



## Donor 6908

### Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 02/05/24

Donor Reported Ancestry: German, Welsh, French

Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Expanded Genetic Disease Carrier Screening Panel attached- 502 diseases by gene sequencing.  Personalized residual risk by gene is in the attached report.	<p>Carrier: Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (CYP21A2)</p> <p>Carrier: Gitelman Syndrome (SLC12A3)</p> <p>Carrier: Glycogen Storage Disease, Type II (GAA)</p> <p>Carrier: Phenylalanine Hydroxylase Deficiency (PAH)</p> <p>Carrier: Zellweger Syndrome Spectrum (PEX1-Related)</p> <p>Negative for other genes sequenced.</p>	Partner testing recommended before using this donor.

\*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

\*\*Donor residual risk is the chance the donor is still a carrier after testing negative.

**Patient Information**

Name: Donor 6908  
 Date of Birth: [REDACTED]  
 Sema4 ID: [REDACTED]  
 Client ID: [REDACTED]  
 Indication: Carrier Screening

**Specimen Information**

Specimen Type: Blood  
 Date Collected: 08/10/2022  
 Date Received: 08/11/2022  
 Final Report: 08/23/2022

**Referring Provider**

[REDACTED]  
 Fairfax Cryobank, Inc.  
 [REDACTED]  
 [REDACTED]

Expanded Carrier Screen (502 genes)  
 with Personalized Residual Risk

**SUMMARY OF RESULTS AND RECOMMENDATIONS**

⊕ Positive	⊖ Negative
<p><b>Carrier of Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (AR)</b>            Associated gene(s): <i>CYP21A2</i>            Variant(s) Detected: c.290-13C&gt;G, Pathogenic, Heterozygous (one copy)</p> <p><b>Carrier of Gitelman Syndrome (AR)</b>            Associated gene(s): <i>SLC12A3</i>            Variant(s) Detected: c.2581C&gt;T, p.R861C, Pathogenic, Heterozygous (one copy)</p> <p><b>Carrier of Glycogen Storage Disease, Type II (AR)</b>            Associated gene(s): <i>GAA</i>            Variant(s) Detected: c.1082C&gt;T, p.P361L, Pathogenic, Heterozygous (one copy)</p> <p><b>Carrier of Phenylalanine Hydroxylase Deficiency (AR)</b>            Associated gene(s): <i>PAH</i>            Variant(s) Detected: c.890G&gt;A, p.R297H, Pathogenic, Heterozygous (one copy)</p> <p><b>Carrier of Zellweger Syndrome Spectrum (PEX1-Related) (AR)</b>            Associated gene(s): <i>PEX1</i>            Variant(s) Detected: c.1952_1960dupCAGTGTGGA, p.W653_M654insTVW, Likely Pathogenic, Heterozygous (one copy)</p>	<p><b>Negative for all other genes tested</b>            To view a full list of genes and diseases tested please see Table 1 in this report</p>

AR=Autosomal recessive; XL=X-linked

## Recommendations

- Testing the partner for the above positive disorder(s) and genetic counseling are recommended.
- Please note that for female carriers of X-linked diseases, follow-up testing of a male partner is not indicated.
- CGG repeat analysis of *FMR1* for fragile X syndrome is not performed on males as repeat expansion of premutation alleles is not expected in the male germline.
- Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.
- Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder. Please note that residual risks for X-linked diseases (including full repeat expansions for Fragile X syndrome) may not be accurate for males and the actual residual risk is likely to be lower.

## Interpretation of positive results

### Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (AR)

#### Results and Interpretation

*CYP21A2* copy number: 2

No pathogenic copy number variants detected

*CYP21A2* sequencing: c.290-13C>G, Pathogenic, Heterozygous (one copy)

**Genes analyzed:** *CYP21A2* (NM\_000500.6)

**Inheritance:** Autosomal Recessive

A heterozygous (one copy) pathogenic intronic variant, c.290-13C>G, was detected in the *CYP21A2* gene (NM\_000500.6). Please note that this variant is reported to be causative for the classic salt-wasting/severe virilizing form of congenital adrenal hyperplasia (PMID: 29450859). Variants associated with the classic form usually cause classic congenital adrenal hyperplasia when found in trans with a second classic allele, or non-classic congenital adrenal hyperplasia when found in trans with a non-classic allele (PMID: 29450859). Therefore, this individual is expected to be at least a carrier for congenital adrenal hyperplasia. Heterozygous carriers are not expected to exhibit symptoms of this disease.

#### What is congenital adrenal hyperplasia (due to 21-hydroxylase deficiency)?

Congenital adrenal hyperplasia (CAH) is a group of autosomal recessive disorders resulting from deficiency in the enzymes involved in cortisol biosynthesis. The majority (95%) of CAH cases are due to 21-hydroxylase deficiency (21-OHD CAH), which is caused by homozygous or compound heterozygous pathogenic variants in the gene *CYP21A2*. Approximately 20% of mutant alleles have deletions of 30 kb that have been generated by unequal meiotic crossing-over between the two genes. Another 75% of mutant alleles are due to gene conversion events, where an inactivating mutation from the *CYP21A1P* pseudogene is introduced into one copy of the *CYP21A2* gene, thus making the gene non-functional. Three different forms of 21-OHD CAH have been reported: a classic salt wasting form, a classic simple virilizing form, and a non-classic form.

- The classic salt wasting form results from a nonfunctional enzyme and is the most severe. The phenotype includes prenatal onset of virilization and inadequate adrenal aldosterone secretion that can result in fatal salt-wasting crises.
- The classic simple virilizing form results from low levels of functional enzyme and involves prenatal virilization but no salt-wasting.
- The non-classic form, which results from a mild enzyme deficiency, occurs postnatally and involves phenotypes associated with hyperandrogenism, such as hirsutism, delayed menarche, and infertility.

Treatment for the classic forms of the disorder include glucocorticoid and mineralocorticoid replacement therapy, as well as the possibility of feminizing genitoplasty, while patients with the non-classic form usually do not require treatment. The life expectancy for this disorder can be normal with treatment, however the occurrence of salt-wasting crises can be fatal.

### Gitelman Syndrome (AR)

#### Results and Interpretation

A heterozygous (one copy) pathogenic missense variant, c.2581C>T, p.R861C, was detected in the *SLC12A3* gene (NM\_000339.2). When this variant is present in trans with a pathogenic variant, it is considered to be causative for Gitelman syndrome. Therefore, this individual is expected to be at least a carrier for Gitelman syndrome. Heterozygous carriers may have decreased blood pressure compared to the general population, but are not expected to develop any symptoms of disease.

### What is Gitelman Syndrome?

Gitelman syndrome is an autosomal recessive, pan-ethnic disease caused by pathogenic variants in the gene *SLC12A3*. In this disease, the kidney does not retain necessary ions, causing an imbalance in the body. Symptoms usually begin in late childhood or adolescence, and include muscle spasms or cramps, tingling sensations, joint pain and fatigue. Most patients have mild symptoms, but severe ion imbalances could lead to seizures or heart arrhythmias. With treatment, including dietary management, patients have a normal life expectancy. It is not currently possible to predict the severity of symptoms based on the variants inherited.

### Glycogen Storage Disease, Type II (AR)

#### Results and Interpretation

A heterozygous (one copy) pathogenic missense variant, c.1082C>T, p.P361L, was detected in the *GAA* gene (NM\_000152.3). When this variant is present in trans with a pathogenic variant, it is considered to be causative for glycogen storage disease, type II. Therefore, this individual is expected to be at least a carrier for glycogen storage disease, type II. Heterozygous carriers are not expected to exhibit symptoms of this disease.

### What is Glycogen Storage Disease, Type II?

Glycogen storage disease type II, also known as Pompe disease, is an autosomal recessive disorder that is caused by pathogenic variants in the gene *GAA*. While it is found in populations worldwide, it is most prevalent in individuals of Ashkenazi Jewish or African descent. Symptoms can manifest in infancy or later in life. There are two main forms of the disease.

- Infantile-onset disease is characterized by poor feeding and failure to thrive, hypotonia, and an enlarged heart. If untreated, the cardiac manifestations usually cause death in the first year of life. A variant form of the infantile-onset disease has a slower progression, with death occurring in childhood. Enzyme replacement therapy may slow the disease progression.
- Late-onset disease can begin any time after infancy. These patients do not usually have cardiac problems, but have muscle weakness and difficulty breathing. Patients may reach adulthood, but life expectancy is reduced.

Specific variants have been associated with the infantile-onset or late-onset forms; however, the correlation is not always absolute, and some variants do not have a known genotype-phenotype correlation.

### Phenylalanine Hydroxylase Deficiency (AR)

#### Results and Interpretation

A heterozygous (one copy) pathogenic missense variant, c.890G>A, p.R297H, was detected in the *PAH* gene (NM\_000277.1). This variant is considered to be a mild allele and usually causes non-phenylketonuria hyperphenylalanemia when found in trans with a pathogenic variant. When this variant is present in trans with a pathogenic variant, it is considered to be causative for phenylalanine hydroxylase deficiency. Therefore, this individual is expected to be at least a carrier for phenylalanine hydroxylase deficiency. Heterozygous carriers are not expected to exhibit symptoms of this disease.

### What is Phenylalanine Hydroxylase Deficiency?

Phenylalanine hydroxylase deficiency is an autosomal recessive disorder caused by pathogenic variants in the gene *PAH*. While it is found in many different ethnicities, it is particularly prevalent in Sephardic Jewish, Sicilian, Irish, and Turkish individuals, as well as Caucasians. Pathogenic *PAH* variants result in loss of function of the phenylalanine hydroxylase enzyme, which breaks down the amino acid phenylalanine. The most severe form of the disease is called phenylketonuria. If untreated, buildup of phenylalanine will result in irreversible brain damage and severe intellectual disability. Treatment involves the removal of phenylalanine from the diet. Even with strict adherence to the treatment, some neurologic deficiencies have been noticed in long-term survivors. Psychological problems, including anxiety, depression, phobias and panic attacks may occur in adults who do not comply well to their treatment. Some patients have a milder form of hyperphenylalaninemia and may tolerate higher levels of phenylalanine in their diet. Depending on the genotype, patients may be responsive to BH<sub>4</sub>, which can direct their treatment. However, it is not always possible to predict the severity of the disease based on genotype.

## Zellweger Syndrome Spectrum (*PEX1*-Related) (AR)

### Results and Interpretation

A heterozygous (one copy) likely pathogenic inframe insertion, c.1952\_1960dupCAGTGTGGA, p.W653\_M654insTVW, was detected in the *PEX1* gene (NM\_000466.2). When this variant is present in trans with a pathogenic variant, it is considered to be causative for Zellweger syndrome spectrum (*PEX1*-related). Therefore, this individual is expected to be at least a carrier for Zellweger syndrome spectrum (*PEX1*-related). Heterozygous carriers are not expected to exhibit symptoms of this disease.

### What is Zellweger Syndrome Spectrum (*PEX1*-Related)?

Zellweger syndrome spectrum (*PEX1*-related) is an autosomal recessive, pan-ethnic disease of peroxisome biogenesis. It is comprised of three diseases that make up a continuum of severity, from the most severe, known as Zellweger syndrome, to neonatal adrenoleukodystrophy, to infantile Refsum disease, which is the mildest.

- Zellweger syndrome is characterized by demyelination of structures in the brain leading to leukodystrophy, resulting in seizures and vision loss. Clinical features also include dysmorphic features, hypotonia, cardiac problems, and dysfunction of the liver and kidneys. Death typically occurs in the first year of life.
- Neonatal adrenoleukodystrophy and infantile Refsum disease share many overlapping features. Onset of symptoms may be in infancy, or may be noticed later in childhood. Features include developmental delay and loss of vision and hearing; some children present with bleeding in the brain. The severity and course of the disease can vary between individuals; some may learn to walk and talk, and rarely, patients may survive until adulthood; others never walk or talk. Many patients do not survive childhood. Symptoms tend to progress in severity over the course of the patient's life.

Specific *PEX1* variants have been reported to be associated with a more severe or milder phenotype; additionally, frameshift variants have been to be associated with more severe disease presentations.

## Test description

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see Table 1 for a list of genes and diseases tested with the patient's personalized residual risk. If personalized residual risk is not provided, please see the complete residual risk table at [go.sema4.com/residualrisk](https://go.sema4.com/residualrisk). Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

*Hongli Zhan*

**Hongli Zhan, Ph.D., Director**

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D

## Genes and diseases tested

The personalized residual risks listed below are specific to this individual. The complete residual risk table is available at [go.sema4.com/residualrisk](https://go.sema4.com/residualrisk)

Table 1: List of genes and diseases tested with detailed results

Disease	Gene	Inheritance Pattern	Status	Detailed Summary
<b>Positive</b>				
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency	<i>CYP21A2</i>	AR	Carrier	<i>CYP21A2</i> copy number: 2 No pathogenic copy number variants detected <i>CYP21A2</i> sequencing: c.290-13C>G, Pathogenic, Heterozygous (one copy)
Gitelman Syndrome	<i>SLC12A3</i>	AR	Carrier	c.2581C>T, p.R861C, Pathogenic, Heterozygous (one copy)
Glycogen Storage Disease, Type II	<i>GAA</i>	AR	Carrier	c.1082C>T, p.P361L, Pathogenic, Heterozygous (one copy)
Phenylalanine Hydroxylase Deficiency	<i>PAH</i>	AR	Carrier	c.890G>A, p.R297H, Pathogenic, Heterozygous (one copy)
Zellweger Syndrome Spectrum ( <i>PEX1</i> -Related)	<i>PEX1</i>	AR	Carrier	c.1952_1960dupCAGTGTGGA, p.W653_M654insTVW, Likely Pathogenic, Heterozygous (one copy)
<b>Negative</b>				
2-Methylbutyrylglycinuria	<i>ACADSB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,800
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	<i>HSD3B2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,300
3-Methylcrotonyl-CoA Carboxylase Deficiency ( <i>MCCC1</i> -Related)	<i>MCCC1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,400
3-Methylcrotonyl-CoA Carboxylase Deficiency ( <i>MCCC2</i> -Related)	<i>MCCC2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
3-Methylglutaconic Aciduria, Type III	<i>OPA3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 50,000
3-Phosphoglycerate Dehydrogenase Deficiency	<i>PHGDH</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 63,000
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	<i>PTS</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,800
<i>CD59</i> -Mediated Hemolytic Anemia	<i>CD59</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 415,000
Abetalipoproteinemia	<i>MTTP</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,200
Achalasia-Addisonianism-Alacrimia Syndrome	<i>AAAS</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,500
Achromatopsia ( <i>CNGA3</i> -Related)	<i>CNGA3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 830
Achromatopsia ( <i>CNGB3</i> -related)	<i>CNGB3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,600
Acrodermatitis Enteropathica	<i>SLC39A4</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Acute Infantile Liver Failure	<i>TRMU</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,400
Acyl-CoA Oxidase I Deficiency	<i>ACOX1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 39,000
Adams-Oliver Syndrome 4	<i>EOGT</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 44,000
Adenosine Deaminase Deficiency	<i>ADA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,100
Adrenocorticotrophic Hormone Deficiency	<i>TBX19</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 35,000
Adrenoleukodystrophy, X-Linked	<i>ABCD1</i>	XL	Reduced Risk	Personalized Residual Risk: 1 in 19,000
Agammaglobulinemia	<i>BTK</i>	XL	Reduced Risk	Personalized Residual Risk: 1 in 250,000
Agenesis of the Corpus Callosum	<i>FRMD4A</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,393,000
Aicardi-Goutieres Syndrome ( <i>RNASEH2C</i> -Related)	<i>RNASEH2C</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Aicardi-Goutieres Syndrome ( <i>SAMHD1</i> -Related)	<i>SAMHD1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 10,000
Aicardi-Goutieres Syndrome ( <i>TREX1</i> -Related)	<i>TREX1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,200
Albinism, Oculocutaneous, Type III	<i>TYRP1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,500



Alkaptonuria	<i>HGD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Alpha-Mannosidosis	<i>MAN2B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,200
Alpha-Thalassemia	<i>HBA1/HBA2</i>	AR	Reduced Risk	<i>HBA1</i> Copy Number: 2 <i>HBA2</i> Copy Number: 2 No pathogenic copy number variants detected <i>HBA1/HBA2</i> Sequencing: Negative <b>Personalized Residual Risk:</b> 1 in 10,000
Alpha-Thalassemia Intellectual Disability Syndrome	<i>ATRX</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 48,000
Alport Syndrome ( <i>COL4A3</i> -Related)	<i>COL4A3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Alport Syndrome ( <i>COL4A4</i> -Related)	<i>COL4A4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Alport Syndrome ( <i>COL4A5</i> -Related)	<i>COL4A5</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 150,000
Alstrom Syndrome	<i>ALMS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,800
Andermann Syndrome	<i>SLC12A6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 151,000
Antley-Bixler Syndrome ( <i>POR</i> -Related)	<i>POR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,000
Argininemia	<i>ARG1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,500
Argininosuccinic Aciduria	<i>ASL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Aromatase Deficiency	<i>CYP19A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,400
Arthrogryposis, Intellectual Disability, and Seizures	<i>SLC35A3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 454,000
Asparagine Synthetase Deficiency	<i>ASNS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 202,000
Aspartylglycosaminuria	<i>AGA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 13,000
Ataxia With Isolated Vitamin E Deficiency	<i>TTPA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 61,000
Ataxia-Telangiectasia	<i>ATM</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,300
Ataxia-Telangiectasia-Like Disorder 1	<i>MRE11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,500
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	<i>SACS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,600
Bardet-Biedl Syndrome ( <i>ARL6</i> -Related)	<i>ARL6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 29,000
Bardet-Biedl Syndrome ( <i>BBS10</i> -Related)	<i>BBS10</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Bardet-Biedl Syndrome ( <i>BBS12</i> -Related)	<i>BBS12</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,900
Bardet-Biedl Syndrome ( <i>BBS1</i> -Related)	<i>BBS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,400
Bardet-Biedl Syndrome ( <i>BBS2</i> -Related)	<i>BBS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Bardet-Biedl Syndrome ( <i>BBS4</i> -Related)	<i>BBS4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 22,000
Bare Lymphocyte Syndrome, Type II	<i>CIITA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 35,000
Barth Syndrome	<i>TAZ</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 183,000
Bartter Syndrome, Type 3	<i>CLCNKB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 740
Bartter Syndrome, Type 4A	<i>BSND</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 91,000
Bernard-Soulier Syndrome, Type A1	<i>GP1BA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 42,000
Bernard-Soulier Syndrome, Type C	<i>GP9</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,300
Beta-Globin-Related Hemoglobinopathies	<i>HBB</i>	AR	Reduced Risk	<b>Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies):</b> 1 in 2,000 <b>Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbS Variant):</b> 1 in 790,000 <b>Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbC Variant):</b> 1 in 2,107,000
Beta-Ketothiolase Deficiency	<i>ACAT1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,400
Beta-Mannosidosis	<i>MANBA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,100
BH4-Deficient Hyperphenylalaninemia C	<i>QDPR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,100
BH4-Deficient Hyperphenylalaninemia D	<i>PCBD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,000
Bilateral Frontoparietal Polymicrogyria	<i>GPR56</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 203,000
Biotinidase Deficiency	<i>BTBD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 500
Bloom Syndrome	<i>BLM</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,400

Canavan Disease	ASPA	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 4,000</b>
Carbamoylphosphate Synthetase I Deficiency	CPS1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,100</b>
Carnitine Acylcarnitine Translocase Deficiency	SLC25A20	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 4,100</b>
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 24,000</b>
Carnitine Palmitoyltransferase II Deficiency	CPT2	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 670</b>
Carpenter Syndrome	RAB23	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 21,000</b>
Cartilage-Hair Hypoplasia	RMRP	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 960</b>
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 5,900</b>
Central Hypothyroidism and Testicular Enlargement	IGSF1	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 781,000</b>
Cerebral Creatine Deficiency Syndrome 1	SLC6A8	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 208,000</b>
Cerebral Creatine Deficiency Syndrome 2	GAMT	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,100</b>
Cerebral Creatine Deficiency Syndrome 3	GATM	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 7,900</b>
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,730,000</b>
Cerebrotendinous Xanthomatosis	CYP27A1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,900</b>
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 730,000</b>
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome	PRPS1	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 114,000</b>
Charcot-Marie-Tooth Disease, X-Linked	GB1	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 11,000</b>
Chediak-Higashi Syndrome	LYST	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 7,100</b>
Chondrodysplasia Punctata	ARSE	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 862,000</b>
Choreoacanthocytosis	VPS13A	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 13,000</b>
Choroideremia	CHM	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 125,000</b>
Chronic Granulomatous Disease (CYBA-Related)	CYBA	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 5,000</b>
Chronic Granulomatous Disease (CYBB-Related)	CYBB	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 294,000</b>
Citrin Deficiency	SLC25A13	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 12,000</b>
Citrullinemia, Type 1	ASS1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,500</b>
Cockayne Syndrome, Type A	ERCC8	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 8,900</b>
Cockayne Syndrome, Type B and other ERCC6-Related Disorders	ERCC6	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 8,100</b>
Cohen Syndrome	VPS13B	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,400</b>
Combined Factor V and VIII Deficiency	LMAN1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 102,000</b>
Combined Malonic and Methylmalonic Aciduria	ACSF3	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,400</b>
Combined Oxidative Phosphorylation Deficiency 1	GFM1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 13,000</b>
Combined Oxidative Phosphorylation Deficiency 3	TTFM	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 27,000</b>
Combined Pituitary Hormone Deficiency 1	POU1F1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,900</b>
Combined Pituitary Hormone Deficiency 2	PROP1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,800</b>
Combined Pituitary Hormone Deficiency 3	LHX3	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 140,000</b>
Combined SAP Deficiency	PSAP	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 44,000</b>
Cone-Rod Dystrophy 6 / Leber Congenital Amaurosis 1	GUCY2D	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,200</b>
Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency	CYP11B1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 520</b>
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency	CYP17A1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,800</b>
Congenital Adrenal Hypoplasia (NR0B1-Related)	NR0B1	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 353,000</b>
Congenital Adrenal Insufficiency (CYP11A1-Related)	CYP11A1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,100</b>
Congenital Amegakaryocytic Thrombocytopenia	MPL	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,100</b>
Congenital Bile Acid Synthesis Defect (AKR1D1-Related)	AKR1D1	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,900</b>



<b>Congenital Bile Acid Synthesis Defect (HSD3B7-Related)</b>	<i>HSD3B7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,900
<b>Congenital Disorder of Deglycosylation</b>	<i>NGLY1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 14,000
<b>Congenital Disorder of Glycosylation, Type Ia</b>	<i>PMM2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 540
<b>Congenital Disorder of Glycosylation, Type Ib</b>	<i>MPI</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,600
<b>Congenital Disorder of Glycosylation, Type Ic</b>	<i>ALG6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,100
<b>Congenital Disorder of Glycosylation, Type Im</b>	<i>DOLK</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 134,000
<b>Congenital Dyserythropoietic Anemia Type 2</b>	<i>SEC23B</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,000
<b>Congenital Dyserythropoietic Anemia, Type Ia</b>	<i>CDAN1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 470
<b>Congenital Ichthyosis 4A and 4B</b>	<i>ABCA12</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,100
<b>Congenital Insensitivity to Pain with Anhidrosis</b>	<i>NTRK1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,700
<b>Congenital Muscular Dystrophy (LAMA2-Related)</b>	<i>LAMA2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 640
<b>Congenital Myasthenic Syndrome (CHAT-Related)</b>	<i>CHAT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,100
<b>Congenital Myasthenic Syndrome (CHRNE-Related)</b>	<i>CHRNE</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,100
<b>Congenital Myasthenic Syndrome (DOK7-Related)</b>	<i>DOK7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
<b>Congenital Myasthenic Syndrome (RAPSN-Related)</b>	<i>RAPSN</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,900
<b>Congenital Neutropenia (HAX1-Related)</b>	<i>HAX1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 82,000
<b>Congenital Neutropenia (VPS45-Related)</b>	<i>VPS45</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 163,000
<b>Congenital Nongoitrous Hypothyroidism 1</b>	<i>TSHR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,000
<b>Congenital Nongoitrous Hypothyroidism 4</b>	<i>TSHB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 118,000
<b>Congenital Secretory Chloride Diarrhea 1</b>	<i>SLC26A3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,400
<b>Corneal Dystrophy and Perceptive Deafness</b>	<i>SLC4A11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,600
<b>Corticosterone Methyloxidase Deficiency</b>	<i>CYP11B2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
<b>Cystic Fibrosis</b>	<i>CFTR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 440
<b>Cystinosis</b>	<i>CTNS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,700
<b>Cystinuria (SLC3A1-Related)</b>	<i>SLC3A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 590
<b>Cytochrome C Oxidase Deficiency / Leigh Syndrome (COX15-Related)</b>	<i>COX15</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,300
<b>D-Bifunctional Protein Deficiency</b>	<i>HSD17B4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,000
<b>Deafness, Autosomal Recessive 3</b>	<i>MYO15A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 240
<b>Deafness, Autosomal Recessive 59</b>	<i>PJVK</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 57,000
<b>Deafness, Autosomal Recessive 7</b>	<i>TMC1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
<b>Deafness, Autosomal Recessive 76</b>	<i>SYNE4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 43,000
<b>Deafness, Autosomal Recessive 77</b>	<i>LOXHD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,700
<b>Deafness, Autosomal Recessive 8/10</b>	<i>TMPRSS3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 510
<b>Deafness, Autosomal Recessive 9</b>	<i>OTOF</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,400
<b>Desbuquois Dysplasia 1</b>	<i>CANT1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 24,000
<b>Desmosterolosis</b>	<i>DHCR24</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 27,000
<b>Diaphanospondylodysostosis</b>	<i>BMPER</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 18,000
<b>Distal Renal Tubular Acidosis and other SLC4A1-related Disorders</b>	<i>SLC4A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,000
<b>Duchenne Muscular Dystrophy / Becker Muscular Dystrophy</b>	<i>DMD</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
<b>Dyskeratosis Congenita (DKC1-related)</b>	<i>DKC1</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,259,000
<b>Dyskeratosis Congenita (RTEL1-Related)</b>	<i>RTEL1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,800
<b>Dystrophic Epidermolysis Bullosa</b>	<i>COL7A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 900
<b>Ehlers-Danlos Syndrome, Type VI</b>	<i>PLOD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 20,000
<b>Ehlers-Danlos Syndrome, Type VIIC</b>	<i>ADAMTS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 243,000
<b>Ellis-Van Creveld Syndrome (EVC2-Related)</b>	<i>EVC2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,300

Ellis-van Creveld Syndrome (EVC-Related)	<i>EVC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,200
Emery-Dreifuss Myopathy 1	<i>EMD</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 833,000
Enhanced S-Cone Syndrome	<i>NR2E3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,600
Ethylmalonic Encephalopathy	<i>ETHE1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,400
Fabry Disease	<i>GLA</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,700
Factor IX Deficiency	<i>F9</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,100
Factor VII Deficiency	<i>F7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 450
Factor XI Deficiency	<i>F11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
Familial Autosomal Recessive Hypercholesterolemia	<i>LDLRAP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 136,000
Familial Dysautonomia	<i>IKBKAP</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 51,000
Familial Hypercholesterolemia	<i>LDLR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 280
Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	<i>HADH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,200
Familial Hyperinsulinism (ABCC8-Related)	<i>ABCC8</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 450
Familial Hyperinsulinism (KCNJ11-Related)	<i>KCNJ11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,300
Familial Hyperphosphatemic Tumoral Calcinosis	<i>GALNT3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,800
Familial Mediterranean Fever	<i>MEFV</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Fanconi Anemia, Group A	<i>FANCA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Fanconi Anemia, Group C	<i>FANCC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Fanconi Anemia, Group G	<i>FANCG</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 28,000
Fanconi-Bickel Syndrome	<i>SLC2A2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,000
Fragile X Syndrome	<i>FMR1</i>	XL	Reduced Risk	FMR1 CGG repeat sizes: Not Performed FMR1 Sequencing: Negative Fragile X CGG triplet repeat expansion testing was not performed at this time, as the patient has either been previously tested or is a male. <b>Personalized Residual Risk:</b> 1 in 19,000
Fructose-1,6-Bisphosphatase Deficiency	<i>FBP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,600
Fucosidosis	<i>FUCA1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,200
Fumarase Deficiency	<i>FH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,500
Fundus Albipunctatus	<i>RDH5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,000
Galactokinase Deficiency	<i>GALK1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Galactose Epimerase Deficiency	<i>GALE</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,600
Galactosemia	<i>GALT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,200
Galactosialidosis	<i>CTSA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,900
Gaucher Disease	<i>GBA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,300
Generalized Thyrotropin-Releasing Hormone Resistance	<i>TRHR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 104,000
Geroderma Osteodysplasticum	<i>GORAB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 70,000
Glanzmann Thrombasthenia (ITGA2B-Related)	<i>ITGA2B</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Glanzmann Thrombasthenia (ITGB3-Related)	<i>ITGB3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,600
Glutaric Acidemia, Type I	<i>GCDH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Glutaric Acidemia, Type IIa	<i>ETFA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,700
Glutaric Acidemia, Type IIb	<i>ETFB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,900
Glutaric Acidemia, Type IIc	<i>ETFDH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,700
Glutathione Synthetase Deficiency	<i>GSS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,500
Glycine Encephalopathy (AMT-Related)	<i>AMT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,700
Glycine Encephalopathy (GLDC-Related)	<i>GLDC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 760
Glycogen Storage Disease, Type 0	<i>GYS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Glycogen Storage Disease, Type Ia	<i>G6PC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,300
Glycogen Storage Disease, Type Ib	<i>SLC37A4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,300
Glycogen Storage Disease, Type III	<i>AGL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,600

Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease	<i>GBE1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,400
Glycogen Storage Disease, Type IXb	<i>PHKB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,600
Glycogen Storage Disease, Type V	<i>PYGM</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Glycogen Storage Disease, Type VI	<i>PYGL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,600
Glycogen Storage Disease, Type VII	<i>PFKM</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,300
GRACILE Syndrome and Other <i>BCS1L</i> -Related Disorders	<i>BCS1L</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,900
Gray Platelet Syndrome	<i>NBEAL2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,800
Growth Hormone Deficiency, Type IB	<i>GHRHR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,900
Hemochromatosis, Type 2A	<i>HFE2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Hemochromatosis, Type 3	<i>TFR2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 11,000
Hereditary Fructose Intolerance	<i>ALDOB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
Hereditary Spastic Paraparesis 49	<i>TECPR2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 116,000
Hermansky-Pudlak Syndrome, Type 1	<i>HPS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,500
Hermansky-Pudlak Syndrome, Type 3	<i>HPS3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 49,000
Hermansky-Pudlak Syndrome, Type 4	<i>HPS4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 35,000
Hermansky-Pudlak Syndrome, Type 6	<i>HPS6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 87,000
HMG-CoA Lyase Deficiency	<i>HMGCL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Hmg-CoA Synthase 2 Deficiency	<i>HMGCS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,000
Holocarboxylase Synthetase Deficiency	<i>HLCS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,500
Homocystinuria ( <i>CBS</i> -Related)	<i>CBS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,400
Homocystinuria due to <i>MTHFR</i> Deficiency	<i>MTHFR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,300
Homocystinuria, cblE Type	<i>MTRR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,600
Homocystinuria-Megaloblastic Anemia, Cobalamin G Type	<i>MTR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,100
Hydrocephalus	<i>L1CAM</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 40,000
Hydroletharus Syndrome	<i>HYLS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 52,000
Hyper-Igm Syndrome	<i>CD40LG</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,167,000
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	<i>SLC25A15</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,700
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	<i>SARS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 23,000
Hypohidrotic Ectodermal Dysplasia 1	<i>EDA</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 22,000
Hypomagnesemia 1	<i>TRPM6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 11,000
Hypomyelinating Leukodystrophy 3	<i>AIMP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 341,000
Hypomyelinating Leukodystrophy 12	<i>VPS11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 72,000
Hypoparathyroidism-Retardation-Dysmorphic Syndrome	<i>TBCE</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 21,000
Hypophosphatasia	<i>ALPL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 790
Hypophosphatemic Rickets with Hypercalciuria	<i>SLC34A3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Hypotrichosis 8 / Autosomal Recessive Woolly Hair 1	<i>LPAR6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 27,000
Immunodeficiency 18	<i>CD3E</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 73,000
Immunodeficiency 19	<i>CD3D</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 46,000
Inclusion Body Myopathy 2	<i>GNE</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,000
Infantile Cerebral and Cerebellar Atrophy	<i>MED17</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 129,000
Infantile Neuroaxonal Dystrophy 1 and other <i>PLA2G6</i> -Related Disorders	<i>PLA2G6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 690
Intellectual Disability, Autosomal Recessive 3	<i>CC2D1A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 220,000
Intrahepatic Cholestasis	<i>ATP8B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,400
Isovaleric Acidemia	<i>IVD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,000
Joubert Syndrome 2	<i>TMEM216</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 152,000

Joubert Syndrome 4 / Senior-Loken Syndrome 1 / Juvenile Nephronophthisis 1	<i>NPHP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 21,000
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome	<i>RPGRIPL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 32,000
Junctional Epidermolysis Bullosa ( <i>COL17A1</i> -Related)	<i>COL17A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 25,000
Junctional Epidermolysis Bullosa ( <i>ITGA6</i> -Related)	<i>ITGA6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 125,000
Junctional Epidermolysis Bullosa ( <i>ITGB4</i> -Related)	<i>ITGB4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,400
Junctional Epidermolysis Bullosa ( <i>LAMA3</i> -Related)	<i>LAMA3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 21,000
Junctional Epidermolysis Bullosa ( <i>LAMB3</i> -Related)	<i>LAMB3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
Junctional Epidermolysis Bullosa ( <i>LAMC2</i> -Related)	<i>LAMC2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 77,000
Kohlschutter-Tonz Syndrome	<i>ROGDI</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,300
Krabbe Disease	<i>GALC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 860
Lamellar Ichthyosis, Type 1	<i>TGM1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
Laron Dwarfism	<i>GHR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,700
Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies	<i>CEP290</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Leber Congenital Amaurosis 13	<i>RDH12</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,500
Leber Congenital Amaurosis 15 / Retinitis Pigmentosa 14	<i>TULP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,800
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20	<i>RPE65</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,500
Leber Congenital Amaurosis 4	<i>AIP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,100
Leber Congenital Amaurosis 5	<i>LCA5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 14,000
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy	<i>CRB1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 990
Leigh Syndrome ( <i>NDUFS7</i> -Related)	<i>NDUFS7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 26,000
Leigh Syndrome ( <i>SURF1</i> -Related)	<i>SURF1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,400
Leigh Syndrome, French-Canadian Type	<i>LRPPRC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 32,000
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogyposis with Anterior Horn Cell Disease	<i>GLE1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Lethal Congenital Contracture Syndrome 2	<i>ERBB3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 96,000
Lethal Congenital Contracture Syndrome 3	<i>PIP5K1C</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 318,000
Leukoencephalopathy with Vanishing White Matter	<i>EIF2B5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,300
Limb-Girdle Muscular Dystrophy, Type 2A	<i>CAPN3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 960
Limb-Girdle Muscular Dystrophy, Type 2B	<i>DYSF</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Limb-Girdle Muscular Dystrophy, Type 2C	<i>SGCG</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,900
Limb-Girdle Muscular Dystrophy, Type 2D	<i>SGCA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,500
Limb-Girdle Muscular Dystrophy, Type 2E	<i>SGCB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 31,000
Limb-Girdle Muscular Dystrophy, Type 2F	<i>SGCD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 52,000
Limb-Girdle Muscular Dystrophy, Type 2H	<i>TRIM32</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Limb-Girdle Muscular Dystrophy, Type 2I	<i>FKRP</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,400
Limb-Girdle Muscular Dystrophy, Type 2L	<i>ANO5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 660
Lipoamide Dehydrogenase Deficiency	<i>DLD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 14,000
Lipoid Adrenal Hyperplasia	<i>STAR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,600
Lipoprotein Lipase Deficiency	<i>LPL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,400
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	<i>HADHA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,900
Lowe Syndrome	<i>OCRL</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,375,000
Lysinuric Protein Intolerance	<i>SLC7A7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,000

Malonyl-CoA Decarboxylase Deficiency	<i>MLYCD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,800
Maple Syrup Urine Disease, Type 1a	<i>BCKDHA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,100
Maple Syrup Urine Disease, Type 1b	<i>BCKDHB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Maple Syrup Urine Disease, Type 2	<i>DBT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,600
Meckel Syndrome 1 / Bardet-Biedl Syndrome 13	<i>MKS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,700
Medium Chain Acyl-CoA Dehydrogenase Deficiency	<i>ACADM</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
MEDNIK Syndrome	<i>AP1S1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 211,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	<i>MLC1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,300
Megaloblastic Anemia 1	<i>AMN</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,300
Menkes Disease	<i>ATP7A</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 172,000
Metachromatic Leukodystrophy	<i>ARSA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,000
Methionine Adenosyltransferase I/III Deficiency	<i>MAT1A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
Methylmalonic Acidemia (MMAA-Related)	<i>MMAA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 15,000
Methylmalonic Acidemia (MMAB-Related)	<i>MMAB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Methylmalonic Acidemia (MUT-Related)	<i>MUT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,300
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type	<i>MMACHC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,800
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type	<i>MMADHC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 219,000
Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type	<i>LMBRD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,600
Methylmalonyl-CoA Epimerase Deficiency	<i>MCEE</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 98,000
Microphthalmia / Anophthalmia	<i>VSX2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 40,000
Mitochondrial Complex I Deficiency (ACAD9-Related)	<i>ACAD9</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Mitochondrial Complex I Deficiency (NDUFA11-Related)	<i>NDUFA11</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 414,000
Mitochondrial Complex I Deficiency (NDUFAF5-Related)	<i>NDUFAF5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 98,000
Mitochondrial Complex I Deficiency (NDUFS6-Related)	<i>NDUFS6</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 353,000
Mitochondrial Complex I Deficiency (NDUFV1-Related)	<i>NDUFV1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 870
Mitochondrial Complex I Deficiency / Leigh Syndrome (FOXRED1-Related)	<i>FOXRED1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 13,000
Mitochondrial Complex I Deficiency / Leigh Syndrome (NDUFAF2-Related)	<i>NDUFAF2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 168,000
Mitochondrial Complex I Deficiency / Leigh Syndrome (NDUFS4-Related)	<i>NDUFS4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 41,000
Mitochondrial Complex IV Deficiency (COX20-related)	<i>COX20</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 42,000
Mitochondrial Complex IV Deficiency (COX6B1-related)	<i>COX6B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,116,000
Mitochondrial Complex IV Deficiency (APOPT1-Related)	<i>APOPT1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,200
Mitochondrial Complex IV Deficiency (PET100-Related)	<i>PET100</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 469,000
Mitochondrial Complex IV Deficiency (SCO1-related)	<i>SCO1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 13,000
Mitochondrial Complex IV Deficiency / Leigh Syndrome (COX10-Related)	<i>COX10</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,200
Mitochondrial DNA Depletion Syndrome 2	<i>TK2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,900
Mitochondrial DNA Depletion Syndrome 3	<i>DGUOK</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,200
Mitochondrial DNA Depletion Syndrome 4A and 4B and other POLG-Related Disorders	<i>POLG</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 320
Mitochondrial DNA Depletion Syndrome 5	<i>SUCLA2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 78,000
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy	<i>MPV17</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,400

<b>Mitochondrial Myopathy and Sideroblastic Anemia 1</b>	<i>PUS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 449,000</b>
<b>Mitochondrial Trifunctional Protein Deficiency (HADHB-Related)</b>	<i>HADHB</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,000</b>
<b>Molybdenum Cofactor Deficiency A</b>	<i>MOCS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 4,700</b>
<b>Mucopolipidosis II / IIIA</b>	<i>GNPTAB</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,100</b>
<b>Mucopolipidosis III Gamma</b>	<i>GNPTG</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 68,000</b>
<b>Mucopolipidosis IV</b>	<i>MCOLN1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 9,400</b>
<b>Mucopolysaccharidosis Type I</b>	<i>IDUA</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,300</b>
<b>Mucopolysaccharidosis Type II</b>	<i>IDS</i>	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 76,000</b>
<b>Mucopolysaccharidosis Type IIIA</b>	<i>SGSH</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,700</b>
<b>Mucopolysaccharidosis Type IIIB</b>	<i>NAGLU</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 950</b>
<b>Mucopolysaccharidosis Type IIIC</b>	<i>HGSNAT</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,200</b>
<b>Mucopolysaccharidosis Type IIID</b>	<i>GNS</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 137,000</b>
<b>Mucopolysaccharidosis Type IVa</b>	<i>GALNS</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 690</b>
<b>Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis</b>	<i>GLB1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,700</b>
<b>Mucopolysaccharidosis type IX</b>	<i>HYAL1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 149,000</b>
<b>Mucopolysaccharidosis type VI</b>	<i>ARSB</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,300</b>
<b>Mucopolysaccharidosis VII</b>	<i>GUSB</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,600</b>
<b>Mulibrey Nanism</b>	<i>TRIM37</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 31,000</b>
<b>Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1</b>	<i>PIGN</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,800</b>
<b>Multiple Pterygium Syndrome</b>	<i>CHRNA3</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 9,900</b>
<b>Multiple Sulfatase Deficiency</b>	<i>SUMF1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 69,000</b>
<b>Muscle-Eye-Brain Disease and Other POMGNT1-Related Congenital Muscular Dystrophy-Dystroglycanopathies</b>	<i>POMGNT1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 4,200</b>
<b>Myoneurogastrointestinal Encephalopathy</b>	<i>TYMP</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,100</b>
<b>Myotubular Myopathy 1</b>	<i>MTM1</i>	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 192,000</b>
<b>N-Acetylglutamate Synthase Deficiency</b>	<i>NAGS</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,200</b>
<b>Nemaline Myopathy 2</b>	<i>NEB</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 2,400</b>
<b>Nephrogenic Diabetes insipidus (AVPR2-related)/ Nephrogenic Syndrome of Inappropriate Antidiuresis</b>	<i>AVPR2</i>	XL	Reduced Risk	<b>Personalized Residual Risk: 1 in 471,000</b>
<b>Nephrogenic Diabetes Insipidus, Type II</b>	<i>AQP2</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,400</b>
<b>Nephronophthisis 2</b>	<i>INVS</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 56,000</b>
<b>Nephrotic Syndrome (NPHS1-Related) / Congenital Finnish Nephrosis</b>	<i>NPHS1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 920</b>
<b>Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome</b>	<i>NPHS2</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 780</b>
<b>Neurodegeneration due to Cerebral Folate Transport Deficiency</b>	<i>FOLR1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 5,300</b>
<b>Neurodevelopmental Disorder with Progressive Microcephaly, Spasticity, and Brain Anomalies</b>	<i>PLAA</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 229,000</b>
<b>Neuronal Ceroid-Lipofuscinosis (CLN3-Related)</b>	<i>CLN3</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 9,200</b>
<b>Neuronal Ceroid-Lipofuscinosis (CLN5-Related)</b>	<i>CLN5</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 4,300</b>
<b>Neuronal Ceroid-Lipofuscinosis (CLN6-Related)</b>	<i>CLN6</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 8,600</b>
<b>Neuronal Ceroid-Lipofuscinosis (CLN8-Related)</b>	<i>CLN8</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,100</b>
<b>Neuronal Ceroid-Lipofuscinosis (MFSD8-Related)</b>	<i>MFSD8</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,200</b>
<b>Neuronal Ceroid-Lipofuscinosis (PPT1-Related)</b>	<i>PPT1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 7,500</b>
<b>Neuronal Ceroid-Lipofuscinosis (TPP1-Related)</b>	<i>TPP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,300</b>
<b>Niemann-Pick Disease (SMPD1-Related)</b>	<i>SMPD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,800</b>
<b>Niemann-Pick Disease, Type C (NPC1-Related)</b>	<i>NPC1</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 690</b>
<b>Niemann-Pick Disease, Type C (NPC2-Related)</b>	<i>NPC2</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,600</b>

Nijmegen Breakage Syndrome	NBN	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 14,000
Non-Syndromic Hearing Loss (GJB2-Related)	GJB2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 600
Oculocutaneous Albinism, Type IA / IB	TYR	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 240
Oculocutaneous Albinism, Type IV	SLC45A2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 830
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	WNT10A	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
Omenn Syndrome (RAG2-Related)	RAG2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 17,000
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type	DCLRE1C	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,500
Omenn Syndrome and other RAG1-Related Disorders	RAG1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 850
Ornithine Aminotransferase Deficiency	OAT	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,400
Ornithine Transcarbamylase Deficiency	OTC	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 103,000
Osteogenesis Imperfecta, Type XI	FKBP10	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,500
Osteopetrosis 1	TCIRG1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,700
Osteopetrosis 8	SNX10	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 16,000
Otospondylomegapiphyseal Dysplasia / Deafness / Fibrochondrogenesis 2	COL11A2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Papillon-Lefevre Syndrome	CTSC	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,000
Pendred Syndrome	SLC26A4	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 390
Peroxisome Biogenesis Disorder 3A and 3B	PEX12	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 30,000
Peroxisome Biogenesis Disorder 7A and 7B	PEX26	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 70,000
Polycystic Kidney Disease, Autosomal Recessive	PKHD1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 450
Polyglandular Autoimmune Syndrome, Type 1	AIRE	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,300
Pontocerebellar Hypoplasia, Type 1A	VRK1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 25,000
Pontocerebellar Hypoplasia, Type 1B	EXOSC3	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Pontocerebellar Hypoplasia, Type 2A and Type 4	TSEN54	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,700
Pontocerebellar Hypoplasia, Type 2E	VPS53	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 139,000
Pontocerebellar Hypoplasia, Type 6	RARS2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,600
Primary Carnitine Deficiency	SLC22A5	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
Primary Ciliary Dyskinesia (CCDC103-Related)	CCDC103	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 27,000
Primary Ciliary Dyskinesia (CCDC151-Related)	CCDC151	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 59,000
Primary Ciliary Dyskinesia (CCDC39-Related)	CCDC39	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Primary Ciliary Dyskinesia (DNAH5-Related)	DNAH5	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
Primary Ciliary Dyskinesia (DNAI1-Related)	DNAI1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,000
Primary Ciliary Dyskinesia (DNAI2-Related)	DNAI2	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 76,000
Primary Ciliary Dyskinesia (RSPH9-Related)	RSPH9	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 253,000
Primary Coenzyme Q10 Deficiency 7	COQ4	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Primary Congenital Glaucoma 3A	CYP1B1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 880
Primary Hyperoxaluria, Type 1	AGXT	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
Primary Hyperoxaluria, Type 2	GRHPR	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 11,000
Primary Hyperoxaluria, Type 3	HOGA1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,400
Progressive Cerebello-Cerebral Atrophy	SEPSECS	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,400
Progressive Familial Intrahepatic Cholestasis, Type 2	ABCB11	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 950
Progressive Myoclonic Epilepsy, Type 1B	PRICKLE1	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 98,000
Progressive Pseudorheumatoid Dysplasia	WISP3	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,600
Prolidase Deficiency	PEPD	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 30,000
Propionic Acidemia (PCCA-Related)	PCCA	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,600
Propionic Acidemia (PCCB-Related)	PCCB	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
Pulmonary Surfactant Dysfunction	ABCA3	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200

Pycnodysostosis	<i>CTSK</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,100
Pyridoxamine 5'-Phosphate Oxidase Deficiency	<i>PNPO</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Pyridoxine-Dependent Epilepsy	<i>ALDH7A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,100
Pyruvate Carboxylase Deficiency	<i>PC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,000
Pyruvate Dehydrogenase E1-Alpha Deficiency	<i>PDHA1</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 139,000
Pyruvate Dehydrogenase E1-Beta Deficiency	<i>PDHB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 15,000
Renal Tubular Acidosis and Deafness	<i>ATP6V1B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,600
Retinitis Pigmentosa 25	<i>EYS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Retinitis Pigmentosa 26	<i>CERKL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 13,000
Retinitis Pigmentosa 28	<i>FAM161A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 34,000
Retinitis Pigmentosa 36	<i>PRCD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 304,000
Retinitis Pigmentosa 59	<i>DHDDS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 601,000
Retinitis Pigmentosa 64 / Bardet-Biedl Syndrome 21 / Cone-Rod Dystrophy 16	<i>C8ORF37</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 168,000
Rh Deficiency Syndrome	<i>RHAG</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 46,000
Rhizomelic Chondrodysplasia Punctata, Type 1	<i>PEX7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Rhizomelic Chondrodysplasia Punctata, Type 3	<i>AGPS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 620,000
Roberts Syndrome	<i>ESCO2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 139,000
Salla Disease	<i>SLC17A5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,400
Salt and Pepper Developmental Regression Syndrome	<i>ST3GAL5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 25,000
Sandhoff Disease	<i>HEXB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
Schimke Immunoosseous Dysplasia	<i>SMARCAL1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,800
Seckel Syndrome 5 / Microcephaly 9	<i>CEP152</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,700
Segawa Syndrome	<i>TH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,100
Sepiapterin Reductase Deficiency	<i>SPR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 35,000
Severe Combined Immunodeficiency ( <i>IL7R</i> -Related)	<i>IL7R</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 20,000
Severe Combined Immunodeficiency ( <i>JAK3</i> -Related)	<i>JAK3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,100
Severe Combined Immunodeficiency ( <i>PTPRC</i> -Related)	<i>PTPRC</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 8,500
Severe Congenital Neutropenia 4	<i>G6PC3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
Severe Neonatal Hyperparathyroidism	<i>CASR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,700
Short Stature, Onychodysplasia, Facial Dysmorphism, and Hypotrichosis	<i>POC1A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 108,000
Short-Chain Acyl-CoA Dehydrogenase Deficiency	<i>ACADS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 660
Shwachman-Diamond Syndrome	<i>SBDS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,700
Sialidosis, Type I and Type II	<i>NEU1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,000
Sjogren-Larsson Syndrome	<i>ALDH3A2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,500
Smith-Lemli-Opitz Syndrome	<i>DHCR7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 750
Spastic Paraplegia 15	<i>ZFYVE26</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 46,000
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly	<i>SLC1A4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 85,000
Spherocytosis, Type 5	<i>EPB42</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,200
Spinal Muscular Atrophy	<i>SMN1</i>	AR	Reduced Risk	SMN1 copy number: 2 SMN2 copy number: 0 c.*3+80T>G: Negative SMN1 Sequencing: Negative <b>Personalized Residual Risk:</b> 1 in 1,107
Spinal Muscular Atrophy with Respiratory Distress 1 / Charcot-Marie-Tooth Disease, Type 2S	<i>IGHMBP2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
Spinocerebellar Ataxia with Axonal Neuropathy 3	<i>COA7</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000



<b>Spodylocostal Dysostosis 1</b>	<i>DLL3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,200
<b>Spodylometaeiphyseal Dysplasia (DDR2-Related)</b>	<i>DDR2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 236,000
<b>Spodylothoracic Dysostosis</b>	<i>MESP2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 382,000
<b>Steel Syndrome</b>	<i>COL27A1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 93,000
<b>Stuve-Wiedemann Syndrome</b>	<i>LIFR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,000
<b>Sulfate Transporter-Related Osteochondrodysplasia</b>	<i>SLC26A2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
				Tay-Sachs disease enzyme: Non-carrier
				White blood cells: Non-carrier
				<ul style="list-style-type: none"> <li>Hex A%: 64.3% (Non-carrier : 55.0 - 72.0%; Carrier: &lt;50%)</li> <li>Total hexosaminidase activity: 2160 nmol/hr/mg</li> </ul>
<b>Tay-Sachs Disease</b>	<i>HEXA</i>	AR	Reduced Risk	Plasma: Non-carrier <ul style="list-style-type: none"> <li>Hex A%: 64.9 (Non-carrier : 58.0 - 72.0%; Carrier: &lt;54%)</li> <li>Total hexosaminidase activity: 460 nmol/hr/ml</li> </ul> HEXA Sequencing: Negative <b>Personalized Residual Risk:</b> 1 in 1,400
<b>Thiamine-Responsive Megaloblastic Anemia Syndrome</b>	<i>SLC19A2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 11,000
<b>Thyroid Dysmorphogenesis 1</b>	<i>SLC5A5</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 45,000
<b>Thyroid Dysmorphogenesis 2A</b>	<i>TPO</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 910
<b>Thyroid Dysmorphogenesis 3</b>	<i>TG</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 850
<b>Thyroid Dysmorphogenesis 4</b>	<i>MYD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
<b>Thyroid Dysmorphogenesis 5</b>	<i>DUOXA2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 29,000
<b>Thyroid Dysmorphogenesis 6</b>	<i>DUOX2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 190
<b>Trichohepatoenteric Syndrome 1</b>	<i>TTC37</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 14,000
<b>Tyrosinemia, Type I</b>	<i>FAH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,900
<b>Tyrosinemia, Type II</b>	<i>TAT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,800
<b>Tyrosinemia, Type III</b>	<i>HPD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 266,000
<b>Usher Syndrome, Type IB</b>	<i>MYO7A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,000
<b>Usher Syndrome, Type IC</b>	<i>USH1C</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,600
<b>Usher Syndrome, Type ID</b>	<i>CDH23</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,400
<b>Usher Syndrome, Type IF</b>	<i>PCDH15</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,800
<b>Usher Syndrome, Type IIA</b>	<i>USH2A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 290
<b>Usher Syndrome, Type III</b>	<i>CLRN1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,300
<b>Very Long Chain Acyl-CoA Dehydrogenase Deficiency</b>	<i>ACADVL</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 920
<b>Vitamin D-Dependent Rickets, Type I</b>	<i>CYP27B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,900
<b>Vitamin D-Resistant Rickets, Type IIA</b>	<i>VDR</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 17,000
<b>Walker-Warburg Syndrome and Other FKTN-Related Dystrophies</b>	<i>FKTN</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 4,200
<b>Werner Syndrome</b>	<i>WRN</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 9,200
<b>Wilson Disease</b>	<i>ATP7B</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 350
<b>Wiskott-Aldrich Syndrome (WAS-Related)</b>	<i>WAS</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,203,000
<b>Wolcott-Rallison Syndrome</b>	<i>EIF2AK3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 22,000
<b>Wolman Disease / Cholesteryl Ester Storage Disease</b>	<i>LIPA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,200
<b>Woodhouse-Sakati Syndrome</b>	<i>DCAF17</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 81,000
<b>X-Linked Juvenile Retinoschisis</b>	<i>RS1</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 40,000
<b>X-Linked Severe Combined Immunodeficiency</b>	<i>IL2RG</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 250,000

<b>Xeroderma Pigmentosum (POLH-Related)</b>	<i>POLH</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 5,900</b>
<b>Xeroderma Pigmentosum, Group A</b>	<i>XPA</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 11,000</b>
<b>Xeroderma Pigmentosum, Group C</b>	<i>XPC</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 12,000</b>
<b>Xeroderma Pigmentosum, Group G</b>	<i>ERCC5</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 3,000</b>
<b>Zellweger Syndrome Spectrum (PEX10-Related)</b>	<i>PEX10</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 6,300</b>
<b>Zellweger Syndrome Spectrum (PEX2-Related)</b>	<i>PEX2</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 77,000</b>
<b>Zellweger Syndrome Spectrum (PEX6-Related)</b>	<i>PEX6</i>	AR	Reduced Risk	<b>Personalized Residual Risk: 1 in 1,600</b>

AR=Autosomal recessive; XL=X-linked

## Test methods and comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

### Fragile X CGG Repeat Analysis (Analytical Detection Rate >99%)

PCR amplification using Asuragen, Inc. AmplideX<sup>®</sup> *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* premutations and full mutations greater than 90 CGG repeats in length were further analyzed by Southern blot analysis or methylation PCR to assess the size and methylation status of the *FMR1* CGG repeat. Additional testing to determine the status of AGG interruptions within the *FMR1* CGG repeat will be automatically performed for premutation alleles ranging from 55 to 90 repeats. These results, which may modify risk for expansion, will follow in a separate report.

### Genotyping (Analytical Detection Rate >99%)

Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY<sup>®</sup> System were used to identify certain recurrent variants that are complex in nature or are present in low copy repeats. Rare sequence variants may interfere with assay performance.

### Multiplex Ligation-Dependent Probe Amplification (MLPA) (Analytical Detection Rate >99%)

MLPA<sup>®</sup> probe sets and reagents from MRC-Holland were used for copy number analysis of specific targets versus known control samples. False positive or negative results may occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

For alpha thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. This test is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome, may not be detected. With the exception of triplications, other benign alpha-globin gene polymorphisms will not be reported. Analyses of *HBA1* and *HBA2* are performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For Duchenne muscular dystrophy, the copy numbers of all *DMD* exons were analyzed. Potentially pathogenic single exon deletions and duplications are confirmed by a second method. Analysis of *DMD* is performed in association with sequencing of the coding regions.

For congenital adrenal hyperplasia, the copy number of the *CYP21A2* gene was analyzed. This analysis can detect large deletions typically due to unequal meiotic crossing-over between *CYP21A2* and the pseudogene *CYP21A1P*. Classic 30-kb deletions make up approximately 20% of *CYP21A2* pathogenic alleles. This test may also identify certain point mutations in *CYP21A2* caused by gene conversion events between *CYP21A2* and *CYP21A1P*. Some carriers may not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *CYP21A2* gene on one chromosome and loss of *CYP21A2* (deletion) on the other chromosome. Analysis of *CYP21A2* is performed in association with long-range PCR of the coding regions followed by short-read sequencing.

For spinal muscular atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals diagnosed with SMA have a causative *SMN1* variant that occurred de novo, and therefore cannot be picked up by carrier screening in the parents. Analysis of *SMN1* is performed in association with short-read sequencing of exons 2a-7, followed by confirmation using long-range PCR (described below).

In individuals with two copies of *SMN1* with Ashkenazi Jewish, East Asian, African American, Native American or Caucasian ancestry, the presence or absence of c. \*3+80T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

MLPA for Gaucher disease (*GBA*), cystic fibrosis (*CFTR*), and non-syndromic hearing loss (*GJB2/GJB6*) will only be performed if indicated for confirmation of detected CNVs. If *GBA* analysis was performed, the copy numbers of exons 1, 3, 4, and 6 - 10 of the *GBA* gene (of 11 exons total)

were analyzed. If *CFTR* analysis was performed, the copy numbers of all 27 *CFTR* exons were analyzed. If *GJB2/GJB6* analysis was performed, the copy number of the two *GJB2* exons were analyzed, as well as the presence or absence of the two upstream deletions of the *GJB2* regulatory region, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854).

#### Next Generation Sequencing (NGS) (Analytical Detection Rate >95%)

NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect™XT Low Input technology was used with a custom capture library to target the exonic regions and intron/exon splice junctions of the relevant genes, as well as a number of UTR, intronic or promoter regions that contain previously reported mutations. Libraries were pooled and sequenced on the Illumina NovaSeq 6000 platform, using paired-end 100 bp reads. The sequencing data was analyzed using a custom bioinformatics algorithm designed and validated in house.

The coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage (minimum of 20X) and data quality threshold values. Most exons not meeting a minimum of >20X read depth across the exon are further analyzed by Sanger sequencing. Please note that several genomic regions present difficulties in mapping or obtaining read depth >20X. These regions, which are described below, will not be reflexed to Sanger sequencing if the mapping quality or coverage is poor. Any variants identified during testing in these regions are confirmed by a second method and reported if determined to be pathogenic or likely pathogenic. However, as there is a possibility of false negative results within these regions, detection rates and residual risks for these genes have been calculated with the presumption that variants in these exons will not be detected, unless included in the MassARRAY® genotyping platform.

**Exceptions:** *ABCD1* (NM\_000033.3) exons 8 and 9; *ACADSB* (NM\_001609.3) chr10:124,810,695-124,810,707 (partial exon 9); *ADA* (NM\_000022.2) exon 1; *ADAMTS2* (NM\_014244.4) exon 1; *AGPS* (NM\_003659.3) chr2:178,257,512-178,257,649 (partial exon 1); *ALDH7A1* (NM\_001182.4) chr5:125,911,150-125,911,163 (partial exon 7) and chr5:125,896,807-125,896,821 (partial exon 10); *ALMS1* (NM\_015120.4) chr2:73,612,990-73,613,041 (partial exon 1); *APOPT1* (NM\_032374.4) chr14:104,040,437-104,040,455 (partial exon 3); *CDAN1* (NM\_138477.2) exon 2; *CEP152* (NM\_014985.3) chr15:49,061,146-49,061,165 (partial exon 14) and exon 22; *CEP290* (NM\_025114.3) exon 5, exon 7, chr12:88,519,017-88,519,039 (partial exon 13), chr12:88,514,049-88,514,058 (partial exon 15), chr12:88,502,837-88,502,841 (partial exon 23), chr12:88,481,551-88,481,589 (partial exon 32), chr12:88,471,605-88,471,700 (partial exon 40); *CFTR* (NM\_000492.3) exon 10; *COL4A4* (NM\_000092.4) chr2:227,942,604-227,942,619 (partial exon 25); *COX10* (NM\_001303.3) exon 6; *CYP11B1* (NM\_000497.3) exons 3-7; *CYP11B2* (NM\_000498.3) exons 3-7; *DNAI2* (NM\_023036.4) chr17:72,308,136-72,308,147 (partial exon 12); *DOK7* (NM\_173660.4) chr4:3,465,131-3,465,161 (partial exon 1) and exon 2; *DUOX2* (NM\_014080.4) exons 6-8; *EIF2AK3* (NM\_004836.5) exon 8; *EVC* (NM\_153717.2) exon 1; *F5* (NM\_000130.4) chr1:169,551,662-169,551,679 (partial exon 2); *FH* (NM\_000143.3) exon 1; *GAMT* (NM\_000156.5) exon 1; *GLDC* (NM\_000170.2) exon 1; *GNPTAB* (NM\_024312.4) chr17:4,837,000-4,837,400 (partial exon 2); *GNPTG* (NM\_032520.4) exon 1; *GHR* (NM\_000163.4) exon 3; *GYS2* (NM\_021957.3) chr12:21,699,370-21,699,409 (partial exon 12); *HGSNAT* (NM\_152419.2) exon 1; *IDS* (NM\_000202.6) exon 3; *ITGB4* (NM\_000213.4) chr17:73,749,976-73,750,060 (partial exon 33); *JAK3* (NM\_000215.3) chr19:17,950,462-17,950,483 (partial exon 10); *LIFR* (NM\_002310.5) exon 19; *LMBRD1* (NM\_018368.3) chr6:70,459,226-70,459,257 (partial exon 5), chr6:70,447,828-70,447,836 (partial exon 7) and exon 12; *LYST* (NM\_000081.3) chr1:235,944,158-235,944,176 (partial exon 16) and chr1:235,875,350-235,875,362 (partial exon 43); *MLYCD* (NM\_012213.2) chr16:83,933,242-83,933,282 (partial exon 1); *MTR* (NM\_000254.2) chr1:237,024,418-237,024,439 (partial exon 20) and chr1:237,038,019-237,038,029 (partial exon 24); *NBEAL2* (NM\_015175.2) chr3:47,021,385-47,021,407 (partial exon 1); *NEB* (NM\_001271208.1) exons 82-105; *NPC1* (NM\_000271.4) chr18:21,123,519-21,123,538 (partial exon 14); *NPHP1* (NM\_000272.3) chr2:110,937,251-110,937,263 (partial exon 3); *OCRL* (NM\_000276.3) chrX:128,674,450-128,674,460 (partial exon 1); *PHKB* (NM\_000293.2) exon 1 and chr16:47,732,498-47,732,504 (partial exon 30); *PIGN* (NM\_176787.4) chr18:59,815,547-59,815,576 (partial exon 8); *PIP5K1C* (NM\_012398.2) exon 1 and chr19:3637602-3637616 (partial exon 17); *POU1F1* (NM\_000306.3) exon 5; *PTPRC* (NM\_002838.4) exons 11 and 23; *PUS1* (NM\_025215.5) chr12:132,414,446-132,414,532 (partial exon 2); *RPGRIP1L* (NM\_015272.2) exon 23; *SGSH* (NM\_000199.3) chr17:78,194,022-78,194,072 (partial exon 1); *SLC6A8* (NM\_005629.3) exons 3 and 4; *ST3GAL5* (NM\_003896.3) exon 1; *SURF1* (NM\_003172.3) chr9:136,223,269-136,223,307 (partial exon 1); *TRPM6* (NM\_017662.4) chr9:77,362,800-77,362,811 (partial exon 31); *TSEN54* (NM\_207346.2) exon 1; *TYR* (NM\_000372.4) exon 5; *VWF* (NM\_000552.3) exons 24-26, chr12:6,125,675-6,125,684 (partial exon 30), chr12:6,121,244-6,121,265 (partial exon 33), and exon 34.

This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions may not be detected, including, but not limited to, UTRs, promoters, and deep intronic areas, or regions that fall into the Exceptions mentioned above. This technology may not detect all small insertion/deletions and is not diagnostic for repeat expansions and structural genomic variation. In addition, a mutation(s) in a gene not included on the panel could be present in this patient.

Variant interpretation and classification was performed based on the American College of Medical Genetics Standards and Guidelines for the Interpretation of Sequence Variants (Richards et al, 2015). All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis will not be reported.

#### Next Generation Sequencing for *SMN1*

Exonic regions and intron/exon splice junctions of *SMN1* and *SMN2* were captured, sequenced, and analyzed as described above. Any variants located within exons 2a-7 and classified as pathogenic or likely pathogenic were confirmed to be in either *SMN1* or *SMN2* using gene-specific long-range PCR analysis followed by Sanger sequencing. Variants located in exon 1 cannot be accurately assigned to either *SMN1* or *SMN2* using our current methodology, and so these variants are not reported.

#### Copy Number Variant Analysis (Analytical Detection Rate >95%)

Large duplications and deletions were called from the relative read depths on an exon-by-exon basis using a custom exome hidden Markov model (XHMM) algorithm. Deletions or duplications determined to be pathogenic or likely pathogenic were confirmed by either a custom arrayCGH platform, quantitative PCR, or MLPA (depending on CNV size and gene content). While this algorithm is designed to pick up deletions and duplications of 2 or more exons in length, potentially pathogenic single-exon CNVs will be confirmed and reported, if detected. Deletions and duplications near the lower limit of detection may not be detected due to run variability.

#### Exon Array (Confirmation method) (Accuracy >99%)

The customized oligonucleotide microarray (Oxford Gene Technology) is a highly-targeted exon-focused array capable of detecting medically relevant microdeletions and microduplications at a much higher resolution than traditional aCGH methods. Each array matrix has approximately 180,000 60-mer oligonucleotide probes that cover the entire genome. This platform is designed based on human genome NCBI Build 37 (hg19) and the CGH probes are enriched to target the exonic regions of the genes in this panel.

#### Quantitative PCR (Confirmation method) (Accuracy >99%)

The relative quantification PCR is utilized on a Roche Universal Library Probe (UPL) system, which relates the PCR signal of the target region in one group to another. To test for genomic imbalances, both sample DNA and reference DNA is amplified with primer/probe sets that specific to the target region and a control region with known genomic copy number. Relative genomic copy numbers are calculated based on the standard  $\Delta\Delta C_t$  formula.

#### Long-Range PCR (Analytical Detection Rate >99%)

Long-range PCR was performed to generate locus-specific amplicons for *CYP21A2*, *HBA1* and *HBA2* and *GBA*. The PCR products were then prepared for short-read NGS sequencing and sequenced. Sequenced reads were mapped back to the original genomic locus and run through the bioinformatics pipeline. If indicated, copy number from MLPA was correlated with the sequencing output to analyze the results. Please note that in rare cases, allele drop-out may occur, which has the potential to lead to false negative results. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where multiple copies of *CYP21A2* are located on the same chromosome in tandem, only the last copy will be amplified and assessed for potentially pathogenic variants, due to size limitations of the PCR reaction. However, because these alleles contain at least two copies of the *CYP21A2* gene in tandem, it is expected that this patient has at least one functional gene in the tandem allele and this patient is therefore less likely to be a carrier. A *CYP21A1P/CYP21A2* hybrid gene detected only by MLPA but not by long-range PCR will not be reported when the long-range PCR indicates the presence of two full *CYP21A2* gene copies (one on each chromosome), as the additional hybrid gene is nonfunctional. Classic 30-kb deletions are identified by MLPA and are also identified by the presence of multiple common pathogenic *CYP21A2* variants by long-range PCR. Since multiple pseudogene-derived variants are detected in all cases with the classic 30kb deletion, we cannot rule out the possibility that some variant(s) detected could be present in trans with the chimeric *CYP21A1P/CYP21A2* gene created by the 30kb deletion. When an individual carries both a duplication allele and a pathogenic variant, or multiple pathogenic variants, the current analysis may not be able to determine the phase (cis/trans configuration) of the *CYP21A2* alleles identified. Family studies may be required in certain scenarios where phasing is required to determine the carrier status.

#### Residual Risk Calculations

Carrier frequencies and detection rates for each ethnicity were calculated through the combination of internal curations of >30,000 variants and genomic frequency data from >138,000 individuals across seven ethnic groups in the gnomAD database. Additional variants in HGMD and novel deleterious variants were also incorporated into the calculation. Residual risk values are calculated using a Bayesian analysis combining the a priori risk of being a pathogenic mutation carrier (carrier frequency) and the detection rate. They are provided only as a guide for assessing approximate risk given a negative result, and values will vary based on the exact ethnic background of an individual. This report does not represent medical advice but should be interpreted by a genetic counselor, medical geneticist or physician skilled in genetic result interpretation and the relevant medical literature.

#### Personalized Residual Risk Calculations

Agilent SureSelect<sup>TM</sup>XT Low-Input technology was utilized in order to create whole-genome libraries for each patient sample. Libraries were then pooled and sequenced on the Illumina NovaSeq platform. Each sequencing lane was multiplexed to achieve 0.4-2x genome coverage, using paired-end 100 bp reads. The sequencing data underwent ancestral analysis using a customized, licensed bioinformatics algorithm that was validated in house. Identified sub-ethnic groupings were binned into one of 7 continental-level groups (African, East Asian, South Asian,

Non-Finnish European, Finnish, Native American, and Ashkenazi Jewish) or, for those ethnicities that matched poorly to the continental-level groups, an 8<sup>th</sup> "unassigned" group, which were then used to select residual risk values for each gene. For individuals belonging to multiple high-level ethnic groupings, a weighting strategy was used to select the most appropriate residual risk. For genes that had insufficient data to calculate ethnic-specific residual risk values, or for sub-ethnic groupings that fell into the "unassigned" group, a "worldwide" residual risk was used. This "worldwide" residual risk was calculated using data from all available continental-level groups.

Several genes have multiple residual risks associated to reflect the likelihood of the tested individual being a carrier for different diseases that are attributed to non-overlapping pathogenic variants in that gene. When calculating the couples' combined reproductive risk, the highest residual risk for each patient was selected.

#### **Sanger Sequencing (Confirmation method) (Accuracy >99%)**

Sanger sequencing, as indicated, was performed using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage (<20 reads) or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

#### **Tay-Sachs Disease (TSD) Enzyme Analysis (Analytical Detection Rate ≥98%)**

Hexosaminidase activity and Hex A% activity were measured by a standard heat-inactivation, fluorometric method using artificial 4-MU-β-N-acetyl glucosaminide (4-MUG) substrate. This assay is highly sensitive and accurate in detecting Tay-Sachs carriers and individuals affected with TSD. Normal ranges of Hex A% activity are 55.0-72.0 for white blood cells and 58.0-72.0 for plasma. It is estimated that less than 0.5% of Tay-Sachs carriers have non-carrier levels of percent Hex A activity, and therefore may not be identified by this assay. In addition, this assay may detect individuals that are carriers of or are affected with Sandhoff disease. False positive results may occur if benign variants, such as pseudodeficiency alleles, interfere with the enzymatic assay. False negative results may occur if both *HEXA* and *HEXB* pathogenic or pseudodeficiency variants are present in the same individual.

Please note that it is not possible to perform Tay-Sachs disease enzyme analysis on saliva samples, buccal swabs, tissue samples, semen samples, or on samples received as extracted DNA.

This test was developed, and its performance characteristics determined by Sema4 Opco, Inc. It has not been cleared or approved by the US Food and Drug Administration. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

## **SELECTED REFERENCES**

### **Carrier Screening**

Grody W et al. ACMG position statement on prenatal/preconception expanded carrier screening. *Genet Med*. 2013 15:482-3.

### **Fragile X syndrome:**

Chen L et al. An information-rich CGG repeat primed PCR that detects the full range of Fragile X expanded alleles and minimizes the need for Southern blot analysis. *J Mol Diag* 2010 12:589-600.

### **Spinal Muscular Atrophy:**

Luo M et al. An Ashkenazi Jewish *SMN1* haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. *Genet Med*. 2014 16:149-56.

### **Ashkenazi Jewish Disorders:**

Scott SA et al. Experience with carrier screening and prenatal diagnosis for sixteen Ashkenazi Jewish Genetic Diseases. *Hum. Mutat*. 2010 31:1-11.

Akler G et al. Towards a unified approach for comprehensive reproductive carrier screening in the Ashkenazi, Sephardi, and Mizrahi Jewish populations. *Mol Genet Genomic Med*. 2020 Feb 8(2):e1053.

### **Duchenne Muscular Dystrophy:**

Flanigan KM et al. Mutational spectrum of *DMD* mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat*. 2009 30:1657-66.



**Variant Classification:**

Richards S et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med.* 2015 May;17(5):405-24

Additional disease-specific references available upon request.



Patient Information	Specimen Information	Client Information
<b>6908, DONOR</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Phone: NG Patient ID: [REDACTED]	Specimen: [REDACTED] Requisition: [REDACTED] Lab Ref #: [REDACTED]  Collected: 08/10/2022 Received: 08/11/2022 / 20:50 EDT Reported: 08/18/2022 / 09:36 EDT	Client #: [REDACTED] NYNJMAIL GENOMICS, SEMA4 SEMA4 62 SOUTHFIELD AVE STAMFORD, CT 06902-7229

Ward: FFXCB

**Cytogenetic Report**

**CHROMOSOME ANALYSIS, BLOOD - 14596** **Lab: EZ**

**CHROMOSOME ANALYSIS, BLOOD**

Order ID: [REDACTED]  
 Specimen Type: Blood  
 Clinical Indication: Encounter of male for testing for disease carrier status for procrea management

**RESULT:**  
 NORMAL MALE KARYOTYPE

**INTERPRETATION:**  
 Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

**NOMENCLATURE:**  
 46,XY

**ASSAY INFORMATION:**

Method: G-Band (Digital Analysis: MetaSyst)  
 Cells Counted: 20  
 Band Level: 450  
 Cells Analyzed: 5  
 Cells Karyotyped: 5

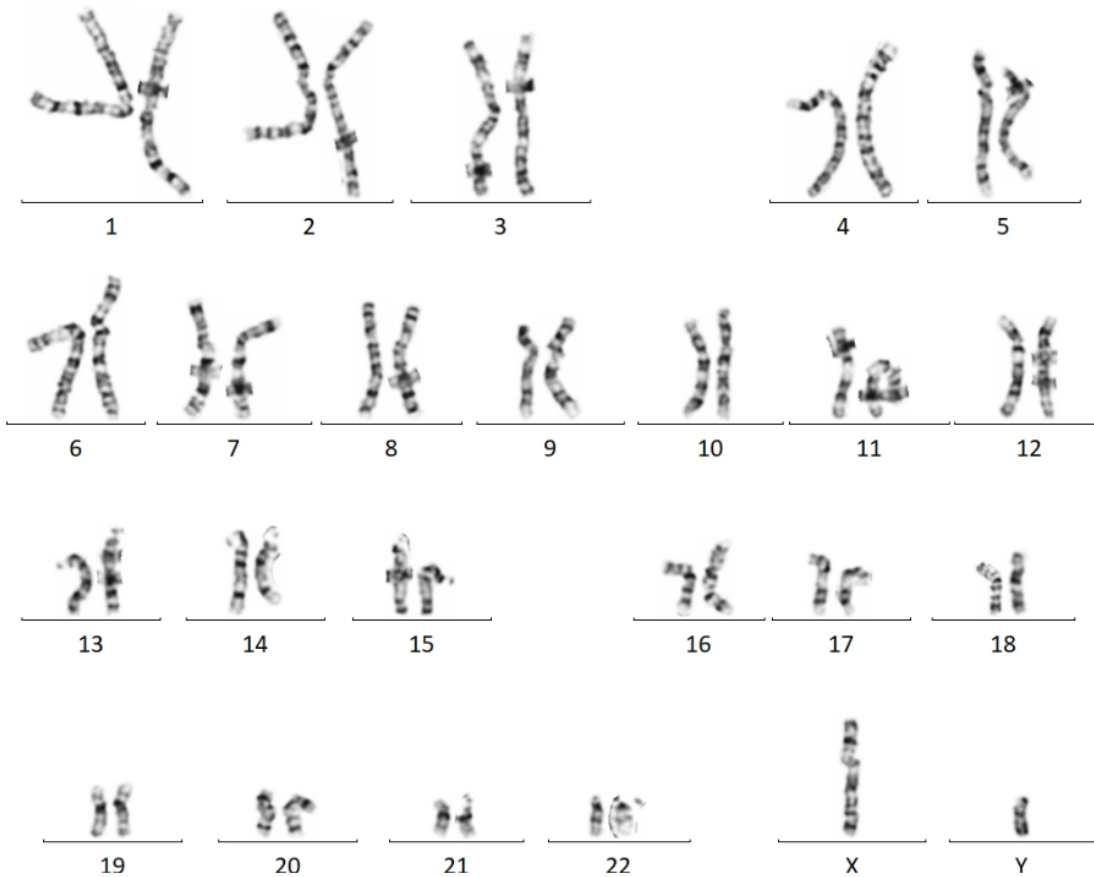
This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements.

Mark A. Micale, PhD, FACMG

Electronic Signature: 8/18/2022 6:30 AM



Patient Information	Specimen Information	Client Information
<b>6908, DONOR</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Patient ID: [REDACTED]	Specimen: [REDACTED] Collected: 08/10/2022 Received: 08/11/2022 / 20:50 EDT Reported: 08/18/2022 / 09:36 EDT	Client #: [REDACTED] GENOMICS, SEMA4



**PERFORMING SITE:**

EZ QUEST DIAGNOSTICS/NICHOLS SJ, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: IRINA MARAMICA, MD, PHD, MBA, CLIA: 05D0643352





Patient Information	Specimen Information	Client Information
<b>6908, DONOR</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Phone: NG Patient ID: [REDACTED]	Specimen: [REDACTED] Requisition: [REDACTED] Lab Ref #: [REDACTED]  Collected: 08/10/2022 Received: 08/11/2022 / 20:49 EDT Reported: 08/13/2022 / 09:29 EDT	Client #: [REDACTED] NYNJMAIL GENOMICS, SEMA4 SEMA4 62 SOUTHFIELD AVE STAMFORD, CT 06902-7229

Ward: FFXCB

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.45		4.20-5.80 Million/uL	Z99
HEMOGLOBIN	15.1		13.2-17.1 g/dL	
HEMATOCRIT	45.7		38.5-50.0 %	
MCV	83.9		80.0-100.0 fL	
MCH	27.7		27.0-33.0 pg	
RDW	13.1		11.0-15.0 %	
HEMOGLOBIN A	97.5		>96.0 %	Z99
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.5		2.2-3.2 %	
INTERPRETATION	*			
Normal phenotype.				

**PERFORMING SITE:**

Z99 QUEST DIAGNOSTICS CLIFTON, 1 INSIGHTS DRIVE, CLIFTON, NJ 07012-2355 Laboratory Director: SHELLA K MONGIA,MD, CLIA: 31D0696246