



RESULTS RECIPIENT  
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 Report Date: 10/08/2021

MALE  
**DONOR 14270**  
 DOB:  
 Ethnicity: Mixed or Other  
 Caucasian  
 Sample Type: EDTA Blood  
 Date of Collection: 09/29/2021  
 Date Received: 10/01/2021  
 Date Tested: 10/08/2021  
 Barcode: 11004512827351  
 Accession ID:  
 CSLCNHXJ3WH3Q9G  
 Indication: Egg or sperm donor

FEMALE  
 N/A

# Foresight® Carrier Screen

**POSITIVE: CARRIER**

## ABOUT THIS TEST

The **Myriad Foresight Carrier Screen** utilizes sequencing, maximizing coverage across all DNA regions tested, to help you learn about your chance to have a child with a genetic disease.

## RESULTS SUMMARY

Risk Details	DONOR 14270	Partner
Panel Information	Foresight Carrier Screen Universal Panel Fundamental Plus Panel Fundamental Panel <b>(175 conditions tested)</b>	N/A
<b>POSITIVE: CARRIER</b> <b>Familial Mediterranean Fever</b> <b>Reproductive Risk: 1 in 450</b> <b>Inheritance: Autosomal Recessive</b>	<b>⚠ CARRIER*</b> NM_000243.2(MEFV):c. 2084A>G(K695R) heterozygote †	The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group. Carrier testing should be considered. See "Next Steps".

†Likely to have a negative impact on gene function.

\*Carriers generally do not experience symptoms.

No disease-causing mutations were detected in any other gene tested. A complete list of all conditions tested can be found on page 7.

## CLINICAL NOTES

- None

## NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner.
- Patients are recommended to discuss reproductive risks with their health care provider or a genetic counselor. Patients may also wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.

## POSITIVE: CARRIER

# Familial Mediterranean Fever

**Reproductive risk: 1 in 450**  
 Risk before testing: 1 in 50,000

**Gene:** MEFV | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 14270	No partner tested
<b>Result</b>	Carrier	N/A
<b>Variant(s)</b>	NM_000243.2(MEFV):c.2084A>G(K695R) heterozygote †	N/A
<b>Methodology</b>	Sequencing with copy number analysis (v3.1)	N/A
<b>Interpretation</b>	This individual is a carrier of familial Mediterranean fever. Carriers generally do not experience symptoms. In the absence of a known personal and/or family history of inflammatory disease, the clinical significance of MEFV mutation status is uncertain.	N/A
<b>Detection rate</b>	>99%	N/A
<b>Exons tested</b>	NM_000243:1-10.	N/A

†Likely to have a negative impact on gene function.

## What is Familial Mediterranean Fever?

Familial Mediterranean fever (FMF) is a condition caused by harmful genetic changes (mutations) in the *MEFV* gene. Mutations in the *MEFV* gene affect the production of a protein called pyrin, which is involved in controlling inflammation within the body. Uncontrolled inflammation can lead to repeated (recurrent) episodes (attacks) of pain and fever in patients with FMF.

Symptoms during an attack can include headache, fever, and pain, which are caused by inflammation of the membranes surrounding the abdomen (peritonitis), joints (synovitis), and lungs (pleuritis), or other organs. Some individuals also develop patches of red, tender, and swollen skin (Erysipelas-like erythema). The recurrent attacks usually last for one to three days and vary in severity. Between attacks, an affected individual typically feels normal, and these symptom-free periods can last for days or even years. Factors such as infection, hormone cycles, and stress may contribute to the timing of these episodes. In most individuals, the first attack occurs before the age of 20. Children who have FMF may experience episodic fever as the only symptom.

Some individuals with FMF develop a protein buildup (amyloidosis) in various parts of the body, notably the kidney. If left untreated, this can lead to life-threatening kidney failure. Individuals who do not experience the characteristic attacks of FMF can still develop this form of kidney failure.

Untreated FMF can also result in decreased fertility.

Some individuals with two mutations in the *MEFV* gene may never have any signs or symptoms, while some individuals with only one mutation in the *MEFV* gene may develop symptoms. Consequently, in the absence of a personal and/or family history of inflammatory disease, *MEFV* mutation status cannot be used with certainty to predict an individual's risk for symptoms.

## How common is Familial Mediterranean Fever?

FMF is most common among ethnic groups from the Mediterranean region, notably individuals of Armenian, Arab, Turkish, Iraqi Jewish, and North African Jewish ancestry, in which the prevalence is approximately 1 in 200 to 1 in 1,000 individuals. In all other populations, the prevalence of FMF is lower.

## How is Familial Mediterranean Fever treated?

There is no cure for FMF, but in most patients, a daily dose of the drug colchicine is effective in preventing the disease's characteristic attacks. Colchicine also prevents the dangerous buildup of proteins in the kidneys, which could otherwise lead to kidney failure.

Treatment for episodic attacks is supportive in nature, such as when medications are used to help ease pain and inflammation. Individuals who do develop serious kidney failure may require kidney transplantation.

## What is the prognosis for an individual with Familial Mediterranean Fever?

With early and regular treatment, individuals with FMF can live a normal lifespan and may even be free of symptoms. The disease has the potential to be life-threatening if the patient develops kidney failure (which may result when a person is untreated or does not respond to treatment).

## Methods and Limitations

**DONOR 14270 [Foresight Carrier Screen]:** Sequencing with copy number analysis, spinal muscular atrophy, analysis of homologous regions, and alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis (Assay(s): DTS v3.2).

### Sequencing with copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to analyze the genes listed in the Conditions Tested section of the report. Except where otherwise noted, the region of interest (ROI) comprises the indicated coding regions and 20 non-coding bases flanking each region. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected non-coding bases are excluded from the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. Select genes or regions for which pseudogenes or other regions of homology impede reliable variant detection may be assayed using alternate technology, or they may be excluded from the ROI. *CFTR* and *DMD* testing includes analysis for exon-level deletions and duplications with an average sensitivity of ~99%. Only exon-level deletions are assayed for other genes on the panel and such deletions are detected with a sensitivity of  $\geq 75\%$ . Selected founder deletions may be detected at slightly higher sensitivity. Affected exons and/or breakpoints of copy number variants are estimated from junction reads, where available, or using the positions of affected probes. Only exons known to be included in the region affected by a copy number variant are provided in the variant nomenclature. In some cases, the copy number variant may be larger or smaller than indicated. If *GJB2* is tested, large upstream deletions involving the *GJB6* and/or *CRYL1* genes that may affect the expression of *GJB2* are also analyzed.

### Spinal muscular atrophy

Targeted copy number analysis via high-throughput sequencing is used to determine the copy number of exon 7 of the *SMN1* gene. Other genetic variants may interfere with this analysis. Some individuals with two copies of *SMN1* are "silent" carriers with both *SMN1* genes on one chromosome and no copies of the gene on the other chromosome. This is more likely in individuals who have two copies of the *SMN1* gene and are positive for the g.27134T>G single-nucleotide polymorphism (SNP) (PMID: 9199562, 23788250, and 28676062), which affects the reported residual risk; Ashkenazi Jewish or Asian patients with this genotype have a high post-test likelihood of being carriers for SMA and are reported as carriers. The g.27134T>G SNP is only reported in individuals who have two copies of *SMN1*.

### Analysis of homologous regions

A combination of high-throughput sequencing, read-depth-based copy number analysis, and targeted genotyping is used to determine the number of functional gene copies and/or the presence of selected loss-of-function variants in certain genes that have homology to other genomic regions. The precise breakpoints of large deletions in these genes cannot be determined but are instead estimated from copy number analysis. Pseudogenes may interfere with this analysis, especially when many pseudogene copies are present.

If *CYP21A2* is tested, patients who have one or more additional copies of the *CYP21A2* gene and a pathogenic variant may or may not be a carrier of 21-hydroxylase deficient CAH, depending on the chromosomal location of the variants (phase). Benign *CYP21A2* gene duplications and/or triplications will only be reported in this context. Some individuals with two functional *CYP21A2* gene copies may be "silent" carriers, with two gene copies resulting from a duplication on one chromosome and a gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay. Given that the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are based only on the published incidence for classic CAH. However, the published prevalence of non-classic CAH is highest in individuals of Ashkenazi Jewish, Hispanic, Italian, and Yugoslav descent. Therefore, the residual and reproductive risks are likely an underestimate for CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

## Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis

High-throughput sequencing and read-depth-based copy number analysis are used to identify sequence variation and functional gene copies within the region of interest (ROI) of *HBA1* and *HBA2*, which includes the listed exons plus 20 intronic flanking bases. In a minority of cases where genomic features (e.g., long homopolymers) compromise calling fidelity, the affected intronic bases are not included in the ROI. The ROI is sequenced to a minimum acceptable read depth, and the sequences are compared to a reference genomic sequence (Genome Reference Consortium Human Build 37 [GRCh37]/hg19). On average, 99% of all bases in the ROI are sequenced at a read depth that is greater than the minimum read depth. Sequence variants may not be detected in areas of lower sequence coverage. Insertions and deletions may not be detected as accurately as single-nucleotide variants. For large deletions or duplications in these genes, the precise breakpoints cannot be determined but are instead estimated from copy number analysis. This assay has been validated to detect up to two additional copies of each alpha globin gene. In rare instances where assay results suggest greater than two additional copies are present, this will be noted but the specific number of gene copies observed will not be provided.

Extensive sequence homology exists between *HBA1* and *HBA2*. This sequence homology can prevent certain variants from being localized to one gene over the other. In these instances, variant nomenclature will be provided for both genes. If follow-up testing is indicated for patients with the nomenclature provided for both genes, both *HBA1* and *HBA2* should be tested. Some individuals with four functional alpha globin gene copies may be "silent" carriers, with three gene copies resulting from triplication on one chromosome and a single gene deletion on the other chromosome. This and other similar rare carrier states, where complementary changes exist between the chromosomes, may not be detected by the assay.

## Interpretation of reported variants

The classification and interpretation of all variants identified in this assay reflects the current state of Myriad's scientific understanding at the time this report was issued. Variants are classified according to internally defined criteria, which are compatible with the ACMG Standards and Guidelines for the Interpretation of Sequence Variants (PMID: 25741868). Variants that have been determined by Myriad to be disease-causing or likely disease-causing (i.e. pathogenic or likely pathogenic) are reported. Benign variants, variants of uncertain clinical significance (VUS), and variants not directly associated with the specified disease phenotype(s) are not reported. Variant classification and interpretation may change for a variety of reasons, including but not limited to, improvements to classification techniques, availability of additional scientific information, and observation of a variant in more patients. If the classification of one or more variants identified in this patient changes, an updated report reflecting the new classification generally will not be issued. If an updated report is issued, the variants reported may change based on their current classification. This can include changes to the variants displayed in gene specific 'variants tested' sections. Healthcare providers may contact Myriad directly to request updated variant classification information specific to this test result.

## Limitations

The MWH Foresight Carrier Screen is designed to detect and report germline (constitutional) alterations. Mosaic (somatic) variation may not be detected, and if it is detected, it may not be reported. If more than one variant is detected in a gene, additional studies may be necessary to determine if those variants lie on the same chromosome or different chromosomes (phase). This test is not designed to detect sex-chromosome copy number variations. If present, sex-chromosome abnormalities may significantly reduce test sensitivity for X-linked conditions. Variant interpretation and residual and reproductive risk estimations assume a normal karyotype and may be different for individuals with abnormal karyotypes. The test does not fully address all inherited forms of intellectual disability, birth defects, or heritable diseases. Furthermore, not all forms of genetic variation are detected by this assay (i.e., duplications [except in specified genes], chromosomal rearrangements, structural abnormalities, etc.). Additional testing may be appropriate for some individuals. Pseudogenes and other regions of homology may interfere with this analysis. In an unknown number of cases, other genetic variation may interfere with variant detection. Rare carrier states where complementary changes exist between the chromosomes may not be detected by the assay. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions, and technical or analytical errors.

Detection rates are determined using published scientific literature and/or reputable databases, when available, to estimate the fraction of disease alleles, weighted by frequency, that the methodology is predicted to be able or unable to detect. Detection rates are approximate and only account for analytical sensitivity. Certain variants that have been previously described in the literature may not be reported, if there is insufficient evidence for pathogenicity. Detection rates do not account for the disease specific rates of *de novo* variation.

This test was developed, and its performance characteristics determined by, Myriad Women's Health, Inc. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA does not require this test to go through premarket review. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high-complexity clinical testing. These results are adjunctive to the ordering physician's evaluation. CLIA Number: #05D1102604.



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MALE  
**DONOR 14270**  
DOB:  
Ethnicity: Mixed or Other  
Caucasian  
Barcode: 11004512827351

FEMALE  
N/A

### Incidental Findings

Unless otherwise indicated, these results and interpretations are limited to the specific disease panel(s) requested by the ordering healthcare provider. In some cases, standard data analyses may identify genetic findings beyond the region(s) of interest specified by the test, and such findings may not be reported. These findings may include genomic abnormalities with major, minor, or no, clinical significance.

If you have questions or would like more information about any of the test methods or limitations, please contact (888) 268-6795.

## Resources

**GENOME CONNECT** | <http://www.genomeconnect.org>

Patients can share their reports using research registries such as Genome Connect, an online research registry building a genetics and health knowledge base. Genome Connect provides patients, physicians, and researchers an opportunity to share genetic information to support the study of the impact of genetic variation on health conditions.

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### SENIOR LABORATORY DIRECTOR

A handwritten signature in black ink that reads "Karla R. Bowles".

Karla R. Bowles, PhD, FACMG, CGMB

Report content approved by Heather Labreche, PhD, FACMG, CGMBS on Oct 8, 2021

# Conditions Tested

**6-pyruvoyl-tetrahydropterin Synthase Deficiency** - Gene: PTS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000317:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Adenosine Deaminase Deficiency** - Gene: ADA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000022:1-12. **Detection Rate:** Mixed or Other Caucasian 98%.

**Alpha Thalassemia, HBA1/HBA2-related** - Genes: HBA1, HBA2. Autosomal Recessive. Alpha thalassemia (HBA1/HBA2) sequencing with targeted copy number analysis. Exons: NM\_000517:1-3; NM\_000558:1-3. Variants (16): -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, Poly(A) AATAAA>AATA--, Poly(A) AATAAA>AATAAG, Poly(A) AATAAA>AATGAA, anti3.7, anti4.2, del HS-40. **Detection Rate:** Not calculated due to rarity of disease in this individual's reported ethnicity.

**Alpha-mannosidosis** - Gene: MAN2B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000528:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.

**Alpha-sarcoglycanopathy** - Gene: SGCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000023:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Alstrom Syndrome** - Gene: ALMS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015120:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.

**Andermann Syndrome** - Gene: SLC12A6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_133647:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.

**Argininemia** - Gene: ARG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000045:1-8. **Detection Rate:** Mixed or Other Caucasian 97%.

**Argininosuccinic Aciduria** - Gene: ASL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001024943:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.

**Aspartylglucosaminuria** - Gene: AGA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000027:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Ataxia with Vitamin E Deficiency** - Gene: TTPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000370:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

**Ataxia-telangiectasia** - Gene: ATM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000051:2-63. **Detection Rate:** Mixed or Other Caucasian 96%.

**ATP7A-related Disorders** - Gene: ATP7A. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000052:2-23. **Detection Rate:** Mixed or Other Caucasian 90%.

**Autoimmune Polyglandular Syndrome Type 1** - Gene: AIRE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000383:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Autosomal Recessive Osteopetrosis Type 1** - Gene: TCIRG1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006019:2-20. **Detection Rate:** Mixed or Other Caucasian 96%.

**Autosomal Recessive Polycystic Kidney Disease, PKHD1-related** - Gene: PKHD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_138694 2-67. **Detection Rate:** Mixed or Other Caucasian >99%.

**Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay** - Gene: SACS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_014363 2-10. **Detection Rate:** Mixed or Other Caucasian 99%.

**Bardet-Biedl Syndrome, BBS1-related** - Gene: BBS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024649:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.

**Bardet-Biedl Syndrome, BBS10-related** - Gene: BBS10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024685:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

**Bardet-Biedl Syndrome, BBS12-related** - Gene: BBS12. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_152618:2. **Detection Rate:** Mixed or Other Caucasian >99%.

**Bardet-Biedl Syndrome, BBS2-related** - Gene: BBS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_031885:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.

**BCS1L-related Disorders** - Gene: BCS1L. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004328:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Beta-sarcoglycanopathy** - Gene: SGCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000232:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Biotinidase Deficiency** - Gene: BTD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000060:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.

**Bloom Syndrome** - Gene: BLM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000057:2-22. **Detection Rate:** Mixed or Other Caucasian >99%.

**Calpainopathy** - Gene: CAPN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000070:1-24. **Detection Rate:** Mixed or Other Caucasian 99%.

**Canavan Disease** - Gene: ASPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000049:1-6. **Detection Rate:** Mixed or Other Caucasian 98%.

**Carbamoylphosphate Synthetase I Deficiency** - Gene: CPS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001875:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.

**Carnitine Palmitoyltransferase IA Deficiency** - Gene: CPT1A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001876:2-19. **Detection Rate:** Mixed or Other Caucasian >99%.

**Carnitine Palmitoyltransferase II Deficiency** - Gene: CPT2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000098:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

**Cartilage-hair Hypoplasia** - Gene: RMRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NR\_003051:1. **Detection Rate:** Mixed or Other Caucasian >99%.

**Cerebrotendinous Xanthomatosis** - Gene: CYP27A1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000784:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Citrullinemia Type 1** - Gene: ASS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000050:3-16. **Detection Rate:** Mixed or Other Caucasian >99%.

**CLN3-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001042432 2-16. **Detection Rate:** Mixed or Other Caucasian >99%.

**CLN5-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006493:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.

**CLN8-related Neuronal Ceroid Lipofuscinosis** - Gene: CLN8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_018941:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Cohen Syndrome** - Gene: VPS13B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017890:2-62. **Detection Rate:** Mixed or Other Caucasian 97%.



**COL4A3-related Alport Syndrome** - Gene: COL4A3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000091:1-52. **Detection Rate:** Mixed or Other Caucasian 94%.

**COL4A4-related Alport Syndrome** - Gene: COL4A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000092:2-48. **Detection Rate:** Mixed or Other Caucasian >99%.

**Combined Pituitary Hormone Deficiency, PROP1-related** - Gene: PROP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006261:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Congenital Adrenal Hyperplasia, CYP11B1-related** - Gene: CYP11B1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000497:1-9. **Detection Rate:** Mixed or Other Caucasian 97%.

**Congenital Adrenal Hyperplasia, CYP21A2-related** - Gene: CYP21A2. Autosomal Recessive. Analysis of homologous regions. Variants (13): CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111Vfs\*21, I173N, L308Ffs\*6, P31L, Q319\*, Q319\*+CYP21A2dup, R357W, V282L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Mixed or Other Caucasian 96%.

**Congenital Disorder of Glycosylation Type Ia** - Gene: PMM2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000303:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Congenital Disorder of Glycosylation Type Ic** - Gene: ALG6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_013339:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

**Congenital Disorder of Glycosylation, MPI-related** - Gene: MPI. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002435:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Costeff Optic Atrophy Syndrome** - Gene: OPA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_025136:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

**Cystic Fibrosis** - Gene: CFTR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000492:1-27. IVS5-8T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Mixed or Other Caucasian >99%.

**Cystinosis** - Gene: CTNS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004937:3-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**D-bifunctional Protein Deficiency** - Gene: HSD17B4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000414:1-24. **Detection Rate:** Mixed or Other Caucasian 98%.

**Delta-sarcoglycanopathy** - Gene: SGCD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000337:2-9. **Detection Rate:** Mixed or Other Caucasian 96%.

**Dihydroloipoamide Dehydrogenase Deficiency** - Gene: DLD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000108:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Dysferlinopathy** - Gene: DYSF. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003494:1-55. **Detection Rate:** Mixed or Other Caucasian 98%.

**Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)** - Gene: DMD. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_004006:1-79. **Detection Rate:** Mixed or Other Caucasian 99%.

**ERCC6-related Disorders** - Gene: ERCC6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000124:2-21. **Detection Rate:** Mixed or Other Caucasian 96%.

**ERCC8-related Disorders** - Gene: ERCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000082:1-12. **Detection Rate:** Mixed or Other Caucasian 97%.

**EVC-related Ellis-van Creveld Syndrome** - Gene: EVC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_153717:1-21. **Detection Rate:** Mixed or Other Caucasian 96%.

**EVC2-related Ellis-van Creveld Syndrome** - Gene: EVC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_147127:1-22. **Detection Rate:** Mixed or Other Caucasian 98%.

**Fabry Disease** - Gene: GLA. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000169:1-7. **Detection Rate:** Mixed or Other Caucasian 98%.

**Familial Dysautonomia** - Gene: ELP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003640:2-37. **Detection Rate:** Mixed or Other Caucasian >99%.

**Familial Hyperinsulinism, ABCC8-related** - Gene: ABCC8. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000352:1-39. **Detection Rate:** Mixed or Other Caucasian >99%.

**Familial Hyperinsulinism, KCNJ11-related** - Gene: KCNJ11. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_000525:1. **Detection Rate:** Mixed or Other Caucasian >99%.

**Familial Mediterranean Fever** - Gene: MEFV. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000243:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

**Fanconi Anemia Complementation Group A** - Gene: FANCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000135:1-43. **Detection Rate:** Mixed or Other Caucasian 92%.

**Fanconi Anemia, FANCC-related** - Gene: FANCC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000136:2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

**FKRP-related Disorders** - Gene: FKRP. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_024301:4. **Detection Rate:** Mixed or Other Caucasian >99%.

**FKTN-related Disorders** - Gene: FKTN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001079802:3-11. **Detection Rate:** Mixed or Other Caucasian >99%.

**Free Sialic Acid Storage Disorders** - Gene: SLC17A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_012434:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.

**Galactokinase Deficiency** - Gene: GALK1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000154:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Galactosemia** - Gene: GALT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000155:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

**Gamma-sarcoglycanopathy** - Gene: SGCG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000231:2-8. **Detection Rate:** Mixed or Other Caucasian 87%.

**Gaucher Disease** - Gene: GBA. Autosomal Recessive. Analysis of homologous regions. Variants (10): D409V, D448H, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L, p.L29Afs\*18. **Detection Rate:** Mixed or Other Caucasian 60%.

**GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness** - Gene: GJB2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004004:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

**GLB1-related Disorders** - Gene: GLB1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000404:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.

**GLDC-related Glycine Encephalopathy** - Gene: GLDC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000170:1-25. **Detection Rate:** Mixed or Other Caucasian 94%.

**Glutaric Acidemia, GCDH-related** - Gene: GCDH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000159:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Glycine Encephalopathy, AMT-related** - Gene: AMT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000481:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Glycogen Storage Disease Type Ia** - Gene: G6PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000151:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

**Glycogen Storage Disease Type Ib** - Gene: SLC37A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001164277 3-11. **Detection Rate:** Mixed or Other Caucasian >99%.

**Glycogen Storage Disease Type III** - Gene: AGL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000642:2-34. **Detection Rate:** Mixed or Other Caucasian >99%.



**GNE Myopathy** - Gene: GNE. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001128227:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**GNPTAB-related Disorders** - Gene: GNPTAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_024312:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.

**HADHA-related Disorders** - Gene: HADHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000182:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

**Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)** - Gene: HBB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000518:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Hereditary Fructose Intolerance** - Gene: ALDOB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000035:2-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Hexosaminidase A Deficiency (Including Tay-Sachs Disease)** - Gene: HEXA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000520:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**HMG-CoA Lyase Deficiency** - Gene: HMGCL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000191:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Holocarboxylase Synthetase Deficiency** - Gene: HLCS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000411:4-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Homocystinuria, CBS-related** - Gene: CBS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000071:3-17. **Detection Rate:** Mixed or Other Caucasian >99%.

**Hydrolethalus Syndrome** - Gene: HYL51. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_145014:4. **Detection Rate:** Mixed or Other Caucasian >99%.

**Hypophosphatasia** - Gene: ALPL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000478:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Isovaleric Acidemia** - Gene: IVD. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002225:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Joubert Syndrome 2** - Gene: TMEM216. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001173990:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

**Junctional Epidermolysis Bullosa, LAMA3-related** - Gene: LAMA3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000227:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.

**Junctional Epidermolysis Bullosa, LAMB3-related** - Gene: LAMB3. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000228:2-23. **Detection Rate:** Mixed or Other Caucasian >99%.

**Junctional Epidermolysis Bullosa, LAMC2-related** - Gene: LAMC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_005562:1-23. **Detection Rate:** Mixed or Other Caucasian >99%.

**Krabbe Disease** - Gene: GALC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000153:1-17. **Detection Rate:** Mixed or Other Caucasian >99%.

**Leigh Syndrome, French-Canadian Type** - Gene: LRPPRC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_133259:1-38. **Detection Rate:** Mixed or Other Caucasian >99%.

**Lipoid Congenital Adrenal Hyperplasia** - Gene: STAR. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000349:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.

**Lysosomal Acid Lipase Deficiency** - Gene: LIPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000235:2-10. **Detection Rate:** Mixed or Other Caucasian 98%.

**Maple Syrup Urine Disease Type Ia** - Gene: BCKDHA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000709:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Maple Syrup Urine Disease Type Ib** - Gene: BCKDHB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_183050:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

**Maple Syrup Urine Disease Type II** - Gene: DBT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001918:1-11. **Detection Rate:** Mixed or Other Caucasian 97%.

**Medium Chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADM. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000016:1-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Megalencephalic Leukoencephalopathy with Subcortical Cysts** - Gene: MLC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015166 2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**Metachromatic Leukodystrophy** - Gene: ARSA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000487:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Methylmalonic Acidemia, cblA Type** - Gene: MMAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_172250:2-7. **Detection Rate:** Mixed or Other Caucasian >99%.

**Methylmalonic Acidemia, cblB Type** - Gene: MMAB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_052845:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Methylmalonic Aciduria and Homocystinuria, cblC Type** - Gene: MMACHC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015506:1-4. **Detection Rate:** Mixed or Other Caucasian >99%.

**MKS1-related Disorders** - Gene: MKS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017777:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.

**Mucopolipidosis III Gamma** - Gene: GNPTG. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_032520:1-11. **Detection Rate:** Mixed or Other Caucasian 98%.

**Mucopolipidosis IV** - Gene: MCOLN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_020533:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Mucopolysaccharidosis Type I** - Gene: IDUA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000203:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Mucopolysaccharidosis Type II** - Gene: IDS. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000202:1-9. **Detection Rate:** Mixed or Other Caucasian 89%.

**Mucopolysaccharidosis Type IIIA** - Gene: SGSH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000199:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Mucopolysaccharidosis Type IIIB** - Gene: NAGLU. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000263:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Mucopolysaccharidosis Type IIIC** - Gene: HGSNAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_152419:1-18. **Detection Rate:** Mixed or Other Caucasian >99%.

**Muscular Dystrophy, LAMA2-related** - Gene: LAMA2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000426:1-43,45-65. **Detection Rate:** Mixed or Other Caucasian 98%.

**MUT-related Methylmalonic Acidemia** - Gene: MUT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000255:2-13. **Detection Rate:** Mixed or Other Caucasian >99%.

**MYO7A-related Disorders** - Gene: MYO7A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000260:2-49. **Detection Rate:** Mixed or Other Caucasian >99%.

**NEB-related Nemaline Myopathy** - Gene: NEB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001271208:3-80,117-183. **Detection Rate:** Mixed or Other Caucasian 92%.

**Nephrotic Syndrome, NPHS1-related** - Gene: NPHS1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004646:1-29. **Detection Rate:** Mixed or Other Caucasian >99%.

**Nephrotic Syndrome, NPHS2-related** - Gene: NPHS2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_014625:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Neuronal Ceroid Lipofuscinosis, CLN6-related** - Gene: CLN6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017882:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.

**Niemann-Pick Disease Type C1** - Gene: NPC1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000271:1-25. **Detection Rate:** Mixed or Other Caucasian >99%.

**Niemann-Pick Disease Type C2** - Gene: NPC2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_006432:1-5. **Detection Rate:** Mixed or Other Caucasian >99%.

**Niemann-Pick Disease, SMPD1-related** - Gene: SMPD1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000543:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Nijmegen Breakage Syndrome** - Gene: NBN. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_002485:1-16. **Detection Rate:** Mixed or Other Caucasian >99%.

**Ornithine Transcarbamylase Deficiency** - Gene: OTC. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000531:1-10. **Detection Rate:** Mixed or Other Caucasian 97%.

**PCCA-related Propionic Acidemia** - Gene: PCCA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000282:1-24. **Detection Rate:** Mixed or Other Caucasian 95%.

**PCCB-related Propionic Acidemia** - Gene: PCCB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000532:1-15. **Detection Rate:** Mixed or Other Caucasian >99%.

**PCDH15-related Disorders** - Gene: PCDH15. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_033056:2-33. **Detection Rate:** Mixed or Other Caucasian 93%.

**Pendred Syndrome** - Gene: SLC26A4. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000441:2-21. **Detection Rate:** Mixed or Other Caucasian >99%.

**Peroxisome Biogenesis Disorder Type 1** - Gene: PEX1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000466:1-24. **Detection Rate:** Mixed or Other Caucasian >99%.

**Peroxisome Biogenesis Disorder Type 3** - Gene: PEX12. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000286:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Peroxisome Biogenesis Disorder Type 4** - Gene: PEX6. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000287:1-17. **Detection Rate:** Mixed or Other Caucasian 97%.

**Peroxisome Biogenesis Disorder Type 5** - Gene: PEX2. Autosomal Recessive. Sequencing with copy number analysis. Exon: NM\_000318:4. **Detection Rate:** Mixed or Other Caucasian >99%.

**Peroxisome Biogenesis Disorder Type 6** - Gene: PEX10. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_153818:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Phenylalanine Hydroxylase Deficiency** - Gene: PAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000277:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.

**POMGNT-related Disorders** - Gene: POMGNT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_017739:2-22. **Detection Rate:** Mixed or Other Caucasian 96%.

**Pompe Disease** - Gene: GAA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000152:2-20. **Detection Rate:** Mixed or Other Caucasian 98%.

**PPT1-related Neuronal Ceroid Lipofuscinosis** - Gene: PPT1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000310:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Primary Carnitine Deficiency** - Gene: SLC22A5. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_003060:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

**Primary Hyperoxaluria Type 1** - Gene: AGXT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000030:1-11. **Detection Rate:** Mixed or Other Caucasian >99%.

**Primary Hyperoxaluria Type 2** - Gene: GRHRP. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_012203:1-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Primary Hyperoxaluria Type 3** - Gene: HOGA1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_138413:1-7. **Detection Rate:** Mixed or Other Caucasian >99%.

**Pycnodysostosis** - Gene: CTSK. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000396:2-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Pyruvate Carboxylase Deficiency** - Gene: PC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000920:3-22. **Detection Rate:** Mixed or Other Caucasian >99%.

**Rhizomelic Chondrodysplasia Punctata Type 1** - Gene: PEX7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000288:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

**RTEL1-related Disorders** - Gene: RTEL1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_032957:2-35. **Detection Rate:** Mixed or Other Caucasian >99%.

**Sandhoff Disease** - Gene: HEXB. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000521:1-14. **Detection Rate:** Mixed or Other Caucasian 98%.

**Short-chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADS. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000017:1-10. **Detection Rate:** Mixed or Other Caucasian >99%.

**Sjogren-Larsson Syndrome** - Gene: ALDH3A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000382:1-10. **Detection Rate:** Mixed or Other Caucasian 96%.

**SLC26A2-related Disorders** - Gene: SLC26A2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000112:2-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Smith-Lemli-Opitz Syndrome** - Gene: DHCR7. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001360:3-9. **Detection Rate:** Mixed or Other Caucasian >99%.

**Spastic Paraplegia Type 15** - Gene: ZFYVE26. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_015346:2-42. **Detection Rate:** Mixed or Other Caucasian >99%.

**Spinal Muscular Atrophy** - Gene: SMN1. Autosomal Recessive. Spinal muscular atrophy. Variant (1): SMN1 copy number. **Detection Rate:** Mixed or Other Caucasian 95%.

**Spondylothoracic Dysostosis** - Gene: MESP2. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_001039958:1-2. **Detection Rate:** Mixed or Other Caucasian >99%.

**TGM1-related Autosomal Recessive Congenital Ichthyosis** - Gene: TGM1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000359 2-15. **Detection Rate:** Mixed or Other Caucasian >99%.

**TPP1-related Neuronal Ceroid Lipofuscinosis** - Gene: TPP1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000391:1-13. **Detection Rate:** Mixed or Other Caucasian >99%.

**Tyrosine Hydroxylase Deficiency** - Gene: TH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_199292:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Tyrosinemia Type I** - Gene: FAH. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000137:1-14. **Detection Rate:** Mixed or Other Caucasian >99%.

**Tyrosinemia Type II** - Gene: TAT. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000353:2-12. **Detection Rate:** Mixed or Other Caucasian >99%.

**USH1C-related Disorders** - Gene: USH1C. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_005709:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
Attn: Jeffrey Olliffe  
NPI: 1306838271  
Report Date: 10/08/2021

MALE  
**DONOR 14270**  
DOB:  
Ethnicity: Mixed or Other  
Caucasian  
Barcode: 11004512827351

FEMALE  
N/A

**USH2A-related Disorders** - Gene: USH2A. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_206933:2-72. **Detection Rate:** Mixed or Other Caucasian 98%.

**Usher Syndrome Type 3** - Gene: CLRN1. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_174878:1-3. **Detection Rate:** Mixed or Other Caucasian >99%.

**Very-long-chain Acyl-CoA Dehydrogenase Deficiency** - Gene: ACADVL. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000018:1-20. **Detection Rate:** Mixed or Other Caucasian >99%.

**Wilson Disease** - Gene: ATP7B. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000053:1-21. **Detection Rate:** Mixed or Other Caucasian >99%.

**X-linked Adrenal Hypoplasia Congenita** - Gene: NROB1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000475:1-2. **Detection Rate:** Mixed or Other Caucasian 97%.

**X-linked Adrenoleukodystrophy** - Gene: ABCD1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000033:1-6. **Detection Rate:** Mixed or Other Caucasian 77%.

**X-linked Alport Syndrome** - Gene: COL4A5. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000495:1-51. **Detection Rate:** Mixed or Other Caucasian 96%.

**X-linked Juvenile Retinoschisis** - Gene: RS1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000330:1-6. **Detection Rate:** Mixed or Other Caucasian 98%.

**X-linked Myotubular Myopathy** - Gene: MTM1. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000252:2-15. **Detection Rate:** Mixed or Other Caucasian 96%.

**X-linked Severe Combined Immunodeficiency** - Gene: IL2RG. X-linked Recessive. Sequencing with copy number analysis. Exons: NM\_000206:1-8. **Detection Rate:** Mixed or Other Caucasian >99%.

**Xeroderma Pigmentosum Group A** - Gene: XPA. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_000380:1-6. **Detection Rate:** Mixed or Other Caucasian >99%.

**Xeroderma Pigmentosum Group C** - Gene: XPC. Autosomal Recessive. Sequencing with copy number analysis. Exons: NM\_004628:1-16. **Detection Rate:** Mixed or Other Caucasian 97%.

## Risk Calculations

Below are the risk calculations for all conditions tested. Negative results do not rule out the possibility of being a carrier. Residual risk is an estimate of each patient's post-test likelihood of being a carrier, while the reproductive risk represents an estimated likelihood that the patients' future children could inherit each disease. These risks are inherent to all carrier-screening tests, may vary by ethnicity, are predicated on a negative family history, and are present even given a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. In addition, average carrier rates are estimated using incidence or prevalence data from published scientific literature and/or reputable databases, where available, and are incorporated into residual risk calculations for each population/ethnicity. When population-specific data is not available for a condition, average worldwide incidence or prevalence is used. Further, incidence and prevalence data are only collected for the specified phenotypes (which include primarily the classic or severe forms of disease) and may not include alternate or milder disease manifestations associated with the gene. Actual incidence rates, prevalence rates, and carrier rates, and therefore actual residual risks, may be higher or lower than the estimates provided. Carrier rates, incidence/prevalence, and/or residual risks are not provided for some genes with biological or heritable properties that would make these estimates inaccurate. A '+' symbol indicates a positive result. See the full clinical report for interpretation and details. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

Disease	DONOR 14270 Residual Risk	Reproductive Risk
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 22,000	< 1 in 1,000,000
Alpha Thalassemia, HBA1/HBA2-related	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Andermann Syndrome	< 1 in 50,000	< 1 in 1,000,000
Argininemia	1 in 12,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 15,000	< 1 in 1,000,000
Aspartylglucosaminuria	< 1 in 50,000	< 1 in 1,000,000
Ataxia with Vitamin E Deficiency	< 1 in 50,000	< 1 in 1,000,000
Ataxia-telangiectasia	1 in 4,200	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 250,000
Autoimmune Polyglandular Syndrome Type 1	1 in 15,000	< 1 in 1,000,000
Autosomal Recessive Osteopetrosis Type 1	1 in 8,900	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related	1 in 8,100	< 1 in 1,000,000
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	< 1 in 44,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 32,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 42,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS12-related	< 1 in 50,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS2-related	< 1 in 50,000	< 1 in 1,000,000
BCS1L-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Beta-sarcoglycanopathy	1 in 39,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 13,000	1 in 650,000
Bloom Syndrome	< 1 in 50,000	< 1 in 1,000,000
Calpainopathy	1 in 13,000	< 1 in 1,000,000
Canavan Disease	1 in 9,700	< 1 in 1,000,000
Carbamoylphosphate Synthetase I Deficiency	< 1 in 57,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 50,000	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	1 in 25,000	< 1 in 1,000,000
Cartilage-hair Hypoplasia	< 1 in 50,000	< 1 in 1,000,000
Cerebrotendinous Xanthomatosis	1 in 11,000	< 1 in 1,000,000
Citrullinemia Type 1	1 in 14,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 8,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
CLN8-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 3,400	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 35,000	< 1 in 1,000,000
Combined Pituitary Hormone Deficiency, PROP1-related	1 in 6,100	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP11B1-related	1 in 8,400	< 1 in 1,000,000
Congenital Adrenal Hyperplasia, CYP21A2-related	1 in 1,300	1 in 280,000
Congenital Disorder of Glycosylation Type Ia	1 in 16,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation, MPI-related	< 1 in 50,000	< 1 in 1,000,000



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 10/08/2021

MALE  
**DONOR 14270**  
 DOB:  
 Ethnicity: Mixed or Other  
 Caucasian  
 Barcode: 11004512827351

FEMALE  
 N/A

Disease	DONOR 14270 Residual Risk	Reproductive Risk
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 3,000	1 in 360,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000
Delta-sarcoglycanopathy	< 1 in 13,000	< 1 in 1,000,000
Dihydroipoamide Dehydrogenase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Dysferlinopathy	1 in 11,000	< 1 in 1,000,000
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy)	Not calculated	Not calculated
ERCC6-related Disorders	1 in 8,500	< 1 in 1,000,000
ERCC8-related Disorders	< 1 in 16,000	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,800	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	1 in 9,800	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 220,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Hyperinsulinism, ABCC8-related	1 in 17,000	< 1 in 1,000,000
Familial Hyperinsulinism, KCNJ11-related	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	NM_000243.2(MEFV):c.2084A>G(K695R) heterozygote †	1 in 450
Fanconi Anemia Complementation Group A	1 in 2,800	< 1 in 1,000,000
Fanconi Anemia, FANCC-related	< 1 in 50,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 16,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Free Sialic Acid Storage Disorders	< 1 in 30,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 37,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,300	< 1 in 1,000,000
Gaucher Disease	1 in 260	1 in 110,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 2,500	1 in 260,000
GLB1-related Disorders	1 in 17,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	< 1 in 1,000,000
Glutaric Acidemia, GCDH-related	1 in 16,000	< 1 in 1,000,000
Glycine Encephalopathy, AMT-related	1 in 26,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ia	1 in 18,000	< 1 in 1,000,000
Glycogen Storage Disease Type Ib	1 in 35,000	< 1 in 1,000,000
Glycogen Storage Disease Type III	1 in 16,000	< 1 in 1,000,000
GNE Myopathy	1 in 23,000	< 1 in 1,000,000
GNPTAB-related Disorders	1 in 20,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 20,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 3,700	1 in 560,000
Hereditary Fructose Intolerance	1 in 7,900	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 30,000	< 1 in 1,000,000
HMG-CoA Lyase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria, CBS-related	1 in 9,400	< 1 in 1,000,000
Hydrolethals Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia	1 in 30,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 32,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMB3-related	1 in 32,000	< 1 in 1,000,000
Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 14,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	< 1 in 50,000	< 1 in 1,000,000
Lipoid Congenital Adrenal Hyperplasia	< 1 in 50,000	< 1 in 1,000,000
Lysosomal Acid Lipase Deficiency	1 in 14,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ib	1 in 39,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 16,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 4,400	1 in 790,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts	< 1 in 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 16,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	< 1 in 50,000	< 1 in 1,000,000





RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 10/08/2021

MALE  
**DONOR 14270**  
 DOB:  
 Ethnicity: Mixed or Other  
 Caucasian  
 Barcode: 11004512827351

FEMALE  
 N/A

Disease	DONOR 14270 Residual Risk	Reproductive Risk
Methylmalonic Acidemia, cblB Type	1 in 48,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 16,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucopolipidosis III Gamma	< 1 in 20,000	< 1 in 1,000,000
Mucopolipidosis IV	< 1 in 50,000	< 1 in 1,000,000
Mucopolysaccharidosis Type I	1 in 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type II	< 1 in 1,000,000	1 in 300,000
Mucopolysaccharidosis Type IIIA	1 in 19,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 27,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	< 1 in 50,000	< 1 in 1,000,000
Muscular Dystrophy, LAMA2-related	1 in 5,700	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 26,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	1 in 1,200	1 in 400,000
Nephrotic Syndrome, NPHS1-related	< 1 in 50,000	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Neuronal Ceroid Lipofuscinosis, CLN6-related	1 in 20,000	< 1 in 1,000,000
Niemann-Pick Disease Type C1	1 in 19,000	< 1 in 1,000,000
Niemann-Pick Disease Type C2	< 1 in 50,000	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-related	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Ornithine Transcarbamylase Deficiency	< 1 in 1,000,000	1 in 140,000
PCCA-related Propionic Acidemia	1 in 4,200	< 1 in 1,000,000
PCCB-related Propionic Acidemia	1 in 22,000	< 1 in 1,000,000
PCDH15-related Disorders	1 in 3,300	< 1 in 1,000,000
Pendred Syndrome	1 in 8,200	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 1	1 in 16,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 3	1 in 44,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 4	1 in 9,300	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 5	< 1 in 71,000	< 1 in 1,000,000
Peroxisome Biogenesis Disorder Type 6	< 1 in 50,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 4,800	1 in 940,000
POMGNT-related Disorders	< 1 in 12,000	< 1 in 1,000,000
Pompe Disease	1 in 4,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	1 in 7,700	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 11,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 17,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 50,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 3	1 in 13,000	< 1 in 1,000,000
Pycnodysostosis	1 in 43,000	< 1 in 1,000,000
Pyruvate Carboxylase Deficiency	1 in 25,000	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
RTEL1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Sandhoff Disease	1 in 18,000	< 1 in 1,000,000
Short-chain Acyl-CoA Dehydrogenase Deficiency	1 in 11,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	< 1 in 12,000	< 1 in 1,000,000
SLC26A2-related Disorders	1 in 16,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 9,400	< 1 in 1,000,000
Spastic Paraplegia Type 15	< 1 in 50,000	< 1 in 1,000,000
Spinal Muscular Atrophy	Negative for g.27134T>G SNP SMN1: 2 copies 1 in 770	1 in 110,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
TGM1-related Autosomal Recessive Congenital Ichthyosis	1 in 22,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 30,000	< 1 in 1,000,000
Tyrosine Hydroxylase Deficiency	< 1 in 50,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 16,000	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 30,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 4,100	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 41,000	< 1 in 1,000,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency	1 in 18,000	< 1 in 1,000,000



RESULTS RECIPIENT  
**SEATTLE SPERM BANK**  
 Attn: Jeffrey Olliffe  
 NPI: 1306838271  
 Report Date: 10/08/2021

MALE  
**DONOR 14270**  
 DOB:  
 Ethnicity: Mixed or Other  
 Caucasian  
 Barcode: 11004512827351

FEMALE  
 N/A

Disease	DONOR 14270 Residual Risk	Reproductive Risk
Wilson Disease	1 in 6,500	< 1 in 1,000,000
X-linked Adrenal Hypoplasia Congenita	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 40,000
X-linked Myotubular Myopathy	Not calculated	Not calculated
X-linked Severe Combined Immunodeficiency	< 1 in 1,000,000	1 in 200,000
Xeroderma Pigmentosum Group A	< 1 in 50,000	< 1 in 1,000,000
Xeroderma Pigmentosum Group C	1 in 7,300	< 1 in 1,000,000