



## Donor 6398

### Genetic Testing Summary

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

Last Updated: 12/20/23

Donor Reported Ancestry: Indian

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 on attached report.	<b>Carrier: Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (MKS1)</b>  Negative for other genes sequenced	Partner testing recommended before using this donor.
<b>Special Testing</b>		
Gene: PRF1	Negative by gene sequencing	

\*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: 6398 Donor

Date of Birth: [REDACTED]

Sema4 ID: [REDACTED]

Client ID: [REDACTED]

Indication: Carrier Screening

**Specimen Information**

Specimen Type: Blood

Date Collected: 05/05/2022

Date Received: 05/06/2022



Final Report: 05/18/2022

**Referring Provider**[REDACTED]  
Fairfax Cryobank, Inc.

## Expanded Carrier Screen Minus TSE (502 genes)

with Personalized Residual Risk

**SUMMARY OF RESULTS AND RECOMMENDATIONS**

 <b>Positive</b>	 <b>Negative</b>
<b>Carrier of Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (AR)</b> Associated gene(s): <i>MKS1</i> Variant(s) Detected: c.1301G>A, p.W434X, Likely Pathogenic, Heterozygous (one copy)	<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

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.

## Interpretation of positive results

**Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (AR)****Results and Interpretation**

A heterozygous (one copy) likely pathogenic premature stop codon, c.1301G>A, p.W434X, was detected in the *MKS1* gene (NM\_017777.3). When this variant is present in trans with a pathogenic variant, it is considered to be causative for *MKS1*-associated ciliopathy. Therefore, this individual is expected to be at least a carrier for *MKS1*-associated ciliopathy. Heterozygous carriers are not expected to exhibit symptoms of this disease.

**What is Meckel Syndrome 1 / Bardet-Biedl Syndrome 13?**

*MKS1*-associated ciliopathies include two overlapping disorders, known as Bardet-Biedl syndrome and Meckel syndrome. Both diseases are inherited in an autosomal recessive manner, manifest at birth, and can be caused by pathogenic variants in the gene *MKS1*. While *MKS1*-associated ciliopathies are found in individuals of different ethnicities, they are more commonly diagnosed in individuals of Finnish descent, due to the presence of a founder mutation. Bardet-Biedl syndrome is characterized by obesity, intellectual disability, kidney disease, polydactyly, and loss of vision that begins with loss of night vision and progresses to tunnel vision and blindness. Meckel syndrome, which is

more severe, often manifests before birth. Infants are often born with a neural tube defect called occipital encephalocele, brain malformations, facial dysmorphism, renal agenesis, and extra digits. Life expectancy varies according to the phenotype; for many patients with Bardet-Biedl syndrome, it may only be reduced for those with a more severe phenotype, but death in infancy is expected in patients with Meckel syndrome. Currently, it is not possible to predict which phenotype a patient will have based on the inherited variants.

## 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.



**Christie Buchovecky, Ph.D., Assistant Director, Reproductive Genomic**

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>				
<b>Meckel Syndrome 1 / Bardet-Biedl Syndrome 13</b>	<i>MKS1</i>	AR	Carrier	c.1301G>A, p.W434X, Likely Pathogenic, Heterozygous (one copy)
<b>Negative</b>				
<b>2-Methylbutyrylglycinuria</b>	<i>ACADSB</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,800
<b>3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency</b>	<i>HSD3B2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,200
<b>3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC1-Related)</b>	<i>MCCC1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 100,000
<b>3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC2-Related)</b>	<i>MCCC2</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,000
<b>3-Methylglutaconic Aciduria, Type III</b>	<i>OPA3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 29,000
<b>3-Phosphoglycerate Dehydrogenase Deficiency</b>	<i>PHGDH</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,400
<b>6-Pyruvoyl-Tetrahydropterin Synthase Deficiency</b>	<i>PTS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,100
<b>CD59-Mediated Hemolytic Anemia</b>	<i>CD59</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 513,000
<b>Abetalipoproteinemia</b>	<i>MTTP</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 103,000
<b>Achalasia-Addisonianism-Alacrimia Syndrome</b>	<i>AAAS</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 6,400
<b>Achromatopsia (CNGA3-Related)</b>	<i>CNGA3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 490
<b>Achromatopsia (CNGB3-related)</b>	<i>CNGB3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,700
<b>Acrodermatitis Enteropathica</b>	<i>SLC39A4</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 55,000
<b>Acute Infantile Liver Failure</b>	<i>TRMU</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,500
<b>Acyl-CoA Oxidase I Deficiency</b>	<i>ACOX1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 192,000
<b>Adams-Oliver Syndrome 4</b>	<i>EOGT</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 12,000
<b>Adenosine Deaminase Deficiency</b>	<i>ADA</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,100
<b>Adrenocorticotrophic Hormone Deficiency</b>	<i>TBX19</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 10,000
<b>Adrenoleukodystrophy, X-Linked</b>	<i>ABCD1</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 19,000
<b>Agammaglobulinemia</b>	<i>BTK</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 250,000
<b>Agenesis of the Corpus Callosum</b>	<i>FRMD4A</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 348,000
<b>Aicardi-Goutieres Syndrome (RNASEH2C-Related)</b>	<i>RNASEH2C</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 7,000
<b>Aicardi-Goutieres Syndrome (SAMHD1-Related)</b>	<i>SAMHD1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 385,000
<b>Aicardi-Goutieres Syndrome (TREX1-Related)</b>	<i>TREX1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 1,200
<b>Albinism, Oculocutaneous, Type III</b>	<i>TYRP1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 90,000
<b>Alkaptonuria</b>	<i>HGD</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 5,800
<b>Alpha-Mannosidosis</b>	<i>MAN2B1</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 2,500
<b>Alpha-Thalassemia</b>	<i>HBA1/HBA2</i>	AR	Reduced Risk	HBA1 Copy Number: 2 HBA2 Copy Number: 2 No pathogenic copy number variants detected HBA1/HBA2 Sequencing: Negative <b>Personalized Residual Risk:</b> 1 in 380
<b>Alpha-Thalassemia Intellectual Disability Syndrome</b>	<i>ATRX</i>	XL	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 48,000
<b>Alport Syndrome (COL4A3-Related)</b>	<i>COL4A3</i>	AR	Reduced Risk	<b>Personalized Residual Risk:</b> 1 in 3,500

Alport Syndrome (COL4A4-Related)	COL4A4	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,700
Alport Syndrome (COL4A5-Related)	COL4A5	XL	Reduced Risk	Personalized Residual Risk: 1 in 150,000
Alstrom Syndrome	ALMS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,400
Andermann Syndrome	SLC12A6	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Antley-Bixler Syndrome (POR-Related)	POR	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,300
Argininemia	ARG1	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Argininosuccinic Aciduria	ASL	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,900
Aromatase Deficiency	CYP19A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,200
Arthrogryposis, Intellectual Disability, and Seizures	SLC35A3	AR	Reduced Risk	Personalized Residual Risk: 1 in 399,000
Asparagine Synthetase Deficiency	ASNS	AR	Reduced Risk	Personalized Residual Risk: 1 in 307,000
Aspartylglycosaminuria	AGA	AR	Reduced Risk	Personalized Residual Risk: 1 in 220,000
Ataxia With Isolated Vitamin E Deficiency	TTPA	AR	Reduced Risk	Personalized Residual Risk: 1 in 240,000
Ataxia-Telangiectasia	ATM	AR	Reduced Risk	Personalized Residual Risk: 1 in 900
Ataxia-Telangiectasia-Like Disorder 1	MRE11	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
BH4-Deficient Hyperphenylalaninemia C	QDPR	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
BH4-Deficient Hyperphenylalaninemia D	PCBD1	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Bardet-Biedl Syndrome (ARL6-Related)	ARL6	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,600
Bardet-Biedl Syndrome (BBS10-Related)	BBS10	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,400
Bardet-Biedl Syndrome (BBS12-Related)	BBS12	AR	Reduced Risk	Personalized Residual Risk: 1 in 170,000
Bardet-Biedl Syndrome (BBS1-Related)	BBS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,400
Bardet-Biedl Syndrome (BBS2-Related)	BBS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Bardet-Biedl Syndrome (BBS4-Related)	BBS4	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Bare Lymphocyte Syndrome, Type II	CIITA	AR	Reduced Risk	Personalized Residual Risk: 1 in 220,000
Barth Syndrome	TAZ	XL	Reduced Risk	Personalized Residual Risk: 1 in 183,000
Bartter Syndrome, Type 3	CLCNKB	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,700
Bartter Syndrome, Type 4A	BSND	AR	Reduced Risk	Personalized Residual Risk: 1 in 73,000
Bernard-Soulier Syndrome, Type A1	GP1BA	AR	Reduced Risk	Personalized Residual Risk: 1 in 66,000
Bernard-Soulier Syndrome, Type C	GP9	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Beta-Globin-Related Hemoglobinopathies	HBB	AR	Reduced Risk	Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies): 1 in 980 Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbS Variant): 1 in 81,000 Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbC Variant): 1 in 42,000
Beta-Ketothiolase Deficiency	ACAT1	AR	Reduced Risk	Personalized Residual Risk: 1 in 138,000
Beta-Mannosidosis	MANBA	AR	Reduced Risk	Personalized Residual Risk: 1 in 100,000
Bilateral Frontoparietal Polymicrogyria	GPR56	AR	Reduced Risk	Personalized Residual Risk: 1 in 64,000
Biotinidase Deficiency	BTBD	AR	Reduced Risk	Personalized Residual Risk: 1 in 370
Bloom Syndrome	BLM	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Canavan Disease	ASPA	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Carbamoylphosphate Synthetase I Deficiency	CPS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Carnitine Acylcarnitine Translocase Deficiency	SLC25A20	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Carnitine Palmitoyltransferase IA Deficiency	CPT1A	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Carnitine Palmitoyltransferase II Deficiency	CPT2	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Carpenter Syndrome	RAB23	AR	Reduced Risk	Personalized Residual Risk: 1 in 28,000
Cartilage-Hair Hypoplasia	RMRP	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000

Central Hypothyroidism and Testicular Enlargement	IGSF1	XL	Reduced Risk	Personalized Residual Risk: 1 in 781,000
Cerebral Creatine Deficiency Syndrome 1	SLC6A8	XL	Reduced Risk	Personalized Residual Risk: 1 in 208,000
Cerebral Creatine Deficiency Syndrome 2	GAMT	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,800
Cerebral Creatine Deficiency Syndrome 3	GATM	AR	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	AR	Reduced Risk	Personalized Residual Risk: 1 in 154,000
Cerebrotendinous Xanthomatosis	CYP27A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 960
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	AR	Reduced Risk	Personalized Residual Risk: 1 in 479,000
Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome	PRPS1	XL	Reduced Risk	Personalized Residual Risk: 1 in 114,000
Charcot-Marie-Tooth Disease, X-Linked	GJB1	XL	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Chediak-Higashi Syndrome	LYST	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Chondrodysplasia Punctata	ARSE	XL	Reduced Risk	Personalized Residual Risk: 1 in 862,000
Choreoacanthocytosis	VPS13A	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,900
Choroideremia	CHM	XL	Reduced Risk	Personalized Residual Risk: 1 in 125,000
Chronic Granulomatous Disease (CYBA-Related)	CYBA	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,800
Chronic Granulomatous Disease (CYBB-Related)	CYBB	XL	Reduced Risk	Personalized Residual Risk: 1 in 294,000
Citrin Deficiency	SLC25A13	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,600
Citrullinemia, Type 1	ASS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Cockayne Syndrome, Type A	ERCC8	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Cockayne Syndrome, Type B and other ERCC6-Related Disorders	ERCC6	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,500
Cohen Syndrome	VPS13B	AR	Reduced Risk	Personalized Residual Risk: 1 in 16,000
Combined Factor V and VIII Deficiency	LMAN1	AR	Reduced Risk	Personalized Residual Risk: 1 in 168,000
Combined Malonic and Methylmalonic Aciduria	ACSF3	AR	Reduced Risk	Personalized Residual Risk: 1 in 340
Combined Oxidative Phosphorylation Deficiency 1	GFM1	AR	Reduced Risk	Personalized Residual Risk: 1 in 77,000
Combined Oxidative Phosphorylation Deficiency 3	TSFM	AR	Reduced Risk	Personalized Residual Risk: 1 in 21,000
Combined Pituitary Hormone Deficiency 1	POU1F1	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Combined Pituitary Hormone Deficiency 2	PROP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,300
Combined Pituitary Hormone Deficiency 3	LHX3	AR	Reduced Risk	Personalized Residual Risk: 1 in 197,000
Combined SAP Deficiency	PSAP	AR	Reduced Risk	Personalized Residual Risk: 1 in 78,000
Cone-Rod Dystrophy 6 / Leber Congenital Amaurosis 1	GUCY2D	AR	Reduced Risk	Personalized Residual Risk: 1 in 920
Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency	CYP11B1	AR	Reduced Risk	Personalized Residual Risk: 1 in 510
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency	CYP17A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,000
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency	CYP21A2	AR	Reduced Risk	CYP21A2 copy number: 2 CYP21A2 sequencing: Negative <b>Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Non-Classic)): 1 in 300</b> <b>Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Classic)): 1 in 1,200</b>
Congenital Adrenal Hypoplasia (NR0B1-Related)	NR0B1	XL	Reduced Risk	Personalized Residual Risk: 1 in 353,000
Congenital Adrenal Insufficiency (CYP11A1-Related)	CYP11A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,100
Congenital Amegakaryocytic Thrombocytopenia	MPL	AR	Reduced Risk	Personalized Residual Risk: 1 in 62,000
Congenital Bile Acid Synthesis Defect (AKR1D1-Related)	AKR1D1	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Congenital Bile Acid Synthesis Defect (HSD3B7-Related)	HSD3B7	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Congenital Disorder of Deglycosylation	NGLY1	AR	Reduced Risk	Personalized Residual Risk: 1 in 101,000
Congenital Disorder of Glycosylation, Type Ia	PMM2	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,000

Congenital Disorder of Glycosylation, Type Ib	MPI	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Congenital Disorder of Glycosylation, Type Ic	ALG6	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Congenital Disorder of Glycosylation, Type Im	DOLK	AR	Reduced Risk	Personalized Residual Risk: 1 in 385,000
Congenital Dyserythropoietic Anemia Type 2	SEC23B	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,900
Congenital Dyserythropoietic Anemia, Type Ia	CDAN1	AR	Reduced Risk	Personalized Residual Risk: 1 in 900
Congenital Ichthyosis 4A and 4B	ABCA12	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,900
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Congenital Muscular Dystrophy (LAMA2-Related)	LAMA2	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Congenital Myasthenic Syndrome (CHAT-Related)	CHAT	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,900
Congenital Myasthenic Syndrome (CHRNE-Related)	CHRNE	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,700
Congenital Myasthenic Syndrome (DOK7-Related)	DOK7	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,400
Congenital Myasthenic Syndrome (RAPSN-Related)	RAPSN	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Congenital Neutropenia (HAX1-Related)	HAX1	AR	Reduced Risk	Personalized Residual Risk: 1 in 513,000
Congenital Neutropenia (VPS45-Related)	VPS45	AR	Reduced Risk	Personalized Residual Risk: 1 in 170,000
Congenital Nongoitrous Hypothyroidism 1	TSHR	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Congenital Nongoitrous Hypothyroidism 4	TSHB	AR	Reduced Risk	Personalized Residual Risk: 1 in 227,000
Congenital Secretory Chloride Diarrhea 1	SLC26A3	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Corneal Dystrophy and Perceptive Deafness	SLC4A11	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,500
Corticosterone Methyloxidase Deficiency	CYP11B2	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,200
Cystic Fibrosis	CFTR	AR	Reduced Risk	Personalized Residual Risk: 1 in 800
Cystinosis	CTNS	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,900
Cystinuria (SLC3A1-Related)	SLC3A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Cytochrome C Oxidase Deficiency / Leigh Syndrome (COX15-Related)	COX15	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
D-Bifunctional Protein Deficiency	HSD17B4	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,200
Deafness, Autosomal Recessive 3	MYO15A	AR	Reduced Risk	Personalized Residual Risk: 1 in 240
Deafness, Autosomal Recessive 59	PJVK	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,700
Deafness, Autosomal Recessive 7	TMC1	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,700
Deafness, Autosomal Recessive 76	SYNE4	AR	Reduced Risk	Personalized Residual Risk: 1 in 76,000
Deafness, Autosomal Recessive 77	LOXHD1	AR	Reduced Risk	Personalized Residual Risk: 1 in 35,000
Deafness, Autosomal Recessive 8/10	TMPRSS3	AR	Reduced Risk	Personalized Residual Risk: 1 in 360
Deafness, Autosomal Recessive 9	OTOF	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,100
Desbuquois Dysplasia 1	CANT1	AR	Reduced Risk	Personalized Residual Risk: 1 in 163,000
Desmosterolosis	DHCR24	AR	Reduced Risk	Personalized Residual Risk: 1 in 28,000
Diaphanospondylodysostosis	BMPER	AR	Reduced Risk	Personalized Residual Risk: 1 in 385,000
Distal Renal Tubular Acidosis and other SLC4A1-related Disorders	SLC4A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,700
Duchenne Muscular Dystrophy / Becker Muscular Dystrophy	DMD	XL	Reduced Risk	Personalized Residual Risk: 1 in 10,000
Dyskeratosis Congenita (DKC1-related)	DKC1	XL	Reduced Risk	Personalized Residual Risk: 1 in 9,259,000
Dyskeratosis Congenita (RTEL1-Related)	RTEL1	AR	Reduced Risk	Personalized Residual Risk: 1 in 73,000
Dystrophic Epidermolysis Bullosa	COL7A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 980
Ehlers-Danlos Syndrome, Type VI	PLOD1	AR	Reduced Risk	Personalized Residual Risk: 1 in 269,000
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 380,000
Ellis-Van Creveld Syndrome (EVC2-Related)	EVC2	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,700
Ellis-van Creveld Syndrome (EVC-Related)	EVC	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,500
Emery-Dreifuss Myopathy 1	EMD	XL	Reduced Risk	Personalized Residual Risk: 1 in 833,000
Enhanced S-Cone Syndrome	NR2E3	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,100
Ethylmalonic Encephalopathy	ETHE1	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000



Fabry Disease	GLA	XL	Reduced Risk	Personalized Residual Risk: 1 in 7,700
Factor IX Deficiency	F9	XL	Reduced Risk	Personalized Residual Risk: 1 in 5,100
Factor VII Deficiency	F7	AR	Reduced Risk	Personalized Residual Risk: 1 in 290
Factor XI Deficiency	F11	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Familial Autosomal Recessive Hypercholesterolemia	LDLRAP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000
Familial Dysautonomia	IKBKAP	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Familial Hypercholesterolemia	LDLR	AR	Reduced Risk	Personalized Residual Risk: 1 in 270
Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADH	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,000
Familial Hyperinsulinism (ABCC8-Related)	ABCC8	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Familial Hyperinsulinism (KCNJ11-Related)	KCNJ11	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Familial Hyperphosphatemic Tumoral Calcinosis	GALNT3	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Familial Mediterranean Fever	MEFV	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Fanconi Anemia, Group A	FANCA	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Fanconi Anemia, Group C	FANCC	AR	Reduced Risk	Personalized Residual Risk: 1 in 102,000
Fanconi Anemia, Group G	FANCG	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
Fanconi-Bickel Syndrome	SLC2A2	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Fragile X Syndrome	FMR1	XL	Reduced Risk	<p>FMR1 CGG repeat sizes: Not Performed</p> <p>FMR1 Sequencing: Negative</p> <p>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.</p> <p>Personalized Residual Risk: 1 in 17,000</p>
Fructose-1,6-Bisphosphatase Deficiency	FBP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,800
Fucosidosis	FUCA1	AR	Reduced Risk	Personalized Residual Risk: 1 in 48,000
Fumarase Deficiency	FH	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,900
Fundus Albipunctatus	RDH5	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
GRACILE Syndrome and Other BCS1L-Related Disorders	BCS1L	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,800
Galactokinase Deficiency	GALK1	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,700
Galactose Epimerase Deficiency	GALE	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Galactosemia	GALT	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,500
Galactosialidosis	CTSA	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Gaucher Disease	GBA	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Generalized Thyrotropin-Releasing Hormone Resistance	TRHR	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000
Geroderma Osteodysplasticum	GORAB	AR	Reduced Risk	Personalized Residual Risk: 1 in 308,000
Gitelman Syndrome	SLC12A3	AR	Reduced Risk	Personalized Residual Risk: 1 in 460
Glanzmann Thrombasthenia (ITGA2B-Related)	ITGA2B	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Glanzmann Thrombasthenia (ITGB3-Related)	ITGB3	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,900
Glutaric Acidemia, Type I	GCDH	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,400
Glutaric Acidemia, Type IIa	ETFA	AR	Reduced Risk	Personalized Residual Risk: 1 in 37,000
Glutaric Acidemia, Type IIb	ETFB	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,200
Glutaric Acidemia, Type IIc	ETFDH	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,400
Glutathione Synthetase Deficiency	GSS	AR	Reduced Risk	Personalized Residual Risk: 1 in 73,000
Glycine Encephalopathy (AMT-Related)	AMT	AR	Reduced Risk	Personalized Residual Risk: 1 in 90,000
Glycine Encephalopathy (GLDC-Related)	GLDC	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Glycogen Storage Disease, Type 0	GYS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,100
Glycogen Storage Disease, Type II	GAA	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,500
Glycogen Storage Disease, Type III	AGL	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease	GBE1	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,600
Glycogen Storage Disease, Type IXb	PHKB	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,400



Glycogen Storage Disease, Type Ia	G6PC	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Glycogen Storage Disease, Type Ib	SLC37A4	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Glycogen Storage Disease, Type V	PYGM	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,600
Glycogen Storage Disease, Type VI	PYGL	AR	Reduced Risk	Personalized Residual Risk: 1 in 820
Glycogen Storage Disease, Type VII	PFKM	AR	Reduced Risk	Personalized Residual Risk: 1 in 308,000
Gray Platelet Syndrome	NBEAL2	AR	Reduced Risk	Personalized Residual Risk: 1 in 191,000
Growth Hormone Deficiency, Type IB	GHRHR	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,800
HMG-CoA Lyase Deficiency	HMGCL	AR	Reduced Risk	Personalized Residual Risk: 1 in 64,000
Hemochromatosis, Type 2A	HFE2	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,800
Hemochromatosis, Type 3	TFR2	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Hereditary Fructose Intolerance	ALDOB	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,700
Hereditary Spastic Paraparesis 49	TECPR2	AR	Reduced Risk	Personalized Residual Risk: 1 in 112,000
Hermansky-Pudlak Syndrome, Type 1	HPS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 154,000
Hermansky-Pudlak Syndrome, Type 3	HPS3	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,800
Hermansky-Pudlak Syndrome, Type 4	HPS4	AR	Reduced Risk	Personalized Residual Risk: 1 in 368,000
Hermansky-Pudlak Syndrome, Type 6	HPS6	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,400
Hmg-CoA Synthase 2 Deficiency	HMGCS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,500
Holocarboxylase Synthetase Deficiency	HLCS	AR	Reduced Risk	Personalized Residual Risk: 1 in 110,000
Homocystinuria (CBS-Related)	CBS	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Homocystinuria due to MTHFR Deficiency	MTHFR	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,700
Homocystinuria, cblE Type	MTRR	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Homocystinuria-Megaloblastic Anemia, Cobalamin G Type	MTR	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,700
Hydrocephalus	L1CAM	XL	Reduced Risk	Personalized Residual Risk: 1 in 40,000
Hydroletharus Syndrome	HYLS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 220,000
Hyper-Igm Syndrome	CD40LG	XL	Reduced Risk	Personalized Residual Risk: 1 in 1167,000
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	SARS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 136,000
Hypohidrotic Ectodermal Dysplasia 1	EDA	XL	Reduced Risk	Personalized Residual Risk: 1 in 22,000
Hypomagnesemia 1	TRPM6	AR	Reduced Risk	Personalized Residual Risk: 1 in 64,000
Hypomyelinating Leukodystrophy 3	AIMP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 308,000
Hypomyelinating Leukodystrophy 12	VPS11	AR	Reduced Risk	Personalized Residual Risk: 1 in 94,000
Hypoparathyroidism-Retardation-Dysmorphic Syndrome	TBCE	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Hypophosphatasia	ALPL	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,500
Hypophosphatemic Rickets with Hypercalciuria	SLC34A3	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,300
Hypotrichosis 8 / Autosomal Recessive Woolly Hair 1	LPAR6	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,500
Immunodeficiency 18	CD3E	AR	Reduced Risk	Personalized Residual Risk: 1 in 120,000
Immunodeficiency 19	CD3D	AR	Reduced Risk	Personalized Residual Risk: 1 in 49,000
Inclusion Body Myopathy 2	GNE	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,600
Infantile Cerebral and Cerebellar Atrophy	MED17	AR	Reduced Risk	Personalized Residual Risk: 1 in 308,000
Infantile Neuroaxonal Dystrophy 1 and other PLA2G6-Related Disorders	PLA2G6	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,400
Intellectual Disability, Autosomal Recessive 3	CC2D1A	AR	Reduced Risk	Personalized Residual Risk: 1 in 150,000
Intrahepatic Cholestasis	ATP8B1	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Isovaleric Acidemia	IVD	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,000
Joubert Syndrome 2	TMEM216	AR	Reduced Risk	Personalized Residual Risk: 1 in 353,000
Joubert Syndrome 4 / Senior-Loken Syndrome 1 / Juvenile Nephronophthisis 1	NPHP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,800
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome	RPGRIP1L	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,800

Junctional Epidermolysis Bullosa ( <i>COL17A1</i> -Related)	<i>COL17A1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Junctional Epidermolysis Bullosa ( <i>ITGA6</i> -Related)	<i>ITGA6</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 165,000
Junctional Epidermolysis Bullosa ( <i>ITGB4</i> -Related)	<i>ITGB4</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Junctional Epidermolysis Bullosa ( <i>LAMA3</i> -Related)	<i>LAMA3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 81,000
Junctional Epidermolysis Bullosa ( <i>LAMB3</i> -Related)	<i>LAMB3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 63,000
Junctional Epidermolysis Bullosa ( <i>LAMC2</i> -Related)	<i>LAMC2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 171,000
Kohlschutter-Tonz Syndrome	<i>ROGDI</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 147,000
Krabbe Disease	<i>GALC</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 390
Lamellar Ichthyosis, Type 1	<i>TGM1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,600
Laron Dwarfism	<i>GHR</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,800
Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies	<i>CEP290</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,800
Leber Congenital Amaurosis 13	<i>RDH12</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Leber Congenital Amaurosis 15 / Retinitis Pigmentosa 14	<i>TULP1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,500
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20	<i>RPE65</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 490
Leber Congenital Amaurosis 4	<i>AIPL1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,500
Leber Congenital Amaurosis 5	<i>LCA5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,800
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy	<i>CRB1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Leigh Syndrome ( <i>NDUFS7</i> -Related)	<i>NDUFS7</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 38,000
Leigh Syndrome ( <i>SURF1</i> -Related)	<i>SURF1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,000
Leigh Syndrome, French-Canadian Type	<i>LRPPRC</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 76,000
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease	<i>GLE1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Lethal Congenital Contracture Syndrome 2	<i>ERBB3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 118,000
Lethal Congenital Contracture Syndrome 3	<i>PIP5K1C</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 151,000
Leukoencephalopathy with Vanishing White Matter	<i>EIF2B5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,600
Limb-Girdle Muscular Dystrophy, Type 2A	<i>CAPN3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Limb-Girdle Muscular Dystrophy, Type 2B	<i>DYSF</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 930
Limb-Girdle Muscular Dystrophy, Type 2C	<i>SGCG</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 37,000
Limb-Girdle Muscular Dystrophy, Type 2D	<i>SGCA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Limb-Girdle Muscular Dystrophy, Type 2E	<i>SGCB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 19,000
Limb-Girdle Muscular Dystrophy, Type 2F	<i>SGCD</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 36,000
Limb-Girdle Muscular Dystrophy, Type 2H	<i>TRIM32</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 308,000
Limb-Girdle Muscular Dystrophy, Type 2I	<i>FKRP</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,000
Limb-Girdle Muscular Dystrophy, Type 2L	<i>ANO5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,400
Lipoamide Dehydrogenase Deficiency	<i>DLD</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 118,000
Lipoid Adrenal Hyperplasia	<i>STAR</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,400
Lipoprotein Lipase Deficiency	<i>LPL</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 900
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	<i>HADHA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 73,000
Lowe Syndrome	<i>OCRL</i>	XL	Reduced Risk	Personalized Residual Risk: 1 in 1,375,000
Lysinuric Protein Intolerance	<i>SLC7A7</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
MEDNIK Syndrome	<i>AP1S1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 294,000
Malonyl-CoA Decarboxylase Deficiency	<i>MLYCD</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,400
Maple Syrup Urine Disease, Type 1a	<i>BCKDHA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 53,000

Maple Syrup Urine Disease, Type 1b	<i>BCKDHB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,400
Maple Syrup Urine Disease, Type 2	<i>DBT</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Medium Chain Acyl-CoA Dehydrogenase Deficiency	<i>ACADM</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 550
Megalencephalic Leukoencephalopathy with Subcortical Cysts	<i>MLC1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
Megaloblastic Anemia 1	<i>AMN</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 29,000
Menkes Disease	<i>ATP7A</i>	XL	Reduced Risk	Personalized Residual Risk: 1 in 172,000
Metachromatic Leukodystrophy	<i>ARSA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,100
Methionine Adenosyltransferase I/III Deficiency	<i>MAT3A</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Methylmalonic Acidemia (MMAA-Related)	<i>MMAA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Methylmalonic Acidemia (MMAB-Related)	<i>MMAB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Methylmalonic Acidemia (MUT-Related)	<i>MUT</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type	<i>MMACHC</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,800
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type	<i>MMADHC</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type	<i>LMBRD1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 44,000
Methylmalonyl-CoA Epimerase Deficiency	<i>MCEE</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 168,000
Microphthalmia / Anophthalmia	<i>VSX2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 396,000
Mitochondrial Complex I Deficiency (ACAD9-Related)	<i>ACAD9</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,500
Mitochondrial Complex I Deficiency (NDUFA11-Related)	<i>NDUFA11</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 244,000
Mitochondrial Complex I Deficiency (NDUFAF5-Related)	<i>NDUFAF5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 118,000
Mitochondrial Complex I Deficiency (NDUFS6-Related)	<i>NDUFS6</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 216,000
Mitochondrial Complex I Deficiency (NDUFV1-Related)	<i>NDUFV1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 600
Mitochondrial Complex I Deficiency / Leigh Syndrome (FOXRED1-Related)	<i>FOXRED1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,700
Mitochondrial Complex I Deficiency / Leigh Syndrome (NDUFAF2-Related)	<i>NDUFAF2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 161,000
Mitochondrial Complex I Deficiency / Leigh Syndrome (NDUFS4-Related)	<i>NDUFS4</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Mitochondrial Complex IV Deficiency (COX20-related)	<i>COX20</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 78,000
Mitochondrial Complex IV Deficiency (COX6B1-related)	<i>COX6B1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,231,000
Mitochondrial Complex IV Deficiency (APOPT1-Related)	<i>APOPT1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,200
Mitochondrial Complex IV Deficiency (PET100-Related)	<i>PET100</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 284,000
Mitochondrial Complex IV Deficiency (SCO1-related)	<i>SCO1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 96,000
Mitochondrial Complex IV Deficiency / Leigh Syndrome (COX10-Related)	<i>COX10</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,800
Mitochondrial DNA Depletion Syndrome 2	<i>TK2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 140,000
Mitochondrial DNA Depletion Syndrome 3	<i>DGUOK</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000
Mitochondrial DNA Depletion Syndrome 4A and 4B and other POLG-Related Disorders	<i>POLG</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 290
Mitochondrial DNA Depletion Syndrome 5	<i>SUCLA2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy	<i>MPV17</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Mitochondrial Myopathy and Sideroblastic Anemia 1	<i>PUS1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 513,000
Mitochondrial Trifunctional Protein Deficiency (HADHB-Related)	<i>HADHB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 10,000
Molybdenum Cofactor Deficiency A	<i>MOCS1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,200

Mucopolipidosis II / IIIA	GNPTAB	AR	Reduced Risk	Personalized Residual Risk: 1 in 10,000
Mucopolipidosis III Gamma	GNPTG	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,400
Mucopolipidosis IV	MCOLN1	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Mucopolysaccharidosis Type I	IDUA	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,300
Mucopolysaccharidosis Type II	IDS	XL	Reduced Risk	Personalized Residual Risk: 1 in 76,000
Mucopolysaccharidosis Type IIIA	SGSH	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Mucopolysaccharidosis Type IIIB	NAGLU	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Mucopolysaccharidosis Type IIIC	HGSNAT	AR	Reduced Risk	Personalized Residual Risk: 1 in 74,000
Mucopolysaccharidosis Type IIID	GNS	AR	Reduced Risk	Personalized Residual Risk: 1 in 201,000
Mucopolysaccharidosis Type IVa	GALNS	AR	Reduced Risk	Personalized Residual Risk: 1 in 530
Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis	GLB1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Mucopolysaccharidosis VII	GUSB	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,300
Mucopolysaccharidosis type IX	HYAL1	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
Mucopolysaccharidosis type VI	ARSB	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Mulibrey Nanism	TRIM37	AR	Reduced Risk	Personalized Residual Risk: 1 in 385,000
Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1	PIGN	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Multiple Pterygium Syndrome	CHRNA3	AR	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Multiple Sulfatase Deficiency	SUMF1	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Muscle-Eye-Brain Disease and Other POMGNT1-Related Congenital Muscular Dystrophy-Dysglycanopathies	POMGNT1	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,300
Myoneurogastrointestinal Encephalopathy	TYMP	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Myotubular Myopathy 1	MTM1	XL	Reduced Risk	Personalized Residual Risk: 1 in 192,000
N-Acetylglutamate Synthase Deficiency	NAGS	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,300
Nemaline Myopathy 2	NEB	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,200
Nephrogenic Diabetes Insipidus, Type II	AQP2	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,600
Nephrogenic Diabetes insipidus (AVPR2-related) / Nephrogenic Syndrome of Inappropriate Antidiuresis	AVPR2	XL	Reduced Risk	Personalized Residual Risk: 1 in 471,000
Nephronophthisis 2	INVS	AR	Reduced Risk	Personalized Residual Risk: 1 in 51,000
Nephrotic Syndrome (NPHS1-Related) / Congenital Finnish Nephrosis	NPHS1	AR	Reduced Risk	Personalized Residual Risk: 1 in 620
Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome	NPHS2	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Neurodegeneration due to Cerebral Folate Transport Deficiency	FOLR1	AR	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Neurodevelopmental Disorder with Progressive Microcephaly, Spasticity, and Brain Anomalies	PLAA	AR	Reduced Risk	Personalized Residual Risk: 1 in 217,000
Neuronal Ceroid-Lipofuscinosis (CLN3-Related)	CLN3	AR	Reduced Risk	Personalized Residual Risk: 1 in 255,000
Neuronal Ceroid-Lipofuscinosis (CLN5-Related)	CLN5	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Neuronal Ceroid-Lipofuscinosis (CLN6-Related)	CLN6	AR	Reduced Risk	Personalized Residual Risk: 1 in 13,000
Neuronal Ceroid-Lipofuscinosis (CLN8-Related)	CLN8	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,500
Neuronal Ceroid-Lipofuscinosis (MFSD8-Related)	MFSD8	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,700
Neuronal Ceroid-Lipofuscinosis (PPT1-Related)	PPT1	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Neuronal Ceroid-Lipofuscinosis (TPP1-Related)	TPP1	AR	Reduced Risk	Personalized Residual Risk: 1 in 220,000
Niemann-Pick Disease (SMPD1-Related)	SMPD1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Niemann-Pick Disease, Type C (NPC1-Related)	NPC1	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,000
Niemann-Pick Disease, Type C (NPC2-Related)	NPC2	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Nijmegen Breakage Syndrome	NBN	AR	Reduced Risk	Personalized Residual Risk: 1 in 102,000
Non-Syndromic Hearing Loss (GJB2-Related)	GJB2	AR	Reduced Risk	Personalized Residual Risk: 1 in 970
Oculocutaneous Albinism, Type IA / IB	TYR	AR	Reduced Risk	Personalized Residual Risk: 1 in 580
Oculocutaneous Albinism, Type IV	SLC45A2	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000

Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome	<i>WNT10A</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,400
Omenn Syndrome ( <i>RAG2</i> -Related)	<i>RAG2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 96,000
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type	<i>DCLRE1C</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,900
Omenn Syndrome and other <i>RAG1</i> -Related Disorders	<i>RAG1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Ornithine Aminotransferase Deficiency	<i>OAT</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,700
Ornithine Transcarbamylase Deficiency	<i>OTC</i>	XL	Reduced Risk	Personalized Residual Risk: 1 in 103,000
Osteogenesis Imperfecta, Type XI	<i>FKBP10</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 168,000
Osteopetrosis 1	<i>TCIRG1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,500
Osteopetrosis 8	<i>SNX10</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 34,000
Otospondylomegapiphyseal Dysplasia / Deafness / Fibrochondrogenesis 2	<i>COL11A2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,700
Papillon-Lefevre Syndrome	<i>CTSC</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,200
Pendred Syndrome	<i>SLC26A4</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 430
Peroxisome Biogenesis Disorder 3A and 3B	<i>PEX12</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Peroxisome Biogenesis Disorder 7A and 7B	<i>PEX26</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 160,000
Phenylalanine Hydroxylase Deficiency	<i>PAH</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 640
Polycystic Kidney Disease, Autosomal Recessive	<i>PKHD1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Polyglandular Autoimmune Syndrome, Type 1	<i>AIRE</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,000
Pontocerebellar Hypoplasia, Type 1A	<i>VRK1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 47,000
Pontocerebellar Hypoplasia, Type 1B	<i>EXOSC3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Pontocerebellar Hypoplasia, Type 2A and Type 4	<i>TSEN54</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 91,000
Pontocerebellar Hypoplasia, Type 2E	<i>VPS53</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Pontocerebellar Hypoplasia, Type 6	<i>RARS2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 67,000
Primary Carnitine Deficiency	<i>SLC22A5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Primary Ciliary Dyskinesia ( <i>CCDC103</i> -Related)	<i>CCDC103</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 16,000
Primary Ciliary Dyskinesia ( <i>CCDC151</i> -Related)	<i>CCDC151</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 255,000
Primary Ciliary Dyskinesia ( <i>CCDC39</i> -Related)	<i>CCDC39</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 70,000
Primary Ciliary Dyskinesia ( <i>DNAH5</i> -Related)	<i>DNAH5</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,500
Primary Ciliary Dyskinesia ( <i>DNAI1</i> -Related)	<i>DNAI1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 118,000
Primary Ciliary Dyskinesia ( <i>DNAI2</i> -Related)	<i>DNAI2</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 67,000
Primary Ciliary Dyskinesia ( <i>RSPH9</i> -Related)	<i>RSPH9</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 171,000
Primary Coenzyme Q10 Deficiency 7	<i>COQ4</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,600
Primary Congenital Glaucoma 3A	<i>CYP1B1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 650
Primary Hyperoxaluria, Type 1	<i>AGXT</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,900
Primary Hyperoxaluria, Type 2	<i>GRHPR</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 11,000
Primary Hyperoxaluria, Type 3	<i>HOGA1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,000
Progressive Cerebello-Cerebral Atrophy	<i>SEPSECS</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Progressive Familial Intrahepatic Cholestasis, Type 2	<i>ABCB11</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,500
Progressive Myoclonic Epilepsy, Type 1B	<i>PRICKLE1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 136,000
Progressive Pseudorheumatoid Dysplasia	<i>WISP3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000
Prolidase Deficiency	<i>PEPD</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,000
Propionic Acidemia ( <i>PCCA</i> -Related)	<i>PCCA</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,300
Propionic Acidemia ( <i>PCCB</i> -Related)	<i>PCCB</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,400
Pulmonary Surfactant Dysfunction	<i>ABCA3</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,700
Pycnodysostosis	<i>CTSK</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 450
Pyridoxamine 5'-Phosphate Oxidase Deficiency	<i>PNPO</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,600
Pyridoxine-Dependent Epilepsy	<i>ALDH7A1</i>	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300



Pyruvate Carboxylase Deficiency	PC	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Pyruvate Dehydrogenase E1-Alpha Deficiency	PDHA1	XL	Reduced Risk	Personalized Residual Risk: 1 in 139,000
Pyruvate Dehydrogenase E1-Beta Deficiency	PDHB	AR	Reduced Risk	Personalized Residual Risk: 1 in 206,000
Renal Tubular Acidosis and Deafness	ATP6V1B1	AR	Reduced Risk	Personalized Residual Risk: 1 in 128,000
Retinitis Pigmentosa 25	EYS	AR	Reduced Risk	Personalized Residual Risk: 1 in 600
Retinitis Pigmentosa 26	CERKL	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Retinitis Pigmentosa 28	FAM161A	AR	Reduced Risk	Personalized Residual Risk: 1 in 79,000
Retinitis Pigmentosa 36	PRCD	AR	Reduced Risk	Personalized Residual Risk: 1 in 422,000
Retinitis Pigmentosa 59	DHDDS	AR	Reduced Risk	Personalized Residual Risk: 1 in 201,000
Retinitis Pigmentosa 64 / Bardet-Biedl Syndrome 21 / Cone-Rod Dystrophy 16	C8ORF37	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Rh Deficiency Syndrome	RHAG	AR	Reduced Risk	Personalized Residual Risk: 1 in 94,000
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	AR	Reduced Risk	Personalized Residual Risk: 1 in 228,000
Rhizomelic Chondrodysplasia Punctata, Type 3	AGPS	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,024,000
Roberts Syndrome	ESCO2	AR	Reduced Risk	Personalized Residual Risk: 1 in 138,000
Salla Disease	SLC17A5	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Salt and Pepper Developmental Regression Syndrome	ST3GAL5	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Sandhoff Disease	HEXB	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Schimke Immunoosseous Dysplasia	SMARCA1	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Seckel Syndrome 5 / Microcephaly 9	CEP152	AR	Reduced Risk	Personalized Residual Risk: 1 in 740
Segawa Syndrome	TH	AR	Reduced Risk	Personalized Residual Risk: 1 in 214,000
Sepiapterin Reductase Deficiency	SPR	AR	Reduced Risk	Personalized Residual Risk: 1 in 49,000
Severe Combined Immunodeficiency (IL7R-Related)	IL7R	AR	Reduced Risk	Personalized Residual Risk: 1 in 51,000
Severe Combined Immunodeficiency (JAK3-Related)	JAK3	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Severe Combined Immunodeficiency (PTPRC-Related)	PTPRC	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,500
Severe Congenital Neutropenia 4	G6PC3	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,200
Severe Neonatal Hyperparathyroidism	CASR	AR	Reduced Risk	Personalized Residual Risk: 1 in 5,000
Short Stature, Onychodysplasia, Facial Dysmorphism, and Hypotrichosis	POC1A	AR	Reduced Risk	Personalized Residual Risk: 1 in 256,000
Short-Chain Acyl-CoA Dehydrogenase Deficiency	ACADS	AR	Reduced Risk	Personalized Residual Risk: 1 in 700
Shwachman-Diamond Syndrome	SBDS	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,200
Sialidosis, Type I and Type II	NEU1	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,400
Sjogren-Larsson Syndrome	ALDH3A2	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Smith-Lemli-Opitz Syndrome	DHCR7	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,100
Spastic Paraplegia 15	ZFYVE26	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly	SLC1A4	AR	Reduced Risk	Personalized Residual Risk: 1 in 385,000
Spherocytosis, Type 5	EPB42	AR	Reduced Risk	Personalized Residual Risk: 1 in 27,000
Spinal Muscular Atrophy	SMN1	AR	Reduced Risk	SMN1 copy number: >=3 SMN2 copy number: 1 c.*3+80T>G: Negative SMN1 Sequencing: Negative Personalized Residual Risk: 1 in 757 As additional gene copies are present, the patient's residual risk is expected to be lower than displayed
Spinal Muscular Atrophy with Respiratory Distress 1 / Charcot-Marie-Tooth Disease, Type 2S	IGHMBP2	AR	Reduced Risk	Personalized Residual Risk: 1 in 880
Spinocerebellar Ataxia with Axonal Neuropathy 3	COA7	AR	Reduced Risk	Personalized Residual Risk: 1 in 127,000
Spondylocostal Dysostosis 1	DLL3	AR	Reduced Risk	Personalized Residual Risk: 1 in 143,000

Spondylometaepiphyseal Dysplasia (DDR2-Related)	DDR2	AR	Reduced Risk	Personalized Residual Risk: 1 in 220,000
Spondylothoracic Dysostosis	MESP2	AR	Reduced Risk	Personalized Residual Risk: 1 in 306,000
Steel Syndrome	COL27A1	AR	Reduced Risk	Personalized Residual Risk: 1 in 513,000
Stuve-Wiedemann Syndrome	LIFR	AR	Reduced Risk	Personalized Residual Risk: 1 in 51,000
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	AR	Reduced Risk	Personalized Residual Risk: 1 in 85,000
Tay-Sachs Disease	HEXA	AR	Reduced Risk	Personalized Residual Risk: 1 in 400
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	AR	Reduced Risk	Personalized Residual Risk: 1 in 192,000
Thyroid Dysmorphogenesis 1	SLC5A5	AR	Reduced Risk	Personalized Residual Risk: 1 in 63,000
Thyroid Dysmorphogenesis 2A	TPO	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,800
Thyroid Dysmorphogenesis 3	TG	AR	Reduced Risk	Personalized Residual Risk: 1 in 12,000
Thyroid Dysmorphogenesis 4	IYD	AR	Reduced Risk	Personalized Residual Risk: 1 in 3,800
Thyroid Dysmorphogenesis 5	DUOXA2	AR	Reduced Risk	Personalized Residual Risk: 1 in 28,000
Thyroid Dysmorphogenesis 6	DUOX2	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Trichohepatoenteric Syndrome 1	TTC37	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,100
Tyrosinemia, Type I	FAH	AR	Reduced Risk	Personalized Residual Risk: 1 in 59,000
Tyrosinemia, Type II	TAT	AR	Reduced Risk	Personalized Residual Risk: 1 in 6,300
Tyrosinemia, Type III	HPD	AR	Reduced Risk	Personalized Residual Risk: 1 in 103,000
Usher Syndrome, Type IB	MYO7A	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,200
Usher Syndrome, Type IC	USH1C	AR	Reduced Risk	Personalized Residual Risk: 1 in 930
Usher Syndrome, Type ID	CDH23	AR	Reduced Risk	Personalized Residual Risk: 1 in 640
Usher Syndrome, Type IF	PCDH15	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,600
Usher Syndrome, Type IIA	USH2A	AR	Reduced Risk	Personalized Residual Risk: 1 in 190
Usher Syndrome, Type III	CLRN1	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,400
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,300
Vitamin D-Dependent Rickets, Type I	CYP27B1	AR	Reduced Risk	Personalized Residual Risk: 1 in 14,000
Vitamin D-Resistant Rickets, Type IIA	VDR	AR	Reduced Risk	Personalized Residual Risk: 1 in 15,000
Walker-Warburg Syndrome and Other FKTN-Related Dystrophies	FKTN	AR	Reduced Risk	Personalized Residual Risk: 1 in 8,300
Werner Syndrome	WRN	AR	Reduced Risk	Personalized Residual Risk: 1 in 1,500
Wilson Disease	ATP7B	AR	Reduced Risk	Personalized Residual Risk: 1 in 200
Wiskott-Aldrich Syndrome (WAS-Related)	WAS	XL	Reduced Risk	Personalized Residual Risk: 1 in 1,203,000
Wolcott-Rallison Syndrome	EIF2AK3	AR	Reduced Risk	Personalized Residual Risk: 1 in 385,000
Wolman Disease / Cholesteryl Ester Storage Disease	LIPA	AR	Reduced Risk	Personalized Residual Risk: 1 in 38,000
Woodhouse-Sakati Syndrome	DCAF17	AR	Reduced Risk	Personalized Residual Risk: 1 in 249,000
X-Linked Juvenile Retinoschisis	RS1	XL	Reduced Risk	Personalized Residual Risk: 1 in 40,000
X-Linked Severe Combined Immunodeficiency	IL2RG	XL	Reduced Risk	Personalized Residual Risk: 1 in 250,000
Xeroderma Pigmentosum (POLH-Related)	POLH	AR	Reduced Risk	Personalized Residual Risk: 1 in 24,000
Xeroderma Pigmentosum, Group A	XPA	AR	Reduced Risk	Personalized Residual Risk: 1 in 65,000
Xeroderma Pigmentosum, Group C	XPC	AR	Reduced Risk	Personalized Residual Risk: 1 in 9,600
Xeroderma Pigmentosum, Group G	ERCC5	AR	Reduced Risk	Personalized Residual Risk: 1 in 4,700
Zellweger Syndrome Spectrum (PEX10-Related)	PEX10	AR	Reduced Risk	Personalized Residual Risk: 1 in 7,100
Zellweger Syndrome Spectrum (PEX1-Related)	PEX1	AR	Reduced Risk	Personalized Residual Risk: 1 in 10,000
Zellweger Syndrome Spectrum (PEX2-Related)	PEX2	AR	Reduced Risk	Personalized Residual Risk: 1 in 96,000
Zellweger Syndrome Spectrum (PEX6-Related)	PEX6	AR	Reduced Risk	Personalized Residual Risk: 1 in 2,100

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. AmpliX<sup>®</sup> *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* CGG repeats in the premutation and full mutation size range were further analyzed by Southern blot analysis to assess the size and methylation status of the *FMR1* CGG repeat.

### 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).

The presence of the c.\*3+80T>G (chr5:70,247,901T>G) variant allele in an individual with Ashkenazi Jewish or Asian ancestry is typically indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, c.\*3+80T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic 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<sup>™</sup>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 9000 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 chr19:363,760-363,761 (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\_020734.6.2) 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 considered to be of uncertain significance and 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.

#### 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. For *CYP21A2*, a certain percentage of healthy individuals carry a duplication of the *CYP21A2* gene, which has no clinical consequences. In cases where two copies of a gene are located on the same chromosome in tandem, only the second 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. 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.

#### 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.

Please note these tests were developed and their performance characteristics were determined by Sema4 Opco, Inc. They have not been cleared or approved by the FDA. 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.

### 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>DONOR, 6398</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Phone: NG Patient ID: [REDACTED]	Specimen: [REDACTED] Requisition: [REDACTED] Lab Ref #: [REDACTED]  Collected: 05/05/2022 Received: 05/06/2022 / 21:53 EDT Reported: 05/17/2022 / 00:55 EDT	Client #: [REDACTED] NYNJMAIL GENOMICS, SEMA4 SEMA4 62 SOUTHFIELD AVE STAMFORD, CT 06902-7229

Ward: FFAXCB

## Cytogenetic Report

### CHROMOSOME ANALYSIS, BLOOD - 14596

Lab:EZ

### CHROMOSOME ANALYSIS, BLOOD

Order ID: [REDACTED]  
Specimen Type: Blood  
Clinical Indication: RULE OUT CHROMOSOME ABNORMALITY

**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:

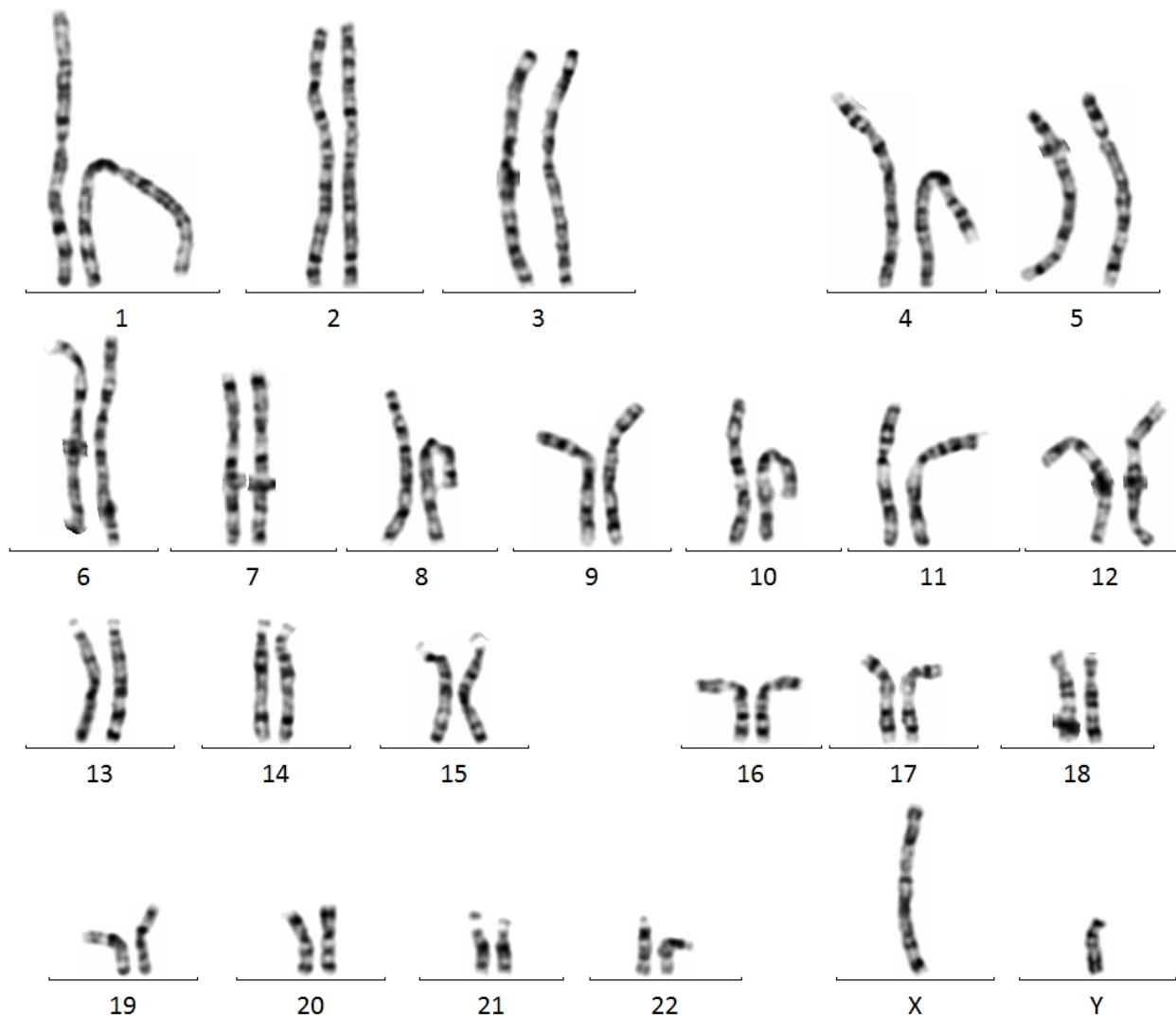
Method: G-Band (Digital Analysis: MetaSyst)  
Cells Counted: 20  
Band Level: 550  
Cells Analyzed: 5  
Cells Karyotyped: 3

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.

Reha M. Toydemir, MD, PhD, FACMG

Electronic Signature: 5/16/2022 11:56 PM

Patient Information	Specimen Information	Client Information
<b>DONOR, 6398</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Patient ID: [REDACTED]	Specimen: [REDACTED] Collected: [REDACTED] Received: 05/06/2022 / 21:53 EDT Reported: 05/17/2022 / 00:55 EDT	Client #: [REDACTED] GENOMICS, SEMA4



**PERFORMING SITE:**

EZ QUEST DIAGNOSTICS/NICHOLS SJC, 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>DONOR, 6398</b>  <b>DOB:</b> [REDACTED] <b>AGE:</b> [REDACTED] Gender: M Phone: NG Patient ID: [REDACTED]	Specimen: [REDACTED] Requisition: [REDACTED] Lab Ref #: [REDACTED]  Collected: 05/05/2022 Received: 05/06/2022 / 21:15 EDT Reported: 05/09/2022 / 09:21 EDT	Client #: 48041578      NYNJMAIL GENOMICS, SEMA4 SEMA4 62 SOUTHFIELD AVE STAMFORD, CT 06902-7229

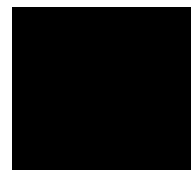
Ward: FFXCB

Test Name	In Range	Out Of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
RED BLOOD CELL COUNT	5.19		4.20-5.80 Million/uL	Z99
HEMOGLOBIN	14.7		13.2-17.1 g/dL	
HEMATOCRIT	44.2		38.5-50.0 %	
MCV	85.2		80.0-100.0 fL	
MCH	28.3		27.0-33.0 pg	
RDW	13.1		11.0-15.0 %	
HEMOGLOBIN A	97.6		>96.0 %	Z99
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.4		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





Patient Information:

**6398, Donor**

**DOB:** [REDACTED]

**Sex:** M

**MR#:** 6398

**Patient#:** [REDACTED]

Accession:

**Test#:** [REDACTED]

**Order#:** [REDACTED]

**Ext Test#:** [REDACTED]

**Ext Order#:** [REDACTED]

**Specimen Type:** DNA

**Collected:** Not provided

**Received Date:** Dec 08,2023

**Authorized Date:** Dec 13,2023

Physician:

**Seitz, Suzanne**

**ATTN:** Seitz, Suzanne

**Fairfax Cryobank**

**3015 Williams Drive**

**Fairfax, VA 22031**

**Phone:**

**Fax:**

Laboratory:

**Fulgent Genetics**

**CAP#:** 8042697

**CLIA#:** 05D2043189

**Laboratory Director:**

**Dr. Hanlin (Harry) Gao**

**Report Date:** Dec 18,2023

Final Report

TEST PERFORMED

**PRF1 Single Gene**

(1 Gene Panel: *PRF1*; gene sequencing with deletion and duplication analysis)

RESULTS:

**No clinically significant sequence or copy-number variants were identified in the submitted specimen.**

**A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations of the sort not queried by this test or in areas not reliably assessed by this test.**

INTERPRETATION:

Notes and Recommendations:

- As requested, this report only includes variants classified as Pathogenic, Likely Pathogenic, or Risk Allele at the time of analysis. If detected, this report does not include variants classified as of uncertain significance.
- Gene specific notes and limitations may be present. See below.
- These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)
- Guide to Interpreting Genomic Reports: A Genomics Toolkit (CSER Consortium; February 2017) (<https://www.genome.gov/For-Health-Professionals/Provider-Genomics-Education-Resources#hep>)

GENES TESTED:

**PRF1 Single Gene**

1 genes tested (100.00% at >20x).

*PRF1*

Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

METHODS:

**Patient:** 6398, Donor; **Sex:** M;  
**DOB:** [REDACTED] **MR#:** 6398

**Accession#:** FT [REDACTED] **FD Patient#:** [REDACTED]  
**DocID:** FT [REDACTED]



Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications identified by NGS are confirmed by an orthogonal method (qPCR or MLPA), unless exceeding an internally specified and validated quality score, beyond which deletions and duplications are considered real without further confirmation. New York patients: diagnostic findings are confirmed by Sanger, MLPA, or qPCR; exception SNV variants in genes for which confirmation of NGS results has been performed  $\geq 10$  times may not be confirmed if identified with high quality by NGS. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

#### LIMITATIONS:

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to this individual's phenotype, and negative results do not rule out a genetic cause for the indication for testing. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is designed and validated for detection of germline variants only. It is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions (eg. trinucleotide or hexanucleotide repeat expansion). DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm for copy number variants, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which are two or more contiguous exons in size; single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

#### SIGNATURE:

A handwritten signature in black ink, appearing to read 'Yan Meng'.

**Yan Meng, Ph.D., CGMB, FACMG** on 12/18/2023 08:41 PM PST  
Electronically signed



#### DISCLAIMER:

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This test was developed and its performance characteristics determined by **Fulgent Genetics**. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or **[info@fulgentgenetics.com](mailto:info@fulgentgenetics.com)**. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.