Counsyl

RESULTS RECIPIENT
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Report Date: 05/07/2018

MALE
DONOR 10229
DOB: [redacted]
Ethnicity: French Canadian or Cajun
Sample Type: EDTA Blood
Date of Collection: 04/27/2018
Date Received: 04/30/2018
Date Tested: 05/07/2018
Barcode: 11004212319624
Accession ID: CSLQKWHNMG9XL9L
Indication: Egg or sperm donor

FEMALE
N/A

Foresight™ Carrier Screen

POSITIVE: CARRIER

ABOUT THIS TEST

The **Counsyl 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 10229	Partner
Panel Information	Foresight Carrier Screen Universal Panel **(175 conditions tested)**	N/A
POSITIVE: CARRIER Biotinidase Deficiency **Reproductive Risk: 1 in 630** **Inheritance: Autosomal Recessive**	**CARRIER*** NM_000060.2(BTD):c.1330G>C (D444H) 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.

POSITIVE: CARRIER

Biotinidase Deficiency

Reproductive risk: 1 in 630
Risk before testing: 1 in 4,800

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

Patient	DONOR 10229	No partner tested
Result	Carrier	N/A
Variant(s)	NM_000060.2(BTD):c.1330G>C(D444H) heterozygote	N/A
Methodology	Sequencing with copy number analysis	N/A
Interpretation	This individual is a carrier of biotinidase deficiency. Carriers generally do not experience symptoms. D444H is a partial biotinidase deficiency mutation.	N/A
Detection rate	>99%	N/A
Exons tested	NM_000060:1-4.	N/A

What is Biotinidase Deficiency?

Biotinidase deficiency is a highly-treatable inherited disease in which the body cannot process the vitamin biotin due to a deficiency in a particular enzyme. If left untreated, the disease can cause numerous life-threatening complications. By taking daily supplements of biotin before symptoms occur, however, all symptoms of the disease can be avoided. With early detection and treatment, a person with biotinidase deficiency can live a completely normal life.

PROFOUND BIOTINIDASE DEFICIENCY

People who have less than 10% of the normal amount of the enzyme biotinidase are said to have profound biotinidase deficiency. Without treatment, their symptoms tend to be significant. People with biotinidase deficiency can experience seizures, poor muscle tone, difficulty with movement and balance, vision and/or hearing loss, skin rashes, breathing problems, fungal infections, and intellectual and/or developmental delays. These symptoms often begin after the first few weeks or months of life and can be life-threatening if untreated.

If symptoms have already appeared, treatment with biotin can reverse damage to the body already done by the disease. Vision loss, hearing loss, and developmental delay are irreversible.

PARTIAL BIOTINIDASE DEFICIENCY

People who have between 10 and 30% of the normal amounts of biotinidase have a milder form of the disease known as partial biotinidase deficiency. They may experience less severe symptoms, or may be asymptomatic until periods of illness or stress.

How common is Biotinidase Deficiency?

Profound biotinidase deficiency occurs in about 1 in 137,000 births. Studies report that the milder partial biotinidase deficiency occurs in about 1 in 110,000 people. Counsyl's internal data suggests that partial biotinidase deficiency is more common.

How is Biotinidase Deficiency treated?

Biotinidase deficiency is treated with a biotin pill taken daily by mouth. A physician can determine the proper dosage and adjust that dosage over time if necessary. This treatment is lifelong and highly effective. Biotin is non-toxic, so it is recommended that people with partial biotinidase deficiency also take biotin supplements.

If treatment is begun after symptoms appear, some symptoms, such as skin problems and hair loss, will disappear. If the disease has already caused irreversible hearing or vision loss, low vision aids or hearing aids may be helpful. Learning specialists can assist with any irreversible developmental deficits.

What is the prognosis for a person with Biotinidase Deficiency?

With early diagnosis and treatment, people with biotinidase deficiency can live completely normal lives with no symptoms. Those in whom the disease is not detected early may experience permanent damage to their hearing, vision, or intellect. In cases where the disease is entirely unrecognized, it can be life-threatening.

Methods and Limitations

DONOR 10229 [Foresight Carrier Screen]: sequencing with copy number analysis, spinal muscular atrophy, and analysis of homologous regions.

Sequencing with copy number analysis

High-throughput sequencing and read depth-based copy number analysis are 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. The region of interest (ROI) of the test comprises these regions, in addition to the 20 intronic bases flanking each exon. 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 high coverage and the sequences are compared to standards and references of normal variation. More than 99% of all bases in the ROI are sequenced at greater than the minimum read depth. Mutations may not be detected in areas of lower sequence coverage. Small insertions and deletions may not be as accurately determined as single nucleotide variants. Genes that have closely related pseudogenes may be addressed by a different method. *CFTR* and *DMD* testing includes analysis for both large (exon-level) deletions and duplications with an average sensitivity of 99%, while other genes are only analyzed for large deletions with a sensitivity of >75%. However, the sensitivity may be higher for selected founder deletions. If *GJB2* is tested, two large upstream deletions which overlap *GJB6* and affect the expression of *GJB2*, del(*GJB6*-D13S1830) and del(*GJB6*-D13S1854), are also analyzed. Mosaicism or somatic variants present at low levels may not be detected. If detected, these may not be reported.

Detection rates are determined by using literature to estimate the fraction of disease alleles, weighted by frequency, that the methodology is unable to detect. Detection rates only account for analytical sensitivity and 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 mutations.

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 "likely" pathogenic are reported. Likely pathogenic variants are described elsewhere in the report as "likely to have a negative impact on gene function". Likely pathogenic variants are evaluated and classified by assessing the nature of the variant and reviewing reports of allele frequencies in cases and controls, functional studies, variant annotation and effect prediction, and segregation studies. Exon level duplications are assumed to be in tandem and are classified according to their predicted effect on the reading frame. Benign variants, variants of uncertain significance, and variants not directly associated with the intended disease phenotype are not reported. Curation summaries of reported variants are available upon request.

Spinal muscular atrophy

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. This is more likely in individuals who have 2 copies of the *SMN1* gene and are positive for the g.27134T>G SNP, 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 2 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 mutations in certain genes that have homology to other regions. The precise breakpoints of large deletions in these genes cannot be determined, but are estimated from copy number analysis. High numbers of pseudogene copies may interfere with this analysis.

If *CYP21A2* is tested, 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). 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. If *HBA1*/*HBA2* are tested, some individuals with four alpha globin genes may be carriers, with three genes on one chromosome and a deletion on the other chromosome. This and similar, but rare, carrier states, where complementary changes exist in both the gene and a pseudogene, may not be detected by the assay.

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. This test is designed to detect and report germline alterations. While somatic variants present at low levels may be detected, these 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. The test 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*).

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**.

LABORATORY DIRECTOR

Hyunseok Kang

H. Peter Kang, MD, MS, FCAP

Report content approved by Rebecca Mar-Heyming, PhD, FACMG on May 7, 2018

Conditions Tested

11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia - **Gene:** CYP11B1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000497:1-9. **Detection Rate:** French Canadian or Cajun 94%.

21-hydroxylase-deficient Congenital Adrenal Hyperplasia - **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, V281L, [I237N;V238E;M240K], c.293-13C>G. **Detection Rate:** French Canadian or Cajun 96%.

6-pyruvoyl-tetrahydropterin Synthase Deficiency - **Gene:** PTS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000317:1-6. **Detection Rate:** French Canadian or Cajun >99%.

ABCC8-related Hyperinsulinism - **Gene:** ABCC8. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000352:1-39. **Detection Rate:** French Canadian or Cajun >99%.

Adenosine Deaminase Deficiency - **Gene:** ADA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000022:1-12. **Detection Rate:** French Canadian or Cajun >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-mannosidosis - **Gene:** MAN2B1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000528:1-23. **Detection Rate:** French Canadian or Cajun >99%.

Alpha-sarcoglycanopathy - **Gene:** SGCA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000023:1-9. **Detection Rate:** French Canadian or Cajun >99%.

Alstrom Syndrome - **Gene:** ALMS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_015120:1-23. **Detection Rate:** French Canadian or Cajun >99%.

AMT-related Glycine Encephalopathy - **Gene:** AMT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000481:1-9. **Detection Rate:** French Canadian or Cajun >99%.

Andermann Syndrome - **Gene:** SLC12A6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_133647:1-25. **Detection Rate:** French Canadian or Cajun >99%.

Argininemia - **Gene:** ARG1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001244438:1-8. **Detection Rate:** French Canadian or Cajun 97%.

Argininosuccinic Aciduria - **Gene:** ASL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001024943:1-16. **Detection Rate:** French Canadian or Cajun >99%.

ARSACS - **Gene:** SACS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_014363:2-10. **Detection Rate:** French Canadian or Cajun 99%.

Aspartylglycosaminuria - **Gene:** AGA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000027:1-9. **Detection Rate:** French Canadian or Cajun >99%.

Ataxia with Vitamin E Deficiency - **Gene:** TTPA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000370:1-5. **Detection Rate:** French Canadian or Cajun >99%.

Ataxia-telangiectasia - **Gene:** ATM. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000051:2-63. **Detection Rate:** French Canadian or Cajun >99%.

ATP7A-related Disorders - **Gene:** ATP7A. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000052:2-23. **Detection Rate:** French Canadian or Cajun 92%.

Autosomal Recessive Osteopetrosis Type 1 - **Gene:** TCIRG1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006019:2-20. **Detection Rate:** French Canadian or Cajun >99%.

Bardet-Biedl Syndrome, BBS1-related - **Gene:** BBS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_024649:1-17. **Detection Rate:** French Canadian or Cajun >99%.

Bardet-Biedl Syndrome, BBS10-related - **Gene:** BBS10. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_024685:1-2. **Detection Rate:** French Canadian or Cajun >99%.

Bardet-Biedl Syndrome, BBS12-related - **Gene:** BBS12. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NM_152618:2. **Detection Rate:** French Canadian or Cajun >99%.

Bardet-Biedl Syndrome, BBS2-related - **Gene:** BBS2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_031885:1-17. **Detection Rate:** French Canadian or Cajun >99%.

Beta-sarcoglycanopathy - **Gene:** SGCB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000232:1-6. **Detection Rate:** French Canadian or Cajun >99%.

Biotinidase Deficiency - **Gene:** BTD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000060:1-4. **Detection Rate:** French Canadian or Cajun >99%.

Bloom Syndrome - **Gene:** BLM. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000057:2-22. **Detection Rate:** French Canadian or Cajun >99%.

Calpainopathy - **Gene:** CAPN3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000070:1-24. **Detection Rate:** French Canadian or Cajun >99%.

Canavan Disease - **Gene:** ASPA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000049:1-6. **Detection Rate:** French Canadian or Cajun 98%.

Carbamoylphosphate Synthetase I Deficiency - **Gene:** CPS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001875:1-38. **Detection Rate:** French Canadian or Cajun >99%.

Carnitine Palmitoyltransferase IA Deficiency - **Gene:** CPT1A. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001876:2-19. **Detection Rate:** French Canadian or Cajun >99%.

Carnitine Palmitoyltransferase II Deficiency - **Gene:** CPT2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000098:1-5. **Detection Rate:** French Canadian or Cajun >99%.

Cartilage-hair Hypoplasia - **Gene:** RMRP. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NR_003051:1. **Detection Rate:** French Canadian or Cajun >99%.

Cerebrotendinous Xanthomatosis - **Gene:** CYP27A1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000784:1-9. **Detection Rate:** French Canadian or Cajun >99%.

Citrullinemia Type 1 - **Gene:** ASS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000050:3-16. **Detection Rate:** French Canadian or Cajun >99%.

CLN3-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001042432:2-16. **Detection Rate:** French Canadian or Cajun >99%.

CLN5-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN5. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006493:1-4. **Detection Rate:** French Canadian or Cajun >99%.

CLN6-related Neuronal Ceroid Lipofuscinosis - **Gene:** CLN6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017882:1-7. **Detection Rate:** French Canadian or Cajun >99%.

Cohen Syndrome - **Gene:** VPS13B. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017890:2-62. **Detection Rate:** French Canadian or Cajun 97%.

COL4A3-related Alport Syndrome - **Gene:** COL4A3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000091:1-52. **Detection Rate:** French Canadian or Cajun 97%.

COL4A4-related Alport Syndrome - **Gene:** COL4A4. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000092:2-48. **Detection Rate:** French Canadian or Cajun 98%.

Congenital Disorder of Glycosylation Type Ia - **Gene:** PMM2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000303:1-8. **Detection Rate:** French Canadian or Cajun >99%.

Congenital Disorder of Glycosylation Type Ib - **Gene:** MPI. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_002435:1-8. **Detection Rate:** French Canadian or Cajun >99%.

Congenital Disorder of Glycosylation Type Ic - **Gene:** ALG6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_013339:2-15. **Detection Rate:** French Canadian or Cajun >99%.
Congenital Finnish Nephrosis - **Gene:** NPHS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004646:1-29. **Detection Rate:** French Canadian or Cajun >99%.
Costeff Optic Atrophy Syndrome - **Gene:** OPA3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_025136:1-2. **Detection Rate:** French Canadian or Cajun >99%.
Cystic Fibrosis - **Gene:** CFTR. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000492:1-27. IVS8-5T allele analysis is only reported in the presence of the R117H mutation. **Detection Rate:** French Canadian or Cajun >99%.
Cystinosis - **Gene:** CTNS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004937:3-12. **Detection Rate:** French Canadian or Cajun >99%.
D-bifunctional Protein Deficiency - **Gene:** HSD17B4. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000414:1-24. **Detection Rate:** French Canadian or Cajun 98%.
Delta-sarcoglycanopathy - **Gene:** SGCD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000337:2-9. **Detection Rate:** French Canadian or Cajun 99%.
Dysferlinopathy - **Gene:** DYSF. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001130987:1-56. **Detection Rate:** French Canadian or Cajun 98%.
Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) - **Gene:** DMD. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004006:1-79. **Detection Rate:** French Canadian or Cajun >99%.
ERCC6-related Disorders - **Gene:** ERCC6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000124:2-21. **Detection Rate:** French Canadian or Cajun 99%.
ERCC8-related Disorders - **Gene:** ERCC8. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000082:1-12. **Detection Rate:** French Canadian or Cajun 95%.
EVC-related Ellis-van Creveld Syndrome - **Gene:** EVC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_153717:1-21. **Detection Rate:** French Canadian or Cajun 96%.
EVC2-related Ellis-van Creveld Syndrome - **Gene:** EVC2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_147127:1-22. **Detection Rate:** French Canadian or Cajun >99%.
Fabry Disease - **Gene:** GLA. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000169:1-7. **Detection Rate:** French Canadian or Cajun 98%.
Familial Dysautonomia - **Gene:** IKBKAP. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_003640:2-37. **Detection Rate:** French Canadian or Cajun >99%.
Familial Mediterranean Fever - **Gene:** MEFV. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000243:1-10. **Detection Rate:** French Canadian or Cajun >99%.
Fanconi Anemia Complementation Group A - **Gene:** FANCA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000135:1-43. **Detection Rate:** French Canadian or Cajun 92%.
Fanconi Anemia Type C - **Gene:** FANCC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000136:2-15. **Detection Rate:** French Canadian or Cajun >99%.
FKRP-related Disorders - **Gene:** FKRP. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NM_024301:4. **Detection Rate:** French Canadian or Cajun >99%.
FKTN-related Disorders - **Gene:** FKTN. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001079802:3-11. **Detection Rate:** French Canadian or Cajun >99%.
Galactokinase Deficiency - **Gene:** GALK1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000154:1-8. **Detection Rate:** French Canadian or Cajun >99%.
Galactosemia - **Gene:** GALT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000155:1-11. **Detection Rate:** French Canadian or Cajun >99%.
Gamma-sarcoglycanopathy - **Gene:** SGCG. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000231:2-8. **Detection Rate:** French Canadian or Cajun 88%.
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:** French Canadian or Cajun 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:** French Canadian or Cajun >99%.
GLB1-related Disorders - **Gene:** GLB1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000404:1-16. **Detection Rate:** French Canadian or Cajun >99%.
GLDC-related Glycine Encephalopathy - **Gene:** GLDC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000170:1-25. **Detection Rate:** French Canadian or Cajun 94%.
Glutaric Acidemia Type 1 - **Gene:** GCDH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000159:2-12. **Detection Rate:** French Canadian or Cajun >99%.
Glycogen Storage Disease Type Ia - **Gene:** G6PC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000151:1-5. **Detection Rate:** French Canadian or Cajun >99%.
Glycogen Storage Disease Type Ib - **Gene:** SLC37A4. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001164277:3-11. **Detection Rate:** French Canadian or Cajun >99%.
Glycogen Storage Disease Type III - **Gene:** AGL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000642:2-34. **Detection Rate:** French Canadian or Cajun >99%.
GNPTAB-related Disorders - **Gene:** GNPTAB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_024312:1-21. **Detection Rate:** French Canadian or Cajun >99%.
GRACILE Syndrome - **Gene:** BCS1L. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004328:3-9. **Detection Rate:** French Canadian or Cajun >99%.
HADHA-related Disorders - **Gene:** HADHA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000182:1-20. **Detection Rate:** French Canadian or Cajun >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:** French Canadian or Cajun >99%.
Hereditary Fructose Intolerance - **Gene:** ALDOB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000035:2-9. **Detection Rate:** French Canadian or Cajun >99%.
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related - **Gene:** LAMA3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000227:1-38. **Detection Rate:** French Canadian or Cajun >99%.
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related - **Gene:** LAMB3. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000228:2-23. **Detection Rate:** French Canadian or Cajun >99%.
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related - **Gene:** LAMC2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_005562:1-23. **Detection Rate:** French Canadian or Cajun >99%.
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) - **Gene:** HEXA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000520:1-14. **Detection Rate:** French Canadian or Cajun >99%.
HMG-CoA Lyase Deficiency - **Gene:** HMGCL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000191:1-9. **Detection Rate:** French Canadian or Cajun 98%.
Holocarboxylase Synthetase Deficiency - **Gene:** HLCS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000411:4-12. **Detection Rate:** French Canadian or Cajun >99%.
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency - **Gene:** CBS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000071:3-17. **Detection Rate:** French Canadian or Cajun >99%.
Hydrolethalus Syndrome - **Gene:** HYLS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NM_001134793:3. **Detection Rate:** French Canadian or Cajun >99%.
Hypophosphatasia, Autosomal Recessive - **Gene:** ALPL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000478:2-12. **Detection Rate:** French Canadian or Cajun >99%.
Inclusion Body Myopathy 2 - **Gene:** GNE. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001128227:1-12. **Detection Rate:** French Canadian or Cajun >99%.
Isovaleric Acidemia - **Gene:** IVD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_002225:1-12. **Detection Rate:** French Canadian or Cajun >99%.

Joubert Syndrome 2 - **Gene:** TMEM216. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001173990:1-5. **Detection Rate:** French Canadian or Cajun >99%.
KCNJ11-related Familial Hyperinsulinism - **Gene:** KCNJ11. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NM_000525:1. **Detection Rate:** French Canadian or Cajun >99%.
Krabbe Disease - **Gene:** GALC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000153:1-17. **Detection Rate:** French Canadian or Cajun >99%.
LAMA2-related Muscular Dystrophy - **Gene:** LAMA2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000426:1-65. **Detection Rate:** French Canadian or Cajun >99%.
Leigh Syndrome, French-Canadian Type - **Gene:** LRPPRC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_133259:1-38. **Detection Rate:** French Canadian or Cajun >99%.
Lipoamide Dehydrogenase Deficiency - **Gene:** DLD. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000108:1-14. **Detection Rate:** French Canadian or Cajun >99%.
Lipoid Congenital Adrenal Hyperplasia - **Gene:** STAR. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000349:1-7. **Detection Rate:** French Canadian or Cajun >99%.
Lysosomal Acid Lipase Deficiency - **Gene:** LIPA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000235:2-10. **Detection Rate:** French Canadian or Cajun >99%.
Maple Syrup Urine Disease Type 1B - **Gene:** BCKDHB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_183050:1-10. **Detection Rate:** French Canadian or Cajun >99%.
Maple Syrup Urine Disease Type Ia - **Gene:** BCKDHA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000709:1-9. **Detection Rate:** French Canadian or Cajun >99%.
Maple Syrup Urine Disease Type II - **Gene:** DBT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001918:1-11. **Detection Rate:** French Canadian or Cajun 95%.
Medium Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADM. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000016:1-12. **Detection Rate:** French Canadian or Cajun >99%.
Megalencephalic Leukoencephalopathy with Subcortical Cysts - **Gene:** MLC1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_015166:2-12. **Detection Rate:** French Canadian or Cajun >99%.
Metachromatic Leukodystrophy - **Gene:** ARSA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000487:1-8. **Detection Rate:** French Canadian or Cajun >99%.
Methylmalonic Acidemia, cblA Type - **Gene:** MMAA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_172250:2-7. **Detection Rate:** French Canadian or Cajun >99%.
Methylmalonic Acidemia, cblB Type - **Gene:** MMAB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_052845:1-9. **Detection Rate:** French Canadian or Cajun >99%.
Methylmalonic Aciduria and Homocystinuria, cblC Type - **Gene:** MMACHC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_015506:1-4. **Detection Rate:** French Canadian or Cajun >99%.
MKS1-related Disorders - **Gene:** MKS1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017777:1-18. **Detection Rate:** French Canadian or Cajun >99%.
Mucolipidosis III Gamma - **Gene:** GNPTG. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_032520:1-11. **Detection Rate:** French Canadian or Cajun >99%.
Mucolipidosis IV - **Gene:** MCOLN1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_020533:1-14. **Detection Rate:** French Canadian or Cajun >99%.
Mucopolysaccharidosis Type I - **Gene:** IDUA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000203:1-14. **Detection Rate:** French Canadian or Cajun >99%.
Mucopolysaccharidosis Type II - **Gene:** IDS. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000202:1-9. **Detection Rate:** French Canadian or Cajun 88%.
Mucopolysaccharidosis Type IIIA - **Gene:** SGSH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000199:1-8. **Detection Rate:** French Canadian or Cajun >99%.
Mucopolysaccharidosis Type IIIB - **Gene:** NAGLU. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000263:1-6. **Detection Rate:** French Canadian or Cajun >99%.
Mucopolysaccharidosis Type IIIC - **Gene:** HGSNAT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_152419:1-18. **Detection Rate:** French Canadian or Cajun >99%.
Muscle-eye-brain Disease - **Gene:** POMGNT1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_017739:2-22. **Detection Rate:** French Canadian or Cajun 96%.
MUT-related Methylmalonic Acidemia - **Gene:** MUT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000255:2-13. **Detection Rate:** French Canadian or Cajun >99%.
MYO7A-related Disorders - **Gene:** MYO7A. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000260:2-49. **Detection Rate:** French Canadian or Cajun >99%.
NEB-related Nemaline Myopathy - **Gene:** NEB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001271208:3-80,117-183. **Detection Rate:** French Canadian or Cajun 92%.
Nephrotic Syndrome, NPHS2-related - **Gene:** NPHS2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_014625:1-8. **Detection Rate:** French Canadian or Cajun >99%.
Niemann-Pick Disease Type C - **Gene:** NPC1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000271:1-25. **Detection Rate:** French Canadian or Cajun >99%.
Niemann-Pick Disease Type C2 - **Gene:** NPC2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006432:1-5. **Detection Rate:** French Canadian or Cajun >99%.
Niemann-Pick Disease, SMPD1-associated - **Gene:** SMPD1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000543:1-6. **Detection Rate:** French Canadian or Cajun >99%.
Nijmegen Breakage Syndrome - **Gene:** NBN. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_002485:1-16. **Detection Rate:** French Canadian or Cajun >99%.
Northern Epilepsy - **Gene:** CLN8. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_018941:2-3. **Detection Rate:** French Canadian or Cajun >99%.
Ornithine Transcarbamylase Deficiency - **Gene:** OTC. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000531:1-10. **Detection Rate:** French Canadian or Cajun 97%.
PCCA-related Propionic Acidemia - **Gene:** PCCA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000282:1-24. **Detection Rate:** French Canadian or Cajun 95%.
PCCB-related Propionic Acidemia - **Gene:** PCCB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001178014:1-16. **Detection Rate:** French Canadian or Cajun >99%.
PCDH15-related Disorders - **Gene:** PCDH15. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_033056:2-33. **Detection Rate:** French Canadian or Cajun 93%.
Pendred Syndrome - **Gene:** SLC26A4. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000441:2-21. **Detection Rate:** French Canadian or Cajun >99%.
Peroxisome Biogenesis Disorder Type 3 - **Gene:** PEX12. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000286:1-3. **Detection Rate:** French Canadian or Cajun >99%.
Peroxisome Biogenesis Disorder Type 4 - **Gene:** PEX6. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000287:1-17. **Detection Rate:** French Canadian or Cajun 97%.
Peroxisome Biogenesis Disorder Type 5 - **Gene:** PEX2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exon:** NM_000318:4. **Detection Rate:** French Canadian or Cajun >99%.
Peroxisome Biogenesis Disorder Type 6 - **Gene:** PEX10. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_153818:1-6. **Detection Rate:** French Canadian or Cajun >99%.
PEX1-related Zellweger Syndrome Spectrum - **Gene:** PEX1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000466:1-24. **Detection Rate:** French Canadian or Cajun >99%.
Phenylalanine Hydroxylase Deficiency - **Gene:** PAH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000277:1-13. **Detection Rate:** French Canadian or Cajun >99%.

PKHD1-related Autosomal Recessive Polycystic Kidney Disease - **Gene:** PKHD1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_138694:2-67. **Detection Rate:** French Canadian or Cajun >99%.
Polyglandular Autoimmune Syndrome Type 1 - **Gene:** AIRE. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000383:1-14. **Detection Rate:** French Canadian or Cajun >99%.
Pompe Disease - **Gene:** GAA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000152:2-20. **Detection Rate:** French Canadian or Cajun >99%.
PPT1-related Neuronal Ceroid Lipofuscinosis - **Gene:** PPT1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000310:1-9. **Detection Rate:** French Canadian or Cajun >99%.
Primary Carnitine Deficiency - **Gene:** SLC22A5. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_003060:1-10. **Detection Rate:** French Canadian or Cajun >99%.
Primary Hyperoxaluria Type 1 - **Gene:** AGXT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000030:1-11. **Detection Rate:** French Canadian or Cajun >99%.
Primary Hyperoxaluria Type 2 - **Gene:** GRHPR. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_012203:1-9. **Detection Rate:** French Canadian or Cajun >99%.
Primary Hyperoxaluria Type 3 - **Gene:** HOGA1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_138413:1-7. **Detection Rate:** French Canadian or Cajun >99%.
PROP1-related Combined Pituitary Hormone Deficiency - **Gene:** PROP1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_006261:1-3. **Detection Rate:** French Canadian or Cajun >99%.
Pycnodysostosis - **Gene:** CTSK. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000396:2-8. **Detection Rate:** French Canadian or Cajun >99%.
Pyruvate Carboxylase Deficiency - **Gene:** PC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_022172:2-21. **Detection Rate:** French Canadian or Cajun >99%.
Rhizomelic Chondrodysplasia Punctata Type 1 - **Gene:** PEX7. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000288:1-10. **Detection Rate:** French Canadian or Cajun >99%.
RTEL1-related Disorders - **Gene:** RTEL1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_032957:2-35. **Detection Rate:** French Canadian or Cajun >99%.
Salla Disease - **Gene:** SLC17A5. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_012434:1-11. **Detection Rate:** French Canadian or Cajun 98%.
Sandhoff Disease - **Gene:** HEXB. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000521:1-14. **Detection Rate:** French Canadian or Cajun 99%.
Segawa Syndrome - **Gene:** TH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000360:1-13. **Detection Rate:** French Canadian or Cajun >99%.
Short Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADS. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000017:1-10. **Detection Rate:** French Canadian or Cajun >99%.
Sjogren-Larsson Syndrome - **Gene:** ALDH3A2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000382:1-10. **Detection Rate:** French Canadian or Cajun 97%.
Smith-Lemli-Opitz Syndrome - **Gene:** DHCR7. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001360:3-9. **Detection Rate:** French Canadian or Cajun >99%.
Spastic Paraplegia Type 15 - **Gene:** ZFYVE26. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_015346:2-42. **Detection Rate:** French Canadian or Cajun >99%.
Spinal Muscular Atrophy - **Gene:** SMN1. Autosomal Recessive. Spinal Muscular Atrophy. **Variant (1):** SMN1 copy number. **Detection Rate:** French Canadian or Cajun 94%.
Spondylothoracic Dysostosis - **Gene:** MESP2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_001039958:1-2. **Detection Rate:** French Canadian or Cajun >99%.
Sulfate Transporter-related Osteochondrodysplasia - **Gene:** SLC26A2. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000112:2-3. **Detection Rate:** French Canadian or Cajun >99%.
TGM1-related Autosomal Recessive Congenital Ichthyosis - **Gene:** TGM1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000359:2-15. **Detection Rate:** French Canadian or Cajun >99%.
TPP1-related Neuronal Ceroid Lipofuscinosis - **Gene:** TPP1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000391:1-13. **Detection Rate:** French Canadian or Cajun >99%.
Tyrosinemia Type I - **Gene:** FAH. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000137:1-14. **Detection Rate:** French Canadian or Cajun >99%.
Tyrosinemia Type II - **Gene:** TAT. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000353:2-12. **Detection Rate:** French Canadian or Cajun >99%.
USH1C-related Disorders - **Gene:** USH1C. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_153676:1-27. **Detection Rate:** French Canadian or Cajun >99%.
USH2A-related Disorders - **Gene:** USH2A. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_206933:2-72. **Detection Rate:** French Canadian or Cajun 94%.
Usher Syndrome Type 3 - **Gene:** CLRN1. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_174878:1-3. **Detection Rate:** French Canadian or Cajun >99%.
Very Long Chain Acyl-CoA Dehydrogenase Deficiency - **Gene:** ACADVL. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000018:1-20. **Detection Rate:** French Canadian or Cajun >99%.
Wilson Disease - **Gene:** ATP7B. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000053:1-21. **Detection Rate:** French Canadian or Cajun >99%.
X-linked Adrenoleukodystrophy - **Gene:** ABCD1. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000033:1-6. **Detection Rate:** French Canadian or Cajun 77%.
X-linked Alport Syndrome - **Gene:** COL4A5. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000495:1-51. **Detection Rate:** French Canadian or Cajun 95%.
X-linked Congenital Adrenal Hypoplasia - **Gene:** NR0B1. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000475:1-2. **Detection Rate:** French Canadian or Cajun 99%.
X-linked Juvenile Retinoschisis - **Gene:** RS1. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000330:1-6. **Detection Rate:** French Canadian or Cajun 98%.
X-linked Myotubular Myopathy - **Gene:** MTM1. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000252:2-15. **Detection Rate:** French Canadian or Cajun 98%.
X-linked Severe Combined Immunodeficiency - **Gene:** IL2RG. X-linked Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000206:1-8. **Detection Rate:** French Canadian or Cajun >99%.
Xeroderma Pigmentosum Group A - **Gene:** XPA. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_000380:1-6. **Detection Rate:** French Canadian or Cajun >99%.
Xeroderma Pigmentosum Group C - **Gene:** XPC. Autosomal Recessive. Sequencing with Copy Number Analysis. **Exons:** NM_004628:1-16. **Detection Rate:** French Canadian or Cajun 97%.

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 10229 Residual Risk	Reproductive Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 3,300	< 1 in 1,000,000
21-hydroxylase-deficient Congenital Adrenal Hyperplasia	1 in 1,400	1 in 310,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency	< 1 in 50,000	< 1 in 1,000,000
ABCC8-related Hyperinsulinism	1 in 11,000	< 1 in 1,000,000
Adenosine Deaminase Deficiency	1 in 39,000	< 1 in 1,000,000
Alpha Thalassemia	Alpha globin status: aa/aa.	Not calculated
Alpha-mannosidosis	1 in 35,000	< 1 in 1,000,000
Alpha-sarcoglycanopathy	1 in 45,000	< 1 in 1,000,000
Alstrom Syndrome	< 1 in 50,000	< 1 in 1,000,000
AMT-related Glycine Encephalopathy	1 in 22,000	< 1 in 1,000,000
Andermann Syndrome	1 in 2,200	1 in 210,000
Argininemia	< 1 in 17,000	< 1 in 1,000,000
Argininosuccinic Aciduria	1 in 13,000	< 1 in 1,000,000
ARSACS	1 in 1,900	1 in 160,000
Aspartylglycosaminuria	< 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 16,000	< 1 in 1,000,000
ATP7A-related Disorders	< 1 in 1,000,000	1 in 600,000
Autosomal Recessive Osteopetrosis Type 1	1 in 35,000	< 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
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
Beta-sarcoglycanopathy	< 1 in 50,000	< 1 in 1,000,000
Biotinidase Deficiency	NM_000060.2(BTD):c.1330G>C(D444H) heterozygote †	1 in 630
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 31,000	< 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 50,000	< 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 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 50,000	< 1 in 1,000,000
CLN6-related Neuronal Ceroid Lipofuscinosis	1 in 43,000	< 1 in 1,000,000
Cohen Syndrome	< 1 in 15,000	< 1 in 1,000,000
COL4A3-related Alport Syndrome	1 in 11,000	< 1 in 1,000,000
COL4A4-related Alport Syndrome	1 in 21,000	< 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 50,000	< 1 in 1,000,000
Congenital Disorder of Glycosylation Type Ic	< 1 in 50,000	< 1 in 1,000,000
Congenital Finnish Nephrosis	< 1 in 50,000	< 1 in 1,000,000
Costeff Optic Atrophy Syndrome	< 1 in 50,000	< 1 in 1,000,000
Cystic Fibrosis	1 in 1,500	1 in 87,000
Cystinosis	1 in 22,000	< 1 in 1,000,000
D-bifunctional Protein Deficiency	1 in 9,000	< 1 in 1,000,000

Disease	DONOR 10229 Residual Risk	Reproductive Risk
Delta-sarcoglycanopathy	< 1 in 40,000	< 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 19,000	< 1 in 1,000,000
ERCC8-related Disorders	1 in 7,300	< 1 in 1,000,000
EVC-related Ellis-van Creveld Syndrome	1 in 7,500	< 1 in 1,000,000
EVC2-related Ellis-van Creveld Syndrome	< 1 in 50,000	< 1 in 1,000,000
Fabry Disease	< 1 in 1,000,000	1 in 80,000
Familial Dysautonomia	< 1 in 50,000	< 1 in 1,000,000
Familial Mediterranean Fever	< 1 in 50,000	< 1 in 1,000,000
Fanconi Anemia Complementation Group A	1 in 3,100	< 1 in 1,000,000
Fanconi Anemia Type C	1 in 16,000	< 1 in 1,000,000
FKRP-related Disorders	1 in 19,000	< 1 in 1,000,000
FKTN-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Galactokinase Deficiency	1 in 35,000	< 1 in 1,000,000
Galactosemia	1 in 8,600	< 1 in 1,000,000
Gamma-sarcoglycanopathy	1 in 3,000	< 1 in 1,000,000
Gaucher Disease	1 in 280	1 in 120,000
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness	1 in 4,100	1 in 690,000
GLB1-related Disorders	1 in 19,000	< 1 in 1,000,000
GLDC-related Glycine Encephalopathy	1 in 2,800	< 1 in 1,000,000
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
GNPTAB-related Disorders	1 in 3,900	1 in 610,000
GRACILE Syndrome	< 1 in 50,000	< 1 in 1,000,000
HADHA-related Disorders	1 in 15,000	< 1 in 1,000,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease)	1 in 16,000	< 1 in 1,000,000
Hereditary Fructose Intolerance	1 in 8,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related	< 1 in 50,000	< 1 in 1,000,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related	< 1 in 50,000	< 1 in 1,000,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease)	1 in 3,000	1 in 350,000
HMG-CoA Lyase Deficiency	< 1 in 33,000	< 1 in 1,000,000
Holocarboxylase Synthetase Deficiency	1 in 15,000	< 1 in 1,000,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency	1 in 25,000	< 1 in 1,000,000
Hydrolethalus Syndrome	< 1 in 50,000	< 1 in 1,000,000
Hypophosphatasia, Autosomal Recessive	1 in 16,000	< 1 in 1,000,000
Inclusion Body Myopathy 2	< 1 in 50,000	< 1 in 1,000,000
Isovaleric Acidemia	1 in 25,000	< 1 in 1,000,000
Joubert Syndrome 2	< 1 in 50,000	< 1 in 1,000,000
KCNJ11-related Familial Hyperinsulinism	< 1 in 50,000	< 1 in 1,000,000
Krabbe Disease	1 in 15,000	< 1 in 1,000,000
LAMA2-related Muscular Dystrophy	1 in 17,000	< 1 in 1,000,000
Leigh Syndrome, French-Canadian Type	1 in 2,200	1 in 200,000
Lipoamide Dehydrogenase Deficiency	< 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 30,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type 1B	1 in 25,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type Ia	1 in 29,000	< 1 in 1,000,000
Maple Syrup Urine Disease Type II	1 in 9,600	< 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 50,000	< 1 in 1,000,000
Metachromatic Leukodystrophy	1 in 20,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblA Type	1 in 47,000	< 1 in 1,000,000
Methylmalonic Acidemia, cblB Type	< 1 in 66,000	< 1 in 1,000,000
Methylmalonic Aciduria and Homocystinuria, cblC Type	1 in 23,000	< 1 in 1,000,000
MKS1-related Disorders	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis III Gamma	< 1 in 50,000	< 1 in 1,000,000
Mucolipidosis IV	< 1 in 50,000	< 1 in 1,000,000

Disease	DONOR 10229 Residual Risk	Reproductive Risk
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 16,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIB	1 in 31,000	< 1 in 1,000,000
Mucopolysaccharidosis Type IIIC	1 in 43,000	< 1 in 1,000,000
Muscle-eye-brain Disease	< 1 in 12,000	< 1 in 1,000,000
MUT-related Methylmalonic Acidemia	1 in 38,000	< 1 in 1,000,000
MYO7A-related Disorders	1 in 15,000	< 1 in 1,000,000
NEB-related Nemaline Myopathy	< 1 in 6,700	< 1 in 1,000,000
Nephrotic Syndrome, NPHS2-related	1 in 35,000	< 1 in 1,000,000
Niemann-Pick Disease Type C	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-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 50,000	< 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 5,300	< 1 in 1,000,000
Pendred Syndrome	1 in 7,000	< 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 1,600	1 in 360,000
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
PEX1-related Zellweger Syndrome Spectrum	1 in 11,000	< 1 in 1,000,000
Phenylalanine Hydroxylase Deficiency	1 in 5,000	1 in 990,000
PKHD1-related Autosomal Recessive Polycystic Kidney Disease	1 in 6,100	< 1 in 1,000,000
Polyglandular Autoimmune Syndrome Type 1	< 1 in 50,000	< 1 in 1,000,000
Pompe Disease	1 in 16,000	< 1 in 1,000,000
PPT1-related Neuronal Ceroid Lipofuscinosis	< 1 in 50,000	< 1 in 1,000,000
Primary Carnitine Deficiency	1 in 16,000	< 1 in 1,000,000
Primary Hyperoxaluria Type 1	1 in 35,000	< 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
PROP1-related Combined Pituitary Hormone Deficiency	1 in 11,000	< 1 in 1,000,000
Pycnodysostosis	< 1 in 50,000	< 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
Salla Disease	< 1 in 30,000	< 1 in 1,000,000
Sandhoff Disease	1 in 11,000	< 1 in 1,000,000
Segawa Syndrome	< 1 in 50,000	< 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 9,100	< 1 in 1,000,000
Smith-Lemli-Opitz Syndrome	1 in 10,000	< 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 570	1 in 79,000
Spondylothoracic Dysostosis	< 1 in 50,000	< 1 in 1,000,000
Sulfate Transporter-related Osteochondrodysplasia	1 in 11,000	< 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
Tyrosinemia Type I	1 in 6,500	< 1 in 1,000,000
Tyrosinemia Type II	1 in 25,000	< 1 in 1,000,000
USH1C-related Disorders	1 in 23,000	< 1 in 1,000,000
USH2A-related Disorders	1 in 2,200	< 1 in 1,000,000
Usher Syndrome Type 3	< 1 in 50,000	< 1 in 1,000,000
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	1 in 8,800	< 1 in 1,000,000
Wilson Disease	1 in 8,600	< 1 in 1,000,000
X-linked Adrenoleukodystrophy	1 in 90,000	1 in 42,000
X-linked Alport Syndrome	Not calculated	Not calculated

Counsyl

RESULTS RECIPIENT
SEATTLE SPERM BANK
Attn: Dr. Jeffrey Olliffe
NPI: 1306838271
Report Date: 05/07/2018

MALE
DONOR 10229
DOB: [redacted]
Ethnicity: French Canadian or Cajun
Barcode: 11004212319624

FEMALE
N/A

Disease	DONOR 10229 Residual Risk	Reproductive Risk
X-linked Congenital Adrenal Hypoplasia	< 1 in 1,000,000	< 1 in 1,000,000
X-linked Juvenile Retinoschisis	< 1 in 1,000,000	1 in 50,000
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