

Client/Sending Facility:
Phoenix Sperm Bank

1492 S Mill Ave Suite 306
Tempe, AZ 85281
Ph: (602)888-7255
AZB-45

LCLS Specimen Number: 247-944-0047-0
Patient Name: 10076, DONOR
Date of Birth: [REDACTED]
Gender: M
Patient ID:
Lab Number: YU16-70597 L
Indications: NOT GIVEN

Account Number: [REDACTED]
Ordering Physician: J OLLIFFE
Specimen Type: BLOOD
Client Reference:
Date Collected: 09/03/2016
Date Received: 09/07/2016
Date Reported: 09/15/2016

Test: Chromosome, Blood, Routine

Cells Counted: 20
Cells Analyzed: 20

Cells Karyotyped: 2
Band Resolution: 500

CYTOGENETIC RESULT: 46,XY

INTERPRETATION: NORMAL MALE KARYOTYPE

Cytogenetic analysis of PHA stimulated cultures has revealed a MALE karyotype with an apparently normal GTG banding pattern in all cells observed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.



LabCorp Specialty Testing Group

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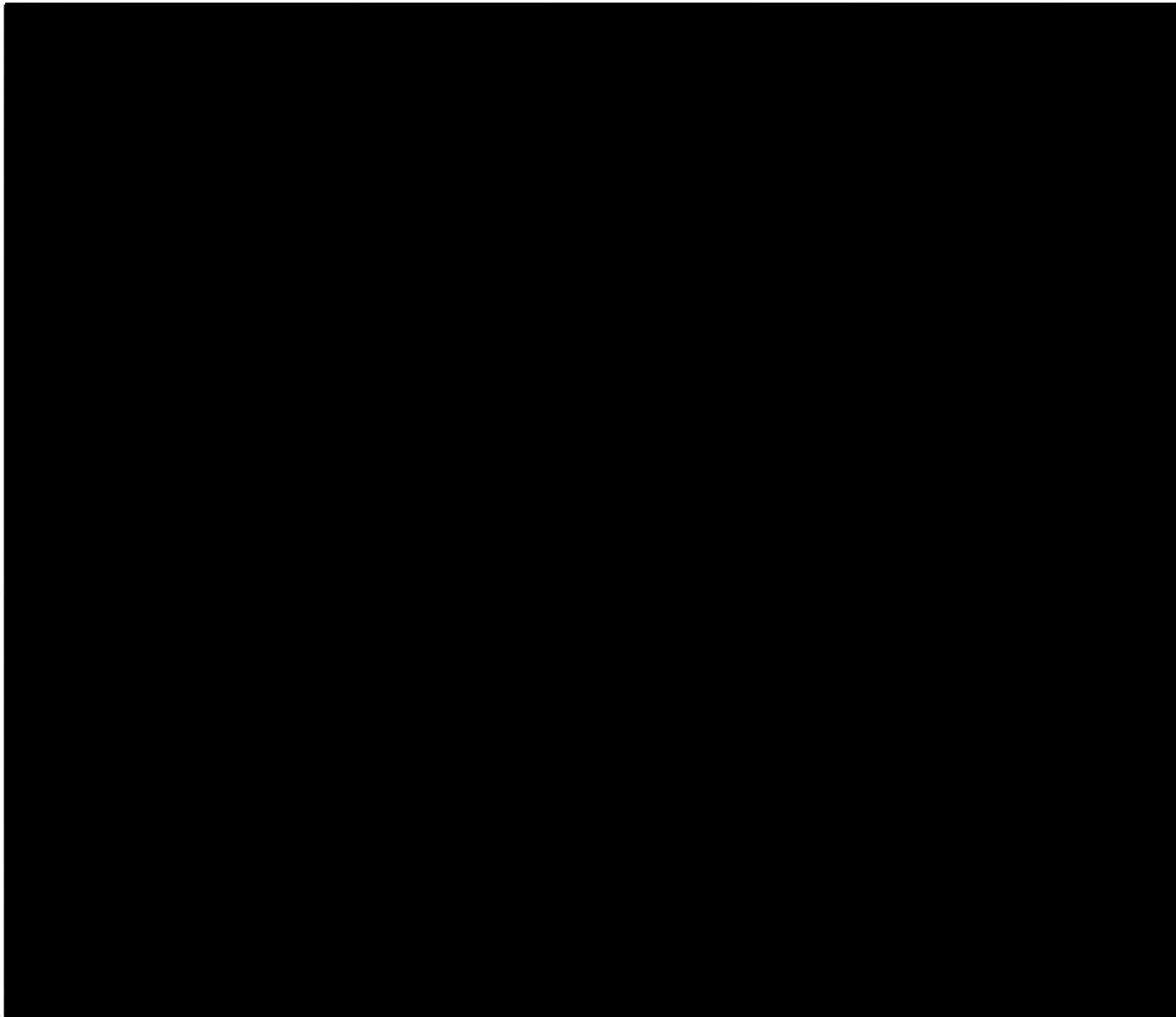
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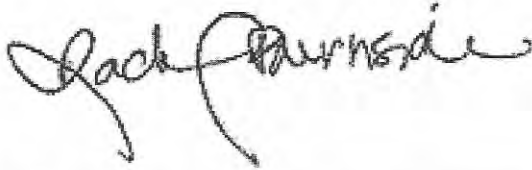
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Rachel Burnside, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive, RTP, NC, 27709-0153 (800) 345-4363

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD.
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

This document contains private and confidential health information **protected by state and federal law.**



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Dr. Jeffrey Olliffe
 4915 25th Ave NE, Suite 204W
 Seattle, WA 98105
 Phone: (206) 588-1484
 Fax: (206) 588-1484
 NPI: 1306838271
 Report Date: 09/12/2016

MALE
DONOR 10076
 DOB: [REDACTED]
 Ethnicity: Mixed or Other
 Caucasian
 Sample Type: EDTA Blood
 Date of Collection: 09/03/2016
 Date Received: 09/06/2016
 Date Tested: 09/12/2016
 Barcode: 11200059662978
 Indication: Egg or sperm donor

FEMALE
 N/A

Family Prep Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The Counsyl Family Prep Screen (version 2.0) 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 10076	Partner
Panel Information	Family Prep Screen 2.0 Universal Panel Minus X-Linked (102 conditions tested)	N/A
POSITIVE: CARRIER Polyglandular Autoimmune Syndrome Type 1 Reproductive Risk: 1 in 570 Inheritance: Autosomal Recessive	⊕ CARRIER* NM_000383.3(AIRE):c.967_979del13 (aka L323Sfs*51) 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".

*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 6.

CLINICAL NOTES

- None

NEXT STEPS

- Carrier testing should be considered for the diseases specified above for the patient's partner, as both parents must be carriers before a child is at high risk of developing the disease.
- Genetic counseling is recommended and patients may wish to discuss any positive results with blood relatives, as there is an increased chance that they are also carriers.



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FEMALE
N/A

POSITIVE: CARRIER

Polyglandular Autoimmune Syndrome Type 1

Reproductive risk: 1 in 570
Risk before testing: 1 in 80,000

Gene: AIRE | Inheritance Pattern: Autosomal Recessive

Patient	DONOR 10076	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000383.3(AIRE):c.967_979del13(aka L323Sfs*51) heterozygote	N/A
Methodology	Sequencing	N/A
Interpretation	This individual is a carrier of polyglandular autoimmune syndrome type 1. Carriers generally do not experience symptoms.	N/A
Detection rate	> 65%	N/A
Exons tested	NM_000383:1-14.	N/A

What is Polyglandular Autoimmune Syndrome Type 1?

Polyglandular Autoimmune Syndrome Type 1 (PAS-1) is an inherited disease in which the body's immune system mistakenly attacks healthy cells, notably the glands that produce the body's hormones. People with PAS-1 have at least two of the disease's main symptoms: fungal infections of the skin and mucous membranes, decreased function in the parathyroid glands, and decreased function in the adrenal glands (Addison disease). Many people with the disease have all three main symptoms. There are also numerous and diverse other symptoms which can occur.

In the majority of people with PAS-1, the first symptom to appear is recurrent and persistent fungal infections of the skin and mucous membranes, such as the in the moist lining of the nose and mouth. These infections typically begin between the ages of 3 and 5, although they can occur any time before one's 30s.

Frequently the second symptom of the disease to appear is an underactive parathyroid gland (hypoparathyroidism). This typically occurs before the age of 10. The parathyroid glands normally secrete a hormone used to regulate the amount of calcium and phosphorous in the bone and blood. An underactive parathyroid gland can cause numerous symptoms including tingling in the lips, fingers, and toes; muscle cramps; pain in the abdomen, face, legs, and feet; weakness or fatigue; and dry hair and skin.

Often the third symptom to appear is underactive adrenal glands, a condition known as Addison disease. This disease typically appears before the age of 15. Addison disease can cause fatigue, muscle weakness, weight loss, low blood pressure, and changes in skin coloration.

There are numerous other symptoms which can also occur in people with PAS-1. These include chronic liver disease, extreme fatigue due to a problem with red blood cells, skin disease, total body hair loss, an underactive pituitary gland, abnormalities in the ovaries and testes, diarrhea, difficulty absorbing nutrients from food, and eye problems, among others.



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The most common pattern with PAS-1 is that the three main symptoms of the disease—fungal infections of the skin, underactive parathyroid gland, and Addison disease—develop in that order before the age of 20. The other symptoms associated with the disease may then begin sporadically over time until one's 50s, when the symptoms typically stabilize. This does not hold true for all people with PAS-1, however. Generally speaking, the earlier in life that the main symptoms appear, the more likely it is that additional symptoms will develop.

How common is Polyglandular Autoimmune Syndrome Type 1?

PAS-1 is a rare condition in the United States, but it is more common among certain ethnic groups. The number of people affected include:

- Iranian Jews: 1 in 6,500 to 9,000
- Sardinians: 1 in 14,000
- Finns: 1 in 25,000
- Slovenians: 1 in 43,000
- Norwegians: 1 in 80,000 to 90,000
- Poles: 1 in 129,000

How is Polyglandular Autoimmune Syndrome Type 1 treated?

There is no cure for PAS-1. Each symptom must be treated as it arises and lifelong regular checkups are necessary to look for any new symptoms. It is important to discover and treat new symptoms as soon as possible to prevent permanent damage to the body.

Physicians often prescribe hormone replacement therapy or intravenous steroids for people with PAS-1. Calcium and vitamin D are often helpful to treat an underactive parathyroid gland. Fungal infections can be treated with medication.

Other symptoms are treated as they arise. For example, people with diabetes can take insulin and monitor their diet.

What is the prognosis for a person with Polyglandular Autoimmune Syndrome Type 1?

The prognosis for a person with PAS-1 varies depending on the number and severity of his or her symptoms. Early detection of the disease and its component symptoms is important for preventing life-threatening scenarios. With careful monitoring, it is possible to have a normal or near-normal lifespan. Women with PAS-1 can give birth, and men with PAS-1 can father healthy children.



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Methods and Limitations

DONOR 10076 [Family Prep Screen 2.0]: sequencing, targeted genotyping, copy number analysis, and analysis of homologous regions.

Sequencing

High-throughput sequencing is used to analyze the listed exons, as well as selected intergenic and intronic regions, of the genes in the Conditions Tested section of the report. These regions are sequenced to high coverage and the sequences are compared to standards and references of normal variation. Mutations may not be detected in areas of lower sequence coverage. On average, more than 99% of all bases in the exons listed for each gene are sequenced at the minimum read depth. Variants discovered in other exons of these genes will also be reported if they meet quality control criteria. Triplet repeats and large deletions and duplications may not be detected. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes are not well analyzed by this method.

High-throughput sequencing detects, on average, 94% of known clinically significant variants. Disease-specific detection rates and residual risks are reported as "greater than (>)" and "less than (<)" the values for targeted genotyping, respectively. More precise values are not currently available, but may become available in the future.

All variants that are a recognized cause of the disease will be reported. In addition, variants that have not previously been established as a recognized cause of disease may be identified. In these cases, only variants classified as "predicted" or "likely" pathogenic are reported. Predicted/likely pathogenic variants are described elsewhere in the report as "predicted/likely to have a negative impact on gene function". In general, predicted pathogenic variants are those which are predicted to be pathogenic based on the nature of the sequence change, while likely pathogenic variants are evaluated by reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Literature citations validating reported variants are available upon request.

Targeted genotyping

Targeted DNA mutation analysis is used to determine the genotypes of the listed variants in the Conditions Tested section of the report. The test is not validated for detection of homozygous mutations, and although rare, asymptomatic individuals affected by the disease may not be genotyped accurately.

Copy number analysis

Targeted copy number analysis is used to determine the copy number of exon 7 of the *SMN1* gene relative to other genes. Other mutations may interfere with this analysis. Some individuals with two copies of *SMN1* are carriers with two *SMN1* genes on one chromosome and a *SMN1* deletion on the other chromosome. In addition, a small percentage of spinal muscular atrophy (SMA) cases are caused by nondeletion mutations in the *SMN1* gene. Thus, a test result of two *SMN1* copies significantly reduces the risk of being a carrier; however, there is still a residual risk of being a carrier and subsequently a small risk of future affected offspring for individuals with two or more *SMN1* gene copies. Some SMA cases arise as the result of *de novo* mutation events which will not be detected by carrier testing.

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 mutations in certain genes that have homology to other regions. The precise breakpoints of large deletions in these regions cannot be determined, but are estimated from copy number analysis. Patients who have one or more additional copies of the *CYP21A2* gene and a loss of function mutation may not actually be a carrier of 21-hydroxylase-deficient congenital adrenal hyperplasia (CAH). In addition, some individuals with four alpha globin genes are carriers with three genes on one chromosome and a deletion on the other chromosome. This and similar carrier states, where complementary changes exist in both the gene and a pseudogene, may not be detected by the assay.



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Because the true incidence of non-classic CAH is unknown, the residual carrier and reproductive risk numbers on the report are only based on published incidences 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 of overall chances for 21-hydroxylase-deficient CAH, especially in the aforementioned populations, as they do not account for non-classic CAH.

Limitations

In an unknown number of cases, nearby genetic variants may interfere with mutation detection. Other possible sources of diagnostic error include sample mix-up, trace contamination, bone marrow transplantation, blood transfusions and technical errors. 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. The Family Prep Screen does not fully address all inherited forms of intellectual disability, birth defects and genetic disease. A family history of any of these conditions may warrant additional evaluation. Furthermore, not all mutations will be identified in the genes analyzed and additional testing may be beneficial for some patients. For example, individuals of African, Southeast Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies, which can be identified by CBC and hemoglobin electrophoresis or HPLC (*ACOG Practice Bulletin No. 78. Obstet. Gynecol. 2007;109:229-37*), and additional Tay-Sachs disease testing can be performed using a biochemical assay (*Gross et al. Genet. Med. 2008;10(1):54-56*).

This test was developed and its performance characteristics determined by Counsyl, 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**.

LAB DIRECTORS

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP



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Conditions Tested

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia - Gene: CYP21A2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** CYP21A2 deletion, CYP21A2 duplication, CYP21A2 triplication, G111VfsX21, I173N, L308FfsX6, P31L, Q319*, Q319*+CYP21A2dup, R357W, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** Mixed or Other Caucasian 96%.

ABCC8-related Hyperinsulinism - Gene: ABCC8. Autosomal Recessive. Sequencing. **Exons:** NM_000352:1-39. **Detection Rate:** Unknown due to rarity of disease.

Achromatopsia - Gene: CNGB3. Autosomal Recessive. Sequencing. **Exons:** NM_019098:1-18. **Detection Rate:** Mixed or Other Caucasian > 62%.

Alkaptonuria - Gene: HGD. Autosomal Recessive. Sequencing. **Exons:** NM_000187:1-14. **Detection Rate:** Mixed or Other Caucasian > 80%.

Alpha Thalassemia - Genes: HBA1, HBA2. Autosomal Recessive. Analysis of Homologous Regions. **Variants (13):** -(alpha)20.5, --BRIT, --MEDI, --MEDII, --SEA, --THAI or --FIL, -alpha3.7, -alpha4.2, HBA1+HBA2 deletion, Hb Constant Spring, anti3.7, anti4.2, del HS-40. **Detection Rate:** Unknown due to rarity of disease.

Alpha-1 Antitrypsin Deficiency - Gene: SERPINA1. Autosomal Recessive. Sequencing. **Exons:** NM_000295:2-5. **Detection Rate:** Mixed or Other Caucasian > 95%.

Alpha-Mannosidosis - Gene: MAN2B1. Autosomal Recessive. Sequencing. **Exons:** NM_000528:1-15,17-24. **Detection Rate:** Mixed or Other Caucasian > 32%.

Andermann Syndrome - Gene: SLC12A6. Autosomal Recessive. Sequencing. **Exons:** NM_133647:1-25. **Detection Rate:** Unknown due to rarity of disease.

ARSACS - Gene: SACS. Autosomal Recessive. Sequencing. **Exons:** NM_014363:2-10. **Detection Rate:** Unknown due to rarity of disease.

Aspartylglycosaminuria - Gene: AGA. Autosomal Recessive. Sequencing. **Exons:** NM_000027:1-9. **Detection Rate:** Unknown due to rarity of disease.

Ataxia With Vitamin E Deficiency - Gene: TTPA. Autosomal Recessive. Sequencing. **Exons:** NM_000370:1-5. **Detection Rate:** Mixed or Other Caucasian > 10%.

Ataxia-Telangiectasia - Gene: ATM. Autosomal Recessive. Sequencing. **Exons:** NM_000051:2-63. **Detection Rate:** Mixed or Other Caucasian > 65%.

Autosomal Recessive Polycystic Kidney Disease - Gene: PKHD1. Autosomal Recessive. Sequencing. **Exons:** NM_138694:2-67. **Detection Rate:** Mixed or Other Caucasian > 18%.

Bardet-Biedl Syndrome, BBS1-related - Gene: BBS1. Autosomal Recessive. Sequencing. **Exons:** NM_024649:1-17. **Detection Rate:** Mixed or Other Caucasian > 79%.

Bardet-Biedl Syndrome, BBS10-related - Gene: BBS10. Autosomal Recessive. Sequencing. **Exons:** NM_024685:1-2. **Detection Rate:** Mixed or Other Caucasian > 46%.

Biotinidase Deficiency - Gene: BTD. Autosomal Recessive. Sequencing. **Exons:** NM_000060:1-4. **Detection Rate:** Mixed or Other Caucasian > 45%.

Bloom Syndrome - Gene: BLM. Autosomal Recessive. Sequencing. **Exons:** NM_000057:2-22. **Detection Rate:** Mixed or Other Caucasian > 10%.

Canavan Disease - Gene: ASPA. Autosomal Recessive. Sequencing. **Exons:** NM_000049:1-6. **Detection Rate:** Mixed or Other Caucasian > 53%.

Carnitine Palmitoyltransferase IA Deficiency - Gene: CPT1A. Autosomal Recessive. Sequencing. **Exons:** NM_001876:2-19. **Detection Rate:** Mixed or Other Caucasian > 10%.

Carnitine Palmitoyltransferase II Deficiency - Gene: CPT2. Autosomal Recessive. Sequencing. **Exons:** NM_000098:1-5. **Detection Rate:** Mixed or Other Caucasian > 80%.

Cartilage-Hair Hypoplasia - Gene: RMRP. Autosomal Recessive. Sequencing. **Exon:** NR_003051:1. **Detection Rate:** Mixed or Other Caucasian > 48%.

Citrullinemia Type 1 - Gene: ASS1. Autosomal Recessive. Sequencing. **Exons:** NM_000050:3-16. **Detection Rate:** Mixed or Other Caucasian > 20%.

CLN3-related Neuronal Ceroid Lipofuscinosis - Gene: CLN3. Autosomal Recessive. Sequencing. **Exons:** NM_001042432:2-16. **Detection Rate:** Mixed or Other Caucasian > 96%.

CLN5-related Neuronal Ceroid Lipofuscinosis - Gene: CLN5. Autosomal Recessive. Sequencing. **Exons:** NM_006493:1-4. **Detection Rate:** Unknown due to rarity of disease.

Cohen Syndrome - Gene: VPS13B. Autosomal Recessive. Sequencing. **Exons:** NM_017890:2-62. **Detection Rate:** Unknown due to rarity of disease.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Autosomal Recessive. Sequencing. **Exons:** NM_000303:1-8. **Detection Rate:** Mixed or Other Caucasian > 72%.

Congenital Disorder of Glycosylation Type Ib - Gene: MPI. Autosomal Recessive. Sequencing. **Exons:** NM_002435:1-8. **Detection Rate:** Unknown due to rarity of disease.

Congenital Finnish Nephrosis - Gene: NPHS1. Autosomal Recessive. Sequencing. **Exons:** NM_004646:2-23,26-27,29. **Detection Rate:** Unknown due to rarity of disease.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing. **Exons:** NM_025136:1-2. **Detection Rate:** Unknown due to rarity of disease.

Cystic Fibrosis - Gene: CFTR. Autosomal Recessive. Sequencing. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** Mixed or Other Caucasian > 91%.

Cystinosis - Gene: CTNS. Autosomal Recessive. Sequencing. **Exons:** NM_004937:3-12. **Detection Rate:** Mixed or Other Caucasian > 67%.

D-Bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing. **Exons:** NM_000414:1-24. **Detection Rate:** Mixed or Other Caucasian > 35%.

Factor XI Deficiency - Gene: F11. Autosomal Recessive. Sequencing. **Exons:** NM_000128:2-15. **Detection Rate:** Mixed or Other Caucasian > 10%.

Familial Dysautonomia - Gene: IKBKAP. Autosomal Recessive. Sequencing. **Exons:** NM_003640:19-20,26. **Detection Rate:** Unknown due to rarity of disease.

Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing. **Exons:** NM_000243:1-10. **Detection Rate:** Unknown due to rarity of disease.

Fanconi Anemia Type C - Gene: FANCC. Autosomal Recessive. Sequencing. **Exons:** NM_000136:2-15. **Detection Rate:** Mixed or Other Caucasian > 54%.

Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing. **Exons:** NM_000155:1-11. **Detection Rate:** Mixed or Other Caucasian > 80%.

Gaucher Disease - Gene: GBA. Autosomal Recessive. Targeted Genotyping. **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. **Exons:** NM_004004:1-2. **Detection Rate:** Mixed or Other Caucasian > 79%.

Glutaric Acidemia Type 1 - Gene: GCDH. Autosomal Recessive. Sequencing. **Exons:** NM_000159:2-12. **Detection Rate:** Mixed or Other Caucasian > 40%.

Glycogen Storage Disease Type Ia - Gene: G6PC. Autosomal Recessive. Sequencing. **Exons:** NM_000151:1-5. **Detection Rate:** Mixed or Other Caucasian > 61%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing. **Exons:** NM_001164277:3-11. **Detection Rate:** Mixed or Other Caucasian > 46%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing. **Exons:** NM_000642:2-34. **Detection Rate:** Mixed or Other Caucasian > 45%.

Glycogen Storage Disease Type V - Gene: PYGM. Autosomal Recessive. Sequencing. **Exons:** NM_005609:1-20. **Detection Rate:** Mixed or Other Caucasian > 80%.

GRACILE Syndrome - Gene: BCS1L. Autosomal Recessive. Sequencing. **Exons:** NM_004328:3-9. **Detection Rate:** Unknown due to rarity of disease.

Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) - Gene: HBB. Autosomal Recessive. Sequencing. **Exons:** NM_000518:1-3. **Detection Rate:** Mixed or Other Caucasian > 83%.

Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing. **Exons:** NM_000035:2-9. **Detection Rate:** Mixed or Other Caucasian > 75%.

Hereditary Thymine-Uraciluria - Gene: DPYD. Autosomal Recessive. Sequencing. **Exons:** NM_000110:1-23. **Detection Rate:** Mixed or Other Caucasian > 52%.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-related - Gene: LAMA3. Autosomal Recessive. Sequencing. **Exons:** NM_000227:1-16,18-38. **Detection Rate:** Mixed or Other Caucasian > 10%.

Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - Gene: LAMB3. Autosomal Recessive. Sequencing. **Exons:** NM_000228:2-23. **Detection Rate:** Mixed or Other Caucasian > 48%.



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FEMALE
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Herlitz Junctional Epidermolysis Bullosa, LAMC2-related - Gene: LAMC2. Autosomal Recessive. Sequencing. Exons: NM_005562:1-23. **Detection Rate:** Unknown due to rarity of disease.

Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Autosomal Recessive. Sequencing. Exons: NM_000520:1-14. **Detection Rate:** Mixed or Other Caucasian > 23%.

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency - Gene: CBS. Autosomal Recessive. Sequencing. Exons: NM_000071:3-17. **Detection Rate:** Mixed or Other Caucasian > 14%.

Hurler Syndrome - Gene: IDUA. Autosomal Recessive. Targeted Genotyping. **Variants (2):** Q70*, W402*. **Detection Rate:** Mixed or Other Caucasian 67%.

Hypophosphatasia, Autosomal Recessive - Gene: ALPL. Autosomal Recessive. Sequencing. Exons: NM_000478:2-12. **Detection Rate:** Mixed or Other Caucasian > 30%.

Inclusion Body Myopathy 2 - Gene: GNE. Autosomal Recessive. Sequencing. Exons: NM_001128227:3-12. **Detection Rate:** Unknown due to rarity of disease.

Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing. Exons: NM_002225:1-12. **Detection Rate:** Mixed or Other Caucasian > 47%.

Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing. Exons: NM_001173990:1-5. **Detection Rate:** Unknown due to rarity of disease.

Krabbe Disease - Gene: GALC. Autosomal Recessive. Sequencing. Exons: NM_000153:1-17. **Detection Rate:** Mixed or Other Caucasian > 58%.

Limb-Girdle Muscular Dystrophy Type 2D - Gene: SGCA. Autosomal Recessive. Sequencing. Exons: NM_000023:1-9. **Detection Rate:** Mixed or Other Caucasian > 32%.

Limb-Girdle Muscular Dystrophy Type 2E - Gene: SGCB. Autosomal Recessive. Sequencing. Exons: NM_000232:1-6. **Detection Rate:** Mixed or Other Caucasian > 12%.

Lipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing. Exons: NM_000108:1-14. **Detection Rate:** Unknown due to rarity of disease.

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency - Gene: HADHA. Autosomal Recessive. Sequencing. Exons: NM_000182:1-20. **Detection Rate:** Mixed or Other Caucasian > 87%.

Maple Syrup Urine Disease Type 1B - Gene: BCKDHB. Autosomal Recessive. Sequencing. Exons: NM_183050:1-10. **Detection Rate:** Unknown due to rarity of disease.

Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. Autosomal Recessive. Sequencing. Exons: NM_000016:1-12. **Detection Rate:** Mixed or Other Caucasian > 78%.

Megalencephalic Leukoencephalopathy With Subcortical Cysts - Gene: MLC1. Autosomal Recessive. Sequencing. Exons: NM_015166:2-12. **Detection Rate:** Mixed or Other Caucasian > 13%.

Metachromatic Leukodystrophy - Gene: ARSA. Autosomal Recessive. Sequencing. Exons: NM_000487:1-8. **Detection Rate:** Mixed or Other Caucasian > 53%.

Mucopolidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing. Exons: NM_020533:1-14. **Detection Rate:** Mixed or Other Caucasian > 10%.

Muscle-Eye-Brain Disease - Gene: POMGNT1. Autosomal Recessive. Sequencing. Exons: NM_017739:2-22. **Detection Rate:** Mixed or Other Caucasian > 75%.

NEB-related Nemaline Myopathy - Gene: NEB. Autosomal Recessive. Sequencing. Exons: NM_004543:7-8,18,25,28,33,36,45,48,54-55,58,61,71,73-74,91,94,101,111-112,114,118-119,122-123,127,129,132-135,138,140,143,146-147. **Detection Rate:** Unknown due to rarity of disease.

Niemann-Pick Disease Type C - Gene: NPC1. Autosomal Recessive. Sequencing. Exons: NM_000271:1-25. **Detection Rate:** Mixed or Other Caucasian > 17%.

Niemann-Pick Disease, SMPD1-associated - Gene: SMPD1. Autosomal Recessive. Sequencing. Exons: NM_000543:1-6. **Detection Rate:** Mixed or Other Caucasian > 38%.

Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing. Exons: NM_002485:1-16. **Detection Rate:** Mixed or Other Caucasian > 78%.

Northern Epilepsy - Gene: CLN8. Autosomal Recessive. Sequencing. Exons: NM_018941:2-3. **Detection Rate:** Unknown due to rarity of disease.

Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing. Exons: NM_000441:2-21. **Detection Rate:** Mixed or Other Caucasian > 69%.

PEX1-related Zellweger Syndrome Spectrum - Gene: PEX1. Autosomal Recessive. Sequencing. Exons: NM_000466:1-24. **Detection Rate:** Mixed or Other Caucasian > 68%.

Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing. Exons: NM_000277:1-13. **Detection Rate:** Mixed or Other Caucasian > 43%.

Polyglandular Autoimmune Syndrome Type 1 - Gene: AIRE. Autosomal Recessive. Sequencing. Exons: NM_000383:1-14. **Detection Rate:** Mixed or Other Caucasian > 65%.

Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing. Exons: NM_000152:2-20. **Detection Rate:** Mixed or Other Caucasian > 67%.

PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing. Exons: NM_000310:1-9. **Detection Rate:** Mixed or Other Caucasian > 53%.

Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing. Exons: NM_003060:1-10. **Detection Rate:** Unknown due to rarity of disease.

Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing. Exons: NM_000030:1-11. **Detection Rate:** Mixed or Other Caucasian > 42%.

Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing. Exons: NM_012203:1-9. **Detection Rate:** Mixed or Other Caucasian > 37%.

PROPI-related Combined Pituitary Hormone Deficiency - Gene: PROPI. Autosomal Recessive. Sequencing. Exons: NM_006261:1-3. **Detection Rate:** Mixed or Other Caucasian > 55%.

Pseudocholinesterase Deficiency - Gene: BCHE. Autosomal Recessive. Sequencing. Exons: NM_000055:2-4. **Detection Rate:** Mixed or Other Caucasian > 83%.

Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing. Exons: NM_000396:2-8. **Detection Rate:** Mixed or Other Caucasian > 10%.

Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing. Exons: NM_000288:1-10. **Detection Rate:** Mixed or Other Caucasian > 70%.

Salla Disease - Gene: SLC17A5. Autosomal Recessive. Sequencing. Exons: NM_012434:1-11. **Detection Rate:** Unknown due to rarity of disease.

Segawa Syndrome - Gene: TH. Autosomal Recessive. Sequencing. Exons: NM_000360:1-13. **Detection Rate:** Mixed or Other Caucasian > 10%.

Short Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing. Exons: NM_000017:1-10. **Detection Rate:** Unknown due to rarity of disease.

Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing. Exons: NM_000382:1-10. **Detection Rate:** Mixed or Other Caucasian > 24%.

Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing. Exons: NM_001360:3-9. **Detection Rate:** Mixed or Other Caucasian > 69%.

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

Steroid-Resistant Nephrotic Syndrome - Gene: NPHS2. Autosomal Recessive. Sequencing. Exons: NM_014625:1-8. **Detection Rate:** Mixed or Other Caucasian > 33%.

Sulfate Transporter-Related Osteochondrodysplasia - Gene: SLC26A2. Autosomal Recessive. Sequencing. Exons: NM_000112:2-3. **Detection Rate:** Mixed or Other Caucasian > 75%.

TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing. Exons: NM_000391:1-13. **Detection Rate:** Mixed or Other Caucasian > 60%.

Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing. Exons: NM_000137:1-14. **Detection Rate:** Mixed or Other Caucasian > 50%.

Usher Syndrome Type 1F - Gene: PCDH15. Autosomal Recessive. Sequencing. Exons: NM_033056:2-33. **Detection Rate:** Unknown due to rarity of disease.

Usher Syndrome Type 3 - Gene: CLRN1. Autosomal Recessive. Sequencing. Exons: NM_174878:1-3. **Detection Rate:** Unknown due to rarity of disease.

Very Long Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing. Exons: NM_000018:1-20. **Detection Rate:** Mixed or Other Caucasian > 20%.

Walker-Warburg Syndrome - Gene: FKTN. Autosomal Recessive. Sequencing. Exons: NM_001079802:3-11. **Detection Rate:** Unknown due to rarity of disease.

Wilson Disease - Gene: ATP7B. Autosomal Recessive. Sequencing. Exons: NM_000053:1-21. **Detection Rate:** Mixed or Other Caucasian > 40%.



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Dr. Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 09/12/2016

MALE
DONOR 10076
 DOB: [REDACTED]
 Ethnicity: Mixed or Other
 Caucasian
 Barcode: 11200059662978

FEMALE
 N/A

Risk Calculations

Below are the risk calculations for all conditions tested. Since negative results do not completely rule out the possibility of being a carrier, the **residual risk** represents the patient's post-test likelihood of being a carrier and the **reproductive risk** represents the likelihood the patient's 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 after a negative test result. Inaccurate reporting of ethnicity may cause errors in risk calculation. The reproductive risk presented is based on a hypothetical pairing with a partner of the same ethnic group.

†Indicates a positive result. See the full clinical report for interpretation and details.

Disease	DONOR 10076 Residual Risk	Reproductive Risk
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
ABCC8-related Hyperinsulinism	< 1 in 110	< 1 in 50,000
Achromatopsia	< 1 in 230	< 1 in 79,000
Alkaptonuria	< 1 in 500	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-1 Antitrypsin Deficiency	< 1 in 680	< 1 in 93,000
Alpha-Mannosidosis	< 1 in 520	< 1 in 730,000
Andermann Syndrome	< 1 in 500	< 1 in 1,000,000
ARSACS	< 1 in 500	< 1 in 1,000,000
Aspartylglycosaminuria	< 1 in 500	< 1 in 1,000,000
Ataxia With Vitamin E Deficiency	< 1 in 500	< 1 in 1,000,000
Ataxia-Telangiectasia	< 1 in 450	< 1 in 290,000
Autosomal Recessive Polycystic Kidney Disease	< 1 in 75	< 1 in 18,000
Bardet-Biedl Syndrome, BBS1-related	< 1 in 750	< 1 in 480,000
Bardet-Biedl Syndrome, BBS10-related	< 1 in 290	< 1 in 180,000
Biotinidase Deficiency	< 1 in 220	< 1 in 110,000
Bloom Syndrome	< 1 in 500	< 1 in 1,000,000
Canavan Disease	< 1 in 500	< 1 in 1,000,000
Carnitine Palmitoyltransferase IA Deficiency	< 1 in 500	< 1 in 1,000,000
Carnitine Palmitoyltransferase II Deficiency	< 1 in 500	< 1 in 1,000,000
Cartilage-Hair Hypoplasia	< 1 in 500	< 1 in 1,000,000
Citrullinemia Type 1	< 1 in 150	< 1 in 70,000
CLN3-related Neuronal Ceroid Lipofuscinosis	< 1 in 5,600	< 1 in 1,000,000
CLN5-related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000
Cohen Syndrome	< 1 in 500	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ia	< 1 in 560	< 1 in 360,000
Congenital Disorder of Glycosylation Type Ib	< 1 in 500	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 500	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 500	< 1 in 1,000,000
Cystic Fibrosis	< 1 in 300	< 1 in 33,000
Cystinosis	< 1 in 670	< 1 in 600,000
D-Bifunctional Protein Deficiency	< 1 in 500	< 1 in 1,000,000
Factor XI Deficiency	< 1 in 500	< 1 in 1,000,000
Familial Dysautonomia	< 1 in 500	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 500	< 1 in 1,000,000
Fanconi Anemia Type C	< 1 in 340	< 1 in 220,000
Galactosemia	< 1 in 430	< 1 in 150,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	< 1 in 160	< 1 in 20,000
Glutaric Acidemia Type 1	< 1 in 170	< 1 in 67,000
Glycogen Storage Disease Type Ia	< 1 in 450	< 1 in 320,000
Glycogen Storage Disease Type Ib	< 1 in 660	< 1 in 930,000
Glycogen Storage Disease Type III	< 1 in 290	< 1 in 180,000
Glycogen Storage Disease Type V	< 1 in 790	< 1 in 500,000
GRACILE Syndrome	< 1 in 500	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	< 1 in 290	< 1 in 58,000
Hereditary Fructose Intolerance	< 1 in 320	< 1 in 100,000



RESULTS RECIPIENT
SEATTLE SPERM BANK
 Attn: Dr. Jeffrey Olliffe
 NPI: 1306838271
 Report Date: 09/12/2016

MALE
DONOR 10076
 DOB: [REDACTED]
 Ethnicity: Mixed or Other
 Caucasian
 Barcode: 11200059662978

FEMALE
 N/A

Disease	DONOR 10076 Residual Risk	Reproductive Risk
Hereditary Thymine-Uraciluria	< 1 in 210	< 1 in 83,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 500	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 500	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 500	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	< 1 in 390	< 1 in 470,000
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	< 1 in 290	< 1 in 290,000
Hurler Syndrome	1 in 480	1 in 300,000
Hypophosphatasia, Autosomal Recessive	< 1 in 230	< 1 in 140,000
Inclusion Body Myopathy 2	< 1 in 500	< 1 in 1,000,000
Isovaleric Acidemia	< 1 in 470	< 1 in 470,000
Joubert Syndrome 2	< 1 in 500	< 1 in 1,000,000
Krabbe Disease	< 1 in 360	< 1 in 210,000
Limb-Girdle Muscular Dystrophy Type 2D	< 1 in 660	< 1 in 1,000,000
Limb-Girdle Muscular Dystrophy Type 2E	< 1 in 500	< 1 in 1,000,000
Lipoamide Dehydrogenase Deficiency	< 1 in 500	< 1 in 1,000,000
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	< 1 in 1,200	< 1 in 690,000
Maple Syrup Urine Disease Type 1B	< 1 in 250	< 1 in 250,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 270	< 1 in 63,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	< 1 in 500	< 1 in 1,000,000
Metachromatic Leukodystrophy	< 1 in 430	< 1 in 340,000
Mucopolidosis IV	< 1 in 500	< 1 in 1,000,000
Muscle-Eye-Brain Disease	< 1 in 500	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 500	< 1 in 1,000,000
Niemann-Pick Disease Type C	< 1 in 230	< 1 in 180,000
Niemann-Pick Disease, SMPD1-associated	< 1 in 400	< 1 in 400,000
Nijmegen Breakage Syndrome	< 1 in 720	< 1 in 450,000
Northern Epilepsy	< 1 in 500	< 1 in 1,000,000
Pendred Syndrome	< 1 in 220	< 1 in 63,000
PEX1-related Zellweger Syndrome Spectrum	< 1 in 350	< 1 in 160,000
Phenylalanine Hydroxylase Deficiency	< 1 in 88	< 1 in 17,000
Polyglandular Autoimmune Syndrome Type 1	NM_000383.3(AIRE):c.967_979del13(aka L323Sfs*51) heterozygote †	1 in 570
Pompe Disease	< 1 in 480	< 1 in 300,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 500	< 1 in 1,000,000
Primary Carnitine Deficiency	< 1 in 500	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	< 1 in 600	< 1 in 850,000
Primary Hyperoxaluria Type 2	< 1 in 500	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	< 1 in 250	< 1 in 110,000
Pseudocholesterase Deficiency	< 1 in 160	< 1 in 18,000
Pycnodysostosis	< 1 in 500	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	< 1 in 530	< 1 in 330,000
Salla Disease	< 1 in 500	< 1 in 1,000,000
Segawa Syndrome	< 1 in 500	< 1 in 1,000,000
Short Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 160	< 1 in 100,000
Sjogren-Larsson Syndrome	< 1 in 330	< 1 in 330,000
Smith-Lemli-Opitz Syndrome	< 1 in 160	< 1 in 32,000
Spinal Muscular Atrophy	SMN1: 2 copies 1 in 610	1 in 84,000
Steroid-Resistant Nephrotic Syndrome	< 1 in 600	< 1 in 950,000
Sulfate Transporter-Related Osteochondrodysplasia	< 1 in 420	< 1 in 180,000
TPP1-related Neuronal Ceroid Lipofuscinosis	< 1 in 740	< 1 in 870,000
Tyrosinemia Type I	< 1 in 350	< 1 in 240,000
Usher Syndrome Type 1F	< 1 in 190	< 1 in 150,000
Usher Syndrome Type 3	< 1 in 500	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 110	< 1 in 39,000
Walker-Warburg Syndrome	< 1 in 500	< 1 in 1,000,000
Wilson Disease	< 1 in 140	< 1 in 50,000