHC mutation other than C282Y is unlikely to cause any health problems.

In men who have not been treated for HFE-HHC, the first symptoms of the disease typically begin between the ages of 30 to 50; for untreated women, symptoms usually begin later, after menopause.
Early symptoms often include weakness, abdominal pain, joint pain, weight loss, loss of interest in sex, chest pain, and a progressive gray or bronze pigmentation to the skin. Liver disease (either fibrosis or the more serious cirrhosis) is a common problem associated with HFE-HHC. Cirrhosis can lead to fatal liver failure and/or an increased likelihood of developing cancer of the liver.

The heart can also be affected by HFE-HHC, seen as an irregular heartbeat and/or congestive heart failure. Other problems caused by HFE-HHC can include diabetes, arthritis, impotence (in men), early menopause (in women), thyroid problems, and adrenal gland problems.

How Common is HFE-Associated Hereditary Hemochromatosis?

HFE-HHC mutations are extremely common, particularly among Caucasians. Roughly one-third (33%) of Caucasians are carriers of the condition, most commonly the H63D mutation. The H63D mutation is almost always associated with asymptomatic cases unless paired with the C282Y mutation. In the general population, 1 in 200 to 400 has two copies of the C282Y genetic mutation, the combination of mutations which is most likely to cause symptoms of HFE-HHC.

Please bear in mind that most people who have these genetic mutations do not develop the disease.

The disease is less common among Hispanics, African Americans, Asians, and Native Americans. Roughly 13% of Hispanics, 8.5% of Asians, and 6% of African Americans is a carrier for the mild mutation, H63D. An additional 3% of Hispanics, 2.3% of African Americans are carriers of the potentially disease-causing C282Y mutation.

How is HFE-Associated Hereditary Hemochromatosis Treated?

Ideally HFE-HHC is treated before the organs of the body are damaged. However, not everyone who has the mutations that cause HFE-HHC develops symptoms or requires treatment. A simple blood test of iron levels in the blood—physicians specifically look at serum ferritin concentration and transferrin-iron saturation levels—can determine whether the body is absorbing too much. When iron reaches a certain threshold, treatment is recommended. If iron levels have not reached that threshold, no treatment is necessary. Blood tests must be repeated periodically to check these iron levels.

If a person has a high level of iron, treatment involves removing a certain quantity of blood at regular intervals. This is known as phlebotomy. Typically phlebotomy is performed frequently—perhaps weekly or twice weekly—until certain iron levels are reached, and then performed less frequently—often 2 to 4 times a year—on an indefinite basis. This treatment is simple, inexpensive, and safe.

If a person is already suffering from symptoms of HFE-HHC, treatment can lessen or relieve some of the symptoms. Cirrhosis is unlikely to improve with treatment, although treatment may slow its progression. If liver disease has reached severe levels, liver transplantation may be an option. Those who have any amount of liver damage are advised to avoid alcohol.

All people with symptoms of HFE-HHC are advised to eat only moderate amounts of iron-rich foods, avoid taking iron supplements or excess vitamin C, and refrain from eating uncooked seafood, as they are highly susceptible to a particular kind of bacterial infection.

What is the Prognosis for Someone With HFE-Associated Hereditary Hemochromatosis?

The prognosis for a person with the genetic mutations that cause HFE-HHC is generally good, as the majority of people in that situation do not develop symptoms of the disease. Most will not have dangerously elevated levels of iron in their blood, and therefore will not have any iron-overload problems.

For those that do have dangerously high iron levels in their blood, beginning treatment before symptoms appear is a critical part of ensuring a long, healthy life. Nearly all symptoms of the disease can be prevented with early and ongoing treatment. If a person with HFE-HHC is treated before he or she develops cirrhosis of the liver, he or she can expect a normal lifespan. Among people who already have cirrhosis associated with HFE-HHC, 72% will survive at least 5 more years and 62% will survive at least 10 more years. People who already have cirrhosis are at an increased risk for developing a type of liver cancer.
What Next Steps Could You Take?

The Universal Genetic Test has indicated that [masked] is a carrier of HFE-associated hereditary hemochromatosis.  

Because carriers of HFE-HHC do not have any symptoms of the disease, there may not be cause for concern. Even if his future children inherit the genes that cause HFE-HHC, it may not necessarily cause them to be sick. Most people with the genetic mutations that cause HFE-HHC do not have symptoms of the disease. Those who do have symptoms can be easily treated when identified early. 

Carriers of HFE-HHC do not face any known health risks, and need not take any further steps to protect their own health.

Consult With a Physician or Genetic Counselor

To schedule an appointment to speak with a genetic counselor at Counsyl, please call (888) COUNSYL or email gc@counsyl.com.

These medical professionals may be able to suggest actions the couple can take to lower the risk of their children developing HFE-associated hereditary hemochromatosis.
Below are the full test results for all diseases on the panel. Noted are the specific genetic mutations for which the patient tested positive or negative. If there was insufficient data to determine the genotype for any variant, this will be noted as "no call." Also listed in this section is the patient's post-test risk of being a carrier of each disease as well as the odds that his future children could inherit each disease.

### ABC8-Related Hyperinsulinism
- **Your child's risk**: 1 in 51,000
- **Risk before testing**: 1 in 50,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 110. 90% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>ABC8</td>
<td>F1388del, V187D, 3992-9G&gt;A</td>
</tr>
</tbody>
</table>

### Achromatopsia
- **Your child's risk**: 1 in 210,000
- **Risk before testing**: 1 in 30,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 86% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>CNGB3</td>
<td>R403Q, E336X, IVS8-3T&gt;G, 819_826del8, T383fs, 886-896del11insT</td>
</tr>
</tbody>
</table>

### Alkaptonuria
- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 84% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>HGD</td>
<td>G161R, G270R, P230S, S47L, M388V, IVS1-1G&gt;A, IVS5+1G&gt;A</td>
</tr>
</tbody>
</table>

### Alpha-1 Antitrypsin Deficiency
- **Your child's risk**: 1 in 740,000
- **Risk before testing**: 1 in 730
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. >99% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>SERPINA1</td>
<td>S allele, Z allele</td>
</tr>
</tbody>
</table>

### Andermann Syndrome
- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>SLC12A6</td>
<td>Thr813fsX813, R675X</td>
</tr>
</tbody>
</table>

### ARSACS
- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 95% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>SACS</td>
<td>6594delT, 5254C&gt;T</td>
</tr>
</tbody>
</table>

### Aspartylglycosaminuria
- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>AGA</td>
<td>199_200delGA, C163S</td>
</tr>
</tbody>
</table>

### Ataxia With Vitamin E Deficiency
- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>TTPA</td>
<td>744delA</td>
</tr>
</tbody>
</table>

This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician's workup.

Laboratory Director: Jessica Jacobson, MD  
CLIA Number: 05D1102604
### Ataxia-Telangiectasia
- **Risk before testing**: 1 in 100,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160.
- **Gene**: ATM. Variants (1) R35X.

### Autosomal Recessive Polycystic Kidney Disease
- **Risk before testing**: 1 in 15,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72.

### Bardet-Biedl Syndrome, BBS1-Related
- **Risk before testing**: 1 in 100,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.
- **Gene**: BBS1. Variants (1) M390R.

### Bardet-Biedl Syndrome, BBS10-Related
- **Risk before testing**: 1 in 100,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290.
- **Gene**: BBS10. Variants (1) C91fs.

### Beta Thalassemia
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.

### Biotinidase Deficiency
- **Risk before testing**: 1 in 55,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.
- **Gene**: BTD. Variants (7) G98 d7i3, A171T, D252G, F403V, Q456H, R538C, D444H.

### Bloom Syndrome
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.
- **Gene**: BLM. Variants (2) 2281del6ins7, 2407insT.

### Canavan Disease
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.
- **Gene**: ASPA. Variants (4) E285A, Y231X, A305E, IVS2-2A>G.

### Carnitine Palmitoyltransferase IA Deficiency
- **Risk before testing**: Less than 1 in 1,000,000
- **Reduced risk**: No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.
- **Gene**: CPT1A. Variants (2) P478L, G710E.
Carnitine Palmitoyltransferase II Deficiency

Your child's risk
Less than 1 in 1,000,000

Risk before testing
1 in 330,000

Reduced risk


Cartilage-Hair Hypoplasia

Your child's risk
Less than 1 in 1,000,000

Risk before testing
Less than 1 in 1,000,000

Reduced risk

Gene: RMRP. Variants (2) 262G>T, g.70A>G.

Choroideremia

Your child's risk
1 in 200,000

Risk before testing
1 in 200,000

Reduced risk

Gene: CHM. Variants (1) IVS13+2dupT.

CLN5-Related Neuronal Ceroid Lipofuscinosis

Your child's risk
Less than 1 in 1,000,000

Risk before testing
Less than 1 in 1,000,000

Reduced risk

Gene: CLN5. Variants (1) 2467AT.

Congenital Disorder of Glycosylation Type Ia

Your child's risk
1 in 160,000

Risk before testing
1 in 100,000

Reduced risk

Gene: PMM2. Variants (2) F119L, R141H.

Congenital Disorder of Glycosylation Type Ib

Your child's risk
Less than 1 in 1,000,000

Risk before testing
Less than 1 in 1,000,000

Reduced risk

Gene: MPI. Variants (1) R295H.

Congenital Finnish Nephrosis

Your child's risk
Less than 1 in 1,000,000

Risk before testing
Less than 1 in 1,000,000

Reduced risk

Gene: NPHS1. Variants (2) 121_122del, R1109X.

Cystic Fibrosis

Your child's risk
1 in 30,000

Risk before testing
1 in 3,100

Reduced risk


Cystinosis

Your child's risk
1 in 240,000

Risk before testing
1 in 200,000

Reduced risk

## Factor V Leiden Thrombophilia

**Non-disease-causing mutations:** H1299R and D2222G. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.

**Gene:** F5. **Variants (3):** R506Q, H1299R, D2222G.

## Factor XI Deficiency

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.


## Familial Dysautonomia

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

**Gene:** KBKAP. **Variants (3):** IVS20+6T>C, R696P, P914L.

## Familial Mediterranean Fever

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 69% detection rate.


## Fanconi Anemia Type C

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 400. 60% detection rate.

**Gene:** FANCC. **Variants (4):** IVS4+4A>T, 322delG, Q13X, R548X.

## Fumarase Deficiency

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 25% detection rate.

**Gene:** FH. **Variants (1):** 1431_1433dupAAA.

## Galactosemia

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 70% detection rate.


## Gaucher Disease

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 390. 71% detection rate.


## GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 79. 46% detection rate.


## Glucose-6-Phosphate Dehydrogenase Deficiency

No mutations detected. This does not rule out the possibility of being affected by untested mutations. The post-test risk of being affected is 1 in 420.

**Gene:** G6PD. **Variants (5):** V68M, S188F, R459P, R459L, N126D.
<table>
<thead>
<tr>
<th>Condition</th>
<th>Your child's risk</th>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glutaric Acidemia Type 1</td>
<td>1 in 48,000</td>
<td>1 in 40,000</td>
<td></td>
</tr>
<tr>
<td>Glycogen Storage Disease Type Ia</td>
<td>1 in 520,000</td>
<td>1 in 130,000</td>
<td></td>
</tr>
<tr>
<td>Glycogen Storage Disease Type Ib</td>
<td>1 in 970,000</td>
<td>1 in 500,000</td>
<td></td>
</tr>
<tr>
<td>Glycogen Storage Disease Type III</td>
<td>1 in 110,000</td>
<td>1 in 100,000</td>
<td></td>
</tr>
<tr>
<td>Glycogen Storage Disease Type V</td>
<td>1 in 320,000</td>
<td>1 in 100,000</td>
<td></td>
</tr>
<tr>
<td>GRACILE Syndrome</td>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td>Hereditary Fructose Intolerance</td>
<td>1 in 81,000</td>
<td>1 in 26,000</td>
<td></td>
</tr>
<tr>
<td>Hereditary Thymine-Uraciluria</td>
<td>1 in 84,000</td>
<td>1 in 40,000</td>
<td></td>
</tr>
<tr>
<td>Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related</td>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td>Disorder</td>
<td>Male</td>
<td>Female</td>
<td></td>
</tr>
<tr>
<td>------------------------------------------------------------------------</td>
<td>----------------------------------------------------------------------</td>
<td>------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related</td>
<td>Your child's risk: Less than 1 in 1,000,000</td>
<td>Risk before testing: Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reduced risk</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000. 52% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene LAMB3. Variants (5) 3024delT, R42X, R144X, Q243X, R635X.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related</td>
<td>Your child's risk: Less than 1 in 1,000,000</td>
<td>Risk before testing: Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reduced risk</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000. &lt;10% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene LAMC2. Variants (1) R95X.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hexosaminidase A Deficiency</td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 52% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>HFE-Associated Hereditary Hemochromatosis</td>
<td>Disease-causing mutations: HFE:p.Cys282Tyr (C282Y) heterozygote. This individual is a carrier of HFE-associated hereditary hemochromatosis. Carriers generally do not experience symptoms. Clinical symptoms are also uncommon in C282Y homozygotes.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency</td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 36% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene CBS. Variants (2) I278T, G307S.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hurler Syndrome</td>
<td>Your child's risk: 1 in 260,000</td>
<td>Risk before testing: 1 in 130,000</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reduced risk</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170,000. 40% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene DUA. Variants (2) A327P, W402X.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome</td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000. &lt;10% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hypophosphatasia, Autosomal Recessive</td>
<td>Your child's risk: 1 in 130,000</td>
<td>Risk before testing: 1 in 100,000</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reduced risk</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130,000. 22% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene ALPL. Variants (5) 1559delT, F310L, D361V, E174K, G317D.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Inclusion Body Myopathy 2</td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000. &lt;10% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene GNE. Variants (1) M712T.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Infantile Refsum Disease</td>
<td>Your child's risk: 1 in 88,000</td>
<td>Risk before testing: 1 in 50,000</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Reduced risk</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 88,000. 45% detection rate.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Gene PEX1. Variants (1) G843D.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Condition</td>
<td>Your child's risk</td>
<td>Risk before testing</td>
<td>Reduced risk</td>
</tr>
<tr>
<td>-------------------------------------------</td>
<td>-----------------------</td>
<td>---------------------</td>
<td>--------------</td>
</tr>
<tr>
<td>Isovaleric Acidemia</td>
<td>1 in 470,000</td>
<td>1 in 250,000</td>
<td></td>
</tr>
<tr>
<td>Krabbe Disease</td>
<td>1 in 120,000</td>
<td>1 in 63,000</td>
<td></td>
</tr>
<tr>
<td>Leigh Syndrome, French-Canadian Type</td>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td>Limb-Girdle Muscular Dystrophy Type 2E</td>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td>Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency</td>
<td>1 in 510,000</td>
<td>1 in 90,000</td>
<td></td>
</tr>
<tr>
<td>Maple Syrup Urine Disease Type 1B</td>
<td>1 in 420,000</td>
<td>1 in 250,000</td>
<td></td>
</tr>
<tr>
<td>Maple Syrup Urine Disease Type 3</td>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td></td>
</tr>
<tr>
<td>Medium Chain Acyl-CoA Dehydrogenase Deficiency</td>
<td>1 in 63,000</td>
<td>1 in 14,000</td>
<td></td>
</tr>
<tr>
<td>Metachromatic Leukodystrophy</td>
<td>1 in 150,000</td>
<td>1 in 81,000</td>
<td></td>
</tr>
</tbody>
</table>

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 470,000. 47% detection rate.

Gene: IVD. Variants (1) A311V.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120,000. 46% detection rate.

Gene: GALC. Variants (3) Ex11-17del, G270D, R168C.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000,000. <10% detection rate.

Gene: LRPPRC. Variants (1) A354V.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 510,000. 82% detection rate.

Gene: HADHA. Variants (2) Q342X, E474Q.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 420,000. 40% detection rate.

Gene: BCKDHB. Variants (3) R183P, G278S, E322X.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 1,000,000. <10% detection rate.

Gene: SGCB. Variants (1) S114F.

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 63,000. 78% detection rate.


No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150,000. 48% detection rate.

Gene: ARSA. Variants (4) P377L, P426L, IVS2+1G>A, T274M.
Mucolipidosis IV

Your child's risk: Less than 1 in 1,000,000
Risk before testing: Less than 1 in 1,000,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 28% detection rate.

Gene: MCOLN1. Variants (2) 511_6944del, IVS3-2A>G.

Muscle-Eye-Brain Disease

Your child's risk: Less than 1 in 1,000,000
Risk before testing: Less than 1 in 1,000,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 28% detection rate.

Gene: POMGNT1. Variants (1) IVS17+1G>A.

MYH-Associated Polyposis

Your child's risk: 1 in 52,000
Risk before testing: 1 in 40,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 23% detection rate.

Gene: MUTYH. Variants (1) Y165C.

Niemann-Pick Disease Type A

Your child's risk: 1 in 260,000
Risk before testing: 1 in 250,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.


Niemann-Pick Disease Type C

Your child's risk: 1 in 170,000
Risk before testing: 1 in 150,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

Gene: NPC1. Variants (1) I1061T.

Nijmegen Breakage Syndrome

Your child's risk: 1 in 480,000
Risk before testing: 1 in 100,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.


Northern Epilepsy

Your child's risk: Less than 1 in 1,000,000
Risk before testing: Less than 1 in 1,000,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

Gene: CLN8. Variants (1) R24G.

Pendred Syndrome

Your child's risk: 1 in 38,000
Risk before testing: 1 in 20,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 130. 44% detection rate.


Phenylalanine Hydroxylase Deficiency

Your child's risk: Less than 1 in 1,000,000
Risk before testing: Less than 1 in 1,000,000

Reduced risk

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. <10% detection rate.

<table>
<thead>
<tr>
<th>Condition</th>
<th>Male</th>
<th>Female</th>
<th>Gene/Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Polyglandular Autoimmune Syndrome Type 1</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 24% detection rate.</td>
</tr>
<tr>
<td>PPT1-Related Neuronal Ceroid Lipofuscinosis</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 35% detection rate.</td>
</tr>
<tr>
<td>Primary Hyperoxaluria Type 1</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 34% detection rate.</td>
</tr>
<tr>
<td>Primary Hyperoxaluria Type 2</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 37% detection rate.</td>
</tr>
<tr>
<td>Pycnodysostosis</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. &lt;10% detection rate.</td>
</tr>
<tr>
<td>Rhizomelic Chondrodysplasia Punctata Type 1</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 370. 57% detection rate.</td>
</tr>
<tr>
<td>Salla Disease</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. 9&lt;10% detection rate.</td>
</tr>
<tr>
<td>Segawa Syndrome</td>
<td></td>
<td></td>
<td>No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is &lt; 1 in 500. &lt;10% detection rate.</td>
</tr>
<tr>
<td>Short Chain Acyl-CoA Dehydrogenase Deficiency</td>
<td></td>
<td></td>
<td>Non-disease-causing mutations: G185S. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. &lt;10% detection rate.</td>
</tr>
</tbody>
</table>

This test was developed and its performance characteristics determined by Counsyl, Inc. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. These results are adjunctive to the ordering physician’s workup.
### Sickle Cell Disease

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
</tr>
</tbody>
</table>

- **Your child's risk**: Less than 1 in 1,000,000
- **Risk before testing**: Less than 1 in 1,000,000

- **Gene**: HBB

### Sjogren-Larsson Syndrome

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 250,000</td>
<td>1 in 330</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 330,000
- **Risk before testing**: 1 in 250,000

- **Gene**: ALDH3A2
- **Variants**: (1) P315S.

### Smith-Lemli-Opitz Syndrome

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 40,000</td>
<td>1 in 250</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 98,000
- **Risk before testing**: 1 in 40,000

- **Gene**: DHCR7

### Spinal Muscular Atrophy

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 4,800</td>
<td>&lt;1 in 500</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 97,000
- **Risk before testing**: 1 in 4,800

- **Gene**: SMN1
- **Variants**: (1) Exon 7 deletion.

### Sulfate Transporter-Related Osteochondrodysplasia

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 18,000</td>
<td>1 in 350</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 60,000
- **Risk before testing**: 1 in 18,000

- **Gene**: SLC26A2
- **Variants**: (5) C653S, R178X, R279W, V340del, IVS1+2T>C.

### Tay-Sachs Disease

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 360,000</td>
<td>&lt;1 in 500</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 690,000
- **Risk before testing**: 1 in 360,000

- **Gene**: HEXA
- **Variants**: (4) 1278insTATC, IVS12+1G>C, IVS7+1G>A, IVS9+1G>A.

### TPP1-Related Neuronal Ceroid Lipofuscinosis

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 350,000</td>
<td>&lt;1 in 500</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 950,000
- **Risk before testing**: 1 in 350,000

- **Gene**: TPP1
- **Variants**: (4) G284V, R208X, IVS5-1G>C, IVS5-1G>A.

### Tyrosinemia Type I

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 120,000</td>
<td>&lt;1 in 500</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 240,000
- **Risk before testing**: 1 in 120,000

- **Gene**: FAH
- **Variants**: (5) IVS8-1G>C, IVS12+5G>A, P261L, W262X, E357X.

### Usher Syndrome Type 1F

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Reduced risk</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 150,000</td>
<td>&lt;1 in 500</td>
</tr>
</tbody>
</table>

- **Your child's risk**: 1 in 160,000
- **Risk before testing**: 1 in 150,000

- **Gene**: PCDH15
- **Variants**: (1) R245X.
### Usher Syndrome Type 3

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Your child's risk</th>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 1 in 1,000,000</td>
<td>Less than 1 in 1,000,000</td>
<td>CLRN1</td>
<td>N48K</td>
</tr>
</tbody>
</table>

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is less than 1 in 500, and the detection rate is 10%.

### Wilson Disease

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Your child's risk</th>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 30,000</td>
<td>1 in 52,000</td>
<td>ATP7B</td>
<td>1340del4, 2337delC, R778G, W779X, H1069Q</td>
</tr>
</tbody>
</table>

No mutations detected. This does not rule out the possibility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150, and the detection rate is 42%.

### X-Linked Juvenile Retinoschisis

<table>
<thead>
<tr>
<th>Risk before testing</th>
<th>Your child's risk</th>
<th>Gene</th>
<th>Variants</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 in 100,000</td>
<td>1 in 100,000</td>
<td>RS1</td>
<td>E72K, G74V, G109R</td>
</tr>
</tbody>
</table>

No mutations detected. This does not rule out the possibility of being affected by untested mutations. The post-test risk of being affected is less than 1 in 500, and the detection rate is 14%.