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fulgent

[redacted]

Patient Information:
[redacted]
DOB: [redacted]
Sex: M
MR#: BFA 0187
Patient#: [redacted]

Partner Information:
Not Tested

Physician:
Shaikly, Valerie
ATTN: Shaikly, Valerie
Fertility Genetics
1 Lanswood Park
Elmstead Market, Essex CO7 7FD GB
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Laboratory:
Fulgent Genetics
CAP#: 8042697
CLIA#: 05D2043189
Laboratory Director:
Dr. Hanlin (Harry) Gao
Report Date: **Jun 28,2023**

Accession:
[redacted]
Test#: [redacted]
Specimen Type: Saliva Swab
Collected: May 15,2023

Accession:
N/A

FINAL RESULTS

Carrier for **ONE** genetic condition
Genetic counseling is recommended.

TEST PERFORMED

176 Matched Fors Male with XL
(177 Gene Panel; gene sequencing with deletion and duplication analysis)

Condition and Gene	Inheritance	[redacted]	Partner
Meckel syndrome 1 *MKS1*	AR	⊕ Carrier c.805dup (p.Ser269Phefs*36)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for a carrier mutation in 1 gene. The risk estimates below are quantified based on general population carrier frequencies. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions:
  - There is a 1/1040 chance of having a child affected with Meckel syndrome 1, a *MKS1*-related condition.
- 31 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.

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MECKEL SYNDROME 1

Patient	[redacted]	Partner
Result	⊕ Carrier	N/A
Variant Details	***MKS1*** (NM_017777.4) c.805dup (p.Ser269Phefs*36)	N/A

What is Meckel syndrome 1?

Meckel syndrome is a group of inherited disorders that affect many parts of the body and is typically lethal. The most common features are enlarged, cystic kidneys, an encephalocele, and extra fingers and toes. These features are usually seen during pregnancy due to their severity. Other features include abnormalities of the liver, heart, lungs, and genitourinary tract; however, the presentation of the other features varies. The different types are distinguished by their genetic contribution.

What is my risk of having an affected child?

Meckel syndrome 1 is inherited in an autosomal recessive manner. The risk to be a carrier of a MKS1-related condition is about 1 in 260. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

What kind of medical management is available?

There is no cure or treatment for Meckel syndrome, and it typically has a fatal outcome prenatally or within the first few weeks of life.

What mutation was detected?

The detected heterozygous variant was NM_017777.4:c.805dup (p.Ser269Phefs*36). This variant is predicted to introduce a premature stop codon at least 50 nucleotides upstream of the canonical donor splice site of the penultimate exon and to result in the loss of function of the protein product due to nonsense-mediated mRNA decay (PubMed: 25741868, 30192042, 27618451, 11532962, 18066079). There's sufficient evidence that loss of function in this gene is a known disease mechanism for MKS1-related conditions (PubMed: 35360848, 29620724, 26490104, 17397051). The laboratory classifies this variant as likely pathogenic.

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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 99.0% 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, BTD, 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, FKRP, 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, 99.27% and 99.01% 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

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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 on 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. *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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Dr. Harry Gao, DABMG, FACMG on **6/28/2023 09:48 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.

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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 glycosylation 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	Bardet-Biedl syndrome 2	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
BBS2	Retinitis Pigmentosa 74	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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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
BCKDHB	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
BCS1L	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BCS1L	Mitochondrial complex III deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
BLM	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
BTD	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
CAPN3	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
CBS	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
CFTR	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
CLN3	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
CLN5	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
CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	AR	General Population	<1 in 500	92%	1 in 6,239	<1 in 10 million
CLN8	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
CLRN1	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
COL4A3	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
COL4A4	Alport syndrome, COL4A4-related	AR	General Population	1 in 267	98%	1 in 13,301	<1 in 10 million
CPS1	Carbamoylphosphate synthetase I deficiency	AR	General Population	1 in 570	98%	1 in 28,451	<1 in 10 million
CPT1A	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
CPT2	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
CRYL1	GJB6-CRYL1 related nonsyndromic hearing loss	UK	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
CTNS	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
CTSK	Pycnodysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
CYP11B1	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
			Morrocan Jewish Population	1 in 35	98%	1 in 1,701	1 in 238,140
CYP21A2	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
CYP27A1	Cerebrotendinous xanthomatosis	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
			Morrocan Jewish Population	1 in 5	98%	1 in 201	1 in 4,020
DBT	Maple syrup urine disease, type II	AR	General Population	1 in 481	98%	1 in 24,001	<1 in 10 million
DHCR7	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
DLD	Dihydrolipoamide 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
DMD	Duchenne Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
DMD	Becker Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
DYSF	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
ELP1	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
ERCC6	De Sanctis-Cacchione syndrome	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
ERCC6	Cockayne syndrome type B	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
ERCC8	Cockayne syndrome type A	AR	General Population	1 in 822	98%	1 in 41,051	<1 in 10 million
EVC	Weyers acrofacial dysostosis, EVC-related	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
EVC	Ellis-van Creveld syndrome, EVC-related	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
EVC2	Weyers acrodental dysostosis, EVC2-related	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
EVC2	Ellis-van Creveld syndrome, EVC2-related	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
FAH	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
FANCA	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
FANCC	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
FKRP	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
FKRP	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FKTN	Muscular dystrophy-dystroglycanopathy, FKTN-related	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
FKTN	Fukuyama congenital muscular dystrophy	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
FKTN	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
FMR1	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
FMR1	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
FMR1	Fragile X Syndrome Full Mutation	XL	General Population	1 in 11,111	99%	1 in 1,111,001	1 in 4,444,004
G6PC	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
GAA	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
GALC	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
GALK1	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
GALT	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
GBA	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
GCDH	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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
GJB2	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
			South Asian/Indian Population	1 in 148	99%	1 in 14,701	1 in 8,702,992
GJB6	GJB6-CRYL1 related nonsyndromic hearing loss	AR	General Population	1 in 423	99%	1 in 42,201	<1 in 10 million
GLA	Fabry disease	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
GLB1	GM1-gangliosidosis	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
GLB1	Mucopolysaccharidosis type IVB (Morquio syndrome B)	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
GLDC	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
GNE	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
GNPTAB	Mucolipidosis II alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucolipidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucolipidosis III gamma	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GRHPR	Primary hyperoxaluria type II	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
HADHA	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
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) 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
HBA1	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
HBA2	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
HBB	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
HBB	Hemoglobin C 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
HBB	Beta thalassemia	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
			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
HEXA	Tay-Sachs disease	AR	General Population	1 in 300	99%	1 in 29,901	<1 in 10 million
			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
HEXB	Sandhoff disease	AR	General Population	1 in 600	98%	1 in 29,951	<1 in 10 million

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Supplemental Table

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HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	AR	General Population Caucasian / European Population	1 in 434 1 in 345	98% 98%	1 in 21,651 1 in 17,201	<1 in 10 million <1 in 10 million
HLCS	Holocarboxylase synthetase deficiency	AR	General Population	1 in 500	98%	1 in 24,951	<1 in 10 million
HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
HOGA1	Primary hyperoxaluria type III	AR	General Population	1 in 184	99%	1 in 18,301	<1 in 10 million
HSD17B4	D-bifunctional protein deficiency	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
HYLS1	Hydrolethalus syndrome	AR	General Population Finnish Population	<1 in 500 1 in 50	98% 98%	1 in 24,951 1 in 2,451	<1 in 10 million 1 in 490,200
IDS	Mucopolysaccharidosis type II (Hunter syndrome)	XL	General Population	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
IDUA	Mucopolysaccharidosis, type I (Hurler syndrome)	AR	General Population Caucasian / European Population	<1 in 500 1 in 153	95% 95%	1 in 9,981 1 in 3,041	<1 in 10 million 1 in 1,861,092
IL2RG	Severe combined immunodeficiency, X-linked	XL	General Population	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
IVD	Isovaleric Acidemia	AR	General Population African/African American Population Caucasian / European Population East Asian Population	1 in 167 1 in 100 1 in 115 1 in 407	90% 90% 90% 90%	1 in 1,661 1 in 991 1 in 1,141 1 in 4,061	1 in 1,109,548 1 in 396,400 1 in 524,860 1 in 6,611,308
KCNJ11	Congenital hyperinsulinism	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
KCNJ11	Permanent neonatal diabetes mellitus	AR	General Population Caucasian / European Population	1 in 423 1 in 232	99% 99%	1 in 42,201 1 in 23,101	<1 in 10 million <1 in 10 million
LAMA2	Muscular dystrophy, LAMA2-related	AR	General Population Caucasian / European Population	<1 in 500 1 in 125	99% 99%	1 in 49,901 1 in 12,401	<1 in 10 million 1 in 6,200,500
LAMA3	Junctional epidermolysis bullosa, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMA3	Laryngo-onycho-cutaneous syndrome	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMB3	Junctional epidermolysis bullosa, LAMB3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LAMC2	Junctional epidermolysis bullosa, LAMC2-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
LIPA	Lysosomal acid lipase deficiency	AR	General Population Caucasian / European Population Iranian Jewish Population	<1 in 500 1 in 112 1 in 26	99% 99% 99%	1 in 49,901 1 in 11,101 1 in 2,501	<1 in 10 million 1 in 4,973,248 1 in 260,104
LRPPRC	Leigh syndrome with Complex IV deficiency	AR	General Population Faroese Population French Canadian Population	1 in 447 1 in 21 1 in 22	98% 98% 98%	1 in 22,301 1 in 1,001 1 in 1,051	<1 in 10 million 1 in 84,084 1 in 92,488
MAN2B1	Alpha-Mannosidosis	AR	General Population Caucasian / European Population	1 in 354 1 in 274	99% 99%	1 in 35,301 1 in 27,301	<1 in 10 million <1 in 10 million
MCOLN1	Mucolipidosis IV	AR	General Population Ashkenazi Jewish Population	1 in 300 1 in 100	99% 99%	1 in 29,901 1 in 9,901	<1 in 10 million 1 in 3,960,400
MEFV	Familial Mediterranean fever	AR	General Population Mediterranean Population	1 in 20 1 in 7	99% 90%	1 in 1,901 1 in 61	1 in 152,080 1 in 1,708
MESP2	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MKS1	Bardet-Biedl syndrome 13	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Joubert syndrome 28	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MKS1	Meckel syndrome 1	AR	General Population Finnish Population	1 in 260 1 in 47	98% 98%	1 in 12,951 1 in 2,301	<1 in 10 million 1 in 432,588
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	AR	General Population Libyan Jewish Population	<1 in 500 1 in 40	99% 99%	1 in 49,901 1 in 3,901	<1 in 10 million 1 in 624,160
MMAA	Methylmalonic aciduria, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
MMAB	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	AR	General Population	1 in 134	90%	1 in 1,331	1 in 713,416
MPI	Congenital disorder of glycosylation type Ib	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
MUT	Methylmalonic aciduria–methylmalonyl–CoA mutase deficiency	AR	General Population	1 in 100	99%	1 in 9,901	1 in 3,960,400
MYO7A	Usher syndrome, type 1B	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
MYO7A	Non-syndromic hearing loss, MYO7A-related	AR	General Population East Asian Population	1 in 206 1 in 62	98% 98%	1 in 10,251 1 in 3,051	1 in 8,446,824 1 in 756,648
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	AR	General Population Caucasian / European Population East Asian Population	<1 in 500 1 in 346 1 in 298	99% 99% 99%	1 in 49,901 1 in 34,501 1 in 29,701	<1 in 10 million <1 in 10 million <1 in 10 million
NBN	Nijmegen breakage syndrome	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032

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NEB	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
NPC1	Niemann-Pick disease, type C1	AR	General Population	1 in 194	90%	1 in 1,931	1 in 1,498,456
NPC2	Niemann-Pick disease, type C2	AR	General Population	1 in 194	99%	1 in 19,301	<1 in 10 million
NPHS1	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
NPHS2	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
NR0B1	Congenital adrenal hypoplasia, X-linked	XL	General Population	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
OPA3	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
OTC	Ornithine transcarbamylase deficiency	XL	General Population	1 in 7,000	90%	1 in 69,991	1 in 279,984
PAH	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
PC	Pyruvate carboxylase deficiency	AR	General Population	1 in 250	95%	1 in 4,981	1 in 4,981,000
PCCA	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
PCCB	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
PCDH15	Non-syndromic hearing loss, PCDH15-related	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
PCDH15	Usher syndrome, type 1F	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
PEX1	Zellweger syndrome, PEX1-related	AR	General Population	1 in 147	95%	1 in 2,921	1 in 1,717,548
PEX10	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
PEX12	Zellweger syndrome, PEX12-related	AR	General Population	1 in 373	95%	1 in 7,441	<1 in 10 million
PEX2	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
PEX6	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
PEX7	Rhizomelic chondrodysplasia punctata, type 1	AR	General Population	1 in 158	99%	1 in 15,701	1 in 9,923,032
PKHD1	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
PMM2	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
POMGNT1	Muscular dystrophy-dystroglycanopathy	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
POMGNT1	Retinitis pigmentosa 76	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
POMGNT1	Walker-Warburg syndrome	AR	General Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
PPT1	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
PROP1	Combined pituitary hormone deficiency 2	AR	General Population	1 in 45	98%	1 in 2,201	1 in 396,180
PTS	Tetrahydrobiopterin deficiency	AR	General Population	1 in 354	96%	1 in 8,826	<1 in 10 million
RMRP	Metaphyseal dysplasia without hypotrichosis	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
RMRP	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 500	99%	1 in 49,901	<1 in 10 million
			Finnish Population	<1 in 500	99%	1 in 49,901	<1 in 10 million
RMRP	Anauxetic dysplasia	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
RMRP	Cartilage-hair hypoplasia	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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
RS1	Juvenile retinoschisis, X-linked	XL	General Population	1 in 2,500	96%	1 in 62,476	1 in 249,956
RTEL1	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
SACS	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
SGCA	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
SGCB	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
SGCD	Limb-girdle muscular dystrophy, type 2F	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
SGCG	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
SGSH	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
SLC12A6	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
SLC17A5	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
SLC22A5	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
SLC26A2	Diastrophic dysplasia	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
SLC26A2	Achondrogenesis, type IB	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
SLC26A2	Multiple epiphyseal dysplasia	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
SLC26A2	Atelosteogenesis II	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
SLC26A4	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
SLC37A4	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
SMN1	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
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
SMPD1	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
STAR	Lipoid congenital adrenal hyperplasia	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
TAT	Tyrosinemia, type II	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TCIRG1	Osteopetrosis 1	AR	General Population	1 in 250	98%	1 in 12,451	<1 in 10 million
TGM1	Congenital ichthyosis	AR	General Population	1 in 224	95%	1 in 4,461	1 in 3,997,056
TH	Segawa syndrome	AR	General Population	1 in 224	98%	1 in 11,151	1 in 9,991,296
TMEM216	Joubert syndrome 2	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
TMEM216	Meckel syndrome 2	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
TPP1	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
TTPA	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

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Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
USH1C	Usher syndrome, type IC	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
USH1C	Non-syndromic hearing loss, USH1C-related	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
USH2A	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
VPS13B	Cohen syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
XPA	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
XPC	Xeroderma pigmentosum, group C	AR	General Population	1 in 500	99%	1 in 49,901	<1 in 10 million
ZFYVE26	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 (αα/α-) is described in rows marked with a dagger symbol. The carrier frequency for alpha thalassemia trait cis (αα/- -) is 1 in 1000.
Abbreviations: AR, autosomal recessive; XL, X-linked