

Patient Information:

DOB:
Sex: M
MR#: BFA0191
Patient#:

Accession:

Test#:
Specimen Type: Saliva Swab
Collected: Jul 13,2023

Partner Information:

Not Tested

Accession:

N/A

Physician:

Shaikly, Valerie
ATTN: Shaikly, Valerie
Fertility Genetics
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Laboratory:

Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Aug 22,2023**

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

176 Matched Fors Male with XL

(177 Gene Panel; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations 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 in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information.
- 30 CGG repeats were observed in this individual.
- Testing for copy number changes in the SMN1 gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. Individuals with negative test results may still have up to a 3-4% risk to have a child with a birth defect due to genetic and/or environmental factors.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Gene specific notes and limitations may be present. See below.
- This report does not include variants of uncertain significance.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.

GENES TESTED:

176 Matched Fors Male with XL - 177 Genes

This analysis was run using the 176 Matched Fors Male with XL gene list. 177 genes were tested with 98.8% of targets sequenced at >20x coverage. For more gene specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

ABCC8, ABCD1, ACADM, ACADS, ACADVL, ADA, AGA, AGL, AGXT, AIRE, ALDH3A2, ALDOB, ALG6, ALMS1, ALPL, AMT, ARG1, ARSA, ASL, ASPA, ASS1, ATM, ATP7A, ATP7B, BBS1, BBS10, BBS12, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BTBD, CAPN3, CBS, CFTR, CLN3, CLN5, CLN6, CLN8, CLRN1, COL4A3, COL4A4, CPS1, CPT1A, CPT2, CRYL1, CTNS, CTSK, CYP11B1, CYP21A2, CYP27A1, DBT, DHCR7, DLD, DMD, DYSF, ELP1, ERCC6, ERCC8, EVC, EVC2, FAH, FANCA, FANCC, FKBP, FKTN, FMR1, G6PC, GAA, GALC, GALK1, GALT, GBA, GCDH, GJB2, GJB6, GLA, GLB1, GLDC, GNE, GNPTAB, GNPTG, GRHPR, HADHA, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HLCS, HMGCL, HOGA1, HSD17B4, HYLS1, IDS, IDUA, IL2RG, IVD, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LIPA, LRPPRC, MAN2B1, MCOLN1, MEFV, MESP2, MKS1, MLC1, MMAA, MMAB, MMACHC, MPI, MUT, MYO7A, NAGLU, NBN, NEB, NPC1, NPC2, NPHS1, NPHS2, NR0B1, OPA3, OTC, PAH, PC, PCCA, PCCB, PCDH15, PEX1, PEX10, PEX12, PEX2, PEX6, PEX7, PKHD1, PMM2, POMGNT1, PPT1, PROP1, PTS, RMRP, RS1, RTEL1, SACS, SGCA, SGCB, SGCD, SGCG, SGSH, SLC12A6, SLC17A5, SLC22A5, SLC26A2, SLC26A4, SLC37A4, SMN1, SMPD1, STAR, TAT, TCIRG1, TGM1, TH, TMEM216, TPP1, TTPA, USH1C, USH2A, VPS13B, XPA, XPC, ZFYVE26

METHODS:

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, 99.25% and 98.82% 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 are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General 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 future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. 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 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 other than specified genes. 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, 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 include one whole

gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); 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.

Gene Specific Notes and Limitations

BTD: If detected, the variant NM_001370658.1:c.1270G>C (p.Asp424His) will not be reported as this variant is associated with low disease penetrance and is primarily associated with reduced enzyme activity when homozygous. **CFTR:** Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. **CRYL1:** As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP21A2:** Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. CYP21A2 variants primarily associated with non-classic congenital adrenal hyperplasia (CAH) are not included in this analysis (PubMed: [23359698](#)). The variants associated with non-classic disease, including but not limited to c.188A>T (p.His63Leu), c.844G>T (p.Val282Leu), c.1174G>A (p.Ala392Thr), and c.1360C>T (p.Pro454Ser) will not be reported. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. **DMD:** Single exon deletion/duplication analysis is limited to exons with >1 patient reported in the UMD database (http://www.umd.be/DMD/W_DMD/index.html), accessed Dec 29, 2020 and all out-of-frame exons after exon 3. This includes deletion of exon 1, and duplication of exon 2, and del/dup for exons 3, 6~8, 11, 12, 17~22, 43~46, 48, 50~56, 58~63, 65~70, 75, 76 and 78. Single-exon detection is limited to blood samples. **FMR1:** The exact size of alleles >200 CGG repeats cannot be determined; these alleles are pathogenic for X-Linked Fragile X Syndrome. Alleles with <10 repeats may fail to amplify; these alleles are benign. The repeat length for this gene may vary by +/- 1 repeat unit. Methylation is not analyzed. RP-PCR analysis of the FMR1 promoter is not routinely performed in males. Small degrees of size mosaicism, including gonadal mosaicism, may not be detected. **GALT:** In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state (PubMed: [25473725](#), [30593450](#)). **GBA:** The current testing method may not be able to reliably detect certain pathogenic variants in the GBA gene due to homologous recombination between the pseudogene and the functional gene. **HBA1:** The phase of heterozygous alterations in the HBA1 gene cannot be determined, but can be confirmed through parental testing. **HBA2:** The phase of heterozygous alterations in the HBA2 gene cannot be determined, but can be confirmed through parental testing. **HSD17B4:** Copy number analysis for exons 4-6 may have reduced sensitivity in the HSD17B4 gene. Confirmation of these exons are limited to individuals with a positive personal history of D-bifunctional protein deficiency and Perrault syndrome and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **NEB:** This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. **NPHS2:** If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. **SMN1:** The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: [20301526](#)). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration, but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported.

SIGNATURE:

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A handwritten signature in black ink, appearing to read 'Jianbo Song'.

Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on 8/22/2023 07:38 PM PDT
Electronically signed

DISCLAIMER:

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. 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.



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ABCC8	Familial hyperinsulinism	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 44	98%	1 in 2,151	1 in 378,576
			Finnish Population	1 in 25	98%	1 in 1,201	1 in 120,100
			Middle-Eastern Population	1 in 25	98%	1 in 1,201	1 in 120,100
ABCD1	Adrenoleukodystrophy, X-linked	XL	General Population	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	AR	General Population	1 in 69	98%	1 in 3,401	1 in 938,676
			Caucasian / European Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			East Asian Population	1 in 198	99%	1 in 19,701	<1 in 10 million
			Native American Population	1 in 43	96%	1 in 1,051	1 in 180,772
ACADS	Short-chain acyl-coA dehydrogenase (SCAD) deficiency	AR	General Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
			African/African American Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			Caucasian / European Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
			Middle-Eastern Population	1 in 52	99%	1 in 5,101	1 in 1,061,008
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	AR	General Population	1 in 118	93%	1 in 1,672	1 in 789,184
			Middle-Eastern Population	1 in 74	93%	1 in 1,044	1 in 309,024
			Native American Population	1 in 61	93%	1 in 858	1 in 209,352
			South Asian/Indian Population	1 in 73	93%	1 in 1,030	1 in 300,760
ADA	Adenosine deaminase deficiency	AR	General Population	1 in 224	93%	1 in 3,187	1 in 2,855,552
AGA	Aspartylglucosaminuria	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 71	98%	1 in 3,501	1 in 994,284
AGL	Glycogen storage disease type III	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Faroese Population	1 in 28	95%	1 in 541	1 in 60,592
			Inuit Population	1 in 25	95%	1 in 481	1 in 48,100
			North African Jewish Population	1 in 37	95%	1 in 721	1 in 106,708
AGXT	Primary hyperoxaluria type 1	AR	General Population	1 in 120	99%	1 in 11,901	1 in 5,712,480
			Caucasian / European Population	1 in 173	99%	1 in 17,201	<1 in 10 million
AIRE	Autoimmune polyendocrinopathy syndrome type I	AR	General Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
			Finnish Population	1 in 79	98%	1 in 3,901	1 in 1,232,716
ALDH3A2	Sjögren-Larsson syndrome	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
ALDOB	Hereditary fructose intolerance	AR	General Population	1 in 122	99%	1 in 12,101	1 in 5,905,288
			African/African American Population	1 in 250	99%	1 in 24,901	<1 in 10 million
			Caucasian / European Population	1 in 67	99%	1 in 6,601	1 in 1,769,068
			Middle-Eastern Population	1 in 97	99%	1 in 9,601	1 in 3,725,188
ALG6	Congenital disorder of glycosylat on type Ic	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
ALMS1	Alstrom syndrome	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
ALPL	Hypophosphatasia	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Caucasian / European Population	1 in 274	95%	1 in 5,461	1 in 5,985,256
			Mennonite Population	1 in 25	95%	1 in 481	1 in 48,100
AMT	Glycine encephalopathy	AR	General Population	1 in 373	98%	1 in 18,601	<1 in 10 million
			Finnish Population	1 in 117	98%	1 in 5,801	1 in 2,714,868
ARG1	Arginase deficiency	AR	General Population	1 in 296	98%	1 in 14,751	<1 in 10 million
ARSA	Metachromatic leukodystrophy	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Caucasian / European Population	1 in 78	99%	1 in 7,701	1 in 2,402,712
			Yemenite Jewish Population	1 in 75	99%	1 in 7,401	1 in 2,220,300
ASL	Argininosuccinate lyase deficiency	AR	General Population	1 in 132	90%	1 in 1,311	1 in 692,208
ASPA	Canavan disease	AR	General Population	1 in 300	97%	1 in 9,968	<1 in 10 million
			Ashkenazi Jewish Population	1 in 55	96%	1 in 1,351	1 in 297,220
ASS1	Citrullinemia	AR	General Population	1 in 119	96%	1 in 2,951	1 in 1,404,676
			East Asian Population	1 in 132	96%	1 in 3,276	1 in 1,729,728
ATM	Ataxia-telangiectasia	AR	General Population	1 in 100	92%	1 in 1,239	1 in 495,600
ATP7A	Menkes disease	XL	General Population	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
ATP7B	Wilson disease	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Caucasian / European Population	1 in 42	98%	1 in 2,051	1 in 344,568
			Ashkenazi Jewish Population	1 in 70	98%	1 in 3,451	1 in 966,280
BBS1	Bardet-Biedl syndrome type 1	AR	General Population	1 in 367	99%	1 in 36,601	<1 in 10 million
BBS10	Bardet-Biedl syndrome type 10	AR	General Population	1 in 395	99%	1 in 39,401	<1 in 10 million
BBS12	Bardet-Biedl syndrome type 12	AR	General Population	1 in 791	99%	1 in 79,001	<1 in 10 million
BBS2	BBS2-related ciliopathies	AR	General Population	1 in 621	99%	1 in 62,001	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	99%	1 in 10,601	1 in 4,537,228
BCKDHA	Maple syrup urine disease type Ia	AR	General Population	1 in 321	98%	1 in 16,001	<1 in 10 million
			Mennonite Population	1 in 10	98%	1 in 451	1 in 18,040



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>BCKDHB</i>	Maple syrup urine disease type Ib	AR	General Population	1 in 364	98%	1 in 18,151	<1 in 10 million
			Ashkenazi Jewish Population	1 in 97	98%	1 in 4,801	1 in 1,862,788
<i>BCS1L</i>	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>BLM</i>	Bloom syndrome	AR	General Population	1 in 800	87%	1 in 6,147	<1 in 10 million
			Ashkenazi Jewish Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
<i>BTD</i>	Biotinidase deficiency	AR	General Population	1 in 124	99%	1 in 12,301	1 in 6,101,296
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
			Latino Population	1 in 136	99%	1 in 13,501	1 in 7,344,544
			Middle-Eastern Population	1 in 55	99%	1 in 5,401	1 in 1,188,220
<i>CAPN3</i>	Limb-girdle muscular dystrophy type 2A	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 103	98%	1 in 5,101	1 in 2,101,612
<i>CBS</i>	Homocystinuria due to cystathionine beta-synthase deficiency	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Caucasian / European Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			Middle-Eastern Population	1 in 21	99%	1 in 2,001	1 in 168,084
<i>CFTR</i>	Cystic Fibrosis	AR	General Population	1 in 32	99%	1 in 3,101	1 in 396,928
			African/African American Population	1 in 61	99%	1 in 6,001	1 in 1,464,244
			Ashkenazi Jewish Population	1 in 24	99%	1 in 2,301	1 in 220,896
			Caucasian / European Population	1 in 25	99%	1 in 2,401	1 in 240,100
			East Asian Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Latino Population	1 in 58	99%	1 in 5,701	1 in 1,322,632
<i>CLN3</i>	Neuronal ceroid lipofuscinosis	AR	General Population	1 in 230	98%	1 in 11,451	<1 in 10 million
			Finnish Population	1 in 72	98%	1 in 3,551	1 in 1,022,688
<i>CLN5</i>	Neuronal ceroid lipofuscinosis 5	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
<i>CLN6</i>	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
<i>CLN8</i>	Neuronal ceroid lipofuscinosis, CLN8-related	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Finnish Population	1 in 135	95%	1 in 2,681	1 in 1,447,740
<i>CLRN1</i>	Usher syndrome, type 3A	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 120	98%	1 in 5,951	1 in 2,856,480
			Finnish Population	1 in 70	98%	1 in 3,451	1 in 966,280
<i>COL4A3</i>	Alport syndrome, COL4A3-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
			Ashkenazi Jewish Population	1 in 188	98%	1 in 9,351	1 in 7,031,952
<i>COL4A4</i>	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
<i>CPS1</i>	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
<i>CPT1A</i>	Carnitine palmitoyltransferase IA deficiency	AR	General Population	1 in 354	90%	1 in 3,531	1 in 4,999,896
			Hutterite Population	1 in 16	90%	1 in 151	1 in 9,664
<i>CPT2</i>	Carnitine palmitoyltransferase II deficiency	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 51	95%	1 in 1,001	1 in 204,204
<i>CRYL1</i>	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
<i>CTNS</i>	Cystinosis	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			British Population	1 in 81	99%	1 in 8,001	1 in 2,592,324
			Moroccan Jewish Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>CTSK</i>	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>CYP11B1</i>	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
			Moroccan Jewish Population	1 in 35	98%	1 in 1,701	1 in 238,140
<i>CYP21A2</i>	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	AR	General Population	1 in 61	99%	1 in 6,001	1 in 1,464,244
			Inuit Population	1 in 9	99%	1 in 801	1 in 28,836
			Middle-Eastern Population	1 in 35	99%	1 in 3,401	1 in 476,140
<i>CYP27A1</i>	Cerebrotendinous xanthomatosis	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Moroccan Jewish Population	1 in 5	98%	1 in 201	1 in 4,020
<i>DBT</i>	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
<i>DHCR7</i>	Smith-Lemli-Opitz syndrome	AR	General Population	1 in 30	96%	1 in 726	1 in 87,120
			African/African American Population	1 in 138	96%	1 in 3,426	1 in 1,891,152
			Ashkenazi Jewish Population	1 in 36	96%	1 in 876	1 in 126,144
<i>DLD</i>	Dihydroliipoamide dehydrogenase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 2,268,828
<i>DMD</i>	Dystrophinopathies	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
<i>DYSF</i>	Limb-girdle muscular dystrophy type 2B	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Japanese Population	1 in 332	95%	1 in 6,621	1 in 8,792,688
			Libyan Jewish Population	1 in 18	95%	1 in 341	1 in 24,552
<i>ELP1</i>	Familial Dysautonomia	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 31	99%	1 in 3,001	1 in 372,124



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>ERCC6</i>	ERCC6-related disorders	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
<i>ERCC8</i>	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
<i>EVC</i>	EVC-related bone growth disorders	AR	General Population	1 in 142	98%	1 in 7,051	1 in 4,004,968
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>EVC2</i>	EVC2-related bone growth disorders	AR	General Population	1 in 240	98%	1 in 11,951	<1 in 10 million
			Amish Population	1 in 7	98%	1 in 301	1 in 8,428
<i>FAH</i>	Tyrosinemia, type 1	AR	General Population	1 in 99	95%	1 in 1,961	1 in 776,556
			Ashkenazi Jewish Population	1 in 150	95%	1 in 2,981	1 in 1,788,600
			Finnish Population	1 in 122	95%	1 in 2,421	1 in 1,181,448
			French Canadian Population	1 in 66	95%	1 in 1,301	1 in 343,464
			South Asian/Indian Population	1 in 172	95%	1 in 3,421	1 in 2,353,648
<i>FANCA</i>	Fanconi anemia group A	AR	General Population	1 in 239	99%	1 in 23,801	<1 in 10 million
			Moroccan Jewish	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Indian Jewish Population	1 in 27	99%	1 in 2,601	1 in 280,908
<i>FANCC</i>	Fanconi anemia group C	AR	General Population	1 in 535	99%	1 in 53,401	<1 in 10 million
			Ashkenazi Jewish Population	1 in 99	99%	1 in 9,801	1 in 3,881,196
<i>FKRP</i>	FKRP Alpha-dystroglycanopathies	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>FKTN</i>	FKTN Alpha-dystroglycanopathies	AR	General Population	1 in 500	99%	1 in 49,901	1 in 10 million
			Ashkenazi Jewish Population	1 in 150	99%	1 in 14,901	1 in 8,940,600
			Japanese Population	1 in 82	99%	1 in 8,101	1 in 2,657,128
<i>FMR1</i>	Fragile X Syndrome Intermediate Allele	XL	General Population	1 in 259	99%	1 in 25,801	1 in 103,204
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
<i>FMR1</i>	Fragile X Syndrome Premutation	XL	General Population	1 in 259	99%	1 in 25,801	1 in 103,204
			Ashkenazi Jewish Population	1 in 115	99%	1 in 11,401	1 in 45,604
<i>FMR1</i>	Fragile X Syndrome Full Mutation	XL	General Population	1 in 11,111	99%	1 in 1,111,001	1 in 4,444,004
<i>G6PC</i>	Glycogen storage disease, type 1a	AR	General Population	1 in 177	95%	1 in 3,521	1 in 2,492,868
			Ashkenazi Jewish Population	1 in 64	95%	1 in 1,261	1 in 322,816
<i>GAA</i>	Pompe disease	AR	General Population	1 in 100	98%	1 in 4,951	1 in 1,980,400
			African/African American Population	1 in 60	98%	1 in 2,951	1 in 708,240
			East Asian Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>GALC</i>	Krabbe disease	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			Israeli Druze Population	1 in 6	99%	1 in 501	1 in 12,024
<i>GALK1</i>	Galactokinase deficiency	AR	General Population	1 in 110	95%	1 in 2,181	1 in 959,640
			Irish Population	1 in 64	95%	1 in 1,261	1 in 322,816
<i>GALT</i>	Galactosemia	AR	General Population	1 in 110	99%	1 in 10,901	1 in 4,796,440
			African/African American Population	1 in 94	99%	1 in 9,301	1 in 3,497,176
			Ashkenazi Jewish Population	1 in 127	99%	1 in 12,601	1 in 6,401,308
<i>GBA</i>	Gaucher disease	AR	General Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			African/African American Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Ashkenazi Jewish Population	1 in 15	99%	1 in 1,401	1 in 84,060
<i>GCDH</i>	Glutaric aciduria, type I	AR	General Population	1 in 87	98%	1 in 4,301	1 in 1,496,748
			Amish Population	1 in 9	98%	1 in 401	1 in 14,436
<i>GJB2</i>	Nonsyndromic hearing loss 1A	AR	General Population	1 in 42	99%	1 in 4,101	1 in 688,968
			African/African American Population	1 in 25	99%	1 in 2,401	1 in 240,100
			Ashkenazi Jewish Population	1 in 21	99%	1 in 2,001	1 in 168,084
			Caucasian / European Population	1 in 33	99%	1 in 3,201	1 in 422,532
			Latino Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
			Middle-Eastern Population	1 in 83	99%	1 in 8,201	1 in 2,722,732
<i>GJB6</i>	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
<i>GLA</i>	Fabry disease	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
<i>GLB1</i>	GLB1-related gangliosidoses	AR	General Population	1 in 134	99%	1 in 13,301	1 in 7,129,336
			Maltese Population	1 in 30	99%	1 in 2,901	1 in 348,120
			Roma Population	1 in 50	99%	1 in 4,901	1 in 980,200
<i>GLDC</i>	Glycine encephalopathy, GLDC-related	AR	General Population	1 in 193	98%	1 in 9,601	1 in 7,411,972
			British Columbia Canadian Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
			Finnish Population	1 in 117	99%	1 in 11,601	1 in 5,429,268
<i>GNE</i>	Inclusion body myopathy type 2 (Nonaka myopathy)	AR	General Population	<1 in 500	99%	1 in 49,901	1 in 99,802,000
			Iranian Jewish Population	1 in 11	99%	1 in 1,001	1 in 44,044
<i>GNPTAB</i>	Mucopolipidosis II & III	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GNPTG</i>	Mucopolipidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
<i>GRHPR</i>	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>HADHA</i>	Trifunctional protein deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 124	98%	1 in 6,151	1 in 3,050,896
<i>HBA1</i>	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
<i>HBA2</i>	Alpha thalassemia	AR	General Population	1 in 1000	98%	1 in 860	1 in 3,440,364
			General Population†	1 in 18	98%	1 in 860	1 in 3,440,364
			Southeast Asian Population	≤1 in 7	98%	≤1 in 305	≤1 in 17,228
			Southeast Asian Population†	≤1 in 14	98%	≤1 in 305	≤1 in 17,228
			Mediterranean Population	≤1 in 6	98%	≤1 in 229	≤1 in 457,556
			Mediterranean Population†	1 in 500	98%	≤1 in 229	≤1 in 457,556
			African/African American Population	1 in 30	98%	1 in 1,451	1 in 5,804,000
<i>HBB</i>	Sickle cell disease	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			African/African American Population	1 in 10	95%	1 in 181	1 in 7,240
			East Asian Population	1 in 50	95%	1 in 981	1 in 196,200
			Latino Population	1 in 128	95%	1 in 2,541	1 in 1,300,992
			Mediterranean Population	1 in 3	95%	1 in 41	1 in 492
			South Asian/Indian Population	1 in 25	95%	1 in 481	1 in 48,100
			<i>HBB</i>	Hemoglobin C disease	AR	General Population	1 in 158
African/African American Population	1 in 10	95%				1 in 181	1 in 7,240
East Asian Population	1 in 50	95%				1 in 981	1 in 196,200
Latino Population	1 in 128	95%				1 in 2,541	1 in 1,300,992
Mediterranean Population	1 in 3	95%				1 in 41	1 in 492
South Asian/Indian Population	1 in 25	95%				1 in 481	1 in 48,100
<i>HBB</i>	Beta thalassemia	AR				General Population	1 in 158
			African/African American Population	1 in 10	99%	1 in 901	1 in 36,040
			East Asian Population	1 in 50	99%	1 in 4,901	1 in 980,200
			Latino Population	1 in 128	99%	1 in 12,701	1 in 6,502,912
			Mediterranean Population	1 in 3	99%	1 in 201	1 in 2,412
			South Asian/Indian Population	1 in 25	99%	1 in 2,401	1 in 240,100
			<i>HEXA</i>	Tay-Sachs disease	AR	General Population	1 in 300
Ashkenazi Jewish Population	1 in 27	99%				1 in 2,601	1 in 280,908
Moroccan Jewish Population	1 in 110	99%				1 in 10,901	1 in 4,796,440
General Population	1 in 600	98%				1 in 29,951	<1 in 10 million
<i>HEXB</i>	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million
<i>HGSNAT</i>	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population	1 in 434	98%	1 in 21,651	<1 in 10 million
			Caucasian / European Population	1 in 345	98%	1 in 17,201	<1 in 10 million
<i>HLCS</i>	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HMGCL</i>	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>HOGA1</i>	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
<i>HSD17B4</i>	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
<i>HYLS1</i>	Hydroletharus syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
<i>IDS</i>	Mucopolysaccharidosis type II (Hunter syndrome)	XL	General Population	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
<i>IDUA</i>	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			Caucasian / European Population	1 in 153	95%	1 in 3,041	1 in 1,861,092
<i>IL2RG</i>	X-linked severe combined immunodeficiency	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
<i>IVD</i>	Isovaleric Acidemia	AR	General Population	1 in 167	90%	1 in 1,661	1 in 1,109,548
			African/African American Population	1 in 100	90%	1 in 991	1 in 396,400
			Caucasian / European Population	1 in 115	90%	1 in 1,141	1 in 524,860
			East Asian Population	1 in 407	90%	1 in 4,061	1 in 6,611,308
<i>KCNJ11</i>	KCNJ11-related hyperinsulinism	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
			Caucasian / European Population	1 in 232	99%	1 in 23,101	<1 in 10 million
<i>LAMA2</i>	Muscular dystrophy, LAMA2-related	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 125	99%	1 in 12,401	1 in 6,200,500
<i>LAMA3</i>	Junctional epidermolysis bullosa 2	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMB3</i>	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMC2</i>	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LIPA</i>	Lysosomal acid lipase deficiency	AR	General Population	1 in 211	99%	1 in 21,001	<1 in 10 million
			Caucasian / European Population	1 in 161	99%	1 in 16,001	1 in 4,973,248
			Iranian Jewish Population	1 in 32	99%	1 in 3,101	1 in 396,928

Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>LRPPRC</i>	Leigh syndrome with Complex IV deficiency	AR	General Population	1 in 447	98%	1 in 22,301	<1 in 10 million
			Faroese Population	1 in 21	98%	1 in 1,001	1 in 84,084
			French Canadian Population	1 in 22	98%	1 in 1,051	1 in 92,488
<i>MAN2B1</i>	Alpha-Mannosidosis	AR	General Population	1 in 354	99%	1 in 35,301	<1 in 10 million
			Caucasian / European Population	1 in 274	99%	1 in 27,301	<1 in 10 million
<i>MCOLN1</i>	Mucopolidosis IV	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>MEFV</i>	Familial Mediterranean fever	AR	General Population	1 in 20	99%	1 in 1,901	1 in 152,080
			Mediterranean Population	1 in 7	90%	1 in 61	1 in 1,708
<i>MESP2</i>	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MKS1</i>	MKS1-related ciliopathies	AR	General Population	1 in 260	98%	1 in 12,951	<1 in 10 million
			Finnish Population	1 in 47	98%	1 in 2,301	1 in 432,588
<i>MLC1</i>	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Libyan Jewish Population	1 in 40	99%	1 in 3,901	1 in 624,160
<i>MMAA</i>	Methylmalonic aciduria, cbIA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
<i>MMAB</i>	Methylmalonic aciduria, cbIB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
<i>MMACHC</i>	Methylmalonic aciduria and homocystinuria, cbIC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
<i>MPI</i>	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MUT</i>	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
<i>MYO7A</i>	MYO7A-related disorders	AR	General Population	1 in 206	98%	1 in 10,251	1 in 8,446,824
			East Asian Population	1 in 62	98%	1 in 3,051	1 in 756,648
<i>NAGLU</i>	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 346	99%	1 in 34,501	<1 in 10 million
			East Asian Population	1 in 298	99%	1 in 29,701	<1 in 10 million
<i>NBN</i>	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
<i>NEB</i>	Nemaline myopathy	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Amish Population	1 in 11	98%	1 in 501	1 in 22,044
			Ashkenazi Jewish Population	1 in 108	98%	1 in 5,351	1 in 2,311,632
			Finnish Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
<i>NPC1</i>	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
<i>NPC2</i>	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
<i>NPHS1</i>	Congenital nephrotic syndrome, type 1	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
<i>NPHS2</i>	Congenital nephrotic syndrome, type 2	AR	General Population	1 in 289	98%	1 in 14,401	<1 in 10 million
			Finnish Population	1 in 50	98%	1 in 2,451	1 in 490,200
<i>NR0B1</i>	Congenital adrenal hypoplasia, X-linked	XL	General Population	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
<i>OPA3</i>	Costeff syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Iraqi Jewish Population	1 in 50	98%	1 in 2,451	1 in 490,200
<i>OTC</i>	Ornithine transcarbamylase deficiency	XL	General Population	1 in 7,000	90%	1 in 69,991	1 in 279,984
<i>PAH</i>	Phenylalanine Hydroxylase deficiency (Phenylketonuria)	AR	General Population	1 in 93	99%	1 in 9,201	1 in 3,422,772
			Caucasian / European Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Middle-Eastern Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
			South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
<i>PC</i>	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
<i>PCCA</i>	Propionic acidemia, PCCA-related	AR	General Population	1 in 224	96%	1 in 5,576	1 in 4,996,096
			Native American Population	1 in 85	96%	1 in 2,101	1 in 714,340
<i>PCCB</i>	Propionic acidemia, PCCB-related	AR	General Population	1 in 224	99%	1 in 22,301	<1 in 10 million
			Native American Population	1 in 85	99%	1 in 8,401	1 in 2,856,340
<i>PCDH15</i>	PCDH15-related sensory loss	AR	General Population	1 in 395	98%	1 in 19,701	1 in 78,804
			Ashkenazi Jewish Population	1 in 72	98%	1 in 3,551	1 in 14,204
<i>PEX1</i>	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
<i>PEX10</i>	Zellweger syndrome, PEX10-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Japanese Population	1 in 354	95%	1 in 7,061	1 in 9,998,376
<i>PEX12</i>	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
<i>PEX2</i>	Zellweger syndrome, PEX2-related	AR	General Population	1 in 500	95%	1 in 9,981	<1 in 10 million
			Ashkenazi Jewish Population	1 in 123	95%	1 in 2,441	1 in 1,200,972
<i>PEX6</i>	Zellweger syndrome, PEX6-related	AR	General Population	1 in 280	99%	1 in 27,901	<1 in 10 million
			Yemenite Jewish Population	1 in 18	99%	1 in 1,701	1 in 122,472
<i>PEX7</i>	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>PKHD1</i>	Polycystic kidney disease, PKHD1-related	AR	General Population	1 in 70	98%	1 in 3,451	1 in 966,280
			Ashkenazi Jewish Population	1 in 107	98%	1 in 5,301	1 in 2,268,828
<i>PMM2</i>	Congenital disorder of glycosylation type 1a	AR	General Population	1 in 63	99%	1 in 6,201	1 in 1,562,652
			Ashkenazi Jewish Population	1 in 57	99%	1 in 5,601	1 in 1,277,028
			Caucasian / European Population	1 in 71	99%	1 in 7,001	1 in 1,988,284
<i>POMGNT1</i>	POMGNT1 Alpha-dystroglycanopathies	AR	General Population	1 in 462	98%	1 in 23,051	<1 in 10 million
			Finnish Population	1 in 111	98%	1 in 5,501	1 in 2,442,444
<i>PPT1</i>	Neuronal ceroid lipofuscinosis, PPT1-related	AR	General Population	1 in 368	98%	1 in 18,351	<1 in 10 million
			Caucasian / European Population	1 in 488	98%	1 in 24,351	<1 in 10 million
			Finnish Population	1 in 75	98%	1 in 3,701	1 in 1,110,300
<i>PROP1</i>	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
<i>PTS</i>	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
<i>RMRP</i>	Cartilage-Hair Hypoplasia Anauxetic Dysplasia Spectrum Disorder	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
			Amish Population	1 in 16	99%	1 in 1,501	1 in 96,064
			Finnish Population	1 in 76	99%	1 in 7,501	1 in 2,280,304
<i>RS1</i>	Juvenile retinoschisis, X-linked	XL	General Population	1 in 2,500	96%	1 in 62,476	1 in 249,956
<i>RTEL1</i>	Dyskeratosis congenita type 5	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Ashkenazi Jewish Population	1 in 203	99%	1 in 20,201	<1 in 10 million
<i>SACS</i>	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
			French Canadian Population	1 in 19	95%	1 in 361	1 in 27,436
<i>SGCA</i>	Limb-girdle muscular dystrophy, type 2D	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 288	98%	1 in 14,351	<1 in 10 million
			Finnish Population	1 in 150	98%	1 in 7,451	1 in 4,470,600
<i>SGCB</i>	Limb-girdle muscular dystrophy, type 2E	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 406	98%	1 in 20,251	<1 in 10 million
<i>SGCD</i>	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>SGCG</i>	Limb-girdle muscular dystrophy, type 2C	AR	General Population	1 in 381	98%	1 in 19,001	<1 in 10 million
			Moroccan Population	1 in 250	98%	1 in 12,451	<1 in 10 million
			Roma / Gypsy Population	1 in 96	98%	1 in 4,751	1 in 1,824,384
<i>SGSH</i>	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	AR	General Population	1 in 454	98%	1 in 22,651	<1 in 10 million
			Caucasian / European Population	1 in 253	98%	1 in 12,601	<1 in 10 million
<i>SLC12A6</i>	Andermann syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			French Canadian Population	1 in 23	99%	1 in 2,201	1 in 202,492
<i>SLC17A5</i>	Sialic acid storage disorder	AR	General Population	<1 in 500	91%	1 in 5,545	<1 in 10 million
			Finnish Population	1 in 100	91%	1 in 1,101	1 in 440,400
<i>SLC22A5</i>	Systemic primary carnitine deficiency	AR	General Population	1 in 129	99%	1 in 12,801	1 in 6,605,316
			African/African American Population	1 in 86	99%	1 in 8,501	1 in 2,924,344
			East Asian Population	1 in 77	99%	1 in 7,601	1 in 2,341,108
			Faroese Population	1 in 9	99%	1 in 801	1 in 28,836
			Pacific Islander Population	1 in 37	99%	1 in 3,601	1 in 532,948
			South Asian/Indian Population	1 in 51	99%	1 in 5,001	1 in 1,020,204
<i>SLC26A2</i>	SLC26A2-related disorders	AR	General Population	1 in 158	90%	1 in 1,571	1 in 992,872
			Finnish Population	1 in 50	90%	1 in 491	1 in 98,200
<i>SLC26A4</i>	Pendred syndrome	AR	General Population	1 in 80	98%	1 in 3,951	1 in 1,264,320
			African/African American Population	1 in 76	98%	1 in 3,751	1 in 1,140,304
			Caucasian / European Population	1 in 88	98%	1 in 4,351	1 in 1,531,552
			East Asian Population	1 in 74	98%	1 in 3,651	1 in 1,080,696
<i>SLC37A4</i>	Glycogen storage disease, type Ib	AR	General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
			Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 397,884
<i>SMN1</i>	Spinal muscular atrophy	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
			African/African American Population	1 in 72	71%	1 in 246	1 in 70,848
			Ashkenazi Jewish Population	1 in 67	91%	1 in 734	1 in 196,712
			Caucasian / European Population	1 in 47	95%	1 in 921	1 in 173,148
			East Asian Population	1 in 59	93%	1 in 830	1 in 195,880
			Latino Population	1 in 68	90%	1 in 671	1 in 182,512
			Sephardic Jewish Population	1 in 34	96%	1 in 826	1 in 112,336
<i>SMN1</i>	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
<i>SMPD1</i>	Niemann-Pick disease, type A/B	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
			Ashkenazi Jewish Population	1 in 115	95%	1 in 2,281	1 in 1,049,260
			Latino Population	1 in 106	95%	1 in 2,101	1 in 890,824
<i>STAR</i>	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>TAT</i>	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>TCIRG1</i>	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
<i>TGM1</i>	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056

Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>TH</i>	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
<i>TMEM216</i>	TMEM216-related ciliopathies	AR	General Population	1 in 141	98%	1 in 7,001	1 in 3,948,564
			Ashkenazi Jewish Population	1 in 92	98%	1 in 4,551	1 in 1,674,768
<i>TPP1</i>	Neuronal ceroid lipofuscinosis, TPP1-related	AR	General Population	1 in 252	97%	1 in 8,368	1 in 8,434,944
			French Canadian Population	1 in 53	97%	1 in 1,734	1 in 367,608
<i>TTPA</i>	Ataxia with isolated vitamin E deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
			Caucasian / European Population	1 in 267	90%	1 in 2,661	1 in 2,841,948
<i>USH1C</i>	USH1C-related disorders	AR	General Population	1 in 353	90%	1 in 3,521	1 in 4,971,652
			French Canadian Population	1 in 227	90%	1 in 2,261	1 in 2,052,988
<i>USH2A</i>	Usher syndrome, type 2A	AR	General Population	1 in 126	96%	1 in 3,126	1 in 1,575,504
			Caucasian / European Population	1 in 73	96%	1 in 1,801	1 in 525,892
			Ashkenazi Jewish Population	1 in 35	99%	1 in 3,401	1 in 476,140
			Iranian Jewish Population	1 in 60	99%	1 in 5,901	1 in 1,416,240
<i>VPS13B</i>	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>XPA</i>	Xeroderma pigmentosum, group A	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
			Japanese Population	1 in 74	99%	1 in 7,301	1 in 2,161,096
<i>XPC</i>	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
<i>ZFYVE26</i>	Spastic paraplegia 15	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million

* For genes that have tested negative

† The carrier frequency for heterozygous alpha thalassemia carriers ($\alpha\alpha/\alpha-$) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis ($\alpha\alpha/-$) is 1 in 1000.

Abbreviations: AR, autosomal recessive; XL, X-linked