TO: ATTN:Seattle Sperm Bank

Integrated GENETICS
LabCorp Specialty Testing Group

Client/Sending Facility:
Seattle Sperm Bank

4915 25th Ave Ne Ste 204
SEATTLE, WA 98105
Ph: (206)588-1484
Fax: (206) 466-4696 WAB-55

LCLS Specimen Number: **273-129-0845-0**
Patient Name: **12101, DONOR**
Date of Birth: [redacted]
Gender: M
Patient ID:
Lab Number: (J16-3743 L

Indications: DONOR

Account Number: [redacted]
Ordering Physician: **J OLLIFFE**
Specimen Type: **BLOOD**
Client Reference:
Date Collected: 09/29/2016
Date Received: 09/30/2016

Date Reported: **10/11/2016**

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.

Chromosome analysis performed by LabCorp, CLIA 45D0674994. 3701 Kirby Dr. Suite 528, Houston, TX 77098. Laboratory Director, Venkateswara R Potluri PhD.

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Hiba Risheg, PhD., FACMG
Board Certified Cytogeneticist

Patricia Kandalaft, MD
Medical Director
Peter Papenhausen, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
550 17th Ave. Suite 200 , SEATTLE , WA , 98122-5789 (800) 676-8033
Professional Component performed by LabCorp/Dynacare CLIA 50D0632667, 550 17th Ave. Suite 200, Seattle WA 98122-5789. Medical Director, Patricia Kandalaft, MD
Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

Counsyl

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: 10/07/2016

MALE
DONOR 12101
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Sample Type: EDTA Blood
Date of Collection: 09/29/2016
Date Received: 09/30/2016
Date Tested: 10/07/2016
Barcode: 11004211675756
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 12101	Partner
Panel Information	Family Prep Screen 2.0 Universal Panel Minus X-Linked **(102 conditions tested)**	N/A
POSITIVE: CARRIER PPT1-related Neuronal Ceroid Lipofuscinosis **Reproductive Risk: 1 in 2,000** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000310.3(PPT1):c.451C>T (R151*) 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.

Counsyl

RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 10/07/2016

MALE
DONOR 12101
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Barcode: 11004211675756

FEMALE
N/A

POSITIVE: CARRIER

PPT1-related Neuronal Ceroid Lipofuscinosis

Reproductive risk: 1 in 2,000
Risk before testing: < 1 in 1,000,000

Gene: PPT1 | **Inheritance Pattern:** Autosomal Recessive

Patient	DONOR 12101	No partner tested
Result	⊕ Carrier	N/A
Variant(s)	NM_000310.3(PPT1):c.451C>T(R151*) heterozygote	N/A
Methodology	Sequencing	N/A
Interpretation	This individual is a carrier of PPT1-related neuronal ceroid lipofuscinosis. Carriers generally do not experience symptoms. The R151* mutation is associated with the infantile form of this disease.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000310:1-9.	N/A

What is PPT1-related Neuronal Ceroid Lipofuscinosis?

PPT1-related neuronal ceroid-lipofuscinosis (NCL) is an inherited disease that causes degeneration of the brain leading to a progressive loss of mental and motor skills. It can also cause blindness, and typically leads to an early death. In the final stages of the disease, affected individuals will be motionless and in a vegetative state.

There are several forms of NCL, largely differentiated by the gene responsible and the age at which symptoms begin. Mutations in the PPT1 gene typically result in the infantile or juvenile form of NCL.

INFANTILE FORM

The infantile form of NCL (INCL) usually begins to cause noticeable symptoms between the ages of 6 months and 24 months. Initially, infants will show developmental delays and experience seizures or jerking movements. Often these infants will have small heads. Blindness and seizures will be present by 24 months, after which mental functions will deteriorate. The child's movement will become spastic and uncontrolled and he or she will develop dementia.

JUVENILE FORM

The symptoms of juvenile NCL (JNCL), also called Batten disease, often begin between the ages of 4 and 10. These children rapidly lose their vision, which is often the first noticeable symptom. They typically become completely blind within two years. People with JNCL often develop periodic seizures between the ages of 5 and 18.

Between the ages of 8 and 14, mental functions typically decline. Children may have difficulty with speech and show behavioral problems. Some people with JNCL also develop psychiatric problems including disturbed thoughts, attention problems, and aggression. These problems can eventually progress to dementia.

People with JNCL also show a decline in motor function and may have difficulty controlling their own movement.

How common is PPT1-related Neuronal Ceroid Lipofuscinosis?

Approximately 1 in 25,000 people globally are affected by some form of NCL. These diseases are most common in Scandinavian countries, but occur elsewhere as well. In the United States, an estimated 25,000 families are affected by some form of NCL. A subset of all NCLs are caused by mutations in the PPT1 gene. The remainder are caused by mutations in multiple other genes.

INCL is most common in Finland, where 1 in 20,000 births is affected by the disease and 1 in 70 people is a carrier. About half the world's cases of INCL are in Finland. Although many genes may be associated with various forms of NCLs, mutations in the PPT1 gene are frequently seen among the Finnish population.

In Iceland, 7 in 100,000 births are affected by JNCL. Other countries experience fewer cases of JNCL. One study showed 0.7 cases per 100,000 births in Germany.

Exactly how many cases of NCL are caused by mutations in the PPT1 gene is unknown.

How is PPT1-related Neuronal Ceroid Lipofuscinosis treated?

There is no treatment for the underlying cause of NCL. Treatments can only address the symptoms as they arise. Various medications can be useful for treating seizures, poor muscle tone, sleep disorders, mood disorders, excessive drooling, and digestion. In some people, a feeding tube is also helpful.

What is the prognosis for a person with PPT1-related Neuronal Ceroid Lipofuscinosis?

The prognosis for a person with NCL depends upon the type of the disease he or she has. People with INCL or JNCL will become blind and will deteriorate mentally. They will eventually enter a vegetative state and become totally dependent on others to care for them.

Among those with INCL, death usually occurs in childhood.

Among those with JNCL, death usually occurs between one's late teens to 30s.

Methods and Limitations

DONOR 12101 [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.

Detection rates are calculated by estimating from literature the fraction of disease alleles that the methodology is unable to detect.

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.

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

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: Mixed or Other Caucasian >99%.

Achromatopsia - Gene: CNGB3. Autosomal Recessive. Sequencing. Exons: NM_019098:1-18. Detection Rate: Mixed or Other Caucasian >99%.

Alkaptonuria - Gene: HGD. Autosomal Recessive. Sequencing. Exons: NM_000187:1-14. Detection Rate: Mixed or Other Caucasian >99%.

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 >99%.

Alpha-Mannosidosis - Gene: MAN2B1. Autosomal Recessive. Sequencing. Exons: NM_000528:1-15,17-24. Detection Rate: Mixed or Other Caucasian >99%.

Andermann Syndrome - Gene: SLC12A6. Autosomal Recessive. Sequencing. Exons: NM_133647:1-25. Detection Rate: Mixed or Other Caucasian >99%.

ARSACS - Gene: SACS. Autosomal Recessive. Sequencing. Exons: NM_014363:2-10. Detection Rate: Mixed or Other Caucasian >99%.

Aspartylglycosaminuria - Gene: AGA. Autosomal Recessive. Sequencing. Exons: NM_000027:1-9. Detection Rate: Mixed or Other Caucasian >99%.

Ataxia With Vitamin E Deficiency - Gene: TTPA. Autosomal Recessive. Sequencing. Exons: NM_000370:1-5. Detection Rate: Mixed or Other Caucasian >99%.

Ataxia-Telangiectasia - Gene: ATM. Autosomal Recessive. Sequencing. Exons: NM_000051:2-63. Detection Rate: Mixed or Other Caucasian 92%.

Autosomal Recessive Polycystic Kidney Disease - Gene: PKHD1. Autosomal Recessive. Sequencing. Exons: NM_138694:2-67. Detection Rate: Mixed or Other Caucasian >99%.

Bardet-Biedl Syndrome, BBS1-related - Gene: BBS1. Autosomal Recessive. Sequencing. Exons: NM_024649:1-17. Detection Rate: Mixed or Other Caucasian >99%.

Bardet-Biedl Syndrome, BBS10-related - Gene: BBS10. Autosomal Recessive. Sequencing. Exons: NM_024685:1-2. Detection Rate: Mixed or Other Caucasian >99%.

Biotinidase Deficiency - Gene: BTD. Autosomal Recessive. Sequencing. Exons: NM_000060:1-4. Detection Rate: Mixed or Other Caucasian >99%.

Bloom Syndrome - Gene: BLM. Autosomal Recessive. Sequencing. Exons: NM_000057:2-22. Detection Rate: Mixed or Other Caucasian >99%.

Canavan Disease - Gene: ASPA. Autosomal Recessive. Sequencing. Exons: NM_000049:1-6. Detection Rate: Mixed or Other Caucasian 94%.

Carnitine Palmitoyltransferase IA Deficiency - Gene: CPT1A. Autosomal Recessive. Sequencing. Exons: NM_001876:2-19. Detection Rate: Mixed or Other Caucasian >99%.

Carnitine Palmitoyltransferase II Deficiency - Gene: CPT2. Autosomal Recessive. Sequencing. Exons: NM_000098:1-5. Detection Rate: Mixed or Other Caucasian >99%.

Cartilage-Hair Hypoplasia - Gene: RMRP. Autosomal Recessive. Sequencing. Exon: NR_003051:1. Detection Rate: Mixed or Other Caucasian >99%.

Citrullinemia Type 1 - Gene: ASS1. Autosomal Recessive. Sequencing. Exons: NM_000050:3-16. Detection Rate: Mixed or Other Caucasian >99%.

CLN3-related Neuronal Ceroid Lipofuscinosis - Gene: CLN3. Autosomal Recessive. Sequencing. Exons: NM_001042432:2-16. Detection Rate: Mixed or Other Caucasian >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - Gene: CLN5. Autosomal Recessive. Sequencing. Exons: NM_006493:1-4. Detection Rate: Mixed or Other Caucasian 98%.

Cohen Syndrome - Gene: VPS13B. Autosomal Recessive. Sequencing. Exons: NM_017890:2-62. Detection Rate: Mixed or Other Caucasian 83%.

Congenital Disorder of Glycosylation Type Ia - Gene: PMM2. Autosomal Recessive. Sequencing. Exons: NM_000303:1-8. Detection Rate: Mixed or Other Caucasian >99%.

Congenital Disorder of Glycosylation Type Ib - Gene: MPI. Autosomal Recessive. Sequencing. Exons: NM_002435:1-8. Detection Rate: Mixed or Other Caucasian >99%.

Congenital Finnish Nephrosis - Gene: NPHS1. Autosomal Recessive. Sequencing. Exons: NM_004646:2-23,26-27,29. Detection Rate: Mixed or Other Caucasian >99%.

Costeff Optic Atrophy Syndrome - Gene: OPA3. Autosomal Recessive. Sequencing. Exons: NM_025136:1-2. Detection Rate: Mixed or Other Caucasian >99%.

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 97%.

Cystinosis - Gene: CTNS. Autosomal Recessive. Sequencing. Exons: NM_004937:3-12. Detection Rate: Mixed or Other Caucasian >99%.

D-Bifunctional Protein Deficiency - Gene: HSD17B4. Autosomal Recessive. Sequencing. Exons: NM_000414:1-24. Detection Rate: Mixed or Other Caucasian >99%.

Factor XI Deficiency - Gene: F11. Autosomal Recessive. Sequencing. Exons: NM_000128:2-15. Detection Rate: Mixed or Other Caucasian >99%.

Familial Dysautonomia - Gene: IKBKAP. Autosomal Recessive. Sequencing. Exons: NM_003640:19-20,26. Detection Rate: Mixed or Other Caucasian >99%.

Familial Mediterranean Fever - Gene: MEFV. Autosomal Recessive. Sequencing. Exons: NM_000243:1-10. Detection Rate: Mixed or Other Caucasian >99%.

Fanconi Anemia Type C - Gene: FANCC. Autosomal Recessive. Sequencing. Exons: NM_000136:2-15. Detection Rate: Mixed or Other Caucasian >99%.

Galactosemia - Gene: GALT. Autosomal Recessive. Sequencing. Exons: NM_000155:1-11. Detection Rate: Mixed or Other Caucasian >99%.

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 98%.

Glutaric Acidemia Type 1 - Gene: GCDH. Autosomal Recessive. Sequencing. Exons: NM_000159:2-12. Detection Rate: Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type Ia - Gene: G6PC. Autosomal Recessive. Sequencing. Exons: NM_000151:1-5. Detection Rate: Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type Ib - Gene: SLC37A4. Autosomal Recessive. Sequencing. Exons: NM_001164277:3-11. Detection Rate: Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type III - Gene: AGL. Autosomal Recessive. Sequencing. Exons: NM_000642:2-34. Detection Rate: Mixed or Other Caucasian >99%.

Glycogen Storage Disease Type V - Gene: PYGM. Autosomal Recessive. Sequencing. Exons: NM_005609:1-20. Detection Rate: Mixed or Other Caucasian >99%.

GRACILE Syndrome - Gene: BCS1L. Autosomal Recessive. Sequencing. Exons: NM_004328:3-9. Detection Rate: Mixed or Other Caucasian >99%.

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 96%.

Hereditary Fructose Intolerance - Gene: ALDOB. Autosomal Recessive. Sequencing. Exons: NM_000035:2-9. Detection Rate: Mixed or Other Caucasian >99%.

Hereditary Thymine-Uraciluria - Gene: DPYD. Autosomal Recessive. Sequencing. Exons: NM_000110:1-23. Detection Rate: Mixed or Other Caucasian >99%.

Herlitz Junctional Epidermolysis Bullosa, LAMA3-related - Gene: LAMA3. Autosomal Recessive. Sequencing. Exons: NM_000227:1-16,18-38. Detection Rate: Mixed or Other Caucasian >99%.

Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - Gene: LAMB3. Autosomal Recessive. Sequencing. Exons: NM_000228:2-23. Detection Rate: Mixed or Other Caucasian >99%.

Herlitz Junctional Epidermolysis Bullosa, LAMC2-related - Gene: LAMC2. Autosomal Recessive. Sequencing. Exons: NM_005562:1-23. Detection Rate: Mixed or Other Caucasian >99%.
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - Gene: HEXA. Autosomal Recessive. Sequencing. Exons: NM_000520:1-14. Detection Rate: Mixed or Other Caucasian >99%.
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency - Gene: CBS. Autosomal Recessive. Sequencing. Exons: NM_000071:3-17. Detection Rate: Mixed or Other Caucasian >99%.
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 >99%.
Inclusion Body Myopathy 2 - Gene: GNE. Autosomal Recessive. Sequencing. Exons: NM_001128227:3-12. Detection Rate: Mixed or Other Caucasian >99%.
Isovaleric Acidemia - Gene: IVD. Autosomal Recessive. Sequencing. Exons: NM_002225:1-12. Detection Rate: Mixed or Other Caucasian >99%.
Joubert Syndrome 2 - Gene: TMEM216. Autosomal Recessive. Sequencing. Exons: NM_001173990:1-5. Detection Rate: Mixed or Other Caucasian >99%.
Krabbe Disease - Gene: GALC. Autosomal Recessive. Sequencing. Exons: NM_000153:1-17. Detection Rate: Mixed or Other Caucasian >99%.
Limb-Girdle Muscular Dystrophy Type 2D - Gene: SGCA. Autosomal Recessive. Sequencing. Exons: NM_000023:1-9. Detection Rate: Mixed or Other Caucasian >99%.
Limb-Girdle Muscular Dystrophy Type 2E - Gene: SGCB. Autosomal Recessive. Sequencing. Exons: NM_000232:1-6. Detection Rate: Mixed or Other Caucasian >99%.
Lipoamide Dehydrogenase Deficiency - Gene: DLD. Autosomal Recessive. Sequencing. Exons: NM_000108:1-14. Detection Rate: Mixed or Other Caucasian >99%.
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency - Gene: HADHA. Autosomal Recessive. Sequencing. Exons: NM_000182:1-20. Detection Rate: Mixed or Other Caucasian >99%.
Maple Syrup Urine Disease Type 1B - Gene: BCKDHB. Autosomal Recessive. Sequencing. Exons: NM_183050:1-10. Detection Rate: Mixed or Other Caucasian >99%.
Medium Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADM. Autosomal Recessive. Sequencing. Exons: NM_000016:1-12. Detection Rate: Mixed or Other Caucasian >99%.
Megalencephalic Leukoencephalopathy With Subcortical Cysts - Gene: MLC1. Autosomal Recessive. Sequencing. Exons: NM_015166:2-12. Detection Rate: Mixed or Other Caucasian >99%.
Metachromatic Leukodystrophy - Gene: ARSA. Autosomal Recessive. Sequencing. Exons: NM_000487:1-8. Detection Rate: Mixed or Other Caucasian >99%.
Mucolipidosis IV - Gene: MCOLN1. Autosomal Recessive. Sequencing. Exons: NM_020533:1-14. Detection Rate: Mixed or Other Caucasian >99%.
Muscle-Eye-Brain Disease - Gene: POMGNT1. Autosomal Recessive. Sequencing. Exons: NM_017739:2-22. Detection Rate: Mixed or Other Caucasian >99%.
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: Mixed or Other Caucasian >99%.
Niemann-Pick Disease Type C - Gene: NPC1. Autosomal Recessive. Sequencing. Exons: NM_000271:1-25. Detection Rate: Mixed or Other Caucasian 96%.
Niemann-Pick Disease, SMPD1-associated - Gene: SMPD1. Autosomal Recessive. Sequencing. Exons: NM_000543:1-6. Detection Rate: Mixed or Other Caucasian >99%.
Nijmegen Breakage Syndrome - Gene: NBN. Autosomal Recessive. Sequencing. Exons: NM_002485:1-16. Detection Rate: Mixed or Other Caucasian >99%.
Northern Epilepsy - Gene: CLN8. Autosomal Recessive. Sequencing. Exons: NM_018941:2-3. Detection Rate: Mixed or Other Caucasian >99%.
Pendred Syndrome - Gene: SLC26A4. Autosomal Recessive. Sequencing. Exons: NM_000441:2-21. Detection Rate: Mixed or Other Caucasian >99%.
PEX1-related Zellweger Syndrome Spectrum - Gene: PEX1. Autosomal Recessive. Sequencing. Exons: NM_000466:1-24. Detection Rate: Mixed or Other Caucasian >99%.
Phenylalanine Hydroxylase Deficiency - Gene: PAH. Autosomal Recessive. Sequencing. Exons: NM_000277:1-13. Detection Rate: Mixed or Other Caucasian 98%.
Polyglandular Autoimmune Syndrome Type 1 - Gene: AIRE. Autosomal Recessive. Sequencing. Exons: NM_000383:1-14. Detection Rate: Mixed or Other Caucasian >99%.
Pompe Disease - Gene: GAA. Autosomal Recessive. Sequencing. Exons: NM_000152:2-20. Detection Rate: Mixed or Other Caucasian 90%.
PPT1-related Neuronal Ceroid Lipofuscinosis - Gene: PPT1. Autosomal Recessive. Sequencing. Exons: NM_000310:1-9. Detection Rate: Mixed or Other Caucasian >99%.
Primary Carnitine Deficiency - Gene: SLC22A5. Autosomal Recessive. Sequencing. Exons: NM_003060:1-10. Detection Rate: Mixed or Other Caucasian >99%.
Primary Hyperoxaluria Type 1 - Gene: AGXT. Autosomal Recessive. Sequencing. Exons: NM_000030:1-11. Detection Rate: Mixed or Other Caucasian >99%.
Primary Hyperoxaluria Type 2 - Gene: GRHPR. Autosomal Recessive. Sequencing. Exons: NM_012203:1-9. Detection Rate: Mixed or Other Caucasian >99%.
PROP1-related Combined Pituitary Hormone Deficiency - Gene: PROP1. Autosomal Recessive. Sequencing. Exons: NM_006261:1-3. Detection Rate: Mixed or Other Caucasian >99%.
Pseudocholinesterase Deficiency - Gene: BCHE. Autosomal Recessive. Sequencing. Exons: NM_000055:2-4. Detection Rate: Mixed or Other Caucasian >99%.
Pycnodysostosis - Gene: CTSK. Autosomal Recessive. Sequencing. Exons: NM_000396:2-8. Detection Rate: Mixed or Other Caucasian >99%.
Rhizomelic Chondrodysplasia Punctata Type 1 - Gene: PEX7. Autosomal Recessive. Sequencing. Exons: NM_000288:1-10. Detection Rate: Mixed or Other Caucasian >99%.
Salla Disease - Gene: SLC17A5. Autosomal Recessive. Sequencing. Exons: NM_012434:1-11. Detection Rate: Mixed or Other Caucasian >99%.
Segawa Syndrome - Gene: TH. Autosomal Recessive. Sequencing. Exons: NM_000360:1-13. Detection Rate: Mixed or Other Caucasian >99%.
Short Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADS. Autosomal Recessive. Sequencing. Exons: NM_000017:1-10. Detection Rate: Mixed or Other Caucasian >99%.
Sjogren-Larsson Syndrome - Gene: ALDH3A2. Autosomal Recessive. Sequencing. Exons: NM_000382:1-10. Detection Rate: Mixed or Other Caucasian >99%.
Smith-Lemli-Opitz Syndrome - Gene: DHCR7. Autosomal Recessive. Sequencing. Exons: NM_001360:3-9. Detection Rate: Mixed or Other Caucasian >99%.
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 >99%.
Sulfate Transporter-Related Osteochondrodysplasia - Gene: SLC26A2. Autosomal Recessive. Sequencing. Exons: NM_000112:2-3. Detection Rate: Mixed or Other Caucasian >99%.
TPP1-related Neuronal Ceroid Lipofuscinosis - Gene: TPP1. Autosomal Recessive. Sequencing. Exons: NM_000391:1-13. Detection Rate: Mixed or Other Caucasian >99%.
Tyrosinemia Type I - Gene: FAH. Autosomal Recessive. Sequencing. Exons: NM_000137:1-14. Detection Rate: Mixed or Other Caucasian >99%.
Usher Syndrome Type 1F - Gene: PCDH15. Autosomal Recessive. Sequencing. Exons: NM_033056:2-33. Detection Rate: Mixed or Other Caucasian 97%.
Usher Syndrome Type 3 - Gene: CLRN1. Autosomal Recessive. Sequencing. Exons: NM_174878:1-3. Detection Rate: Mixed or Other Caucasian >99%.
Very Long Chain Acyl-CoA Dehydrogenase Deficiency - Gene: ACADVL. Autosomal Recessive. Sequencing. Exons: NM_000018:1-20. Detection Rate: Mixed or Other Caucasian >99%.
Walker-Warburg Syndrome - Gene: FKTN. Autosomal Recessive. Sequencing. Exons: NM_001079802:3-11. Detection Rate: Mixed or Other Caucasian >99%.
Wilson Disease - Gene: ATP7B. Autosomal Recessive. Sequencing. Exons: NM_000053:1-21. Detection Rate: Mixed or Other Caucasian >99%.

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RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 10/07/2016

MALE
DONOR 12101
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Barcode: 11004211675756

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 12101 Residual Risk	Reproductive Risk
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Achromatopsia	1 in 8,600	< 1 in 1,000,000
Alkaptonuria	< 1 in 500	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-1 Antitrypsin Deficiency	1 in 3,400	1 in 460,000
Alpha-Mannosidosis	1 in 35,000	< 1 in 1,000,000
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 2,100	< 1 in 1,000,000
Autosomal Recessive Polycystic Kidney Disease	1 in 6,100	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS1-related	1 in 16,000	< 1 in 1,000,000
Bardet-Biedl Syndrome, BBS10-related	1 in 16,000	< 1 in 1,000,000
Biotinidase Deficiency	1 in 12,000	< 1 in 1,000,000
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 12,000	< 1 in 1,000,000
CLN3-related Neuronal Ceroid Lipofuscinosis	1 in 22,000	< 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 16,000	< 1 in 1,000,000
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 910	1 in 99,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
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 16,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 1,700	1 in 220,000
Glutaric Acidemia Type 1	1 in 10,000	< 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
Glycogen Storage Disease Type V	1 in 16,000	< 1 in 1,000,000
GRACILE Syndrome	< 1 in 500	< 1 in 1,000,000
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 1,200	1 in 240,000
Hereditary Fructose Intolerance	1 in 8,000	< 1 in 1,000,000

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RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 10/07/2016

MALE
DONOR 12101
DOB: [redacted]
Ethnicity: Mixed or Other Caucasian
Barcode: 11004211675756

FEMALE
N/A

Disease	DONOR 12101 Residual Risk	Reproductive Risk
Hereditary Thymine-Uraciluria	1 in 10,000	< 1 in 1,000,000
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 30,000	< 1 in 1,000,000
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency	1 in 25,000	< 1 in 1,000,000
Hurler Syndrome	1 in 480	1 in 300,000
Hypophosphatasia, Autosomal Recessive	1 in 16,000	< 1 in 1,000,000
Inclusion Body Myopathy 2	< 1 in 500	< 1 in 1,000,000
Isovaleric Acidemia	1 in 25,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 500	< 1 in 1,000,000
Krabbe Disease	1 in 15,000	< 1 in 1,000,000
Limb-Girdle Muscular Dystrophy Type 2D	1 in 45,000	< 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 15,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	< 1 in 1,000,000
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1 in 5,900	< 1 in 1,000,000
Megalencephalic Leukoencephalopathy With Subcortical Cysts	< 1 in 500	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 20,000	< 1 in 1,000,000
Mucolipidosis 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 5,400	< 1 in 1,000,000
Niemann-Pick Disease, SMPD1-associated	1 in 25,000	< 1 in 1,000,000
Nijmegen Breakage Syndrome	1 in 16,000	< 1 in 1,000,000
Northern Epilepsy	< 1 in 500	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 1 in 1,000,000
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 3,000	1 in 600,000
Polyglandular Autoimmune Syndrome Type 1	1 in 14,000	< 1 in 1,000,000
Pompe Disease	1 in 1,600	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	R151* heterozygote †	1 in 2,000
Primary Carnitine Deficiency	< 1 in 500	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 2	< 1 in 500	< 1 in 1,000,000
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pseudocholinesterase Deficiency	1 in 2,700	1 in 300,000
Pycnodysostosis	< 1 in 500	< 1 in 1,000,000
Rhizomelic Chondrodysplasia Punctata Type 1	1 in 16,000	< 1 in 1,000,000
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 16,000	< 1 in 1,000,000
Sjogren-Larsson Syndrome	1 in 25,000	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 4,900	1 in 970,000
Spinal Muscular Atrophy	SMN1: 2 copies	1 in 84,000
Steroid-Resistant Nephrotic Syndrome	1 in 610	
Sulfate Transporter-Related Osteochondrodysplasia	1 in 40,000	< 1 in 1,000,000
TPP1-related Neuronal Ceroid Lipofuscinosis	1 in 11,000	< 1 in 1,000,000
Tyrosinemia Type I	1 in 30,000	< 1 in 1,000,000
Usher Syndrome Type 1F	1 in 17,000	< 1 in 1,000,000
Usher Syndrome Type 3	1 in 6,600	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	< 1 in 500	< 1 in 1,000,000
Walker-Warburg Syndrome	1 in 8,800	< 1 in 1,000,000
Wilson Disease	< 1 in 500	< 1 in 1,000,000
	1 in 8,600	< 1 in 1,000,000