GENETIC TEST SUMMARY
Donor: 12345

ANCESTRY	JEWISH ANCESTRY?[X]
English, Norwegian	No

IT IS STRONGLY RECOMMENDED THAT YOU DISCUSS THE DONOR'S RESULTS WITH YOUR PHYSICIAN **PRIOR TO SHIPMENT** TO VERIFY THAT THIS DONOR IS SUITABLE FOR YOUR USE.

TEST RESULTS (TOTAL: 284)

GENETIC TEST	RESULTS	DETAILS/ESTIMATED INDIVIDUAL RISK**
Chromosome (karyotype) analysis	Normal male karyo	No evidence of a clinically significant chromosome abnormality
Hemoglobin evaluation	Normal hemoglobin Fractionation and MCH Results	Reduced risk to be a carrier for sickle cell anemia, thalassemias, and other hemoglobinopathies
Expanded Carrier Screening	Attached	Some donors may have positive carrier screening results for one or more of the conditions tested. Please see the following report for details.

ALL people carry genetic mutations for disorders inherited in an autosomal recessive (AR) manner, and some of these mutations can be detected by carrier screening. A person's offspring are not expected to develop that condition unless they inherit mutations for the same AR condition from BOTH parents. Therefore, CCB strongly recommends that all recipients and their physicians discuss a donor's genetic test results PRIOR to shipment of a donor's specimens, to ensure that the results are suitable for the recipient's reproductive plans. CCB also recommends that the recipient meet with a genetic counselor who can help to explain the donor's results and testing options that may be appropriate for the recipient to consider.

Genetic testing can only reduce the chance for specific inherited conditions in a donor's offspring; it cannot eliminate the risks for those specific disorders or other untested conditions. There is always a 3 to 4% chance to have a child with a medical issue, regardless of the screening performed.

[X] Please see the Donor Profile for details on the type of Jewish ancestry (Ashkenazi, Sephardic, maternal, paternal, etc.).

sema4
a Mount Sinai venture

CARRIER SCREENING REPORT

Patient	Sample	Referring Doctor
Patient Name: Donor 12345 **Date of Birth:** [redacted]1994 **Reference #:** [redacted] **Indication:** Carrier Testing **Test Type:** Expanded Carrier Screen (281)	**Specimen Type:** Blood **Lab #:** [redacted] **Date Collected:** 9/29/2017 **Date Received:** 9/29/2017 **Final Report:** 10/13/2017	**Jaime Marie Shamonki, M.D.** **California Cryobank-Genetics Department** **11915 La Grange Ave** **Los Angeles, CA 90025** Fax: **888-317-4725**

RESULT SUMMARY

THIS PATIENT WAS TESTED FOR 281 DISEASES

POSITIVE for Salla disease

A heterozygous (one copy) pathogenic variant, c.1138_1139delGT, p.V380SfsX8, was detected in the *SLC17A5* gene

NEGATIVE for the remaining diseases

Recommendations

Testing the partner for the above positive disorder(s) and genetic counseling are recommended.

Please note that for female carriers of X-linked diseases, follow-up testing of a male partner is not indicated. In addition, CGG repeat analysis of *FMR1* for fragile X syndrome is not performed on males as repeat expansion of premutation alleles is not expected in the male germline.

Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.

Consideration of residual risk by ethnicity after a negative carrier screen is recommended for the other diseases on the panel, especially in the case of a positive family history for a specific disorder.

Interpretation for Salla disease

A heterozygous (one copy) frameshift variant, c.1138_1139delGT, p.V380SfsX8, was detected in the *SLC17A5* gene (NM_012434.4). This variant is considered to be pathogenic and when present *in trans* with a pathogenic variant causative for Salla disease. Therefore, this individual is expected to be at least a carrier for Salla disease. Heterozygous carriers are not expected to exhibit symptoms of this disease.

What is Salla disease?

Salla disease is an autosomal recessive disease that is caused by pathogenic variants in the *SLC17A5* gene. While diagnosed in individuals of various ethnicities, a higher prevalence of this disease is found in individuals of

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Finnish, Swedish, or Canadian Inuit descent. Salla disease is a progressive neurologic disorder that can present clinically with ataxia, psychomotor delay, and intellectual disability. This condition has a severe infantile form that presents prenatally or at birth and a later onset, milder form that presents with hypotonia during the first year of life. While the infantile onset form of this disease is often fatal in early childhood, individuals with the milder form can survive into adulthood. Several specific variants have been associated with more severe or milder phenotypes, and therefore the disease severity may be predicted in some patients based on the inherited variants.

This patient was tested for a panel of diseases using a combination of sequencing, targeted genotyping and copy number analysis. Please note that negative results reduce but do not eliminate the possibility that this individual is a carrier for one or more of the disorders tested. Please see *Table of Residual Risks Based on Ethnicity* for specific detection rates, exons sequenced, number of variants tested and residual risk estimates after a negative screening result. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate. Detection rates were determined based on the exons and list of pathogenic variants that are guaranteed by this testing. Please note that additional variants not guaranteed by this test may be identified by sequencing. Only variants determined to be pathogenic or likely pathogenic are reported in this carrier screening test.

TEST SPECIFIC RESULTS

Alpha-thalassemia

NEGATIVE for alpha-thalassemia
HBA1 copy number: 2
HBA2 copy number: 2
No pathogenic variants detected (aa/aa)
Reduced risk of being an alpha-thalassemia carrier

Genes analyzed: *HBA1* (NM_000558.4) and *HBA2* (NM_000517.4)
Inheritance: Autosomal Recessive

Recommendations
Individuals of Asian, African, Hispanic and Mediterranean ancestry should also be screened for hemoglobinopathies by CBC and hemoglobin electrophoresis.

Interpretation
No pathogenic variants were detected in this patient, suggesting that four copies of the alpha-globin gene are present (aa/aa). Typically, individuals have four functional alpha-globin genes: 2 copies of *HBA1* and 2 copies

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

of *HBA2*, whose expression is regulated by a cis-acting regulatory element HS-40. Alpha-thalassemia carriers have three (silent carrier) or two (carrier of the alpha-thalassemia trait) functional alpha-globin genes with or without a mild phenotype. Individuals with only one functional alpha-globin gene have HbH disease with microcytic, hypochromic hemolytic anemia and hepatosplenomegaly. Loss of all four alpha-globin genes results in Hb Barts syndrome with the accumulation of Hb Barts in red blood cells and hydrops fetalis, which is fatal in utero or shortly after birth.

This individual was negative for all *HBA* deletions, duplications and variants that were tested. These negative results reduce but do not eliminate the possibility that this individual is a carrier. See *Table of Residual Risks Based on Ethnicity*. With individuals of mixed ethnicity, it is recommended to use the highest residual risk estimate.

12345

Table of Residual Risks Based on Ethnicity

Ethnicity	Carrier Frequency	Detection Rate	Residual Risk
Caucasian	1 in 500	90%	1 in 4991
African American	1 in 30	90%	1 in 291
Asian	1 in 20	90%	1 in 191
Worldwide	1 in 25	90%	1 in 241

Spinal Muscular Atrophy

NEGATIVE for spinal muscular atrophy
SMN1 Copy Number: 2
SMN2 Copy Number: 2
g.27134T>G: g.27134T>G negative

Negative copy number result
Decreased risk of being an *SMN1* silent (2+0) carrier (see *SMA Table*)

Genes analyzed: *SMN1* (NM_000344.3) and *SMN2* (NM_017411.3)
Inheritance: Autosomal Recessive

Recommendations
Consideration of residual risk by ethnicity after a negative carrier screen is recommended, especially in the case of a positive family history for spinal muscular atrophy.

Interpretation
This patient is negative for loss of *SMN1* copy number. Complete loss of *SMN1* is causative in spinal muscular atrophy (SMA). Two copies of *SMN1* were detected in this individual, which significantly reduces the risk of being an SMA carrier. Parallel testing to assess the presence of an *SMN1* duplication allele was also performed to detect a single nucleotide polymorphism (SNP), g.27134T>G, in intron 7 of the *SMN1* gene. This individual was found to be negative for this change and is therefore, at a decreased risk of being a silent (2+0) carrier, see *SMA Table* for residual risk estimates based on ethnicity.

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

SMA Table: Carrier detection and residual risk estimates before and after testing for g.27134T>G

Ethnicity	Carrier Frequency	Detection rate	Residual risk after negative result*	Detection rate with *SMN1* g.27134T>G	Residual risk g.27134T>G* negative	Residual risk g.27134T>G* positive
Ashkenazi Jewish	1 in 41	90%	1 in 345	94%	1 in 580	^likely carrier
Asian	1 in 53	92.6%	1 in 628	93.3%	1 in 702	^likely carrier
African American	1 in 66	71.1%	1 in 121	N/A	1 in 396	1 in 34
Hispanic	1 in 117	90.6%	1 in 1061	N/A	1 in 1762	1 in 140
Caucasian	1 in 35	94.9%	1 in 632	N/A	1 in 769	1 in 29

*Residual risk with two copies *SMN1* detected using dosage sensitive methods. The presence of three or more copies of *SMN1* reduces the risk of being an *SMN1* carrier between 5 - 10 fold, depending on ethnicity.
FOR INDIVIDUALS WITH MIXED ETHNICITY, USE HIGHEST RESIDUAL RISK ESTIMATE
^ Parental follow-up will be requested for confirmation

Tay-Sachs Disease Enzyme Analysis

Results: **Non-carrier**

Specimen	Hexosaminidase Activity	Hex A%	Non-Carrier Range	Comment
Tay-Sachs WBC	1890 nmol/hr/mg	66.5	55.0 - 72.0	Non-Carrier
Tay-Sachs Plasma	258 nmol/hr/ml	59.0	58.0 - 72.0	Non-Carrier

Expected Carrier Ranges:
Hex A% <54% (Serum/Plasma), Hex A%<50% (WBC)

Interpretation:

The test was performed in the patient's plasma and white blood cells (WBC). The Hex A% activities are both within the non-carrier range. These findings are consistent with the patient being a **non-carrier** for Tay-Sachs disease.

Fragile X syndrome

Fragile X CGG triplet repeat expansion testing was not performed at this time, as the patient has either been previously tested or is a male. Sequencing of the *FMR1* gene by next generation sequencing did not identify any clinically significant variants.

This case has been reviewed and electronically signed by Ashley Birch, Ph.D., DABMGG, FCCMG, Associate Laboratory Director

Laboratory Medical Consultant: George A. Diaz, M.D., Ph.D.

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Test Methods and Comments

Genomic DNA isolated from this patient was analyzed by one or more of the following methodologies, as applicable:

Fragile X CGG Repeat Analysis
PCR amplification using Asuragen, Inc. AmplideX® *FMR1* PCR reagents followed by capillary electrophoresis for allele sizing was performed. Samples positive for *FMR1* CGG repeats in the premutation and full mutation size range are further analyzed by Southern blot analysis to assess the size and methylation status of the *FMR1* CGG repeat.

Genotyping
Multiplex PCR amplification and allele specific primer extension analyses using the MassARRAY® System or Luminex® xMAP® technology were used to identify variants that are complex in nature or are present in low copy repeat regions and are, therefore, not amenable to Next Generation Sequencing technologies. Rare sequence variants may interfere with assay performance.

Multiplex Ligation-Dependent Probe Amplification (MLPA)
MLPA® probe sets and reagents, MRC-Holland, were used for the analysis of copy number of specific targets versus known control samples. Each target region was assayed with two adjacent oligonucleotide probes which following hybridization were ligated and used as template for subsequent rounds of amplification. Each complete probe within the assay has a unique length and amplicons are separated and identified by capillary electrophoresis. False positive or negative results may also occur due to rare sequence variants in target regions detected by MLPA probes. Analytical sensitivity and specificity of the MLPA method are both 99%.

For Alpha Thalassemia, the copy numbers of the *HBA1* and *HBA2* genes were analyzed. Alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation are assessed. However, it does not detect all known alpha-thalassemia mutations such as point mutations. In addition, carriers of alpha-thalassemia with three or more *HBA* copies on one chromosome, and one or no copies on the other chromosome, will not be detected. This test detects most alpha-globin gene deletions, triplications, and the Constant Spring (CS) mutation using Multiplex Ligation-Dependent Probe Amplification (MLPA). It is expected to detect approximately 90% of all alpha-thalassemia mutations, varying by ethnicity. Therefore, this result reduces, but does not eliminate, the chance that this patient is a carrier of alpha-thalassemia. With the exception of triplications, other benign alpha-globin gene polymorphisms will not be reported.

For Duchenne Muscular Dystrophy, the copy numbers of all *DMD* exons were analyzed. Please note that single-exon deletions and duplications will not be reported unless they are confirmed by NGS data (for example, if breakpoints occurring in an exon can be visualized).

For Spinal Muscular Atrophy (SMA), the copy numbers of the *SMN1* and *SMN2* genes were analyzed. The individual dosage of exons 7 and 8 as well as the combined dosage of exons 1, 4, 6 and 8 of *SMN1* and *SMN2* were assessed. Copy number gains and losses can be detected with this assay.

Depending on ethnicity 6 - 29 % of carriers will not be identified by dosage sensitive methods as this testing cannot detect individuals with two copies (duplication) of the *SMN1* gene on one chromosome and loss of *SMN1* (deletion) on the other chromosome (silent 2+0 carrier) or individuals that carry an intragenic mutation in *SMN1*. Please also note that 2% of individuals with SMA have an *SMN1* mutation that occurred *de novo*. Typically in these cases, only one parent is an SMA carrier.

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

The presence of the g.27134T>G variant allele in an individual with Ashkenazi Jewish or Asian ancestry is indicative of a duplication of *SMN1*. When present in an Ashkenazi Jewish or Asian individual with two copies of *SMN1*, g.27134T>G is likely indicative of a silent (2+0) carrier. In individuals with two copies of *SMN1* with African American, Hispanic or Caucasian ancestry, the presence or absence of g.27134T>G significantly increases or decreases, respectively, the likelihood of being a silent 2+0 silent carrier.

Next Generation Sequencing (NGS)
NGS was performed on a panel of genes for the purpose of identifying pathogenic or likely pathogenic variants.

Agilent SureSelect™QXT technology was used with custom capture library to target the guaranteed list of mutations and exonic regions of the relevant genes. These targeted regions were sequenced using the Illumina HiSeq2500 system with 100 bp paired-end reads. The DNA sequences were mapped to and analyzed in comparison with the published human genome build UCSC hg19 reference sequence. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes were assessed for the average depth of coverage and data quality threshold values. This technology may not detect all small insertion/deletions and is not diagnostic for large duplications/deletions, repeat expansions, and structural genomic variation. This test will detect variants within the exons and the intron-exon boundaries of the target regions. Variants outside these regions will either not be detected or are not guaranteed to be detected. These regions include, but are not limited to, UTRs, promoters, and deep intronic areas or regions that fall within low copy repeat segments. In addition, a mutation(s) in a gene not included on the panel could be present in this patient. All potentially pathogenic variants may be confirmed by either a specific genotyping assay or Sanger sequencing, if indicated. Any benign variants, likely benign variants or variants of uncertain significance identified during this analysis were not reported.

Sanger Sequencing
Sanger sequencing, as indicated, was performed in both directions using BigDye Terminator chemistry with the ABI 3730 DNA analyzer with target specific amplicons. It also may be used to supplement specific guaranteed target regions that fail NGS sequencing due to poor quality or low depth of coverage <20 reads or as a confirmatory method for NGS positive results. False negative results may occur if rare variants interfere with amplification or annealing.

Tay-Sachs Disease (TSD) Enzyme Analysis
Hexosaminidase activity and Hex A% activity were measured by a standard heat-inactivation, fluorometric method using artificial 4-MU-β-N-acetyl glucosaminide (4-MUG) substrate. This assay is highly sensitive and accurate in detecting Tay-Sachs carriers and individuals affected with TSD. It is estimated that less than 0.5% of Tay-Sachs carriers have non-carrier levels of percent Hex A activity, and therefore may not be identified by this assay. In addition, this assay may detect individuals that are carriers of or are affected with Sandhoff Disease. False positive results, such as pseudodeficiency alleles, may occur if benign variants interfere with the enzymatic assay. False negative results may occur if both *HEXA* and *HEXB* pathogenic variants are present in the same individual.

Please note these tests were developed and their performance characteristics were determined by Mount Sinai Genomics, Inc. They have not been cleared or approved by the FDA. These analyses generally provide highly accurate information regarding the patient's carrier or affected status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

SELECTED REFERENCES
Carrier Screening
Grody W et al. ACMG position statement on prenatal/preconception expanded carrier screening. *Genet Med.* 2013 15:482-

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

3.
Umbarger MA. Next-generation carrier screening. *Genet Med*. 2014 16:132-40.

Alpha-thalassemia:

Galanello R et al. Gene test review: Alpha-thalassemia. *Genet Med*. 2011 13:83-8.

Waye JS et al. Diagnostic testing for α-globin gene disorders in a heterogeneous North American population. *Int J Lab Hematol*. 2013 35:306-13.

Cystic Fibrosis:

ACOG Committee Opinion. Number 325, Update on carrier screening for cystic fibrosis. 2005.

Fragile X syndrome:

Chen L et al. An information-rich CGG repeat primed PCR that detects the full range of Fragile X expanded alleles and minimizes the need for Southern blot analysis. *J Mol Diag* 2010 12:589-600.

Spinal Muscular Atrophy:

Hendrickson BC et al. Differences in SMN1 allele frequencies among ethnic groups within North America. *J Med Genet*. 2009 46:641-4.

Ogino S et al. Genetic risk assessment in carrier testing for spinal muscular atrophy. *Am J Med Genet*. 2002 110:301-7.

Luo M et al. An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. *Genet Med*. 2014 16:149-56.

Ashkenazi Jewish Disorders:

Scott SA et al. Experience with carrier screening and prenatal diagnosis for sixteen Ashkenazi Jewish Genetic Diseases. *Hum. Mutat.* 2010 31:1-11.

Duchenne Muscular Dystrophy:

Aartsma-Rus A et al. Entries in the Leiden Duchenne muscular dystrophy mutation database: an overview of mutation types and paradoxical cases that confirm the reading-frame rule. *Muscle Nerve*. 2006b 34:135-44.

Flanigan KM et al. Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat*. 2009 30:1657-66.

Beta Globin-related Disorders:

Cao A et al. Beta-Thalassemia. GeneReviews (http://www.ncbi.nlm.nih.gov/books/NBK1426/)

Modell B et al. Epidemiology of haemoglobin disorders in Europe: an overview. Scand J Clin Lab Invest. 2007 67:39-69.

For further reading:

Orphanet: http://www.orpha.net/consor/cgi-bin/index.php

GeneReviews: <http://www.ncbi.nlm.nih.gov/books/NBK1116/>

For Disease Specific Standards and Guidelines:

https://www.acmg.net/

Additional disease-specific references available upon request.

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Table of Residual Risks by Ethnicity

Please note: This table displays residual risks after a negative result for each of the genes and corresponding disorders. **If a patient is reported to be a carrier of a disease, their residual risk is 1 and this table does not apply for that disease.**

Disease (Inheritance)	Gene	Ethnicity	Carrier Frequency	Detection Rate	Residual Risk	Analytical Detection Rate
Abetalipoproteinemia (AR)	*MTTP*	Caucasian	< 1 in 500	81%	1 in 2627	>95%
NM_000253.3		Ashkenazi Jewish	1 in 186	>95%	1 in 3701	>95%
		Worldwide	< 1 in 500	70%	1 in 1664	95%
Achromatopsia (AR)	*CNGB3*	Caucasian	1 in 91	85%	1 in 601	>95%
NM_019098.4		Worldwide	1 in 98	>95%	1 in 1941	>95%
Acrodermatitis Enteropathica (AR)	*SLC39A4*	Worldwide	1 in 354	75%	1 in 1413	>95%
NM_130849.3						
Acute Infantile Liver Failure (AR)	*TRMU*	Worldwide	< 1 in 500	87%	1 in 3839	>95%
NM_018006.4		Sephardic Jewish - Yemenite	1 in 34	81%	1 in 175	94%
Acyl-CoA Oxidase I Deficiency (AR)	*ACOX1*	Worldwide	< 1 in 500	80%	1 in 2496	91%
NM_004035.6						
Adenosine Deaminase Deficiency (AR)	*ADA*	Worldwide	1 in 337	73%	1 in 1245	>95%
NM_000022.2						
Adrenoleukodystrophy, X-Linked (XL)	*ABCD1*	Worldwide	< 1 in 500	47%	1 in 943	85%
NM_000033.3		Sephardic Jewish	< 1 in 500	73%	1 in 1849	>95%
Exception: Exons 8 and 9						
Aicardi-Goutières Syndrome (*SAMHD1*-Related) (AR)	*SAMHD1*	Worldwide	< 1 in 500	84%	1 in 3120	93%
NM_015474.3						
Alpha-Mannosidosis (AR)	*MAN2B1*	Caucasian	1 in 485	85%	1 in 3228	94%
NM_000528.3		Worldwide	< 1 in 500	81%	1 in 2627	>95%
Alpha-Thalassemia (AR)	*HBA1* / *HBA2*	Caucasian	1 in 500	90%	1 in 4991	90%
NM_000558.4 / NM_000517.4		African American	1 in 30	90%	1 in 291	90%
		Asian	1 in 20	90%	1 in 191	90%
MLPA only (Copy-number analysis)		Worldwide	1 in 25	90%	1 in 241	90%
Alpha-Thalassemia Mental Retardation Syndrome (XL)	*ATRX*	Worldwide	< 1 in 500	56%	1 in 1135	95%
NM_000489.4						
Alport Syndrome (*COL4A3*-Related) (AR)	*COL4A3*	Caucasian	1 in 284	86%	1 in 2022	95%
NM_000091.4		Ashkenazi Jewish	1 in 192	>95%	1 in 3821	>95%
		Worldwide	1 in 354	87%	1 in 2716	>95%
Alport Syndrome (*COL4A4*-Related) (AR)	*COL4A4*	Worldwide	1 in 353	88%	1 in 2934	>95%
NM_000092.4						
Alport Syndrome (*COL4A5*-Related) (XL)	*COL4A5*	Worldwide	< 1 in 500	83%	1 in 2936	88%
NM_000495.3						
Alstrom Syndrome (AR)	*ALMS1*	Worldwide	1 in 500	79%	1 in 2377	>95%
NM_015120.4						

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Andermann Syndrome (AR)	*SLC12A6*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
NM_133647.1		French Canadian - Saguenay Lac-St. Jean	1 in 23	>95%	1 in 441	>95%
Argininosuccinic Aciduria (AR)	*ASL*	Worldwide	1 in 274	62%	1 in 719	>95%
NM_000048.3						
Aromatase Deficiency (AR)	*CYP19A1*	Worldwide	< 1 in 500	72%	1 in 1783	81%
NM_031226.2						
Arthrogryposis, Mental Retardation, and Seizures (AR)	*SLC35A3*	Ashkenazi Jewish	1 in 453	>95%	1 in 9041	>95%
		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
NM_012243.2						
Asparagine Synthetase Deficiency (AR)	*ASNS*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
NM_001673.4		Sephardic Jewish - Iranian	1 in 80	>95%	1 in 1581	>95%
Aspartylglycosaminuria (AR)	*AGA*	Caucasian	< 1 in 500	>95%	1 in 9981	>95%
NM_000027.3		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
		Finnish	1 in 63	>95%	1 in 1241	>95%
Ataxia With Isolated Vitamin E Deficiency (AR)	*TTPA*	Caucasian	< 1 in 500	86%	1 in 3565	90%
NM_000370.3		Worldwide	< 1 in 500	93%	1 in 7130	>95%
Ataxia Telangiectasia (AR)	*ATM*	Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
NM_000051.3		Worldwide	1 in 100	75%	1 in 397	91%
		Sephardic Jewish - Moroccan	1 in 69	>95%	1 in 1361	>95%
Autosomal Recessive Spastic Ataxia Of Charlevoix-Saguenay (AR)	*SACS*	Caucasian	1 in 450	70%	1 in 1498	>95%
		Worldwide	< 1 in 500	86%	1 in 3565	>95%
NM_014363.5		French Canadian - Charlevoix-Saguenay	1 in 21	>95%	1 in 401	>95%
Bardet-Biedl Syndrome (*BBS1*-Related) (AR)	*BBS1*	Worldwide	1 in 392	93%	1 in 5587	>95%
NM_024649.4		Faroese	1 in 30	>95%	1 in 581	>95%
Bardet-Biedl Syndrome (*BBS2*-Related) (AR)	*BBS2*	Ashkenazi Jewish	1 in 140	>95%	1 in 2781	>95%
NM_031885.3		Worldwide	< 1 in 500	69%	1 in 1611	>95%
		Hutterite	1 in 22	>95%	1 in 421	>95%
Bardet-Biedl Syndrome (*BBS10*-Related) (AR)	*BBS10*	Worldwide	1 in 423	78%	1 in 1919	>95%
NM_024685.3						
Bardet-Biedl Syndrome (*BBS12*-Related) (AR)	*BBS12*	Worldwide	< 1 in 500	77%	1 in 2171	>95%
NM_152618.2						
Bare Lymphocyte Syndrome, Type II (AR)	*CIITA*	Worldwide	< 1 in 500	95%	1 in 9981	>95%
NM_000246.3						
Bartter Syndrome, Type 4A (AR)	*BSND*	Worldwide	< 1 in 500	94%	1 in 8318	>95%
NM_057176.2						
Bernard-Soulier Syndrome, Type A1 (AR)	*GP1BA*	Worldwide	< 1 in 500	84%	1 in 3120	>95%
NM_000173.5						
Bernard-Soulier Syndrome, Type C (AR)	*GP9*	Worldwide	< 1 in 500	83%	1 in 2936	>95%
NM_000174.4						
Beta-Globin Related Hemoglobinopathies: Beta- Thalassemia (AR)	*HBB*	Caucasian	1 in 373	>95%	1 in 7441	>95%
		African American	1 in 100	>95%	1 in 1981	>95%
NM_000518.4		Asian	1 in 54	>95%	1 in 1061	>95%
		Worldwide	1 in 158	>95%	1 in 3141	>95%
		Mediterranean	1 in 28	>95%	1 in 541	>95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Beta-Globin Related Hemoglobinopathies:	*HBB*	Caucasian	< 1 in 500	>99%	1 in 49901	
HbC Variant (AR)		African American	1 in 35	>99%	1 in 3401	
NM_000518.4		Hispanic	< 1 in 500	>99%	1 in 49901	>99%
		Asian	< 1 in 500	>99%	1 in 49901	
Variant Tested: c.19G>A, p.E7K		Worldwide	< 1 in 500	>99%	1 in 49901	
Beta-Globin Related Hemoglobinopathies:	*HBB*	Caucasian	< 1 in 500	>99%	1 in 49901	
HbS Variant (Sickle Cell Disease) (AR)		African American	1 in 12	>99%	1 in 1101	
NM_000518.4		Hispanic	1 in 17	>99%	1 in 1601	>99%
		Asian	< 1 in 500	>99%	1 in 49901	
Variant Tested: c.20A>T, p.E7V		Worldwide	1 in 71	>99%	1 in 7001	
3-Beta-Hydroxysteroid Deficiency (AR)	*HSD3B2*	Worldwide	< 1 in 500	58%	1 in 1189	>95%
NM_000198.3						
Beta-Ketothiolase Deficiency (AR)	*ACAT1*	Caucasian	1 in 354	46%	1 in 655	73%
NM_000019.3		Asian	1 in 289	67%	1 in 874	>95%
		Worldwide	1 in 347	60%	1 in 866	>95%
Bilateral Frontoparietal Polymicrogyria (AR)	*GPR56 (ADGRG1)*	Worldwide	< 1 in 500	80%	1 in 2496	>95%
NM_005682.6						
Biotinidase Deficiency (AR)	*BTD*	Caucasian	1 in 12	80%	1 in 56	86%
NM_000060.3		Hispanic	1 in 30	72%	1 in 105	>95%
		Worldwide	1 in 25	74%	1 in 93	>95%
Bloom Syndrome (AR)	*BLM*	Ashkenazi Jewish	1 in 134	>95%	1 in 2661	>95%
NM_000057.2		Worldwide	< 1 in 500	88%	1 in 4159	>95%
Canavan Disease (AR)	*ASPA*	Ashkenazi Jewish	1 in 55	>95%	1 in 1081	>95%
NM_000049.2		Worldwide	1 in 158	82%	1 in 873	95%
Carbamoylphosphate Synthetase I	*CPS1*	Caucasian	1 in 284	25%	1 in 378	86%
Deficiency (AR)		Asian	1 in 447	46%	1 in 827	84%
NM_001875.4		Worldwide	< 1 in 500	54%	1 in 1086	>95%
Carnitine Palmitoyltransferase IA	*CPT1A*	Worldwide	< 1 in 500	56%	1 in 1135	>95%
Deficiency (AR)		Hutterite	1 in 16	>95%	1 in 301	>95%
NM_001876.3						
Carnitine Palmitoyltransferase II	*CPT2*	Caucasian	1 in 200	91%	1 in 2212	>95%
Deficiency (AR)		African	1 in 308	75%	1 in 1229	>95%
NM_000098.2		Asian	< 1 in 500	76%	1 in 2080	>95%
		Ashkenazi Jewish	1 in 45	>95%	1 in 881	>95%
		Worldwide	1 in 182	90%	1 in 1811	>95%
Carpenter Syndrome (AR)	*RAB23*	Caucasian	< 1 in 500	95%	1 in 9981	>95%
NM_001278667.1		Worldwide	< 1 in 500	84%	1 in 3120	>95%
Cartilage-Hair Hypoplasia (AR)	*RMRP*	Worldwide	< 1 in 500	50%	1 in 999	>95%
NR_003051.3		Amish	1 in 19	>95%	1 in 361	>95%
		Finnish	1 in 76	>95%	1 in 1501	>95%
Cerebral Creatine Deficiency Syndrome 1 (XL)	*SLC6A8*	Worldwide	< 1 in 500	76%	1 in 2080	94%
NM_005629.3						
Exception: Exon 3						
Cerebral Creatine Deficiency Syndrome 2 (AR)	*GAMT*	Worldwide	< 1 in 500	71%	1 in 1722	>95%
NM_000156.5		Portuguese	1 in 125	>95%	1 in 2481	>95%
Cerebrotendinous Xanthomatosis (AR)	*CYP27A1*	Worldwide	1 in 112	71%	1 in 384	>95%
NM_000784.3		Sephardic Jewish - Moroccan	1 in 76	>95%	1 in 1501	>95%
Charcot-Marie-Tooth Disease, Type 4D (AR)	*NDRG1*	Roma	1 in 22	>95%	1 in 421	>95%
NM_001135242.1						

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Charcot-Marie-Tooth Disease, Type 5 / Arts Syndrome (XL) NM_002764.3	*PRPS1*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Charcot-Marie-Tooth Disease, X-Linked (XL) NM_000166.5	*GJB1*	Worldwide	< 1 in 500	53%	1 in 1063	>95%
Choreoacanthocytosis (AR) NM_033305.2	*VPS13A*	Worldwide Ashkenazi Jewish	< 1 in 500 N/A	88% >95%	1 in 4159 N/A	95% >95%
Choroideremia (XL) NM_000390.2	*CHM*	Worldwide	< 1 in 500	92%	1 in 6239	94%
Chronic Granulomatous Disease (*CYBA*-Related) (AR) NM_000101.2	*CYBA*	Worldwide Sephardic Jewish - Moroccan	< 1 in 500 1 in 13	57% 83%	1 in 1161 1 in 72	85% >95%
Chronic Granulomatous Disease (*CYBB*-Related) (XL) NM_000397.3	*CYBB*	Worldwide	< 1 in 500	83%	1 in 2936	94%
Citrin Deficiency (AR) NM_014251.2	*SLC25A13*	Caucasian Asian Worldwide	< 1 in 500 1 in 123 < 1 in 500	92% 95% 89%	1 in 6239 1 in 2441 1 in 4537	>95% >95% >95%
Citrullinemia, Type I (AR) NM_000050.4	*ASS1*	Caucasian Asian Worldwide	1 in 195 1 in 123 1 in 119	53% 78% 84%	1 in 414 1 in 556 1 in 739	>95% 91% >95%
Cohen Syndrome (AR) NM_017890.4	*VPS13B*	Worldwide	1 in 500	85%	1 in 3328	90%
Combined Malonic and Methylmalonic Aciduria (AR) NM_001127214.3	*ACSF3*	Worldwide	1 in 86	>95%	1 in 1701	>95%
Combined Oxidative Phosphorylation Deficiency 1 (AR) NM_024996.5	*GFM1*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Combined Oxidative Phosphorylation Deficiency 3 (AR) NM_001172696.1	*TSFM*	Finnish Worldwide	1 in 80 < 1 in 500	83% >95%	1 in 466 1 in 9981	>95% >95%
Combined Pituitary Hormone Deficiency 2 (AR) NM_006261.4	*PROP1*	Worldwide	1 in 141	>95%	1 in 2801	>95%
Combined Pituitary Hormone Deficiency 3 (AR) NM_014564.3	*LHX3*	Worldwide	< 1 in 500	88%	1 in 4159	92%
Combined SAP Deficiency (AR) NM_002778.2	*PSAP*	Worldwide	< 1 in 500	89%	1 in 4537	>95%
Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency (AR) NM_000102.3	*CYP17A1*	Worldwide	< 1 in 500	57%	1 in 1161	>95%
Congenital Amegakaryocytic Thrombocytopenia (AR) NM_005373.2	*MPL*	Caucasian Ashkenazi Jewish Worldwide	1 in 266 1 in 57 1 in 415	69% >95% 79%	1 in 856 1 in 1121 1 in 1972	>95% >95% >95%
Congenital Disorder of Glycosylation, Type Ia (AR) NM_000303.2	*PMM2*	Caucasian Asian Ashkenazi Jewish Worldwide	1 in 42 1 in 449 1 in 61 1 in 124	87% 45% >95% 84%	1 in 316 1 in 816 1 in 1201 1 in 770	>95% >95% >95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Congenital Disorder of Glycosylation, Type Ib (AR) NM_002435.2	*MPI*	Worldwide	< 1 in 500	74%	1 in 1920	>95%
Congenital Disorder of Glycosylation, Type Ic (AR) NM_013339.3	*ALG6*	Worldwide	< 1 in 500	87%	1 in 3839	>95%
Congenital Insensitivity to Pain with Anhidrosis (AR) NM_001012331.1	*NTRK1*	Asian Worldwide Sephardic Jewish - Moroccan	1 in 387 < 1 in 500 N/A	91% 91% >95%	1 in 4290 1 in 5545 N/A	>95% >95% >95%
Congenital Myasthenic Syndrome (*CHRNE*-Related) (AR) NM_000080.3	*CHRNE*	Caucasian Worldwide Southeastern European Roma	1 in 383 1 in 408 1 in 25	65% 85% >95%	1 in 1092 1 in 2714 1 in 481	>95% >95% >95%
Congenital Myasthenic Syndrome (*RAPSN*-Related) (AR) NM_005055.4	*RAPSN*	Caucasian Worldwide Sephardic Jewish - Iraqi and Iranian	1 in 176 1 in 252 N/A	90% 86% >95%	1 in 1751 1 in 1794 N/A	>95% >95% >95%
Congenital Neutropenia (*HAX1*-Related) (AR) NM_006118.3	*HAX1*	Worldwide	< 1 in 500	90%	1 in 4991	95%
Congenital Neutropenia (*VPS45*-Related) (AR) NM_001279354.1	*VPS45*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Corneal Dystrophy and Perceptive Deafness (AR) NM_032034.3	*SLC4A11*	Worldwide	< 1 in 500	60%	1 in 1249	>95%
Corticosterone Methyloxidase Deficiency (AR) NM_000498.3 *Exception:* Exons 3 - 7	*CYP11B2*	Worldwide Sephardic Jewish - Iranian	< 1 in 500 1 in 30	68% >95%	1 in 1560 1 in 581	72% >95%
Cystic Fibrosis (AR) NM_000492.3 *Exception:* Exon 10	*CFTR*	Caucasian African American Hispanic Asian Ashkenazi Jewish Worldwide	1 in 25 1 in 61 1 in 58 1 in 94 1 in 24 1 in 45	94% 87% 87% 65% >95% 88%	1 in 401 1 in 463 1 in 439 1 in 267 1 in 461 1 in 368	>95% >95% >95% >95% >95% >95%
Cystinosis (AR) NM_004937.2 	*CTNS*	Caucasian African Hispanic Asian Worldwide French Canadian - Saguenay-Lac St. Jean Sephardic Jewish - Moroccan	1 in 220 < 1 in 500 < 1 in 500 < 1 in 500 1 in 224 1 in 39 1 in 100	81% 83% 64% 90% 86% 90% 92%	1 in 1154 1 in 2936 1 in 1387 1 in 4991 1 in 1594 1 in 381 1 in 1239	90% >95% 75% >95% >95% 90% >95%
D-Bifunctional Protein Deficiency (AR) NM_000414.3	*HSD17B4*	Worldwide	< 1 in 500	64%	1 in 1387	87%
Deafness, Autosomal Recessive 77 (AR) NM_144612.6	*LOXHD1*	Ashkenazi Jewish Worldwide	1 in 180 < 1 in 500	>95% >95%	1 in 3581 1 in 9981	>95% >95%
Duchenne Muscular Dystrophy/ Becker Muscular Dystrophy (XL) NM_004006.2	*DMD*	Worldwide	< 1 in 500	90%	1 in 4991	>95%
Dyskeratosis Congenita (*RTEL1*-Related) (AR) NM_001283009.1	*RTEL1*	Ashkenazi Jewish Worldwide	1 in 165 < 1 in 500	>95% 78%	1 in 3281 1 in 2269	>95% >95%
Dystrophic Epidermolysis Bullosa (AR) NM_000094.3	*COL7A1*	Worldwide	1 in 370	85%	1 in 2461	>95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Ehlers-Danlos Syndrome, Type VIIC (AR) NM_014244.4	*ADAMTS2*	Ashkenazi Jewish Worldwide	1 in 187 < 1 in 500	>95% 91%	1 in 3721 1 in 5545	>95% 93%
Ellis-van Creveld Syndrome (*EVC*-Related) (AR) NM_153717.2 *Exception:* Exon 1	*EVC*	Worldwide Lancaster County Amish	1 in 345 1 in 12	72% >95%	1 in 1230 1 in 221	90% >95%
Emery-Dreifuss Myopathy 1 (XL) NM_000117.2	*EMD*	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Enhanced S-Cone Syndrome (AR) NM_014249.3	*NR2E3*	Ashkenazi Jewish Worldwide	N/A 1 in 204	>95% 82%	N/A 1 in 1129	>95% >95%
Ethylmalonic Encephalopathy (AR) NM_014297.3	*ETHE1*	Worldwide	< 1 in 500	72%	1 in 1783	94%
Fabry Disease (XL) NM_000169.2	*GLA*	Worldwide	< 1 in 500	74%	1 in 1920	>95%
Factor IX Deficiency (XL) NM_000133.3	*F9*	Worldwide	< 1 in 500	59%	1 in 1218	95%
Factor XI Deficiency (AR) NM_000128.3	*F11*	Caucasian Asian Ashkenazi Jewish Worldwide	1 in 101 1 in 163 1 in 11 1 in 92	36% 65% >95% 51%	1 in 157 1 in 464 1 in 201 1 in 187	>95% >95% >95% >95%
Familial Dysautonomia (AR) NM_003640.3	*IKBKAP*	Ashkenazi Jewish Worldwide	1 in 31 < 1 in 500	>95% >95%	1 in 601 1 in 9981	>95% >95%
Familial Hypercholesterolemia (AR) NM_000527.4	*LDLR*	Caucasian Ashkenazi Jewish Worldwide French Canadian Finnish South African Afrikaner	1 in 200 1 in 69 < 1 in 500 1 in 267 1 in 143 1 in 70	15% 35% 41% 17% 93% 94%	1 in 235 1 in 106 1 in 847 1 in 321 1 in 2030 1 in 1151	85% 85% 86% 26% >95% >95%
Familial Hypercholesterolemia, Autosomal Recessive (AR) NM_015627.2	*LDLRAP1*	Worldwide Sardinian	< 1 in 500 1 in 143	88% >95%	1 in 4159 1 in 2841	95% >95%
Familial Hyperinsulinism (*ABCC8*-Related) (AR) NM_000352.4	*ABCC8*	Ashkenazi Jewish Worldwide Finnish	1 in 52 1 in 167 1 in 100	>95% 56% 50%	1 in 1021 1 in 378 1 in 199	>95% >95% >95%
Familial Hyperinsulinism (*KCNJ11*-Related) (AR) NM_000525.3	*KCNJ11*	Worldwide	1 in 500	26%	1 in 675	>95%
Familial Mediterranean Fever (AR) NM_000243.2	*MEFV*	Ashkenazi Jewish Worldwide Sepharic Jewish Armenian Turkish	1 in 13 1 in 115 1 in 14 1 in 5 1 in 5	>95% 87% >95% >95% 75%	1 in 241 1 in 878 1 in 261 1 in 81 1 in 17	>95% >95% >95% >95% >95%
Fanconi Anemia, Group A (AR) NM_000135.2	*FANCA*	Worldwide Spanish Roma Sephardic Jewish - Moroccan and Tunisian	1 in 345 1 in 64 1 in 133	60% >95% 86%	1 in 861 1 in 1261 1 in 944	76% >95% >95%
Fanconi Anemia, Group C (AR) NM_000136.2	*FANCC*	Ashkenazi Jewish Worldwide	1 in 89 1 in 417	>95% >95%	1 in 1761 1 in 8321	>95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Fanconi Anemia, Group G (AR) *FANCG*	African American	1 in 100	80%	1 in 496	>95%
NM_004629.1	Worldwide	< 1 in 500	92%	1 in 6239	>95%
Fumarase Deficiency (AR) *FH*	Worldwide	< 1 in 500	49%	1 in 979	95%
NM_000143.3					
Galactokinase Deficiency (AR) *GALK1*	Asian	1 in 500	60%	1 in 1249	90%
NM_000154.1	Worldwide	1 in 122	75%	1 in 485	>95%
	Roma	1 in 47	>95%	1 in 921	>95%
Galactosemia (AR) *GALT*	Caucasian	1 in 152	91%	1 in 1679	>95%
NM_000155.3	African	1 in 87	>95%	1 in 1721	>95%
	Hispanic	1 in 305	>95%	1 in 6081	>95%
	Ashkenazi Jewish	1 in 156	>95%	1 in 3101	>95%
	Worldwide	1 in 112	88%	1 in 926	>95%
	Irish Travellers	1 in 11	>95%	1 in 201	>95%
Gaucher Disease (AR) *GBA*	Caucasian	1 in 164	67%	1 in 495	67%
NM_000157.3	Ashkenazi Jewish	1 in 15	>95%	1 in 281	>95%
	Worldwide	1 in 158	56%	1 in 358	56%
Variants tested: p.L29fs, c.115+1G>A, p.N409S, p.L422fs, p.V433L, p.D448H, p.D448V, p.L483P, p.R502C, p.R502H, p.R535C *(Genotyping only)*					
Gitelman Syndrome (AR) *SLC12A3*	Worldwide	1 in 100	51%	1 in 203	>95%
NM_000339.2					
Glutaric Acidemia, Type I (AR) *GCDH*	Caucasian	1 in 172	77%	1 in 744	>95%
NM_000159.3	African	1 in 36	>95%	1 in 701	>95%
	Worldwide	1 in 158	70%	1 in 524	>95%
	Oji-Cree First Nations (N. Manitoba)	1 in 8	>95%	1 in 141	>95%
	Old Order Amish of Pennsylvania	1 in 11	>95%	1 in 201	>95%
	Lumbee Native American	1 in 16	>95%	1 in 301	>95%
Glutaric Acidemia, Type IIa (AR) *ETFA*	Worldwide	< 1 in 500	63%	1 in 1350	95%
NM_000126.3					
Glutaric Acidemia, Type IIc (AR) *ETFDH*	Asian	1 in 87	64%	1 in 240	94%
NM_004453.3	Worldwide	1 in 250	55%	1 in 554	>95%
Glycine Encephalopathy (*AMT*-Related) (AR) *AMT*	Caucasian	1 in 271	67%	1 in 819	94%
NM_000481.3	Worldwide	1 in 319	74%	1 in 1224	>95%
Glycine Encephalopathy (*GLDC*-Related) (AR) *GLDC*	Caucasian	1 in 140	48%	1 in 268	66%
NM_000170.2	Worldwide	1 in 165	41%	1 in 279	83%
Exception: Exon 1					
Glycogen Storage Disease, Type Ia (AR) *G6PC*	Caucasian	1 in 177	88%	1 in 1468	91%
NM_000151.3	Asian	1 in 192	>95%	1 in 3821	>95%
	Ashkenazi Jewish	1 in 71	>95%	1 in 1401	>95%
	Worldwide	1 in 261	95%	1 in 5201	>95%
Glycogen Storage Disease, Type Ib (AR) *SLC37A4*	Caucasian	< 1 in 500	94%	1 in 8318	>95%
NM_001164277.1	Worldwide	1 in 354	78%	1 in 1606	>95%
Glycogen Storage Disease, Type II (AR) *GAA*	Caucasian	1 in 100	68%	1 in 310	89%
NM_000152.3	African	1 in 70	83%	1 in 407	>95%
	Asian	1 in 112	64%	1 in 309	>95%
	Ashkenazi Jewish	1 in 58	>95%	1 in 1141	>95%
	Worldwide	1 in 132	73%	1 in 486	>95%
Glycogen Storage Disease, Type III (AR) *AGL*	Worldwide	1 in 158	95%	1 in 3141	>95%
NM_000028.2	Sephardic Jewish - Moroccan	1 in 34	>95%	1 in 661	>95%
	Faroese	1 in 28	>95%	1 in 541	>95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease (AR) NM_000158.3	*GBE1*	Caucasian Ashkenazi Jewish Worldwide	1 in 144 1 in 68 1 in 387	70% >95% 73%	1 in 478 1 in 1341 1 in 1431	70% >95% 95%
Glycogen Storage Disease, Type V (AR) NM_005609.2	*PYGM*	Caucasian Sephardic Jewish - Kurdish	1 in 191 1 in 84	76% >95%	1 in 793 1 in 1661	94% >95%
Glycogen Storage Disease, Type VII (AR) NM_000289.5	*PFKM*	Ashkenazi Jewish Worldwide	1 in 250 < 1 in 500	>95% 74%	1 in 4981 1 in 1920	>95% >95%
GRACILE Syndrome and Other *BCS1L*-Related Disorders (AR) NM_001257342.1	*BCS1L*	Caucasian Worldwide Finnish	1 in 407 < 1 in 500 1 in 108	64% 95% >95%	1 in 1129 1 in 9981 1 in 2141	>95% >95% >95%
Hemochromatosis, Type 2A (AR) NM_213653.3	*HFE2*	Caucasian Worldwide	< 1 in 500 < 1 in 500	84% 79%	1 in 3120 1 in 2377	>95% >95%
Hemochromatosis, Type 3 (AR) NM_003227.3	*TFR2*	Worldwide	< 1 in 500	73%	1 in 1849	>95%
Hereditary Fructose Intolerance (AR) NM_000035.3	*ALDOB*	Caucasian African Hispanic Worldwide	1 in 80 1 in 406 1 in 275 1 in 121	95% >95% 94% 87%	1 in 1581 1 in 8101 1 in 4568 1 in 924	>95% >95% >95% >95%
Hereditary Spastic Paraparesis 49 (AR) NM_014844.4	*TECPR2*	Sephardic Jewish - Bukharian	1 in 27	>95%	1 in 521	>95%
Hermansky-Pudlak Syndrome, Type 1 (AR) NM_000195.4	*HPS1*	Worldwide Puerto Rican	< 1 in 500 1 in 59	>95% >95%	1 in 9981 1 in 1161	>95% >95%
Hermansky-Pudlak Syndrome, Type 3 (AR) NM_032383.4	*HPS3*	Ashkenazi Jewish Worldwide	1 in 235 < 1 in 500	>95% 87%	1 in 4681 1 in 3839	>95% >95%
HMG-CoA Lyase Deficiency (AR) NM_000191.2	*HMGCL*	Worldwide	< 1 in 500	73%	1 in 1849	94%
Holocarboxylase Synthetase Deficiency (AR) NM_000411.6	*HLCS*	Caucasian Asian Worldwide Faroese	1 in 500 1 in 158 1 in 500 1 in 20	83% 83% 79% >95%	1 in 2936 1 in 925 1 in 2377 1 in 381	>95% 94% >95% >95%
Homocystinuria (*CBS*-Related) (AR) NM_000071.2	*CBS*	Caucasian Worldwide Qatari	1 in 52 1 in 293 1 in 21	74% 78% 86%	1 in 197 1 in 1328 1 in 144	>95% >95% >95%
Homocystinuria due to *MTHFR* Deficiency (AR) NM_005957.4 *Variant tested:* p.G158G *(Genotyping only)*	*MTHFR*	Sephardic Jewish - Bukharian	1 in 39	>95%	1 in 761	>95%
Homocystinuria, cblE Type (AR) NM_002454.2	*MTRR*	Caucasian Worldwide	< 1 in 500 < 1 in 500	93% 73%	1 in 7130 1 in 1849	>95% >95%
Hydrolethalus Syndrome (AR) NM_001134793.1	*HYLS1*	Worldwide Finnish	1 in 455 1 in 50	>95% >95%	1 in 9081 1 in 981	>95% >95%
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome (AR) NM_014252.3	*SLC25A15*	Worldwide Metis - Saskatchewan	< 1 in 500 1 in 19	83% >95%	1 in 2936 1 in 361	>95% >95%
Hypohidrotic Ectodermal Dysplasia 1 (XL) NM_001399.4	*EDA*	Worldwide	< 1 in 500	73%	1 in 1849	95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Hypophosphatasia (AR) NM_000478.4	*ALPL*	Asian Worldwide Mennonite	1 in 192 1 in 345 1 in 25	72% 64% >95%	1 in 683 1 in 957 1 in 481	>95% >95% >95%
Inclusion Body Myopathy 2 (AR) NM_005476.5	*GNE*	Caucasian Asian Ashkenazi Jewish Worldwide Sephardic Jewish - Iranian and Syrian	< 1 in 500 1 in 58 < 1 in 500 1 in 179 1 in 10	86% 83% >95% 82% >95%	1 in 3565 1 in 336 1 in 9981 1 in 990 1 in 181	>95% >95% >95% >95% >95%
Infantile Cerebral and Cerebellar Atrophy (AR) NM_004268.4	*MED17*	Sephardic Jewish - Bukharian and Kurdish	1 in 20	>95%	1 in 381	>95%
Isovaleric Acidemia (AR) NM_002225.3	*IVD*	Caucasian Asian Worldwide	1 in 144 1 in 75 1 in 158	69% 55% 68%	1 in 462 1 in 165 1 in 492	>95% >95% >95%
Joubert Syndrome 2 (AR) NM_001173990.2	*TMEM216*	Ashkenazi Jewish Worldwide	1 in 110 < 1 in 500	>95% >95%	1 in 2181 1 in 9981	>95% >95%
Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome (AR) NM_015272.2	*RPGRIP1L*	Worldwide	1 in 259	81%	1 in 1359	>95%
Junctional Epidermolysis Bullosa (*LAMA3*-Related) (AR) NM_000227.4	*LAMA3*	Worldwide	< 1 in 500	91%	1 in 5545	>95%
Junctional Epidermolysis Bullosa (*LAMB3*-Related) (AR) NM_000228.2	*LAMB3*	Worldwide	1 in 500	92%	1 in 6239	>95%
Junctional Epidermolysis Bullosa (*LAMC2*-Related) (AR) NM_018891.2	*LAMC2*	Worldwide	< 1 in 500	90%	1 in 4991	>95%
Krabbe Disease (AR) NM_000153.3	*GALC*	Worldwide Druze Northern Israel Muslim Arab (Jerusalem)	1 in 158 1 in 6 1 in 6	80% >95% >95%	1 in 786 1 in 101 1 in 101	>95% >95% >95%
Lamellar Ichthyosis, Type 1 (AR) NM_000359.2	*TGM1*	Caucasian Worldwide Norwegian	1 in 253 1 in 301 1 in 151	83% 81% 80%	1 in 1483 1 in 1580 1 in 751	>95% >95% >95%
Leber Congenital Amaurosis 10 and Other *CEP290*-Related Ciliopathies (AR) NM_025114.3	*CEP290*	Worldwide	1 in 185	93%	1 in 2630	>95%
Leber Congenital Amaurosis 13 (AR) NM_152443.2	*RDH12*	Worldwide	1 in 456	59%	1 in 1111	>95%
Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20 (AR) NM_000329.2	*RPE65*	Worldwide Sephardic Jewish - North African	1 in 228 1 in 90	73% >95%	1 in 842 1 in 1781	>95% >95%
Leber Congenital Amaurosis 5 (AR) NM_181714.3	*LCA5*	Worldwide	< 1 in 500	88%	1 in 4159	>95%
Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 (AR) NM_201253.2	*CRB1*	Worldwide	1 in 112	78%	1 in 506	>95%
Leigh Syndrome, French-Canadian Type (AR) NM_133259.3	*LRPPRC*	Worldwide French Canadian - Saguenay-Lac St. Jean	< 1 in 500 1 in 23	>95% >95%	1 in 9981 1 in 441	>95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Lethal Congenital Contracture Syndrome 1 / Cell Lethal Arthrogryposis with Anterior Horn Disease (AR) NM_001003722.1	*GLE1*	Finnish	1 in 100	>95%	1 in 1981	>95%
Leukoencephalopathy with Vanishing White Matter (AR) NM_003907.2	*EIF2B5*	Worldwide	< 1 in 500	72%	1 in 1783	>95%
Limb-Girdle Muscular Dystrophy, Type 2A (AR) NM_000070.2	*CAPN3*	Caucasian Hispanic Asian Worldwide Amish	1 in 130 1 in 260 1 in 238 1 in 158 N/A	56% 68% 80% 73% >95%	1 in 294 1 in 810 1 in 1186 1 in 582 N/A	90% >95% >95% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2B (AR) NM_003494.3	*DYSF*	Worldwide Sephardic Jewish - Libyan, Kavkazi and Yemenite	1 in 311 1 in 14	>95% >95%	1 in 6201 1 in 261	>95% >95%
Limb-Girdle Muscular Dystrophy, Type 2C (AR) NM_000231.2	*SGCG*	Worldwide Moroccan Roma	1 in 354 1 in 250 1 in 96	80% 77% >95%	1 in 1766 1 in 1084 1 in 1901	87% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2D (AR) NM_000023.2	*SGCA*	Caucasian Worldwide Finnish	1 in 290 < 1 in 500 1 in 150	83% 64% >95%	1 in 1701 1 in 1387 1 in 2981	>95% >95% >95%
Limb-Girdle Muscular Dystrophy, Type 2E (AR) NM_000232.4	*SGCB*	Caucasian Worldwide	1 in 406 1 in 500	59% 77%	1 in 989 1 in 2171	64% 93%
Limb-Girdle Muscular Dystrophy, Type 2I (AR) NM_024301.4	*FKRP*	Worldwide Norwegian	1 in 158 1 in 116	87% >95%	1 in 1209 1 in 2301	>95% >95%
Lipoamide Dehydrogenase Deficiency (AR) NM_000108.4	*DLD*	Ashkenazi Jewish Worldwide	1 in 107 < 1 in 500	>95% 74%	1 in 2121 1 in 1920	>95% >95%
Lipoid Adrenal Hyperplasia (AR) NM_000349.2	*STAR*	East Asian Worldwide	1 in 177 < 1 in 500	95% 82%	1 in 3521 1 in 2773	>95% >95%
Lipoprotein Lipase Deficiency (AR) NM_000237.2	*LPL*	Caucasian Asian Worldwide French Canadian - Saguenay - Lac St. Jean French Canadian - Other	< 1 in 500 1 in 189 1 in 500 1 in 46 1 in 139	55% 40% 86% >95% >95%	1 in 1110 1 in 314 1 in 3565 1 in 901 1 in 2761	68% 67% >95% >95% >95%
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (AR) NM_000182.4	*HADHA*	Caucasian Worldwide	1 in 254 1 in 351	93% 93%	1 in 3615 1 in 5001	>95% >95%
Lysinuric Protein Intolerance (AR) NM_001126106.2	*SLC7A7*	Worldwide Japanese Finnish	< 1 in 500 1 in 119 1 in 122	88% 88% >95%	1 in 4159 1 in 984 1 in 2421	>95% >95% >95%
Maple Syrup Urine Disease, Type 1a (AR) NM_000709.3	*BCKDHA*	Caucasian Worldwide Mennonite Portuguese Roma	1 in 320 1 in 289 1 in 10 1 in 71	75% 63% >95% >95%	1 in 1277 1 in 779 1 in 181 1 in 1401	94% >95% >95% >95%
Maple Syrup Urine Disease, Type 1b (AR) NM_000056.3	*BCKDHB*	Caucasian Asian Ashkenazi Jewish Worldwide	1 in 433 1 in 163 1 in 97 1 in 327	56% 57% >95% 72%	1 in 983 1 in 378 1 in 1921 1 in 1165	>95% >95% >95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Meckel Syndrome 1 / Bardet-Biedl Syndrome 13 (AR) NM_017777.3	*MKS1*	Caucasian Worldwide Finnish	1 in 260 1 in 260 1 in 47	83% 91% >95%	1 in 1525 1 in 2879 1 in 921	>95% >95% >95%
Medium Chain Acyl-CoA Dehydrogenase Deficiency (AR) NM_000016.5	*ACADM*	Caucasian Asian Worldwide	1 in 55 1 in 178 1 in 69	88% 53% 81%	1 in 451 1 in 378 1 in 359	93% 83% >95%
Megalencephalic Leukoencephalopathy With Subcortical Cysts (AR) NM_015166.3	*MLC1*	Worldwide Sephardic Jewish - Libyan	< 1 in 500 1 in 40	93% >95%	1 in 7130 1 in 781	>95% >95%
Menkes Disease (XL) NM_000052.6	*ATP7A*	Worldwide	< 1 in 500	71%	1 in 1722	87%
Metachromatic Leukodystrophy (AR) NM_000487.5	*ARSA*	Ashkenazi Jewish Worldwide Sephardic Jewish - Yemenite Navajo	< 1 in 500 1 in 100 1 in 46 1 in 25	>95% 67% >95% >95%	1 in 9981 1 in 301 1 in 901 1 in 481	>95% >95% >95% >95%
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC1*-Related) (AR) NM_020166.4	*MCCC1*	Caucasian Worldwide	1 in 137 1 in 147	77% 75%	1 in 592 1 in 585	88% >95%
3-Methylcrotonyl-CoA Carboxylase Deficiency (*MCCC2*-Related) (AR) NM_022132.4	*MCCC2*	Caucasian Worldwide	1 in 112 1 in 120	59% 69%	1 in 272 1 in 385	91% >95%
3-Methylglutaconic Aciduria, Type III (AR) NM_025136.3	*OPA3*	Worldwide Sephardic Jewish - Iraqi	< 1 in 500 1 in 13	95% >95%	1 in 9981 1 in 241	>95% >95%
Methylmalonic Acidemia (*MMAA*-Related) (AR) NM_172250.2	*MMAA*	Caucasian Worldwide	1 in 316 1 in 316	92% 88%	1 in 3939 1 in 2626	>95% >95%
Methylmalonic Acidemia (*MMAB*-Related) (AR) NM_052845.3	*MMAB*	Caucasian Worldwide	1 in 456 1 in 456	91% 91%	1 in 5057 1 in 5057	>95% >95%
Methylmalonic Acidemia (*MUT*-Related) (AR) NM_000255.3	*MUT*	Caucasian African Hispanic Asian Worldwide	1 in 224 1 in 177 1 in 383 1 in 53 1 in 383	22% 52% 65% 50% 70%	1 in 287 1 in 368 1 in 1092 1 in 105 1 in 1274	>95% >95% >95% >95% >95%
Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type (AR) NM_015506.2	*MMACHC*	Caucasian Asian Worldwide	1 in 138 1 in 113 1 in 138	95% 90% 91%	1 in 2741 1 in 1121 1 in 1523	>95% >95% >95%
Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type (AR) NM_015702.2	*MMADHC*	Caucasian	< 1 in 500	>95%	1 in 9981	>95%
Microphthalmia / Anophthalmia (AR) NM_182894.2	*VSX2*	Worldwide Sephardic Jewish - Iranian and Syrian	< 1 in 500 1 in 145	>95% >95%	1 in 9981 1 in 2881	>95% >95%
Mitochondrial Complex I Deficiency (*ACAD9*-Related) (AR) NM_014049.4	*ACAD9*	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Mitochondrial Complex I Deficiency (*NDUFAF5*-Related) (AR) NM_024120.4	*NDUFAF5*	Ashkenazi Jewish Worldwide	1 in 290 < 1 in 500	>95% >95%	1 in 5781 1 in 9981	>95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Mitochondrial Complex I Deficiency (*NDUFS6*-Related) (AR) NM_004553.4	*NDUFS6*	Worldwide Sephardic Jewish - Caucasus	< 1 in 500 1 in 24	93% >95%	1 in 7130 1 in 461	93% >95%
Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy (AR) NM_002437.4	*MPV17*	Worldwide Navajo	< 1 in 500 1 in 20	88% >95%	1 in 4159 1 in 381	>95% >95%
Mitochondrial Myopathy and Sideroblastic Anemia 1 (AR) NM_025215.5	*PUS1*	Worldwide Sephardic Jewish - Iranian	< 1 in 500 N/A	>95% >95%	1 in 9981 N/A	>95% >95%
Mucolipidosis II / IIIA (AR) NM_024312.4	*GNPTAB*	Caucasian Asian Worldwide	1 in 225 1 in 389 1 in 408	90% 79% 84%	1 in 2241 1 in 1849 1 in 2545	95% 84% >95%
Mucolipidosis III Gamma (AR) NM_032520.4	*GNPTG*	Caucasian Worldwide	1 in 273 < 1 in 500	94% 81%	1 in 4534 1 in 2627	94% >95%
Mucolipidosis IV (AR) NM_020533.2	*MCOLN1*	Ashkenazi Jewish Worldwide	1 in 89 < 1 in 500	>95% 92%	1 in 1761 1 in 6239	>95% >95%
Mucopolysaccharidosis, Type I (AR) NM_000203.4	*IDUA*	Worldwide	1 in 144	72%	1 in 512	>95%
Mucopolysaccharidosis, Type II (XL) NM_000202.6 *Exception:* Exon 1	*IDS*	Worldwide	< 1 in 500	67%	1 in 1513	86%
Mucopolysaccharidosis, Type IIIA (AR) NM_000199.3	*SGSH*	Caucasian Worldwide	1 in 253 1 in 415	68% 56%	1 in 789 1 in 942	95% >95%
Mucopolysaccharidosis, Type IIIB (AR) NM_000263.3	*NAGLU*	Caucasian Asian Worldwide	1 in 346 1 in 298 < 1 in 500	59% 70% 64%	1 in 842 1 in 991 1 in 1387	80% >95% >95%
Mucopolysaccharidosis, Type IIIC (AR) NM_152419.2	*HGSNAT*	Caucasian Asian Worldwide	1 in 259 < 1 in 500 1 in 482	81% >95% 77%	1 in 1359 1 in 9981 1 in 2092	93% >95% >95%
Mucopolysaccharidosis, Type IIID (AR) NM_002076.3	*GNS*	Worldwide	< 1 in 500	90%	1 in 4991	90%
Mucopolysaccharidosis, Type IVb / GM1 Gangliosidosis (AR) NM_000404.2	*GLB1*	Caucasian Worldwide Roma South Brazilian	1 in 278 1 in 158 1 in 50 1 in 58	57% 69% >95% >95%	1 in 645 1 in 507 1 in 981 1 in 1141	>95% >95% >95% >95%
Mucopolysaccharidosis, Type VI (AR) NM_000046.3	*ARSB*	Caucasian Asian Worldwide	1 in 273 1 in 423 1 in 291	67% 53% 54%	1 in 825 1 in 899 1 in 631	>95% >95% >95%
Mucopolysaccharidosis, Type IX (AR) NM_153281.1	*HYAL1*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Multiple Sulfatase Deficiency (AR) NM_182760.3	*SUMF1*	Ashkenazi Jewish Worldwide	1 in 279 < 1 in 500	>95% 65%	1 in 5561 1 in 1427	>95% >95%
Muscle-Eye-Brain Disease and Other *POMGNT1*-Related Congenital Muscular Dystrophy- Dystroglycanopathies (AR) NM_017739.3	*POMGNT1*	Worldwide Finnish	1 in 462 1 in 111	74% >95%	1 in 1774 1 in 2201	95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Myoneurogastrointestinal Encephalopathy (AR) NM_001113755.2	*TYMP*	Caucasian Worldwide Sephardic Jewish - Iranian	1 in 500 < 1 in 500 1 in 158	69% 62% >95%	1 in 1611 1 in 1314 1 in 3141	>95% >95% >95%
Myotubular Myopathy 1 (XL) NM_000252.2	*MTM1*	Worldwide	< 1 in 500	86%	1 in 3565	>95%
N-Acetylglutamate Synthase Deficiency (AR) NM_153006.2	*NAGS*	Worldwide	< 1 in 500	59%	1 in 1218	>95%
Nemaline Myopathy 2 (AR) NM_001271208.1 *Exception:* Exons 82 - 105	*NEB*	Ashkenazi Jewish Worldwide Finnish	1 in 168 1 in 224 1 in 112	>95% 81% 75%	1 in 3341 1 in 1175 1 in 445	>95% 94% 75%
Nephrogenic Diabetes Insipidus, Type II (AR) NM_000486.5	*AQP2*	Worldwide	< 1 in 500	44%	1 in 892	>95%
Nephrotic Syndrome (*NPHS1*-Related) / Congenital Finnish Nephrosis (AR) NM_004646.3	*NPHS1*	Worldwide Finnish Groffdale Conference Mennonites	1 in 325 1 in 45 1 in 12	80% >95% >95%	1 in 1621 1 in 881 1 in 221	>95% >95% >95%
Nephrotic Syndrome (*NPHS2*-Related) / Steroid-Resistant Nephrotic Syndrome (AR) NM_014625.3	*NPHS2*	Worldwide	1 in 377	80%	1 in 1881	>95%
Neuronal Ceroid-Lipofuscinosis (*CLN3*-Related) (AR) NM_000086.2	*CLN3*	Caucasian Worldwide	1 in 188 1 in 233	94% 94%	1 in 3118 1 in 3868	>95% >95%
Neuronal Ceroid-Lipofuscinosis (*CLN5*-Related) (AR) NM_006493.2	*CLN5*	Worldwide Finnish	< 1 in 500 1 in 100	91% >95%	1 in 5545 1 in 1981	>95% >95%
Neuronal Ceroid-Lipofuscinosis (*CLN6*-Related) (AR) NM_017882.2	*CLN6*	Worldwide	< 1 in 500	86%	1 in 3565	>95%
Neuronal Ceroid-Lipofuscinosis (*CLN8*-Related) (AR) NM_018941.3	*CLN8*	Worldwide Finnish	< 1 in 500 1 in 135	84% >95%	1 in 3120 1 in 2681	>95% >95%
Neuronal Ceroid-Lipofuscinosis (*MFSD8*-Related) (AR) NM_152778.2	*MFSD8*	Worldwide	< 1 in 500	82%	1 in 2773	>95%
Neuronal Ceroid-Lipofuscinosis (*PPT1*-Related) (AR) NM_000310.3	*PPT1*	Worldwide Finnish	1 in 368 1 in 70	77% 94%	1 in 1597 1 in 1151	>95% >95%
Neuronal Ceroid-Lipofuscinosis (*TPP1*-Related) (AR) NM_000391.3	*TPP1*	Worldwide Newfoundland	1 in 314 1 in 59	87% >95%	1 in 2409 1 in 1161	>95% >95%
Niemann-Pick Disease, Type A/B (AR) NM_000543.4	*SMPD1*	Caucasian Ashkenazi Jewish Worldwide	1 in 244 1 in 115 1 in 196	37% >95% 85%	1 in 387 1 in 2281 1 in 1301	>95% >95% >95%
Niemann-Pick Disease, Type C (*NPC1*-Related) (AR) NM_000271.4	*NPC1*	Caucasian Asian Worldwide	1 in 185 1 in 404 1 in 282	73% 45% 62%	1 in 682 1 in 734 1 in 740	>95% 85% >95%
Niemann-Pick Disease, Type C (*NPC2*-Related) (AR) NM_006432.3	*NPC2*	Worldwide	< 1 in 500	83%	1 in 2936	>95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Nijmegen Breakage Syndrome (AR)	*NBN*	Caucasian	1 in 155	90%	1 in 1541	>95%
NM_002485.4		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Non-Syndromic Hearing Loss (*GJB2*-Related) (AR)	*GJB2*	Caucasian	1 in 42	88%	1 in 343	>95%
		Asian	1 in 50	83%	1 in 289	>95%
NM_004004.5		Ashkenazi Jewish	1 in 21	>95%	1 in 401	>95%
		Worldwide	1 in 43	82%	1 in 234	>95%
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome (AR)	*WNT10A*	Worldwide	1 in 305	67%	1 in 922	>95%
NM_025216.2						
Omenn Syndrome (*RAG2*-Related) (AR)	*RAG2*	Worldwide	< 1 in 500	51%	1 in 1019	>95%
NM_000536.2		Sephardic Jewish - Iraqi	N/A	88%	N/A	>95%
Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type (AR)	*DCLRE1C*	Worldwide	< 1 in 500	54%	1 in 1086	81%
		Navajo and Apache	1 in 48	>95%	1 in 941	>95%
NM_001033855.1		Native American				
Ornithine Aminotransferase Deficiency (AR)	*OAT*	Worldwide	< 1 in 500	89%	1 in 4537	>95%
NM_000274.3		Finnish	1 in 147	>95%	1 in 2921	>95%
		Sephardic Jewish - Iraqi and Syrian	1 in 177	>95%	1 in 3521	>95%
Ornithine Transcarbamylase Deficiency (XL)	*OTC*	Worldwide	< 1 in 500	70%	1 in 1664	95%
NM_000531.5						
Osteopetrosis 1 (AR)	*TCIRG1*	Ashkenazi Jewish	1 in 350	>95%	1 in 6981	>95%
NM_006019.2		Worldwide	1 in 316	67%	1 in 956	95%
		Costa Rican	1 in 86	>95%	1 in 1701	>95%
		Chuvashiyan	1 in 60	>95%	1 in 1181	>95%
Pendred Syndrome (AR)	*SLC26A4*	Caucasian	1 in 88	86%	1 in 622	93%
NM_000441.1		African	1 in 76	>95%	1 in 1501	>95%
		Asian	1 in 74	83%	1 in 430	>95%
		Worldwide	1 in 80	81%	1 in 417	>95%
Phenylalanine Hydroxylase Deficiency (AR)	*PAH*	Caucasian	1 in 50	94%	1 in 818	>95%
NM_000277.1		African	1 in 143	87%	1 in 1093	>95%
		Asian	1 in 78	79%	1 in 368	>95%
		Ashkenazi Jewish	1 in 225	75%	1 in 897	93%
		Worldwide	1 in 65	77%	1 in 279	>95%
		Turkish	1 in 32	63%	1 in 85	>95%
		Irish	1 in 34	91%	1 in 368	92%
		Sicilian	1 in 26	48%	1 in 49	>95%
		Sephardic Jewish - Iranian, Bukharian, Kavkazi, Tunisian and Moroccan	1 in 18	88%	1 in 143	91%
3-Phosphoglycerate Dehydrogenase Deficiency (AR)	*PHGDH*	Ashkenazi Jewish	1 in 453	>95%	1 in 9041	>95%
		Worldwide	< 1 in 500	>95%	1 in 9981	>95%
NM_006623.3						
Polycystic Kidney Disease, Autosomal Recessive (AR)	*PKHD1*	Caucasian	1 in 100	70%	1 in 331	>95%
		Ashkenazi Jewish	1 in 106	>95%	1 in 2101	>95%
NM_138694.3		Worldwide	1 in 144	69%	1 in 462	>95%
		South African Afrikaner	1 in 52	>95%	1 in 1021	>95%
Polyglandular Autoimmune Syndrome, Type 1 (AR)	*AIRE*	Worldwide	1 in 354	93%	1 in 5044	>95%
		Finnish	1 in 79	>95%	1 in 1561	>95%
NM_000383.2		Sardinian	1 in 60	95%	1 in 1181	95%
		Sephardic Jewish - Iranian	1 in 27	>95%	1 in 521	>95%
Pontocerebellar Hypoplasia, Type 1A (AR)	*VRK1*	Ashkenazi Jewish	1 in 225	>95%	1 in 4481	>95%
NM_003384.2		Worldwide	< 1 in 500	>95%	1 in 9981	>95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Pontocerebellar Hypoplasia, Type 6 (AR) NM_020320.3	*RARS2*	Worldwide Sephardic Jewish - Iraqi, Syrian and Tunisian	< 1 in 500 N/A	>95% >95%	1 in 9981 N/A	>95% >95%
Primary Carnitine Deficiency (AR) NM_003060.2	*SLC22A5*	Caucasian Asian Worldwide Faroese	1 in 110 1 in 100 1 in 200 1 in 20	94% 70% 68% >95%	1 in 1818 1 in 331 1 in 623 1 in 381	94% 81% >95% >95%
Primary Ciliary Dyskinesia (*DNAH5*-Related) (AR) NM_001369.2	*DNAH5*	Ashkenazi Jewish Worldwide	1 in 174 1 in 120	>95% 78%	1 in 3461 1 in 542	>95% >95%
Primary Ciliary Dyskinesia (*DNAI1*-Related) (AR) NM_012144.3	*DNAI1*	Ashkenazi Jewish Worldwide	1 in 352 1 in 182	>95% 92%	1 in 7021 1 in 2264	>95% >95%
Primary Ciliary Dyskinesia (*DNAI2*-Related) (AR) NM_023036.4	*DNAI2*	Ashkenazi Jewish Worldwide	1 in 200 1 in 500	>95% >95%	1 in 3981 1 in 9981	>95% >95%
Primary Hyperoxaluria, Type 1 (AR) NM_000030.2	*AGXT*	Worldwide	1 in 158	77%	1 in 684	>95%
Primary Hyperoxaluria, Type 2 (AR) NM_012203.1	*GRHPR*	Worldwide	< 1 in 500	92%	1 in 6239	>95%
Primary Hyperoxaluria, Type 3 (AR) NM_138413.3	*HOGA1*	Ashkenazi Jewish Worldwide	N/A 1 in 309	>95% 88%	N/A 1 in 2568	>95% >95%
Progressive Cerebello-Cerebral Atrophy (AR) NM_016955.3	*SEPSECS*	Worldwide Sephardic Jewish - Moroccan and Iraqi	< 1 in 500 1 in 41	>95% >95%	1 in 9981 1 in 801	>95% >95%
Progressive Familial Intrahepatic Cholestasis, Type 2 (AR) NM_003742.2	*ABCB11*	Worldwide	1 in 158	61%	1 in 404	>95%
Propionic Acidemia (*PCCA*-Related) (AR) NM_000282.3	*PCCA*	Caucasian Asian Worldwide	1 in 380 1 in 162 1 in 224	54% 72% 52%	1 in 825 1 in 576 1 in 466	73% 90% 85%
Propionic Acidemia (*PCCB*-Related) (AR) NM_000532.4	*PCCB*	Caucasian Asian Worldwide	1 in 202 1 in 145 1 in 224	60% 83% 70%	1 in 504 1 in 848 1 in 744	88% >95% >95%
Pycnodysostosis (AR) NM_000396.3	*CTSK*	Worldwide	1 in 438	60%	1 in 1094	>95%
Pyruvate Dehydrogenase E1-Alpha Deficiency (XL) NM_000284.3	*PDHA1*	Worldwide	< 1 in 500	64%	1 in 1387	94%
Pyruvate Dehydrogenase E1-Beta Deficiency (AR) NM_000925.3	*PDHB*	Worldwide	< 1 in 500	74%	1 in 1920	>95%
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (AR) NM_000317.2	*PTS*	Asian Worldwide	1 in 122 < 1 in 500	87% 71%	1 in 932 1 in 1722	>95% >95%
Renal Tubular Acidosis and Deafness (AR) NM_001692.3	*ATP6V1B1*	Worldwide Sephardic Jewish - Syrian	< 1 in 500 1 in 140	86% >95%	1 in 3565 1 in 2781	>95% >95%
Retinitis Pigmentosa 25 (AR) NM_001142800.1	*EYS*	Caucasian Ashkenazi Jewish Worldwide Sephardic Jewish - Moroccan	1 in 53 < 1 in 500 1 in 129 1 in 42	60% >95% 63% 22%	1 in 131 1 in 9981 1 in 347 1 in 54	72% >95% 92% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Retinitis Pigmentosa 26 (AR) NM_001030311.2	*CERKL*	Worldwide Sephardic Jewish - Yemenite	1 in 137 1 in 24	>95% >95%	1 in 2721 1 in 461	>95% >95%
Retinitis Pigmentosa 28 (AR) NM_032180.2	*FAM161A*	Ashkenazi Jewish Worldwide Sephardic Jewish - Libyan, Moroccan, Tunisian and Bulgarian	1 in 214 1 in 289 1 in 41	>95% >95% >95%	1 in 4261 1 in 5761 1 in 801	>95% >95% >95%
Retinitis Pigmentosa 59 (AR) NM_001243564.1	*DHDDS*	Ashkenazi Jewish	1 in 117	>95%	1 in 2321	>95%
Rhizomelic Chondrodysplasia Punctata, Type 1 (AR) NM_000288.3	*PEX7*	Caucasian Worldwide	1 in 158 < 1 in 500	87% 87%	1 in 1209 1 in 3839	>95% >95%
Rhizomelic Chondrodysplasia Punctata, Type 3 (AR) NM_003659.3	*AGPS*	Worldwide	< 1 in 500	87%	1 in 3839	93%
Roberts Syndrome (AR) NM_001017420.2	*ESCO2*	Worldwide	< 1 in 500	>95%	1 in 9981	>95%
Salla Disease (AR) NM_012434.4	*SLC17A5*	Worldwide Finnish Swedish Canadian Inuit	< 1 in 500 1 in 100 1 in 125 1 in 129	94% >95% >95% >95%	1 in 8318 1 in 1981 1 in 2481 1 in 2561	>95% >95% >95% >95%
Sandhoff Disease (AR) NM_000521.3	*HEXB*	Caucasian Worldwide Northern Saskachetwan Metis Argentinian Creole	1 in 235 1 in 180 1 in 15 1 in 26	26% 82% 75% >95%	1 in 317 1 in 995 1 in 57 1 in 501	>95% >95% >95% >95%
Schimke Immunoosseous Dysplasia (AR) NM_014140.3	*SMARCAL1*	Worldwide	< 1 in 500	81%	1 in 2627	>95%
Segawa Syndrome (AR) NM_000360.3	*TH*	Caucasian Asian Worldwide	1 in 224 1 in 416 < 1 in 500	74% 78% 78%	1 in 859 1 in 1887 1 in 2269	>95% >95% >95%
Sjogren-Larsson Syndrome (AR) NM_000382.2	*ALDH3A2*	Worldwide Swedish	< 1 in 500 1 in 205	87% >95%	1 in 3839 1 in 4081	>95% >95%
Smith-Lemli-Opitz Syndrome (AR) NM_001360.2	*DHCR7*	Caucasian African Asian Ashkenazi Jewish Worldwide	1 in 48 1 in 93 < 1 in 500 1 in 41 1 in 68	81% 67% 84% >95% 80%	1 in 248 1 in 280 1 in 3120 1 in 801 1 in 336	>95% >95% >95% >95% >95%
Spondylothoracic Dysostosis (AR) NM_001039958.1	*MESP2*	Worldwide Puerto Rican	1 in 224 1 in 55	>95% >95%	1 in 4461 1 in 1081	>95% >95%
Steel Syndrome (AR) NM_032888.2 *Variant tested:* p.G697R (Genotyping only)	*COL27A1*	Puerto Rican	< 1 in 500	>95%	1 in 9981	>95%
Stuve-Wiedemann Syndrome (AR) NM_002310.5	*LIFR*	Worldwide	< 1 in 500	94%	1 in 8318	>95%
Sulfate Transporter-Related Osteochondrodysplasia (AR) NM_000112.3	*SLC26A2*	Worldwide Finnish	1 in 158 1 in 50	>95% >95%	1 in 3141 1 in 981	>95% >95%

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

Tay-Sachs Disease (AR)	*HEXA*	Caucasian	1 in 182	91%*	1 in 2012	>95%
NM_000520.4		African American	1 in 271	90%*	1 in 2701	>95%
		Asian	1 in 126	70%*	1 in 418	91%
		Ashkenazi Jewish	1 in 27	>95%*	1 in 521	>95%
		Worldwide	1 in 288	94%*	1 in 4784	>95%
		French Canadian - Gaspesie	1 in 13	>95%*	1 in 241	>95%
		French Canadian - Other	1 in 73	>95%*	1 in 1441	>95%
		Irish	1 in 41	90%*	1 in 401	>95%
		Sephardic Jewish – Moroccan and Iraqi	1 in 125	>95%*	1 in 2481	>95%
Tyrosinemia, Type I (AR)	*FAH*	Caucasian	1 in 333	89%	1 in 3019	>95%
NM_000137.2		African	1 in 478	92%	1 in 5964	>95%
		Asian	< 1 in 500	70%	1 in 1664	>95%
		Ashkenazi Jewish	1 in 143	>95%	1 in 2841	>95%
		Worldwide	< 1 in 500	82%	1 in 2773	>95%
		French Canadian - Saguenay Lac-St. Jean	1 in 25	>95%	1 in 481	>95%
		French Canadian - Other	1 in 66	>95%	1 in 1301	>95%
Usher Syndrome, Type IB (AR)	*MYO7A*	Caucasian	1 in 145	75%	1 in 577	93%
NM_000260.3		African	< 1 in 500	13%	1 in 575	>95%
		Asian	1 in 62	85%	1 in 408	>95%
		Worldwide	1 in 206	73%	1 in 760	>95%
Usher Syndrome, Type IC (AR)	*USH1C*	Worldwide	1 in 353	77%	1 in 1531	>95%
NM_005709.3		French Canadian/Acadian	1 in 227	>95%	1 in 4521	>95%
Usher Syndrome, Type ID (AR)	*CDH23*	Worldwide	1 in 306	66%	1 in 898	>95%
NM_022124.5						
Usher Syndrome, Type IF (AR)	*PCDH15*	Ashkenazi Jewish	1 in 78	>95%	1 in 1541	>95%
NM_001142764.1		Worldwide	1 in 395	76%	1 in 1643	92%
Usher Syndrome, Type IIA (AR)	*USH2A*	Caucasian	1 in 73	77%	1 in 314	88%
NM_206933.2		Worldwide	1 in 126	69%	1 in 404	>95%
		Sephardic Jewish – Iraqi and Iranian	1 in 36	71%	1 in 122	75%
Usher Syndrome, Type III (AR)	*CLRN1*	Ashkenazi Jewish	1 in 120	>95%	1 in 2381	>95%
NM_174878.2		Worldwide	1 in 500	88%	1 in 4159	>95%
		Finnish	1 in 70	>95%	1 in 1381	>95%
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (AR)	*ACADVL*	Caucasian	1 in 88	62%	1 in 230	>95%
		Asian	1 in 194	53%	1 in 412	93%
NM_000018.3		Worldwide	1 in 146	59%	1 in 355	>95%
Walker-Warburg Syndrome and Other *FKTN*-Related Dystrophies (AR)	*FKTN*	Ashkenazi Jewish	1 in 80	>95%	1 in 1581	>95%
		Worldwide	< 1 in 500	14%	1 in 581	20%
NM_001079802.1		Japanese	1 in 188	4%	1 in 196	5%
Wilson Disease (AR)	*ATP7B*	Caucasian	1 in 90	64%	1 in 248	>95%
NM_000053.3		Asian	1 in 50	63%	1 in 133	>95%
		Ashkenazi Jewish	1 in 67	>95%	1 in 1321	>95%
		Worldwide	1 in 90	79%	1 in 425	>95%
		Canary Islands	1 in 25	88%	1 in 201	>95%
		Sardinian	1 in 42	>95%	1 in 821	>95%
		Sephardic Jewish - North African, Iraqi, Yemenite, Iranian and Bukharian	1 in 65	>95%	1 in 1281	>95%
Wolman Disease / Cholesteryl Ester Storage Disease (AR)	*LIPA*	Caucasian	1 in 145	>95%	1 in 2881	>95%
		Ashkenazi Jewish	< 1 in 500	>95%	1 in 9981	>95%
NM_000235.3		Worldwide	< 1 in 500	86%	1 in 3565	>95%
		Sephardic Jewish - Iranian	1 in 26	>95%	1 in 501	>95%
X-Linked Juvenile Retinoschisis (XL)	*RS1*	Worldwide	< 1 in 500	74%	1 in 1920	>95%
NM_000330.3						

Patient:	Donor 12345	DOB:	[redacted]1994	Lab #:	[redacted]

X-Linked Severe Combined Immunodeficiency (XL) NM_000206.2	*IL2RG*	Worldwide	< 1 in 500	89%	1 in 4537	>95%
Zellweger Syndrome Spectrum (*PEX1*-Related) (AR) NM_000466.2	*PEX1*	Caucasian Worldwide	1 in 147 < 1 in 500	89% 80%	1 in 1328 1 in 2496	>95% >95%
Zellweger Syndrome Spectrum (*PEX2*-Related) (AR) NM_000318.2	*PEX2*	Caucasian Ashkenazi Jewish Worldwide	< 1 in 500 1 in 227 < 1 in 500	80% >95% 84%	1 in 2496 1 in 4521 1 in 3120	>95% >95% >95%
Zellweger Syndrome Spectrum (*PEX6*-Related) (AR) NM_000287.3	*PEX6*	Worldwide French Canadian Sephardic Jewish - Yemenite	1 in 280 1 in 55 1 in 18	69% >95% >95%	1 in 901 1 in 1081 1 in 341	>95% >95% >95%
Zellweger Syndrome Spectrum (*PEX10*-Related) (AR) NM_153818.1	*PEX10*	Asian Worldwide	< 1 in 500 < 1 in 500	>95% 89%	1 in 9981 1 in 4537	>95% >95%

*Carrier detection by HEXA enzyme analysis has a detection rate of approximately 98%.
AR: Autosomal Recessive
XL: X-Linked
N/A: Not Available