Counsyl

Results Recipient	Ordering Healthcare Professional	Male Details	Female Details
[redacted] Report Date [redacted]/2010	Attn: Jessica Jacobson, MD Counsyl, Inc. 2200 Bridge Parkway, Suite 103 Redwood City, CA 94065 Phone: 1-888-COUNSYL NPI: [redacted]	[redacted] Ethnicity: Northern European Sample Type: Saliva (OG-300) Date of Collection: [redacted]/2009 Indication: Carrier testing in individual of reproductive age	Not tested

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. *

[redacted]

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

Partner

The child risk presented below is based on a hypothetical pairing with a partner of the same ethnic group.

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
- Medium Chain Acyl-CoA Dehydrogenase Deficiency
- Phenylalanine Hydroxylase Deficiency
- Spinal Muscular Atrophy

* **Limitations:** Interpretation is given as a probability due to the inheritance pattern of hese 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.

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Laboratory Director: Jessica Jacobson, MD
CLIA Number: 05D1102604

Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Mild Disease Summary

The chart below shows [redacted] 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.

Mild Disease	[redacted]
Factor V Leiden Thrombophilia	No disease-causing mutations detected.
Glucose-6-Phosphate Dehydrogenase Deficiency	No disease-causing mutations detected.
HFE-Associated Hereditary Hemochromatosis	HFE:p.Cys282Tyr (C282Y) heterozygote.

For details on HFE-associated hereditary hemochromatosis, see page 3.

Male	Female
[redacted]	Not tested

Mild Disease Positive Report: HFE-Associated Hereditary Hemochromatosis

This disease report is included due to positive result for [redacted]

Patient Results

	[redacted]	No partner tested
Result:	HFE:p.Cys282Tyr (C282Y) heterozygote.	N/A
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.	N/A

Variants on the Counsyl panel 11
Gene HFE. **Variants** H63D, S65C, Q127H, E168Q, E168X, W169X, C282Y, Q283P, V53M, V59M, H63H.

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.

Male	Female
[redacted]	Not tested

What Next Steps Could You Take?

The Universal Genetic Test has indicated that [redacted] 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.

Male	Female
[redacted]	Not tested

Full Results

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.

ABCC8-Related Hyperinsulinism	Your child's risk	Risk before testing	Reduced risk
	1 in 51,000	1 in 50,000	

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 110. <10% detection rate.

Gene ABCC8. **Variants (3)** F1388del, V187D, 3992-9G>A.

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

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

Gene CNGB3. **Variants (6)** R403Q, E336X, IVS8-3T>G, 819_826del8, T383fs, 886-896del11insT.

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

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

Gene HGD. **Variants (7)** G161R, G270R, P230S, S47L, M368V, IVS1-1G>A, IVS5+1G>A.

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

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

Gene SERPINA1. **Variants (2)** S allele, Z allele.

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

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

Gene SLC12A6. **Variants (2)** Thr813fsX813, R675X.

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

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

Gene SACS. **Variants (2)** 6594delT, 5254C>T.

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

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

Gene AGA. **Variants (2)** 199_200delGA, C163S.

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

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

Gene TTPA. **Variants (1)** 744delA.

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CLIA Number: 05D1102604

Counsyl

Male	Female
Name: [redacted] DOB: [redacted]	Not tested

	Your child's risk	Risk before testing	
Ataxia-Telangiectasia	1 in 100,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 160. <10% detection rate.

Gene ATM. **Variants (1)** R35X.

	Your child's risk	Risk before testing	
Autosomal Recessive Polycystic Kidney Disease	1 in 18,000	1 in 15,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 72. 14% detection rate.

Gene PKHD1. **Variants (5)** Leu1965fs, 9689delA, T36M, R496X, V3471G.

	Your child's risk	Risk before testing	
Bardet-Biedl Syndrome, BBS1-Related	1 in 500,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 80% detection rate.

Gene BBS1. **Variants (1)** M390R.

	Your child's risk	Risk before testing	
Bardet-Biedl Syndrome, BBS10-Related	1 in 190,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 46% detection rate.

Gene BBS10. **Variants (1)** C91fs.

	Your child's risk	Risk before testing	
Beta Thalassemia	Less than 1 in 1,000,000	1 in 250,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 80% detection rate.

Gene HBB. **Variants (35)** Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, K17X, Q39X, 619 bp deletion, Pro5fs, Gly16fs, Glu6fs, Phe41fs, Lys8fs, Phe71fs, Ser9fs, IVS-II-654, IVS-II-705, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-II-844, IVS-I-1, IVS-I-1, IVS-II-849, IVS-II-849, Gly24 T>A, -30T>A, -88C>T, -28A>G, -29A>G, CAP+1 A>C, -87C>G, Hb C, Hb E, Hb O-Arab.

	Your child's risk	Risk before testing	
Biotinidase Deficiency	1 in 410,000	1 in 55,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 89% detection rate.

Gene BTD. **Variants (7)** G98 d7i3, A171T, D252G, F403V, Q456H, R538C, D444H.

	Your child's risk	Risk before testing	
Bloom Syndrome	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene BLM. **Variants (2)** 2281del6ins7, 2407insT.

	Your child's risk	Risk before testing	
Canavan Disease	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 53% detection rate.

Gene ASPA. **Variants (4)** E285A, Y231X, A305E, IVS2-2A>G.

	Your child's risk	Risk before testing	
Carnitine Palmitoyltransferase IA Deficiency	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene CPT1A. **Variants (2)** P479L, G710E.

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Counsyl

Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Carnitine Palmitoyltransferase II Deficiency

Your child's risk	Risk before testing	
Less than 1 in 1,000,000	1 in 330,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 94% detection rate.

Gene CPT2. **Variants (13)** S38fs, Leu178_Ile186delinsPhe, Q413fs, P50H, S113L, R124X, P227L, R503C, G549D, Q550R, P604S, Y628S, R631C.

Cartilage-Hair Hypoplasia

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

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

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

Choroideremia

Your child's risk	Risk before testing	
1 in 200,000	1 in 200,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being affected by untested mutations. The post-test risk of being affected is < 1 in 500. <10% detection rate.

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

CLN5-Related Neuronal Ceroid Lipofuscinosis

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

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

Gene CLN5. **Variants (1)** 2467AT.

Congenital Disorder of Glycosylation Type Ia

Your child's risk	Risk before testing	
1 in 160,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 260. 39% detection rate.

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

Congenital Disorder of Glycosylation Type Ib

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

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

Gene MPI. **Variants (1)** R295H.

Congenital Finnish Nephrosis

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

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

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

Cystic Fibrosis

Your child's risk	Risk before testing	
1 in 30,000	1 in 3,100	Reduced risk

[redacted] Non-disease-causing mutations: I506V. 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 270. 90% detection rate.

Gene CFTR. **Variants (109)** G85E, R117H, R334W, R347P, A455E, G542X, G551D, R553X, R560T, R1162X, W1282X, N1303K, F508del, I507del, 2184delA, 3659delC, 621+1G>T, 711+1G>T, 1717-1G>A, 1898+1G>A, 2789+5G>A, 3120+1G>A, 3849+10kbC>T, E60X, R75X, G91R, E92X, R117C, Y122X, G178R, L206W, G330X, T338I, R347H, R352Q, S364P, G480C, Q493X, V520F, C524X, S549I, S549N, S549R, Q552X, A559T, P574H, G622D, R709X, K710X, Q890X, R1066C, R1070Q, W1089X, Y1092X, M1101K, D1152H, R1158X, S1196X, W1204X, S1235R, Q1238X, S1251N, S1255X, R1283M, dele2-3 21kb, 3199del6, F311del, 394delTT, 574delA, 663delT, 935delA, 936delTA, 1078delT, 1161delC, 1609delCA, 1677delTA, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 3171delC, 3667del4, 3821delT, 3876delA, 1288insTA, 2184insA, 2307insA, 2869insG, 3905insT, 296+12T>C, 405+1G>A, 405+3A>C, 406-1G>A, 711+5G>A, 712-1G>T, 1811+1.6kbA>G, 1812-1G>A, 1898+1G>T, 1898+5G>T, 3272-26A>G, 3120G>A, 457TAT>G, 2183AA>G, S549R, W1204X, IVS8-5T, I148T, I506V, F508C.

Cystinosis

Your child's risk	Risk before testing	
1 in 240,000	1 in 200,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 17% detection rate.

Gene CTNS. **Variants (4)** 537del21, W138X, L158P, D205N.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Factor V Leiden Thrombophilia M LD

[redacted] 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

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

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

Gene F11. **Variants (4)** E117X, F283L, IVS14+1G>A, IVS14del14.

Familial Dysautonomia

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

[redacted] No mutations detected. This does not rule out the poss bility 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

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

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

Gene MEFV. **Variants (13)** I692del, T267I, F479L, R653H, M680I, M694I, M694V, K695R, V726A, A744S, R761H, P369S, R408Q.

Fanconi Anemia Type C

Your child's risk	Risk before testing	
1 in 250,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility 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

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

[redacted] No mutations detected. This does not rule out the poss bility 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

Your child's risk	Risk before testing	
1 in 100,000	1 in 30,000	Reduced risk

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

Gene GALT. **Variants (10)** IVS2-2A>G, S135L, T138M, F171S, Q169K, Q188R, L195P, Y209C, K285N, X380R.

Gaucher Disease

Your child's risk	Risk before testing	
1 in 170,000	1 in 50,000	Reduced risk

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

Gene GBA. **Variants (9)** N370S, L444P, 1035insG, IVS2+1G>A, V394L, D409V, R463C, R463H, R496H.

GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness

Your child's risk	Risk before testing	
1 in 13,000	1 in 7,000	Reduced risk

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

Gene GJB2. **Variants (11)** 35delG, 167delT, 313del14, E120del, W24X, V37I, W77R, W77X, Q124X, R184P, M34T.

Glucose-6-Phosphate Dehydrogenase Deficiency M LD

[redacted] No mutations detected. This does not rule out the poss bility 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.

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Laboratory Director: Jessica Jacobson, MD
CLIA Number: 05D1102604

Counsyl

Male	Female
Name: [redacted] DOB: [redacted]	Not tested

	Your child's risk	Risk before testing	
Glutaric Acidemia Type 1	1 in 46,000	1 in 40,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 120. 13% detection rate.

Gene GCDH. **Variants (2)** R402W, A421V.

	Your child's risk	Risk before testing	
Glycogen Storage Disease Type Ia	1 in 520,000	1 in 130,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 76% detection rate.

Gene G6PC. **Variants (10)** 727G>T, F327del, Q27fsdelC, 459insTA, R83H, R83C, G188R, Q242X, G270V, Q347X.

	Your child's risk	Risk before testing	
Glycogen Storage Disease Type Ib	1 in 970,000	1 in 500,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 48% detection rate.

Gene G6PT1. **Variants (4)** 1211delCT, G339C, G339D, A367T.

	Your child's risk	Risk before testing	
Glycogen Storage Disease Type III	1 in 110,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 170. <10% detection rate.

Gene AGL. **Variants (3)** Q6X, 17delAG, 1484delT.

	Your child's risk	Risk before testing	
Glycogen Storage Disease Type V	1 in 320,000	1 in 100,000	Reduced risk

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

Gene PYGM. **Variants (4)** R49X, G204S, K542T, K542X.

	Your child's risk	Risk before testing	
GRACILE Syndrome	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene BCS1L. **Variants (1)** S78G.

	Your child's risk	Risk before testing	
Hereditary Fructose Intolerance	1 in 81,000	1 in 26,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 68% detection rate.

Gene ALDOB. **Variants (4)** Delta4E4, A149P, Y204X, N334K.

	Your child's risk	Risk before testing	
Hereditary Thymine-Uraciluria	1 in 84,000	1 in 40,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 210. 52% detection rate.

Gene DPYD. **Variants (1)** IVS14+1G>A.

	Your child's risk	Risk before testing	
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene LAMA3. **Variants (1)** R650X.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related

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

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 52% detection rate.

Gene LAMB3. **Variants (5)** 3024delT, R42X, R144X, Q243X, R635X.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related

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

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

Gene LAMC2. **Variants (1)** R95X.

Hexosaminidase A Deficiency

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500.

Gene HEXA. **Variants (8)** 1278insTATC, IVS12+1G>C, R178C, R178H, G269S, IVS7+1G>A, IVS9+1G>A, R247W.

HFE-Associated Hereditary Hemochromatosis M LD

[redacted] 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.

Gene HFE. **Variants (11)** H63D, S65C, Q127H, E168Q, E168X, W169X, C282Y, Q283P, V53M, V59M, H63H.

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency

Your child's risk	Risk before testing	
1 in 200,000	1 in 130,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 290. 38% detection rate.

Gene CBS. **Variants (2)** I278T, G307S.

Hurler Syndrome

Your child's risk	Risk before testing	
1 in 170,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 40% detection rate.

Gene DUA. **Variants (2)** A327P, W402X.

Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome

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

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

Gene SLC25A15. **Variants (1)** F188del.

Hypophosphatasia, Autosomal Recessive

Your child's risk	Risk before testing	
1 in 130,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 22% detection rate.

Gene ALPL. **Variants (5)** 1559delT, F310L, D361V, E174K, G317D.

Inclusion Body Myopathy 2

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

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

Gene GNE. **Variants (1)** M712T.

Infantile Refsum Disease

Your child's risk	Risk before testing	
1 in 88,000	1 in 50,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. 43% detection rate.

Gene PEX1. **Variants (1)** G843D.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

	Your child's risk	Risk before testing	
Isovaleric Acidemia	1 in 470,000	1 in 250,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 470. 47% detection rate.

Gene IVD. **Variants (1)** A311V.

	Your child's risk	Risk before testing	
Krabbe Disease	1 in 120,000	1 in 63,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 240. 46% detection rate.

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

	Your child's risk	Risk before testing	
Leigh Syndrome, French-Canadian Type	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene LRPPRC. **Variants (1)** A354V.

	Your child's risk	Risk before testing	
Limb-Girdle Muscular Dystrophy Type 2E	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene SGCB. **Variants (1)** S114F.

	Your child's risk	Risk before testing	
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	1 in 510,000	1 in 90,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 82% detection rate.

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

	Your child's risk	Risk before testing	
Maple Syrup Urine Disease Type 1B	1 in 420,000	1 in 250,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 420. 40% detection rate.

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

	Your child's risk	Risk before testing	
Maple Syrup Urine Disease Type 3	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene DLD. **Variants (2)** 105insA, G229C.

	Your child's risk	Risk before testing	
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 63,000	1 in 14,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 270. 78% detection rate.

Gene ACADM. **Variants (7)** L59F, G170R, G242R, Y42H, K304E, R181C, R181H.

	Your child's risk	Risk before testing	
Metachromatic Leukodystrophy	1 in 150,000	1 in 81,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 260. 45% detection rate.

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

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

	Your child's risk	Risk before testing	
Mucolipidosis IV	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

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

	Your child's risk	Risk before testing	
Muscle-Eye-Brain Disease	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility 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.

	Your child's risk	Risk before testing	
MYH-Associated Polyposis	1 in 52,000	1 in 40,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility 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.

	Your child's risk	Risk before testing	
Niemann-Pick Disease Type A	1 in 260,000	1 in 250,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 260. <10% detection rate.

Gene SMPD1. **Variants (3)** fsP330, L302P, R496L.

	Your child's risk	Risk before testing	
Niemann-Pick Disease Type C	1 in 170,000	1 in 150,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 220. <10% detection rate.

Gene NPC1. **Variants (1)** I1061T.

	Your child's risk	Risk before testing	
Nijmegen Breakage Syndrome	1 in 460,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 78% detection rate.

Gene NBN. **Variants (1)** 657del5.

	Your child's risk	Risk before testing	
Northern Epilepsy	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility 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.

	Your child's risk	Risk before testing	
Pendred Syndrome	1 in 36,000	1 in 20,000	Reduced risk

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

Gene SLC26A4. **Variants (3)** L236P, E384G, T416P.

	Your child's risk	Risk before testing	
Phenylalanine Hydroxylase Deficiency	Less than 1 in 1,000,000	1 in 10,000	Reduced risk

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

Gene PAH. **Variants (11)** IVS-10int-546, IVS12+1G>A, L48S, I65T, R158Q, R252W, R261Q, G272X, R408Q, R408W, Y414C.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Polyglandular Autoimmune Syndrome Type 1

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

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 24% detection rate.

Gene A RE. **Variants (2)** Y85C, R257X.

PPT1-Related Neuronal Ceroid Lipofuscinosis

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

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 58% detection rate.

Gene PPT1. **Variants (4)** L10X, T75P, R122W, R151X.

Primary Hyperoxaluria Type 1

Your child's risk	Risk before testing	
1 in 760,000	1 in 500,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 34% detection rate.

Gene AGXT. **Variants (3)** F152I, G170R, I244T.

Primary Hyperoxaluria Type 2

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

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 37% detection rate.

Gene GRHPR. **Variants (1)** 103delG.

Pycnodysostosis

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

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

Gene CTSK. **Variants (1)** X330W.

Rhizomelic Chondrodysplasia Punctata Type 1

Your child's risk	Risk before testing	
1 in 230,000	1 in 100,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 370. 57% detection rate.

Gene PEX7. **Variants (2)** G217R, L292X.

Salla Disease

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

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

Gene SLC17A5. **Variants (2)** Leu336fsX13, R39C.

Segawa Syndrome

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

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

Gene TH. **Variants (1)** R233H.

Short Chain Acyl-CoA Dehydrogenase Deficiency

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

[redacted] 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. <10% detection rate.

Gene ACADS. **Variants (2)** R107C, G185S.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

	Your child's risk	Risk before testing	
Sickle Cell Disease	Less than 1 in 1,000,000	Less than 1 in 1,000,000	Reduced risk

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

Gene HBB. **Variants (37)** Hb S, Poly A: AATAAA->AATGAA, Poly A: AATAAA->AATAAG, W15X, K17X, Q39X, 619 bp deletion, Pro5fs, Gly16fs, Glu6fs, Phe41fs, Lys8fs, Phe71fs, Ser9fs, IVS-II-654, IVS-II-705, IVS-II-745, IVS-II-850, IVS-I-6, IVS-I-110, IVS-I-5, IVS-II-844, IVS-I-1, IVS-I-1, IVS-II-849, IVS-II-849, Gly24 T>A, -30T>A, -88C>T, -28A>G, -29A>G, CAP+1 A>C, -87C>G, Hb C, Hb E, Hb D-Punjab, Hb O-Arab.

	Your child's risk	Risk before testing	
Sjogren-Larsson Syndrome	1 in 330,000	1 in 250,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 330. 24% detection rate.

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

	Your child's risk	Risk before testing	
Smith-Lemli-Opitz Syndrome	1 in 98,000	1 in 40,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 250. 59% detection rate.

Gene DHCR7. **Variants (11)** IVS8-1G>C, T93M, L109P, W151X, L157P, V326L, R352Q, R352W, C380Y, R404C, W151X.

	Your child's risk	Risk before testing	
Spinal Muscular Atrophy	1 in 97,000	1 in 4,800	Reduced risk

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

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

	Your child's risk	Risk before testing	
Sulfate Transporter-Related Osteochondrodysplasia	1 in 60,000	1 in 18,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 220. 70% detection rate.

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

	Your child's risk	Risk before testing	
Tay-Sachs Disease	1 in 690,000	1 in 360,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 48% detection rate.

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

	Your child's risk	Risk before testing	
TPP1-Related Neuronal Ceroid Lipofuscinosis	1 in 950,000	1 in 350,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is < 1 in 500. 63% detection rate.

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

	Your child's risk	Risk before testing	
Tyrosinemia Type I	1 in 240,000	1 in 120,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 350. 50% detection rate.

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

	Your child's risk	Risk before testing	
Usher Syndrome Type 1F	1 in 160,000	1 in 150,000	Reduced risk

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 200. <10% detection rate.

Gene PCDH15. **Variants (1)** R245X.

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Male	Female
Name: [redacted] DOB: [redacted]	Not tested

Usher Syndrome Type 3	Your child's risk	Risk before testing	Reduced risk
	Less than 1 in 1,000,000	Less than 1 in 1,000,000	

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

Gene CLRN1. **Variants (1)** N48K.

Wilson Disease	Your child's risk	Risk before testing	Reduced risk
	1 in 52,000	1 in 30,000	

[redacted] No mutations detected. This does not rule out the poss bility of being a carrier of untested mutations. The post-test risk of being a carrier is 1 in 150. 42% detection rate.

Gene ATP7B. **Variants (5)** 1340del4, 2337delC, R778G, W779X, H1069Q.

X-Linked Juvenile Retinoschisis	Your child's risk	Risk before testing	Reduced risk
	1 in 100,000	1 in 100,000	

[redacted] No mutations detected. This does not rule out the poss bility of being affected by untested mutations. The post-test risk of being affected is < 1 in 500. 14% detection rate.

Gene RS1. **Variants (3)** E72K, G74V, G109R.

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