



Patient Information:

DOB: [REDACTED]
Sex: M
MR#: BFA0183
Patient#: [REDACTED]

Partner Information:

Not Tested

Physician:

Shaikly, Valerie
ATTN: Shaikly, Valerie
Fertility Genetics
1 Lanswood Park
Elmstead Market, Essex CO7 7FD GB
Phone: 7711197938

Laboratory:

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

Accession:


Test#: [REDACTED]
Specimen Type: Saliva Swab
Collected: Mar 06,2023

Accession:

N/A

FINAL RESULTS

TEST PERFORMED



Carrier for genetic conditions in **multiple** genes.
Genetic counseling is recommended.

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

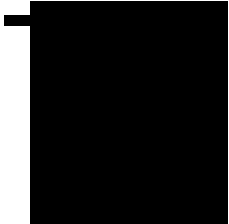
Condition and Gene	Inheritance	Partner		
Wilson disease <i>ATP7B</i>	AR	+	Carrier c.3008C>T (p.Ala1003Val)	N/A
Methylmalonic aciduria and homocystinuria, cbIC type <i>MMACHC</i>	AR	+	Carrier Whole Gene Deletion	N/A
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency <i>CYP21A2</i>	AR	+	Possible Carrier c.955C>T(:)*12C>T + CYP21A2 duplication p.(Gln319*)(:)(?)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for carrier mutations in 3 genes. 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 1/348 chance of having a child affected with Wilson disease, a *ATP7B*-related condition.
 - There is a 1/536 chance of having a child affected with Methylmalonic aciduria and homocystinuria, cbIC type, a *MMACHC*-related condition.
 - There is a 1/244 chance of having a child affected with Congenital adrenal hyperplasia due to 21-hydroxylase deficiency, a *CYP21A2*-related condition.
- Repeat expansion testing for FMR1 indicates that 36 CGG repeats were observed in this individual. This result is within a normal range.
- 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.

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- This report does not include variants of uncertain significance.
- Genetic counseling is recommended. Contact your physician about the available options for genetic counseling.

Patient: [REDACTED] Sex: M;
DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
DocID: [REDACTED] PAGE 2 of 16



WILSON DISEASE

Patient		Partner
Result	⊕ Carrier	N/A
Variant Details	ATP7B (NM_000053.4) c.3008C>T (p.Ala1003Val)	N/A

What is Wilson disease?

Wilson disease is a disorder that affects the liver's ability to remove excess copper in the body. Normally, a healthy amount of copper is absorbed by the body through diet. When the liver is impaired and cannot properly excrete copper, the buildup over time can lead to symptoms such as liver disease, psychiatric disturbance, and neurologic disorder. Ages of onset vary, with symptoms most commonly reported between the ages of 5 to 35.

What is my risk of having an affected child?

Wilson disease is inherited in an autosomal recessive manner. This means that when both parents are carriers for the condition, there is a 1 in 4 (25%) risk of having an affected child. The overall risk of being a carrier for *ATP7B*-related Wilson disease is 1 in 87 in the general population. Individuals of Caucasian/European descent have an increased carrier risk of 1 in 42.

What kind of medical management is available?



Medical management is specific to each individual and should be discussed with your doctor. To treat Wilson disease, lifelong management is required. Administration of chelating agents (medications) to remove excess copper and reducing dietary intake of copper may help to prevent disease progression. Liver transplantation may also be required in severe cases of liver damage. Biannual surveillance is strongly recommended to assess the progression of the disease and/or treatment. Routine monitoring should include blood tests, physical examinations, and liver function tests.

What mutation was detected?

The detected heterozygous variant was NM_000053.4:c.3008C>T (p.Ala1003Val). This missense variant, [p.Ala1003Val], has been reported in homozygous or compound heterozygous in combination with another *ATP7B* variant in individuals affected with Wilson disease (PMID: 31059521, 10544227, 33763395, 17264425). Other variant(s) at this position in the gene (p.Ala1003Thr, p.Ala1003Pro) have been associated with Wilson disease, suggesting that a change at this position adversely affects protein structure and/or function and is potentially disease-causing (PubMed: [21610751](#), [12885331](#), [26799313](#)). This variant is classified as "Pathogenic" or "Likely Pathogenic" in ClinVar, with multiple submitters in agreement (Variation ID:188781) The laboratory classifies this variant as pathogenic.



METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLC TYPE

Patient		Partner
Result	 Carrier	N/A
Variant Details	MMACHC (NM_015506.3) Whole Gene Deletion	N/A

What is Methylmalonic aciduria and homocystinuria, cb1C type?

Methylmalonic aciduria and homocystinuria is a condition in which the body is unable to process certain fats and proteins. When the condition begins early in life, affected individuals typically have failure to thrive, difficulty feeding, and an abnormally pale appearance. Neurological problems are also common in methylmalonic aciduria and homocystinuria, including hypotonia, seizures, microcephaly, delayed development, and intellectual disabilities. The signs and symptoms worsen over time and the condition can be life-threatening if not treated.

What is my risk of having an affected child?

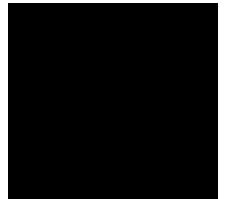
Methylmalonic aciduria and homocystinuria is inherited in an autosomal recessive manner. This means that when both parents are carriers for the same condition, there is a 25% (1 in 4) risk of having an affected child. The overall carrier frequency is estimated to be 1 in 134 in the general population.

What kind of medical management is available?

There is currently no cure for Methylmalonic aciduria and homocystinuria, but the early institution of dietary therapy may reduce but not completely prevent primary symptoms. Avoidance of prolonged fasting and dehydration may reduce episodes of metabolic decompensation. Other options for medical management include the use of certain medications and antibiotics.

What mutation was detected?

The detected heterozygous variant was NM_015506.3:c.(? -170)_(*2219_?)del (p.?). This is an apparent whole-gene deletion which encompasses the genomic region including exons 1-4 and is predicted to result in loss of function of the MMACHC gene. There is sufficient evidence that loss of function in this gene is a known disease mechanism for methylmalonic aciduria and homocystinuria of complementation group cb1C (PubMed: [19370762](#), [16311595](#)). This variant is absent from the general population. The scope of the performed analysis is not designed to determine the exact breakpoints or boundaries of copy number variants. The above whole gene deletion of MMACHC may or may not represent part of a larger deletion involving other potentially clinically relevant genes not assessed by this test. The laboratory classifies this variant as likely pathogenic.



CONGENITAL ADRENAL HYPERPLASIA DUE TO 21-HYDROXYLASE DEFICIENCY

Patient	██████████ ██████████	Partner
Result	⊕ Possible Carrier	N/A
Variant Details	CYP21A2 (NM_000500.9) c.955C>T(;)*12C>T + CYP21A2 duplication p.(Gln319*)(;)(?)	N/A

What is Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is an inherited disorder that affects the adrenal glands and hormone production. Approximately 75 percent of individuals with classic 21-hydroxylase deficiency have the salt-wasting type, whereby the body excretes too much salt in urine. Affected infants present with poor feeding, weight loss, dehydration, and vomiting, all of which can be life-threatening. Females with this condition typically have ambiguous genitalia, while males usually have normal genitalia, but with small testes. Individuals with the simple virilizing form and the non-classic form of the disease do not experience salt loss. Males and females with either the classic or non-classic forms of 21-hydroxylase deficiency tend to have an early growth spurt, but their final adult height is usually shorter than others in their family, and affected individuals may have reduced fertility. Additionally, individuals may have excessive body hair growth, hair loss, and irregular menstruation. Some individuals (male or female) with the non-classic form of the disease may have mild, non-life-threatening symptoms, while others may never develop symptoms of the disorder at all.

What is my risk of having an affected child?

CAH due to 21-hydroxylase deficiency is inherited in an autosomal recessive manner. The risk for being a carrier for CYP21A2-related CAH is 1/61. Individuals of Inuit descent have an increased carrier risk of 1/9. Individuals of Middle-Eastern descent have an increased carrier risk of 1/35. If the patient and the partner are both carriers, the risk for an affected child is 1 in 4 (25%).

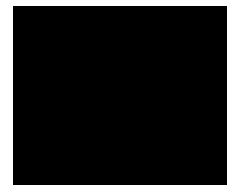
What kind of medical management is available?

Treatment consists of early initiation of hormone replacement therapy and/or surgery for females. Prognosis is good for patients with appropriate medical management and psychological support.

What mutation was detected?

The heterozygous variants c.955C>T (p.Gln319*) and a whole gene duplication of CYP21A2 were detected in this sample. In addition, the benign polymorphism c.*12C>T was also detected. The phase of these variants is unknown but could be determined through parental testing.

The nonsense variant, p.Gln319*, introduces a premature stop codon and is expected to result in the loss of function of the protein product of the CYP21A2 gene, either as the result of protein truncation or of nonsense-mediated mRNA decay. This variant, also reported as Q318*, is a classic 21-hydroxylase-deficient congenital adrenal hyperplasia mutation and has been reported in multiple affected individuals (PubMed: [3267225](#), [12220458](#), [12915679](#)). The variant, p.Gln319*, and the polymorphism c.*12C>T are known to frequently occur in a duplicated copy of the CYP21A2 gene coexisting with a normal copy of CYP21A2 on the same chromosome. This haplotype was identified in approximately 2% of the general population and in ~80% of carriers of p.Gln319*, and such a configuration may represent a benign allele (PubMed: [28401898](#), [19773403](#)). Nonetheless, there is a possibility that p.Gln319* occurs on a chromosome with only a single copy of CYP21A2, in which case it results in a pathogenic allele. If multiple copies of CYP21A2 are present, we cannot be certain if this p.Gln319* variant occurs on a chromosome with one (i.e. pathogenic state) or two (i.e. benign state) copies of CYP21A2. While this combination of variants may represent a benign allele, the laboratory classifies the variant p.Gln319* as likely pathogenic.



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 99.15% 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 99.15% 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

BTB: 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](https://pubmed.ncbi.nlm.nih.gov/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:



Yan Meng, Ph.D., CGMB, FACMG on 4/5/2023 08:41 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

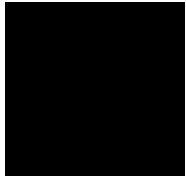
Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
ABCC8	Familial hyperinsulinism	AR	General Population	1 in 112	98%	1 in 5,551	1 in 2,486,848
			Ashkenazi Jewish Population	1 in 44	98%	1 in 2,151	1 in 378,576
			Finnish Population	1 in 25	98%	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 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

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 9 of 16

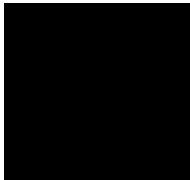
Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<i>BCKDHA</i>	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
<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>	Björnstad syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>BCS1L</i>	GRACILE syndrome	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<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>BTBD</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>DLSD</i>	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
<i>DMD</i>	Duchenne Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260
<i>DMD</i>	Becker Muscular Dystrophy	XL	General Population	1 in 2,350	93%	1 in 33,558	1 in 134,260



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 acrofacial 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
FKRP	Muscular dystrophy-dystroglycanopathy, FKRP-related	AR	General Population	1 in 158	98%	1 in 7,851	1 in 4,961,832
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
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
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



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	Mucopolipidosis II alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTAB	Mucopolipidosis III alpha/beta	AR	General Population	<1 in 500	95%	1 in 9,981	<1 in 10 million
GNPTG	Mucopolipidosis 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
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
			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
			HEXA	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

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 12 of 16



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
<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>	Hydrolethalus 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>	Severe combined immunodeficiency, X-linked	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>	Congenital 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>KCNJ11</i>	Permanent neonatal diabetes mellitus	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, LAMA3-related	AR	General Population	1 in 781	98%	1 in 39,001	<1 in 10 million
<i>LAMA3</i>	Laryngo-onycho-cutaneous syndrome	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 500	99%	1 in 49,901	<1 in 10 million
			Caucasian / European Population	1 in 112	99%	1 in 11,101	1 in 4,973,248
			Iranian Jewish Population	1 in 26	99%	1 in 2,501	1 in 260,104
<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>	Mucopolysaccharidosis 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
<i>MESP2</i>	Spondylocostal dysostosis	AR	General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
<i>MKS1</i>	Bardet-Biedl syndrome 13	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>MKS1</i>	Joubert syndrome 28	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>MKS1</i>	Meckel syndrome 1	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, cblA type	AR	General Population	1 in 301	97%	1 in 10,001	<1 in 10 million
<i>MMAB</i>	Methylmalonic aciduria, cblB type	AR	General Population	1 in 435	98%	1 in 21,701	<1 in 10 million
<i>MMACHC</i>	Methylmalonic aciduria and homocystinuria, cblC 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>	Usher syndrome, type 1B	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>MYO7A</i>	Non-syndromic hearing loss, MYO7A-related	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

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 13 of 16



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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
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
RS1	Juvenile retinoschisis, X-linked	XL	General Population	1 in 2,500	96%	1 in 62,476	1 in 249,956

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 14 of 16

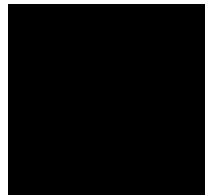


Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
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
			General Population	1 in 158	95%	1 in 3,141	1 in 1,985,112
SLC37A4	Glycogen storage disease, type Ib	AR	Ashkenazi Jewish Population	1 in 71	95%	1 in 1,401	1 in 397,884
			General Population	1 in 54	91%	1 in 590	1 in 127,440
SMN1	Spinal muscular atrophy	AR	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
			General Population	1 in 54	91%	1 in 590	1 in 127,440
SMN1	Spinal muscular atrophy silent carrier	AR	General Population	1 in 54	91%	1 in 590	1 in 127,440
			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
SMPD1	Niemann-Pick disease, type A/B	AR	Latino Population	1 in 106	95%	1 in 2,101	1 in 890,824
			General Population	<1 in 500	98%	1 in 24,951	<1 in 10 million
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
TPPA	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

Patient: [REDACTED] Sex: M;
 DOB: [REDACTED] MR#: BFA0183

Accession#: [REDACTED] FD Patient#: [REDACTED]
 DocID: [REDACTED] PAGE 15 of 16



Supplemental Table

Gene	Condition	Inheritance	Ethnicity	Carrier Rate	Detection Rate	Post-test Carrier Probability*	Residual Risk*
USH1C	Usher syndrome, type 1C	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 ($\alpha\alpha/\alpha-$) 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