![Fairfax Cryobank — The Trusted Choice for Donor Sperm]

Donor 7021

Genetic Testing Summary

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

Last Updated: 03/07/24

Donor Reported Ancestry: Chinese   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.	Carrier of 2 variants: Non-Syndromic Hearing Loss (GJB2-Related) Donor has normal hearing. Donor has 2 copies of same low penetrance variant (c.109G>A). -See report attached. Negative for other genes sequenced.	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.

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Patient Information

Name: Donor 7021

Date of Birth:

Sema4 ID:

Client ID:

Indication: Carrier Screening

Specimen Information

Specimen Type: Blood

Date Collected: 12/08/2022

Date Received: 12/09/2022

Final Report: 01/04/2023

Referring Provider

Fairfax Cryobank, Inc.

Expanded Carrier Screen Minus TSE (502 genes)
with Personalized Residual Risk

SUMMARY OF RESULTS AND RECOMMENDATIONS

⊕ **Positive**	⊖ **Negative**
May Exhibit Clinical Symptoms of Non-Syndromic Hearing Loss (*GJB2*-Related) (AR) Associated gene(s): *GJB2* Variant(s) Detected: c.109G>A, p.V37I, Pathogenic, Homozygous (two copies)	**Negative for all other genes tested** 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.
- Clinical correlation of the patient's *GJB2* genotype and phenotype is 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.
- As genetic technologies may improve and variant classifications may change over time, it is recommended to obtain a new carrier screening test or reanalysis when a new pregnancy is being considered.

Interpretation of positive results

Non-Syndromic Hearing Loss (*GJB2*-Related) (AR)

Results and Interpretation

A homozygous (two copies) missense variant, c.109G>A, p.V37I, was detected in the *GJB2* gene. This variant is considered to be pathogenic and when present in trans with a pathogenic variant causative for non-syndromic hearing loss (*GJB2*-related). Please note that this variant has been reported to have a variable penetrance, and some individuals with a pathogenic variant on the opposite allele may not have hearing loss. Therefore, this individual may exhibit clinical symptoms of non-syndromic hearing loss (*GJB2*-related). Clinical correlation and follow-up is strongly recommended.

What is Non-Syndromic Hearing Loss (*GJB2*-Related)?

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Non-syndromic hearing loss (*GJB2*-related) is an autosomal recessive disorder that is caused by pathogenic variants in the gene *GJB2* . It is found in individuals of many different ethnicities, but it more prevalent in individuals of Ashkenazi Jewish descent, as well as Caucasians and Asians. Patients with this form of hearing loss do not experience any other disease manifestations. Hearing loss is usually present from birth and does not progress in severity over time. The level of hearing loss can vary between patients from mild to profound. Patients with two inactivating variants are more likely to have profound hearing loss, whereas patients with two non-inactivating variants are more likely to have mild hearing loss. However, the variability that exists between patients means that it may not be possible to predict the severity of an individual's hearing loss based on their genotype. Life expectancy is not reduced.

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**. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

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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

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

Disease	Gene	Inheritance Pattern	Status	Detailed Summary
⊕ **Positive**				
Non-Syndromic Hearing Loss (*GJB2*-Related)	*GJB2*	AR	May Exhibit Clinical Symptoms	c.109G>A, p.V37I, Pathogenic, Homozygous (two copies)
⊖ **Negative**				
2-Methylbutyrylglycinuria	*ACADSB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 410
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	*HSD3B2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 181,000
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related)	*MCCC1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 930
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related)	*MCCC2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 500
3-Methylglutaconic Aciduria, Type III	*OPA3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 29,000
3-Phosphoglycerate Dehydrogenase Deficiency	*PHGDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 123,000
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	*PTS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
***CD59*-Mediated Hemolytic Anemia**	*CD59*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 513,000
***WNT10A*-Related Ectodermal Dysplasia**	*WNT10A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 900
Abetalipoproteinemia	*MTTP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,500
Achalasia-Addisonianism-Alacrimia Syndrome	*AAAS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Achromatopsia (*CNGA3*-Related)	*CNGA3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 320
Achromatopsia (*CNGB3*-related)	*CNGB3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 21,000
Acrodermatitis Enteropathica	*SLC39A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 62,000
Acute Infantile Liver Failure	*TRMU*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 55,000
Acyl-CoA Oxidase I Deficiency	*ACOX1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 59,000
Adams-Oliver Syndrome 4	*EOGT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 59,000
Adenosine Deaminase Deficiency	*ADA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 127,000
Adrenocorticotropic Hormone Deficiency	*TBX19*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,500
Adrenoleukodystrophy, X-Linked	*ABCD1*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 19,000
Agammaglobulinemia	*BTK*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 250,000
Agenesis of the Corpus Callosum	*FRMD4A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 348,000
Aicardi-Goutieres Syndrome (*RNASEH2C*-Related)	*RNASEH2C*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Aicardi-Goutieres Syndrome (*SAMHD1*-Related)	*SAMHD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700
Aicardi-Goutieres Syndrome (*TREX1*-Related)	*TREX1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Albinism, Oculocutaneous, Type III	*TYRP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 430
Alkaptonuria	*HGD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,200
Alpha-Mannosidosis	*MAN2B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,000
Alpha-Thalassemia	*HBA1/HBA2*	AR	Reduced Risk	*HBA1* Copy Number: 2 *HBA2* Copy Number: 2 No pathogenic copy number variants detected *HBA1/ HBA2* Sequencing: Negative **Personalized Residual Risk:** 1 in 380
Alpha-Thalassemia Intellectual Disability Syndrome	*ATRX*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 48,000

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Alport Syndrome (*COL4A3*-Related)	*COL4A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Alport Syndrome (*COL4A4*-Related)	*COL4A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 510
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 3,100
Andermann Syndrome	*SLC12A6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Antley-Bixler Syndrome (*POR*-Related)	*POR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 650
Argininemia	*ARG1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,900
Argininosuccinic Aciduria	*ASL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Aromatase Deficiency	*CYP19A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Arthrogryposis, Intellectual Disability, and Seizures	*SLC35A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 240,000
Asparagine Synthetase Deficiency	*ASNS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 178,000
Aspartylglycosaminuria	*AGA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Ataxia With Isolated Vitamin E Deficiency	*TTPA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 20,000
Ataxia-Telangiectasia	*ATM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 540
Ataxia-Telangiectasia-Like Disorder 1	*MRE11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,700
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	*SACS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Bardet-Biedl Syndrome (*ARL6*-Related)	*ARL6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Bardet-Biedl Syndrome (*BBS10*-Related)	*BBS10*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Bardet-Biedl Syndrome (*BBS12*-Related)	*BBS12*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Bardet-Biedl Syndrome (*BBS1*-Related)	*BBS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 10,000
Bardet-Biedl Syndrome (*BBS2*-Related)	*BBS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,400
Bardet-Biedl Syndrome (*BBS4*-Related)	*BBS4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Bare Lymphocyte Syndrome, Type II	*CIITA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 129,000
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 710
Bartter Syndrome, Type 4A	*BSND*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 69,000
Bernard-Soulier Syndrome, Type A1	*GP1BA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Bernard-Soulier Syndrome, Type C	*GP9*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Beta-Globin-Related Hemoglobinopathies	*HBB*	AR	Reduced Risk	**Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies):** 1 in 1,200 **Personalized Residual Risk (Beta-Globin-Related Hemoglobinopathies: HbS Variant):** 1 in 11,000 **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 3,800
Beta-Mannosidosis	*MANBA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 57,000
BH4-Deficient Hyperphenylalaninemia C	*QDPR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,000
BH4-Deficient Hyperphenylalaninemia D	*PCBD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Bilateral Frontoparietal Polymicrogyria	*GPR56*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 143,000
Biotinidase Deficiency	*BTD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Bloom Syndrome	*BLM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 34,000
Canavan Disease	*ASPA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,200
Carbamoylphosphate Synthetase I Deficiency	*CPS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 690
Carnitine Acylcarnitine Translocase Deficiency	*SLC25A20*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,500
Carnitine Palmitoyltransferase IA Deficiency	*CPT1A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 143,000
Carnitine Palmitoyltransferase II Deficiency	*CPT2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 930
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 450

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Disease	Gene	Inheritance	Risk	Result
Catecholaminergic Polymorphic Ventricular Tachycardia	CASQ2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
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 2,300
Cerebral Creatine Deficiency Syndrome 3	GATM	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Cerebral Dysgenesis, Neuropathy, Ichthyosis, and Palmoplantar Keratoderma Syndrome	SNAP29	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 383,000
Cerebrotendinous Xanthomatosis	CYP27A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 750
Charcot-Marie-Tooth Disease, Type 4D	NDRG1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 225,000
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 129,000
Chondrodysplasia Punctata	ARSE	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 862,000
Choreoacanthocytosis	VPS13A	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,700
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 3,700
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 2,200
Citrullinemia, Type 1	ASS1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 81,000
Cockayne Syndrome, Type A	ERCC8	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 32,000
Cockayne Syndrome, Type B and other *ERCC6*-Related Disorders	ERCC6	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,300
Cohen Syndrome	VPS13B	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 13,000
Combined Factor V and VIII Deficiency	LMAN1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 68,000
Combined Malonic and Methylmalonic Aciduria	ACSF3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 23,000
Combined Oxidative Phosphorylation Deficiency 1	GFM1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,100
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 1,900
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 121,000
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 720
Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency	CYP11B1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency	CYP17A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 840
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency	CYP21A2	AR	Reduced Risk	*CYP21A2* copy number: 2 *CYP21A2* sequencing: Negative **Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Non-Classic)):** 1 in 300 **Personalized Residual Risk (Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (Classic)):** 1 in 1,200
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 128,000
Congenital Amegakaryocytic Thrombocytopenia	MPL	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 68,000
Congenital Bile Acid Synthesis Defect (*AKR1D1*-Related)	AKR1D1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
Congenital Bile Acid Synthesis Defect (*HSD3B7*-Related)	HSD3B7	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,700

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Congenital Disorder of Deglycosylation	NGLY1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Congenital Disorder of Glycosylation, Type Ia	PMM2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 550
Congenital Disorder of Glycosylation, Type Ib	MPI	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Congenital Disorder of Glycosylation, Type Ic	ALG6	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Congenital Disorder of Glycosylation, Type Im	DOLK	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 216,000
Congenital Dyserythropoietic Anemia Type 2	SEC23B	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Congenital Dyserythropoietic Anemia, Type Ia	CDAN1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 640
Congenital Ichthyosis 4A and 4B	ABCA12	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Congenital Insensitivity to Pain with Anhidrosis	NTRK1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,000
Congenital Muscular Dystrophy (LAMA2-Related)	LAMA2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 690
Congenital Myasthenic Syndrome (CHAT-Related)	CHAT	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Congenital Myasthenic Syndrome (CHRNE-Related)	CHRNE	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 30,000
Congenital Myasthenic Syndrome (DOK7-Related)	DOK7	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 470
Congenital Myasthenic Syndrome (RAPSN-Related)	RAPSN	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 47,000
Congenital Neutropenia (HAX1-Related)	HAX1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 126,000
Congenital Neutropenia (VPS45-Related)	VPS45	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 110,000
Congenital Nongoitrous Hypothyroidism 1	TSHR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 230
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 40,000
Corneal Dystrophy and Perceptive Deafness	SLC4A11	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,200
Corticosterone Methyloxidase Deficiency	CYP11B2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Cystic Fibrosis	CFTR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Cystinosis	CTNS	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,100
Cystinuria (SLC3A1-Related)	SLC3A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 530
Cytochrome C Oxidase Deficiency / Leigh Syndrome (COX15-Related)	COX15	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 182,000
D-Bifunctional Protein Deficiency	HSD17B4	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700
Deafness, Autosomal Recessive 3	MYO15A	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 100
Deafness, Autosomal Recessive 59	PJVK	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 73,000
Deafness, Autosomal Recessive 7	TMC1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Deafness, Autosomal Recessive 76	SYNE4	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 121,000
Deafness, Autosomal Recessive 77	LOXHD1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
Deafness, Autosomal Recessive 8/10	TMPRSS3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 330
Deafness, Autosomal Recessive 9	OTOF	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 370
Desbuquois Dysplasia 1	CANT1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,800
Desmosterolosis	DHCR24	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 28,000
Diaphanospondylodysostosis	BMPER	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 144,000
Distal Renal Tubular Acidosis and other SLC4A1-related Disorders	SLC4A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 910
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 3,900
Dystrophic Epidermolysis Bullosa	COL7A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Ehlers-Danlos Syndrome, Type VI	PLOD1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,700
Ehlers-Danlos Syndrome, Type VIIC	ADAMTS2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
Ellis-Van Creveld Syndrome (EVC2-Related)	EVC2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,100
Ellis-van Creveld Syndrome (EVC-Related)	EVC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Emery-Dreifuss Myopathy 1	EMD	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 833,000

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Disease	Gene	Inheritance	Risk	Result
Enhanced S-Cone Syndrome	*NR2E3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Ethylmalonic Encephalopathy	*ETHE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
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 300
Factor XI Deficiency	*F11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 440
Familial Autosomal Recessive Hypercholesterolemia	*LDLRAP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 171,000
Familial Dysautonomia	*IKBKAP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 78,000
Familial Hypercholesterolemia	*LDLR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 260
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 240
Familial Hyperinsulinism (*KCNJ11*-Related)	*KCNJ11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Familial Hyperphosphatemic Tumoral Calcinosis	*GALNT3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Familial Mediterranean Fever	*MEFV*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,400
Fanconi Anemia, Group A	*FANCA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Fanconi Anemia, Group C	*FANCC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 34,000
Fanconi Anemia, Group G	*FANCG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Fanconi-Bickel Syndrome	*SLC2A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 295,000
Fragile X Syndrome	*FMR1*	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. **Personalized Residual Risk:** 1 in 222,000
Fructose-1,6-Bisphosphatase Deficiency	*FBP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 11,000
Fucosidosis	*FUCA1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 49,000
Fumarase Deficiency	*FH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,900
Fundus Albipunctatus	*RDH5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 810
Galactokinase Deficiency	*GALK1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Galactose Epimerase Deficiency	*GALE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 850
Galactosemia	*GALT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 390
Galactosialidosis	*CTSA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 11,000
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 296,000
Geroderma Osteodysplasticum	*GORAB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 76,000
Gitelman Syndrome	*SLC12A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 230
Glanzmann Thrombasthenia (*ITGA2B*-Related)	*ITGA2B*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Glanzmann Thrombasthenia (*ITGB3*-Related)	*ITGB3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Glutaric Acidemia, Type I	*GCDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 20,000
Glutaric Acidemia, Type IIa	*ETFA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Glutaric Acidemia, Type IIb	*ETFB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,800
Glutaric Acidemia, Type IIc	*ETFDH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 260
Glutathione Synthetase Deficiency	*GSS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 48,000
Glycine Encephalopathy (*AMT*-Related)	*AMT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 144,000
Glycine Encephalopathy (*GLDC*-Related)	*GLDC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 240
Glycogen Storage Disease, Type 0	*GYS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 29,000
Glycogen Storage Disease, Type Ia	*G6PC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 410
Glycogen Storage Disease, Type Ib	*SLC37A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,900
Glycogen Storage Disease, Type II	*GAA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 280
Glycogen Storage Disease, Type III	*AGL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 55,000

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Disease	Gene	Inheritance	Risk	Residual Risk
Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease	*GBE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 64,000
Glycogen Storage Disease, Type IXb	*PHKB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Glycogen Storage Disease, Type V	*PYGM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,400
Glycogen Storage Disease, Type VI	*PYGL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Glycogen Storage Disease, Type VII	*PFKM*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,900
GM3 Synthase Deficiency	*ST3GAL5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 108,000
GRACILE Syndrome and Other *BCS1L*-Related Disorders	*BCS1L*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 82,000
Gray Platelet Syndrome	*NBEAL2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,400
Growth Hormone Deficiency, Type IB	*GHRHR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 104,000
Hemochromatosis, Type 2A	*HFE2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 740
Hemochromatosis, Type 3	*TFR2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 275,000
Hereditary Fructose Intolerance	*ALDOB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 35,000
Hereditary Spastic Paraparesis 49	*TECPR2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 166,000
Hermansky-Pudlak Syndrome, Type 1	*HPS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 286,000
Hermansky-Pudlak Syndrome, Type 3	*HPS3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 22,000
Hermansky-Pudlak Syndrome, Type 4	*HPS4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Hermansky-Pudlak Syndrome, Type 6	*HPS6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 680
HMG-CoA Lyase Deficiency	*HMGCL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 113,000
Hmg-CoA Synthase 2 Deficiency	*HMGCS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,000
Holocarboxylase Synthetase Deficiency	*HLCS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,900
Homocystinuria (*CBS*-Related)	*CBS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,200
Homocystinuria due to *MTHFR* Deficiency	*MTHFR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,000
Homocystinuria, cblE Type	*MTRR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 16,000
Homocystinuria-Megaloblastic Anemia, Cobalamin G Type	*MTR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 35,000
Hydrocephalus	*L1CAM*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 40,000
Hydrolethalus Syndrome	*HYLS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 296,000
Hyper-Igm Syndrome	*CD40LG*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 1,167,000
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	*SLC25A15*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 30,000
Hyperuricemia, Pulmonary Hypertension, Renal Failure, and Alkalosis	*SARS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 220,000
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 86,000
Hypomyelinating Leukodystrophy 3	*AIMP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 273,000
Hypomyelinating Leukodystrophy 12	*VPS11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 94,000
Hypophosphatasia	*ALPL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,200
Hypophosphatemic Rickets with Hypercalciuria	*SLC34A3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,000
Hypotrichosis 8 / Autosomal Recessive Woolly Hair 1	*LPAR6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 17,000
Immunodeficiency 18	*CD3E*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 120,000
Immunodeficiency 19	*CD3D*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 69,000
Inclusion Body Myopathy 2	*GNE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,600
Infantile Cerebral and Cerebellar Atrophy	*MED17*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 130,000
Infantile Neuroaxonal Dystrophy 1 and other *PLA2G6*-Related Disorders	*PLA2G6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 380
Intellectual Disability, Autosomal Recessive 3	*CC2D1A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 108,000
Intrahepatic Cholestasis	*ATP8B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 580
Isovaleric Acidemia	*IVD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Joubert Syndrome 2	*TMEM216*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 133,000
Joubert Syndrome 4 / Senior-Loken Syndrome 1 / Juvenile Nephronophthisis 1	*NPHP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000

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Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome	RPGRIP1L	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Junctional Epidermolysis Bullosa (*COL17A1*-Related)	COL17A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Junctional Epidermolysis Bullosa (*ITGA6*-Related)	ITGA6	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Junctional Epidermolysis Bullosa (*ITGB4*-Related)	ITGB4	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 26,000
Junctional Epidermolysis Bullosa (*LAMA3*-Related)	LAMA3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 49,000
Junctional Epidermolysis Bullosa (*LAMB3*-Related)	LAMB3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,600
Junctional Epidermolysis Bullosa (*LAMC2*-Related)	LAMC2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 28,000
Kohlschutter-Tonz Syndrome	ROGDI	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Krabbe Disease	GALC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 340
Lamellar Ichthyosis, Type 1	TGM1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Laron Dwarfism	GHR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies	CEP290	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Leber Congenital Amaurosis 13	RDH12	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 88,000
Leber Congenital Amaurosis 15 / Retinitis Pigmentosa 14	TULP1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20	RPE65	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Leber Congenital Amaurosis 4	AIPL1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,100
Leber Congenital Amaurosis 5	LCA5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy	CRB1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 960
Leigh Syndrome (*NDUFS7*-Related)	NDUFS7	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 38,000
Leigh Syndrome (*SURF1*-Related)	SURF1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Leigh Syndrome, French-Canadian Type	LRPPRC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 22,000
Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease	GLE1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,900
Lethal Congenital Contracture Syndrome 2	ERBB3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 52,000
Lethal Congenital Contracture Syndrome 3	PIP5K1C	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 304,000
Leukoencephalopathy with Vanishing White Matter	EIF2B5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,200
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,200
Limb-Girdle Muscular Dystrophy, Type 2B	DYSF	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,000
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,400
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 72,000
Limb-Girdle Muscular Dystrophy, Type 2F	SGCD	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 36,000
Limb-Girdle Muscular Dystrophy, Type 2H	TRIM32	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 123,000
Limb-Girdle Muscular Dystrophy, Type 2I	FKRP	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 460
Limb-Girdle Muscular Dystrophy, Type 2L	ANO5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 920
Lipoamide Dehydrogenase Deficiency	DLD	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 225,000
Lipoid Adrenal Hyperplasia	STAR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 36,000
Lipoprotein Lipase Deficiency	LPL	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 800
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,500
Lowe Syndrome	OCRL	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 1,375,000
Lysinuric Protein Intolerance	SLC7A7	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 72,000
Malonyl-CoA Decarboxylase Deficiency	MLYCD	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Maple Syrup Urine Disease, Type 1a	BCKDHA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000

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Disease	Gene	Inheritance	Risk	Personalized Residual Risk
Maple Syrup Urine Disease, Type 1b	BCKDHB	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Maple Syrup Urine Disease, Type 2	DBT	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 790
Meckel Syndrome 1 / Bardet-Biedl Syndrome 13	MKS1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 28,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	ACADM	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
MEDNIK Syndrome	AP1S1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 294,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 171,000
Megaloblastic Anemia 1	AMN	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 13,000
Menkes Disease	ATP7A	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Metachromatic Leukodystrophy	ARSA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,600
Methionine Adenosyltransferase I/III Deficiency	MAT1A	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Methylmalonic Acidemia (*MMAA*-Related)	MMAA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 216,000
Methylmalonic Acidemia (*MMAB*-Related)	MMAB	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Methylmalonic Acidemia (*MUT*-Related)	MUT	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 830
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type	MMACHC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type	MMADHC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type	LMBRD1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 43,000
Methylmalonyl-CoA Epimerase Deficiency	MCEE	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 168,000
Microphthalmia / Anophthalmia	VSX2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 83,000
Mitochondrial Complex I Deficiency (*ACAD9*-Related)	ACAD9	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,100
Mitochondrial Complex I Deficiency (*NDUFA11*-Related)	NDUFA11	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 548,000
Mitochondrial Complex I Deficiency (*NDUFAF5*-Related)	NDUFAF5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 770
Mitochondrial Complex I Deficiency (*NDUFS6*-Related)	NDUFS6	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 211,000
Mitochondrial Complex I Deficiency (*NDUFV1*-Related)	NDUFV1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,100
Mitochondrial Complex I Deficiency / Leigh Syndrome (*FOXRED1*-Related)	FOXRED1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,900
Mitochondrial Complex I Deficiency / Leigh Syndrome (*NDUFAF2*-Related)	NDUFAF2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 114,000
Mitochondrial Complex I Deficiency / Leigh Syndrome (*NDUFS4*-Related)	NDUFS4	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 31,000
Mitochondrial Complex IV Deficiency (*COX20*-related)	COX20	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 68,000
Mitochondrial Complex IV Deficiency (*COX6B1*-related)	COX6B1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,231,000
Mitochondrial Complex IV Deficiency (*APOPT1*-Related)	APOPT1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 22,000
Mitochondrial Complex IV Deficiency (*PET100*-Related)	PET100	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 546,000
Mitochondrial Complex IV Deficiency (*SCO1*-related)	SCO1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 74,000
Mitochondrial Complex IV Deficiency / Leigh Syndrome (*COX10*-Related)	COX10	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,900
Mitochondrial DNA Depletion Syndrome 2	TK2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,700
Mitochondrial DNA Depletion Syndrome 3	DGUOK	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,500
Mitochondrial DNA Depletion Syndrome 4A and 4B and other *POLG*-Related Disorders	POLG	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 180
Mitochondrial DNA Depletion Syndrome 5	SUCLA2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 152,000
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy	MPV17	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,400
Mitochondrial Myopathy and Sideroblastic Anemia 1	PUS1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 333,000

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Disease	Gene	Inheritance	Result	Risk
Mitochondrial Trifunctional Protein Deficiency (*HADHB*-Related)	*HADHB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Molybdenum Cofactor Deficiency A	*MOCS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 215,000
Mucolipidosis II / IIIA	*GNPTAB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Mucolipidosis III Gamma	*GNPTG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 213,000
Mucolipidosis IV	*MCOLN1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,500
Mucopolysaccharidosis Type I	*IDUA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 630
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 700
Mucopolysaccharidosis Type IIIB	*NAGLU*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 900
Mucopolysaccharidosis Type IIIC	*HGSNAT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 42,000
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 440
Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis	*GLB1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Mucopolysaccharidosis type IX	*HYAL1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
Mucopolysaccharidosis type VI	*ARSB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 144,000
Mucopolysaccharidosis VII	*GUSB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
Mulibrey Nanism	*TRIM37*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 36,000
Multiple Congenital Anomalies-Hypotonia-Seizures Syndrome 1	*PIGN*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 19,000
Multiple Pterygium Syndrome	*CHRNG*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Multiple Sulfatase Deficiency	*SUMF1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 144,000
Muscle-Eye-Brain Disease and Other *POMGNT1*-Related Congenital Muscular Dystrophy-Dystroglycanopathies	*POMGNT1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,100
Myoneurogastrointestinal Encephalopathy	*TYMP*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,400
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 5,700
Nemaline Myopathy 2	*NEB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 300
Nephrogenic Diabetes insipidus (*AVPR2*-related)/ Nephrogenic Syndrome of Inappropriate Antidiuresis	*AVPR2*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 471,000
Nephrogenic Diabetes Insipidus, Type II	*AQP2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,700
Nephronophthisis 2	*INVS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 24,000
Nephrotic Syndrome (*NPHS1*-Related) / Congenital Finnish Nephrosis	*NPHS1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 980
Nephrotic Syndrome (*NPHS2*-Related) / Steroid-Resistant Nephrotic Syndrome	*NPHS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,700
Neurodegeneration due to Cerebral Folate Transport Deficiency	*FOLR1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
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 59,000
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related)	*CLN5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 75,000
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related)	*CLN6*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 91,000
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related)	*CLN8*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,300
Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related)	*MFSD8*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 87,000
Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related)	*PPT1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related)	*TPP1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,000
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 2,600
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 214,000

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Oculocutaneous Albinism, Type IA / IB	*TYR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 220
Oculocutaneous Albinism, Type IV	*SLC45A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 980
Omenn Syndrome (*RAG2*-Related)	*RAG2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 32,000
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type	*DCLRE1C*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 48,000
Omenn Syndrome and other *RAG1*-Related Disorders	*RAG1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 180
Ornithine Aminotransferase Deficiency	*OAT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,900
Ornithine Transcarbamylase Deficiency	*OTC*	XL	Reduced Risk	**Personalized Residual Risk:** 1 in 103,000
Osteogenesis Imperfecta, Type XI	*FKBP10*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 8,100
Osteopetrosis 1	*TCIRG1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,700
Osteopetrosis 8	*SNX10*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 215,000
Otospondylomegaepiphyseal Dysplasia / Deafness / Fibrochondrogenesis 2	*COL11A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 5,800
Papillon-Lefevre Syndrome	*CTSC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,400
Pendred Syndrome	*SLC26A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 72
Peroxisome Biogenesis Disorder 3A and 3B	*PEX12*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 225,000
Peroxisome Biogenesis Disorder 7A and 7B	*PEX26*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 23,000
Phenylalanine Hydroxylase Deficiency	*PAH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 150
Polycystic Kidney Disease, Autosomal Recessive	*PKHD1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 350
Polyglandular Autoimmune Syndrome, Type 1	*AIRE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Pontocerebellar Hypoplasia, Type 1A	*VRK1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 215,000
Pontocerebellar Hypoplasia, Type 1B	*EXOSC3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 54,000
Pontocerebellar Hypoplasia, Type 2A and Type 4	*TSEN54*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,800
Pontocerebellar Hypoplasia, Type 2E	*VPS53*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 146,000
Pontocerebellar Hypoplasia, Type 6	*RARS2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 52,000
Primary Carnitine Deficiency	*SLC22A5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 600
Primary Ciliary Dyskinesia (*CCDC103*-Related)	*CCDC103*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 33,000
Primary Ciliary Dyskinesia (*CCDC151*-Related)	*CCDC151*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 215,000
Primary Ciliary Dyskinesia (*CCDC39*-Related)	*CCDC39*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 84,000
Primary Ciliary Dyskinesia (*DNAH5*-Related)	*DNAH5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 19,000
Primary Ciliary Dyskinesia (*DNAI1*-Related)	*DNAI1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 9,300
Primary Ciliary Dyskinesia (*DNAI2*-Related)	*DNAI2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 144,000
Primary Ciliary Dyskinesia (*RSPH9*-Related)	*RSPH9*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 73,000
Primary Coenzyme Q10 Deficiency 7	*COQ4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 31,000
Primary Congenital Glaucoma 3A	*CYP1B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 130
Primary Hyperoxaluria, Type 1	*AGXT*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,400
Primary Hyperoxaluria, Type 2	*GRHPR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 68,000
Primary Hyperoxaluria, Type 3	*HOGA1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 12,000
Progressive Cerebello-Cerebral Atrophy	*SEPSECS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 247,000
Progressive Familial Intrahepatic Cholestasis, Type 2	*ABCB11*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 390
Progressive Myoclonic Epilepsy, Type 1B	*PRICKLE1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 136,000
Progressive Pseudorheumatoid Dysplasia	*WISP3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 287,000
Prolidase Deficiency	*PEPD*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,900
Propionic Acidemia (*PCCA*-Related)	*PCCA*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,600
Propionic Acidemia (*PCCB*-Related)	*PCCB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 920
Pulmonary Surfactant Dysfunction	*ABCA3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Pycnodysostosis	*CTSK*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,200
Pyridoxamine 5'-Phosphate Oxidase Deficiency	*PNPO*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 6,600
Pyridoxine-Dependent Epilepsy	*ALDH7A1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100

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Pyruvate Carboxylase Deficiency	*PC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 215,000
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 8,300
Renal Tubular Acidosis and Deafness	*ATP6V1B1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 7,800
Retinitis Pigmentosa 25	*EYS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 580
Retinitis Pigmentosa 26	*CERKL*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,000
Retinitis Pigmentosa 28	*FAM161A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 145,000
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 4,300
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 55,000
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 95,000
Salla Disease	*SLC17A5*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Sandhoff Disease	*HEXB*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 680
Sanjad-Sakati Syndrome	*TBCE*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 66,000
Schimke Immunoosseous Dysplasia	*SMARCAL1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 56,000
Seckel Syndrome 5 / Microcephaly 9	*CEP152*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500
Segawa Syndrome	*TH*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,000
Sepiapterin Reductase Deficiency	*SPR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 43,000
Severe Combined Immunodeficiency (*IL7R*-Related)	*IL7R*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 48,000
Severe Combined Immunodeficiency (*JAK3*-Related)	*JAK3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Severe Combined Immunodeficiency (*PTPRC*-Related)	*PTPRC*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Severe Congenital Neutropenia 4	*G6PC3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 296,000
Severe Neonatal Hyperparathyroidism	*CASR*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 216,000
Short Stature, Onychodysplasia, Facial Dysmorphism, and Hypotrichosis	*POC1A*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 98,000
Short-Chain Acyl-CoA Dehydrogenase Deficiency	*ACADS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 340
Shwachman-Diamond Syndrome	*SBDS*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,200
Sialidosis, Type I and Type II	*NEU1*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,700
Sjogren-Larsson Syndrome	*ALDH3A2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,100
Smith-Lemli-Opitz Syndrome	*DHCR7*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,800
Spastic Paraplegia 15	*ZFYVE26*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly	*SLC1A4*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 136,000
Spherocytosis, Type 5	*EPB42*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Spinal Muscular Atrophy	*SMN1*	AR	Reduced Risk	*SMN1* copy number: 2 *SMN2* copy number: 2 c.*3+80T>G: Negative *SMN1* Sequencing: Negative **Personalized Residual Risk:** 1 in 1,115
Spinal Muscular Atrophy with Respiratory Distress 1 / Charcot-Marie-Tooth Disease, Type 2S	*IGHMBP2*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,100
Spinocerebellar Ataxia with Axonal Neuropathy 3	*COA7*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 12,000
Spondylocostal Dysostosis 1	*DLL3*	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 156,000
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 53,000

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Steel Syndrome	COL27A1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 275,000
Stuve-Wiedemann Syndrome	LIFR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 172,000
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,000
Tay-Sachs Disease	HEXA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,700
Thiamine-Responsive Megaloblastic Anemia Syndrome	SLC19A2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 63,000
Thyroid Dyshormonogenesis 1	SLC5A5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,300
Thyroid Dyshormonogenesis 2A	TPO	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 350
Thyroid Dyshormonogenesis 3	TG	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 130
Thyroid Dyshormonogenesis 4	IYD	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,900
Thyroid Dyshormonogenesis 5	DUOXA2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,300
Thyroid Dyshormonogenesis 6	DUOX2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 32
Trichohepatoenteric Syndrome 1	TTC37	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 11,000
Tyrosinemia, Type I	FAH	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,900
Tyrosinemia, Type II	TAT	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 4,200
Tyrosinemia, Type III	HPD	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Usher Syndrome, Type IB	MYO7A	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 180
Usher Syndrome, Type IC	USH1C	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 400
Usher Syndrome, Type ID	CDH23	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 880
Usher Syndrome, Type IF	PCDH15	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,100
Usher Syndrome, Type IIA	USH2A	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 54
Usher Syndrome, Type III	CLRN1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,800
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	ACADVL	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 380
Vitamin D-Dependent Rickets, Type I	CYP27B1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,600
Vitamin D-Resistant Rickets, Type IIA	VDR	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 21,000
Walker-Warburg Syndrome and Other *FKTN*-Related Dystrophies	FKTN	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 390
Werner Syndrome	WRN	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 2,000
Wilson Disease	ATP7B	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 150
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 287,000
Wolman Disease / Cholesteryl Ester Storage Disease	LIPA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 32,000
Woodhouse-Sakati Syndrome	DCAF17	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 59,000
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 4,300
Xeroderma Pigmentosum, Group A	XPA	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 170,000
Xeroderma Pigmentosum, Group C	XPC	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 15,000
Xeroderma Pigmentosum, Group G	ERCC5	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 3,900
Zellweger Syndrome Spectrum (*PEX10*-Related)	PEX10	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 218,000
Zellweger Syndrome Spectrum (*PEX1*-Related)	PEX1	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 740
Zellweger Syndrome Spectrum (*PEX2*-Related)	PEX2	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 108,000
Zellweger Syndrome Spectrum (*PEX6*-Related)	PEX6	AR	Reduced Risk	**Personalized Residual Risk:** 1 in 1,500

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%)

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PCR amplification using Asuragen, Inc. AmplideX$^®$ *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 single-base pair probe extension analyses using the Agena Bioscience iPlex Pro chemistry on a MassARRAY$^®$ 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%)

Conventional MLPA and/or digitalMLPA$^®$ probe sets and reagents from MRC-Holland were used for copy number variations (CNVs) analysis of specific targets versus known control samples. digitalMLPA$^®$ is a semi-quantitative technique, based on the well-established conventional MLPA method, followed by Illumina based sequencing to determine read number for amplicon quantification. False positive or negative results may occur due to rare sequence variants in target regions detected by conventional MLPA or digitalMLPA$^®$ probes. Analytical sensitivity and specificity of both the conventional MLPA method and the digitalMLPA$^®$ method are greater than 99%.

For alpha thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, duplications, 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 precisely specified without phase analysis. With the exception of duplications, 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. Depending on ethnicity, 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot distinguish 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 identify intragenic mutation in *SMN1*. Please also note that 2% of individuals diagnosed with SMA have a causative *SMN1* variant that occurred de novo, 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^{TM}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.

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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_1767874.) 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 (CNV) Analysis (Analytical Detection Rate >98% for CNVs of 3 exons and larger, >90% for CNVs of 2 exons)

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

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and duplications near the lower limit of detection may not be detected due to run variability. Genomic regions with high homology or highly repetitive sequences are excluded from this analysis.

Exon Array Comparative Genomic Hybridization (aCGH) (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 1,000,000 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 SYBR Green reagents on a LightCycler$^®$ 480 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 ΔΔCt 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^{TM}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 8th "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.

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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.

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Additional disease-specific references available upon request.

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Quest Diagnostics

Patient Information	Specimen Information	Client Information
7021, DONOR **DOB:** ■■■ **AGE:** ■ Gender: Phone: N ■■■ Patient ID: ■■■	Specimen: ■■■ Requisition: ■■■ Lab Ref #: ■■■ Collected: 12/08/2022 Received: 12/09/2022 / 21:37 EST Reported: 12/19/2022 / 03:53 EST	Client #: 48041578　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:　　　　　　　　　■■■
Specimen Type:　　　　　　Blood ■■■
Clinical Indication:　　　　　Donor of other specified organs or

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:	500
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.

Peter H. Bui, PhD, FACMG (800) NICHOLS-4307

Electronic Signature:　　　　12/19/2022 3:08 AM

![Quest Diagnostics]

Patient Information	Specimen Information	Client Information
7021, DONOR DOB: ███ AGE: █ Gender: M Patient ID: ███	Specimen: ███ Collected: 12/08/2022 Received: 12/09/2022 / 21:37 EST Reported: 12/19/2022 / 03:53 EST	Client #: 48041578 GENOMICS, SEMA4
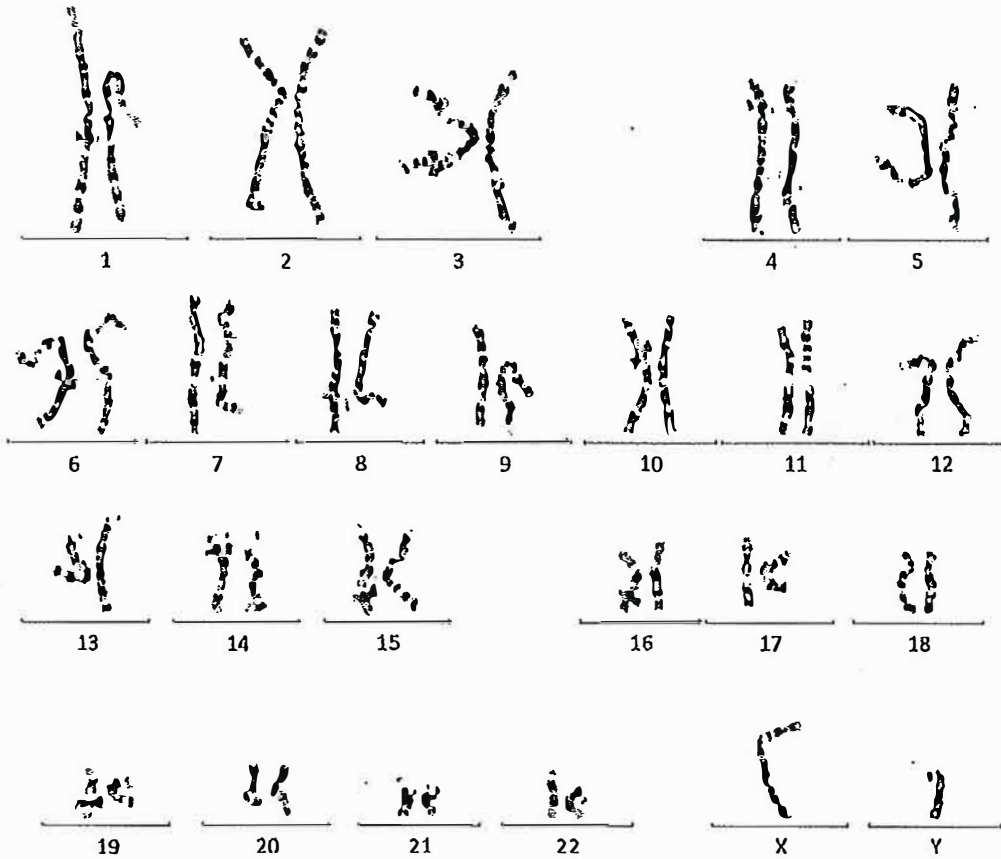
PERFORMING SITE:

BZ   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
7021, DONOR **DOB:** ▉ **AGE:** ▉ Gender: M Phone: NG Patient ID: ▉	Specimen: ▉ Requisition: ▉ Lab Ref #: ▉ Collected: 12/08/2022 Received: 12/09/2022 / 21:26 EST Reported: 12/12/2022 / 21:28 EST	Client #: 48041578 NYNJMAIL GENOMICS, SEMA4 SEMA4 62 SOUTHFIELD AVE STAMFORD, CT 06902-7229

Ward: FFAXCB

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	16.0		13.2-17.1 g/dL	
HEMATOCRIT	47.9		38.5-50.0 %	
MCV	92.3		80.0-100.0 fL	
MCH	30.8		27.0-33.0 pg	
RDW	12.1		11.0-15.0 %	
HEMOGLOBIN A	97.0		>96.0 %	Z99
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	3.0		2.2-3.2 %	
INTERPRETATION	*			
Normal phenotype.				

PERFORMING SITE:

Z99   QUEST DIAGNOSTICS CLIFTON, 1 INSIGHTS DRIVE, CLIFTON, NJ 07012-2355 Laboratory Director: LEZA N GALLO, MD, CLIA: 31D0696246