Donor 2925

Genetic Testing Summary

Fairfax Cryobank recommends reviewing this genetic testing summary with your healthcare provider to determine suitability.

Last Updated: 04/27/22

Donor Reported Ancestry: German, Irish, French, Native American Jewish Ancestry: No

Genetic Test*	Result	Comments/Donor's Residual Risk**
Chromosome analysis (karyotype)	Normal male karyotype	No evidence of clinically significant chromosome abnormalities
Hemoglobin evaluation	Normal hemoglobin fractionation and MCV/MCH results	Reduced risk to be a carrier for sickle cell anemia, beta thalassemia, alpha thalassemia trait (aa/-- and a-/a-) and other hemoglobinopathies
Cystic Fibrosis (CF) carrier screening	Negative by genotyping of 98 mutations in the CFTR gene	1/343
Spinal Muscular Atrophy	Negative for deletions in the SMN1 gene	1/648
Tay Sachs Enzyme Analysis	Non-carrier by Hexosaminidase A analysis	
Carrier testing for 21 genes	Negative by genotyping- see attached	

*No single test can screen for all genetic disorders. A negative screening result significantly reduces, but cannot eliminate, the risk for these conditions in a pregnancy.

**Donor residual risk is the chance the donor is still a carrier after testing negative.

genzyme

Chromosome Analysis

Patient Name: Donor, 2925
Referring Physician: [redacted]
Specimen #: 80207482 **Client #:** 606452
Patient ID: 80148488-6-B1

Fairfax Cryobank
[redacted]

DOB: Not Given
SSN:

Date Collected: 07/02/2008
Date Received: 07/03/2008
Lab ID: 2925-080702
Hospital ID:
Specimen Type: **Peripheral Blood**

Indication: Gamete donor

Metaphases Counted:	20			**Banding Technique:**	GTW
Metaphases Analyzed:	5	**Number of Cultures:**	2	**Banding Resolution:**	550
Metaphases Karyotyped:	2			**Dept. Section:**	B1

RESULTS: 46,XY
Male karyotype

INTERPRETATION:

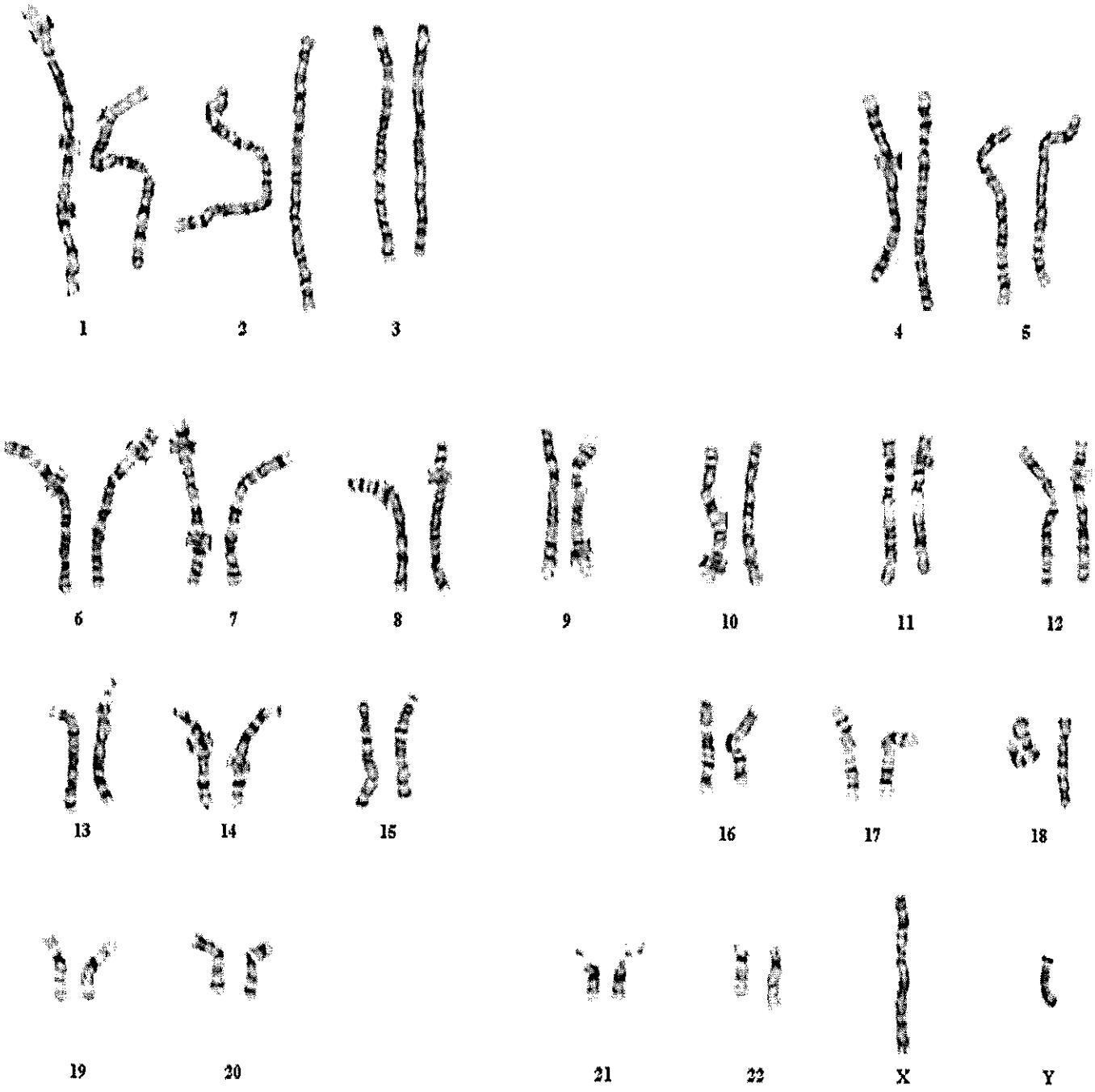
This analysis shows no evidence of clinically significant numerical or structural chromosome abnormalities. The standard cytogenetic methodology utilized in this analysis does not routinely detect small rearrangements and low level mosaicism, and cannot detect microdeletions.

Signed: Jay W. Moore

Jay W. Moore, Ph.D. FFACMG

Date: 07/14/2008

Testing Performed At Genzyme Genetics 10421 University Center Drive Suite 100 Tampa, FL 33612 1-800-966-4440



Specimen #: **80207482 6**
Specimen Type: BLDPER
Patient Name: Donor, 2925
Image ID: DKE1
Karyotype: 46,XY

Dept ID: B1
Date Received: 07/03/2008
Date Reviewed: 07/14/2008
Reviewed By: JWM

genzyme
GENERAL
genetics

genzyme

SMN1 Copy Number Analysis

Patient Name: . Donor 2925
DOB: Age:
SSN #: Gender: M

606452 / 310544
Fairfax Cryobank
[redacted]

Genzyme Specimen #:61060506-06
Case #: 60943119 Patient ID #: 60900233
Date Collected: 06/16/2008 Date Received: 06/17/2008

Referring Physician: Steve Pool
Genetic Counselor:

Client Lab ID #:
Hospital ID #:
Specimen ID #:

Specimen Type: Peripheral Blood

Specimen(s) Received: 2 - Yellow (ACD) 10 ml round bottom tube(s)

Clinical Data: Gamete donor

Ethnicity: Caucasian

RESULTS: SMN1 copy number: 2

INTERPRETATION:

This individual's risk to be a carrier of SMA is reduced from approximately 1/41 to 1/648, based on an SMN1 copy number of two and a negative family history.

COMMENT:

Spinal muscular atrophy (SMA) is an autosomal recessive disease of variable age of onset and severity caused by mutations, most often deletions or gene conversions, resulting in zero copies of the survival motor neuron (SMN1) gene. Approximately 1/41 individuals without a family history of SMA is a carrier. This analysis identifies approximately 94% of carriers. Individuals with one copy of the SMN1 gene are predicted to be carriers of SMA. Individuals with two or more copies of the SMN1 gene have a reduced risk to be carriers of SMA.

This copy number analysis cannot detect the ~6% of individuals who are carriers of SMA as a result of: 1) 2 copies of the SMN1 gene on one chromosome and a deletion or gene conversion of SMN1 gene on the other chromosome or 2) small intragenic mutations within the SMN1 gene. This analysis also will not detect germline mosaicism or mutations in genes other than SMN1. SMA carriers falling into any of these categories have an SMN1 copy number result of 2 by dosage analysis. Additionally, de novo mutations have been reported in approximately 2% of SMA patients. Other false negative or false positive results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation, or erroneous representation of family relationships.

METHOD:
Specimen DNA is isolated and amplified by real-time polymerase chain reaction (PCR) for exon 7 of the SMN1 gene and two reference genes. A mathematical algorithm is used to calculate the number of copies of SMN1. Sequencing of the primer and probe binding sites for the SMN1 real-time PCR reaction is performed on all fetal samples, and on samples from individuals with 1 copy of SMN1 on carrier testing, to rule out the presence of sequence variants which could interfere with analysis and interpretation.

REFERENCES:
Smith M, Calabro V, Chong B, et al. 2007. Eur J Hum Genet 15:759-766.
Online review of SMA: http://www.genereviews.org/profiles/sma

The test was developed and its performance characteristics have been determined by Genzyme. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing. This test must be used in conjunction with clinical assessment when available.

RELEASED 7/15/08

Electronically Signed by: Narasimhan Nagan, Ph.D., FACMG on 06/27/2008

Reported by: MS/aw

genzyme

Carrier Testing

Patient Name: Donor 2925, .
Referring Physician: [redacted]
Specimen #: 61060506
Patient ID: 60900233-6

Client #: 606452
Case #: 60943119

Fairfax Cryobank
Genetics and IVF Institute
[redacted]

DOB: Not Given
Sex: M
SSN:

Date Collected: 06/16/2008
Date Received: 06/17/2008
Lab ID:
Hospital ID:
Specimen Type: **BLDPER**

Ethnicity: Caucasian

Indication: Gamete donor

Disease	Result	Interpretation
Cystic Fibrosis	Negative	Carrier risk reduced from 1/25 (4%) to 1/343 (0.3%).
Tay-Sachs - Enzyme	Hex. Activity: 1470 nmol/mg protein Hex. Percent A: 73.2	Non carrier Plasma/Serum WBC Non carrier range: Hex A >= 55% >= 55% Carrier range : Hex A 20 - 48% 20 - 49%

COMMENTS:

DNA:
The negative results from this analysis cannot eliminate the possibility that this individual carries a mutation not detected by this test. Unless otherwise noted, interpretations are based on a negative family history and the absence of symptoms.

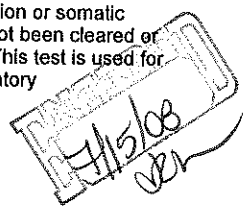
This interpretation is based on the clinical and family relationship information provided and the current understanding of the molecular genetics of this condition.
Enzyme: [White Blood Cells]
This result is within the non-carrier range for Tay-Sachs disease. Less than 0.1% of patients having non-carrier levels of Hexosaminidase-A activity are Tay-Sachs carriers.

NOTE: Maximum sensitivity and specificity for Tay-Sachs disease carrier testing are achieved by using enzymology and DNA mutation analysis together.

METHOD:

DNA is isolated from the sample and amplified for disease specific regions using the polymerase chain reaction (PCR). Mutations are identified by hybridization to allele specific oligonucleotides or by solution-phase multiplex allele-specific primer extension with subsequent mutation-specific hybridization and detection.

False positive or negative results may occur for reasons that include genetic variants, blood transfusions, bone marrow transplantation or somatic heterogeneity of the tissue sample. This test was developed and its performance characteristics determined by Genzyme. It has not been cleared or approved by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is regulated under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) as qualified to perform high complexity clinical testing.



(REPORT CONTINUED ...)

Date: 06/27/2008

Narasimhan Nagan, Ph.D., FACMG
Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

genzyme

Carrier Testing

Patient Name: Donor 2925, . Referring Physician: [redacted] Specimen #: [redacted] Patient ID: [redacted]

MUTATIONS ANALYZED / DETECTION RATE

...Continued From Page 1

Cystic Fibrosis

ΔF311	2043delG	3120+1G>A	4016insT	712-1G>T	G330X	Q359K/T360K	R347P	S549N
ΔF508	2055del9>A	3120G>A	405+1G>A	935delA	G480C	Q493X	R352Q	S549R T>G
ΔI507	2105del13ins5	3171delC	405+3A>C	936delTA	G542X	Q552X	R553X	T338I
1078delT	2108delA	3199del6	406-1G>A	A455E	G551D	Q890X	R560T	V520F
1288insTA	2143delT	3659delC	444delA	A559T	G85E	R1066C	R709X	W1089X
1677delTA	2183delAA>G	3667del4	457TAT>G	C524X	K710X	R1158X	R75X	W1204X
1717-1G>A	2184delA	3791delC	574delA	CFTRdele2,3	L206W	R1162X	R764X	W1282X
1812-1G>A	2184insA	3849+10kbC>T	621+1G>T	D1152H	M1101K	R117C	S1196X	Y1092X C>A
1898+1G>A	2307insA	3876delA	663delT	E60X	N1303K	R117H	S1251N	Y1092X C>G
1898+5G>T	2789+5G>A	3905insT	711+1G>T	E92X	P574H	R334W	S1255X	Y122X
1949del84	2869insG	394delTT	711+5G>A	G178R	Q1238X	R347H	S364P	

This 97 mutation assay discriminates between ΔF508 and the following polymorphisms: F508C, I506V and I507V.

Mutation Detection Rates among Ethnic Groups

Detection rates are based on mutation frequencies in patients affected with cystic fibrosis. Among individuals with an atypical or mild presentation (e.g. congenital absence of the vas deferens, pancreatitis) detection rates may vary from those provided here.

Ethnicity	Carrier risk reduction when no family history	Detection rate	References
African American	1/65 to 1/338	81%	Genet in Med 3:168, 2001
Ashkenazi Jewish	1/26 to 1/834	97%	Am J Hum Genet 51:951, 1994
Asian		Not Provided	Insufficient data
Caucasian	1/25 to 1/343	93%	Genet in Med 3:168, 2001; Genet in Med 4:90, 2002
Hispanic	1/46 to 1/205	78%	Genet in Med 3:168, 2001;www.dhs.ca.gov/pcfh/gdb/html/PDE/CFStudy.htm
Jewish, non-Ashkenazi		Varies by country of origin	Genet Testing 5:47, 2001, Genet Testing, 1:35, 1997
Other or Mixed Ethnicity		Not Provided	Detection rate not determined and varies with ethnicity

Under the direction of:

Narasimhan Nagan, Ph.D., FACMG

Additional approvals by:

Enzyme:Stanford Marenberg, Ph.D.

Molecular Testing Performed At Genzyme Genetics 3400 Computer Drive Westborough, MA 01581 1-800-255-7357
Biochemical Testing Performed At Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505 1-800-848-4436

QUEST DIAGNOSTICS INCORPORATED

PATIENT INFORMATION
DONOR,2925

DOB: Age:
GENDER: M

ID: 2925-080616

REPORT STATUS **Final**

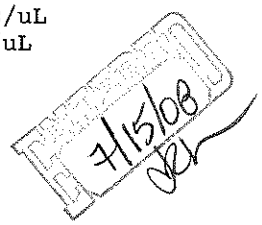
ORDERING PHYSICIAN
[redacted]

CLIENT INFORMATION
41550
FAIRFAX CRYOBANK
[redacted]

SPECIMEN INFORMATION
SPECIMEN: IF671333Y
REQUISITION: 0097116
LAB REF NO:

COLLECTED: 06/16/2008 11:00
RECEIVED: 06/17/2008 04:52
REPORTED: 06/19/2008 02:10

Test Name	In Range	Out of Range	Reference Range	Lab
HEMOGLOBINOPATHY EVALUATION				
HEMOGLOBINOPATHY INDICES				IG
RED BLOOD CELL COUNT	5.03		4.20-5.80 Million/uL	
HEMOGLOBIN	15.9		13.2-17.1 g/dL	
HEMATOCRIT	46.2		38.5-50.0 %	
MCV	91.7		80.0-100.0 fL	
MCH	31.6		27.0-33.0 pg	
RDW	13.6		11.0-15.0 %	
HEMOGLOBINOPATHY EVALUATION				IG
HEMOGLOBIN A1	97.7		>96.0 %	
HEMOGLOBIN F	<1.0		<2.0 %	
HEMOGLOBIN A2 (QUANT)	2.3		1.8-3.5 %	
INTERPRETATION	NORMAL PHENOTYPE.			
CHOLESTEROL, TOTAL	190		125-200 mg/dL	IG
AST	20		10-35 U/L	IG
ALT	15		9-60 U/L	IG
CBC (INCLUDES DIFF/PLT)				IG
WHITE BLOOD CELL COUNT	5.2		3.8-10.8 Thousand/uL	
RED BLOOD CELL COUNT	5.03		4.20-5.80 Million/uL	
HEMOGLOBIN	15.9		13.2-17.1 g/dL	
HEMATOCRIT	46.2		38.5-50.0 %	
MCV	91.7		80.0-100.0 fL	
MCH	31.6		27.0-33.0 pg	
MCHC	34.5		32.0-36.0 g/dL	
RDW	13.6		11.0-15.0 %	
PLATELET COUNT	179		140-400 Thousand/uL	
ABSOLUTE NEUTROPHILS	3104		1500-7800 cells/uL	
ABSOLUTE LYMPHOCYTES	1524		850-3900 cells/uL	
ABSOLUTE MONOCYTES	296		200-950 cells/uL	
ABSOLUTE EOSINOPHILS	244		15-500 cells/uL	
ABSOLUTE BASOPHILS	31		0-200 cells/uL	
NEUTROPHILS	59.7		%	
LYMPHOCYTES	29.3		%	
MONOCYTES	5.7		%	
EOSINOPHILS	4.7		%	
BASOPHILS	0.6		%	



Recombine

CarrierMap™

Ordering Practice:

Practice Code: 926
Fairfax Cryobank
[redacted]
Report Generated: 2015-09-16

Donor 2925 [redacted]

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 29949
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #31387
Specimen Collection: 2015-09-09
Specimen Received: 2015-09-10
Specimen Analyzed: 2015-09-16

Partner Not Tested

SUMMARY OF RESULTS

NO MUTATIONS IDENTIFIED

Donor 2925 [redacted] was not identified to carry any of the mutations tested.

All mutations analyzed were not detected, reducing but not eliminating your chance to be a carrier for the associated genetic diseases. A list of all the diseases and mutations you were screened for is included later in this report. The test does not screen for every possible genetic disease.

For disease information, please visit www.recombine.com/diseases. To speak with a Genetic Counselor, call 855.OUR.GENES.

♂ Male

Panel: Fairfax Cryobank Panel , Diseases Tested: 21, Mutations Tested: 382, Genes Tested: 22, Null Calls: 0

Assay performed by Reprogenetics
CLIA ID: 31D1054821
Lab Technician Bo Chu

Reviewed by Pere Colls, PhD, HCLD, Lab Director

Methods and Limitations

Genotyping: Genotyping is performed using the Illumina Infinium Custom HD Genotyping assay to identify mutations in >200 genes. The assay is not validated for homozygous mutations, and it is possible that individuals affected with disease may not be accurately genotyped.

Spinal Muscular Atrophy: Spinal Muscular Atrophy is tested for via an Identity-by-State shared haplotype comparison algorithm. Detection is limited to haplotypes within our library of known carriers of the most common mutation (deletion of Exon 7).

Limitations: In some cases, genetic variations other than that which is being assayed may interfere with mutation detection, resulting in false-negative or false-positive results. Additional sources of error include, but are not limited to: sample contamination, sample mix-up, bone marrow transplantation, blood transfusions, and technical errors.

The test does not test for all forms of genetic disease, birth defects, and intellectual disability. All results should be interpreted in the context of family history; additional evaluation may be indicated based on a history of these conditions. Additional testing may be necessary to determine mutation phase in individuals identified to carry more than one mutation in the same gene. All mutations included within the genes assayed may not be detected, and additional testing may be appropriate for some individuals.

Diseases & Mutations Assayed

High Impact | Treatment Benefits | X-Linked | Moderate Impact

H	T	X	M	Disease	#	Mutations
●	○	○	○	Alpha Thalassemia	10	♂ Genotyping | SEA deletion, 11.1kb deletion, c.207C>A (p.N69K), c.223G>C (p.D75G), c.2T>C (p.M1T), c.207C>G (p.N69K), c.340_351delCTCCCCGCCGAG (p.L114_ E117del), c.377T>C (p.L126P), c.427T>C (p.X143Qext32), c.*+94A>G
●	●	○	○	Beta Thalassemia	83	♂ Genotyping | c.17_18delCT, c.20delA (p.E7Gfs), c.217insA (p.S73Kfs), c.223+702_444+342del620insAAGTAGA, c.230delC, c.25_26delAA, c.315+1G>A, c.315+2T>C, c.316-197C>T, c.316-146T>G, c.315+745C>G, c.316-1G>A, c.316-1G>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, c.4delG (p.V2Cfs), c.51delC (p.K18Rfs), c.93-21G>A, c.92+1G>A, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>T, c.-50A>C, c.a-78g, c.a-79g, c.a-81g, c.A52T (p.K18X), c.c-137g, c.c-138t, c.c-151t, c.C118T (p.Q40X), c.G169C (p.G57R), c.G295A (p.V99M), c.G34A (p.V12I), c.G415C (p.A139P), c.G47A (p.W16X), c.G48A (p.W16X), c.t-80a, c.T2C (p.M1T), c.T75A (p.G25G), c.444+111A>G, c.g-29a, c.68_74delAAGTTGG, c.G92C (p.R31T), c.27_28insG, c.92+1G>T, c.92+1G>C, c.93-15T>G, c.93-1G>C, c.112delT, c.G113A (p.W38X), c.G114A (p.W38X), c.126delC, c.444+113A>G, c.250delG, c.225delC, c.383_385delAGG (p.Q128_A129delQAinsP), c.321_322insG (p.N109fs), c.316-1G>T, c.316-2A>C, c.316-106C>T, c.287_288insA (p.L97fs), c.271G>T (p.E91X), c.203_204delTG (p.V68Afs), c.154delC (p.P52fs), c.135delC (p.F46fs), c.92+2T>A, c.92+2T>C, c.90C>T (p.G30G), c.59A>G (p.N20S), c.46delT (p.W16Gfs), c.45_46insG (p.L16fs), c.36delT (p.T13fs), c.2T>G (p.M1R), c.1A>G (p.M1V), c.c-137t, c.c-136g, c.c-142t, c.c-140t
●	○	○	○	Bloom Syndrome	24	♂ Genotyping | c.2207_2212delATCTGAinsTAGATTC (p.Y736Lfs), c.2407insT, c.557_559delCAA (p.S186X), c.1284G>A (p.W428X), c.1701G>A (p.W567X), c.1933C>T (p.Q645X), c.C2528T (p.T843I), c.C2695T (p.R899X), c.G3107T (p.C1036F), c.2923delC (p.Q975K), c.3558+1G>T, c.3875-2A>G, c.2074+2T>A, c.2343_2344dupGA (p.781EfsX), c.380delC (p.127Tfs), c.3564delC (p.1188Dfs), c.4008delG (p.1336Rfs), c.C947G (p.S316X), c.2193+1_2193+9del9, c.C1642T (p.Q548X), c.3143delA (p.1048NfsX), c.356_357delTA (p.Cys120Hisfs), c.4076+1delG, c.C3281A (p.S1094X)
●	○	○	○	Canavan Disease	8	♂ Genotyping | c.433-2A>G, c.A854C (p.E285A), c.C693A (p.Y231X), c.C914A (p.A305E), c.A71G (p.E24G), c.C654A (p.C218X), c.T2C (p.M1T), c.G79A (p.G27R)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Cystic Fibrosis	130	♂ Genotyping | c.1029delC, 1153_1154insAT, c.1519_1521delATC (p.507delI), c.1521_1523delCTT (p.508delF), c.1545_1546delTA (p.Y515Xfs), c.1585-1G>A, c.164+12T>C, c.1680-886A>G, c.1680-1G>A, c.1766+1G>A, c.1766+1G>T, c.1766+5G>T, c.1818del84, c.1911delG, c.1923delCTCAAAACTinsA, c.1973delGAAATTCAATCCTinsAGAAA, c.2052delA (p.K684fs), c.2052insA (p.Q685fs), c.2051_2052delAAinsG (p.K684SfsX38), c.2174insA, c.261delTT, c.2657+5G>A, c.273+1G>A, c.273+3A>C, c.274-1G>A, c.2988+1G>A, c.3039delC, c.3140-26A>G, c.325delTATinsG, c.3527delC, c.3535delACCA, c.3691delT, c.3717+12191C>T, c.3744delA, c.3773_3774insT (p.L1258fs), c.442delA, c.489+1G>T, c.531delT, c.579+1G>T, c.579+5G>A (IVS4+5G>A), c.803delA (p.N268fs), c.805_806delAT (p.I269fs), c.933_935delCTT (p.311delF), c.A1645C (p.S549R), c.A2128T (p.K710X), c.C1000T (p.R334W), c.C1013T (p.T338I), c.C1364A (p.A455E), c.C1477T (p.Q493X), c.C1572A (p.C524X), c.C1654T (p.Q552X), c.C1657T (p.R553X), c.C1721A (p.P574H), c.C2125T (p.R709X), c.C223T (p.R75X), c.C2668T (p.Q890X), c.C3196T (p.R1066C), c.C3276G (p.Y1092X), c.C3472T (p.R1158X), c.C3484T (p.R1162X), c.C349T (p.R117C), c.C3587G (p.S1196X), c.C3712T (p.Q1238X), c.C3764A (p.S1255X), c.C3909G (p.N1303K), c.G1040A (p.R347H), c.G1040C (p.R347P), c.G1438T (p.G480C), c.G1624T (p.G542X), c.G1646A (p.S549N), c.G1646T (p.S549I), c.G1652A (p.G551D), c.G1675A (p.A559T), c.G1679C (p.R560T), c.G178T (p.E60X), c.G1865A (p.G622D), c.G254A (p.G85E), c.G271A (p.G91R), c.G274T (p.E92X), c.G3209A (p.R1070Q), c.G3266A (p.W1089X), c.G3454C (p.D1152H), c.G350A (p.R117H), c.G3611A (p.W1204X), c.G3752A (p.S1251N), c.G3846A (p.W1282X), c.G3848T (p.R1283M), c.G532A (p.G178R), c.G988T (p.G330X), c.T1090C (p.S364P), c.T3302A (p.M1101K), c.T617G (p.L206W), c.C14T (p.P5L), c.G19T (p.E7X), c.G171A (p.W57X), c.313delA (p.I105fs), c.G328C (p.D110H), c.580-1G>T, c.G1055A (p.R352Q), c.C1075A (p.Q359K), c.C1079A (p.T360K), c.T1647G (p.S549R), c.1976delA (p.N659fs), c.C2290T (p.R764X), c.2737_2738insG (p.Y913X), c.3067_3072delATAGTG (p.I1023_V1024delT), c.3536_3539delCCAA (p.T1179fs), c.3659delC (p.T1220fs), c.G3808A (p.D1270N), c.G4056C (p.Q1352H), c.C4364G (p.S1455X), c.C4003T (p.L1335F), c.G2538A (p.W846X), c.C200T (p.P67L), c.C4426T (p.Q1476X), c.1116+1G>A, c.1986_1989delAACT (p.T663R), c.2089_2090insA (p.R697Kfs), c.2215delG (p.V739Y), c.T263G (p.L196X), c.3022delG (p.V1008S), c.3908dupA (p.N1303Kfs), c.C658T (p.Q220X), c.C868T (p.Q290X), c.1526delG (p.G509fs), c.2908+1085-3367+260del7201, c.C11A (p.S4X), c.A3700G (p.I1234V), c.A416T (p.H139L), c.T366A (p.Y122X)
●	○	○	○	Familial Dysautonomia	4	♂ Genotyping | c.2204+6T>C, c.C2741T (p.P914L), c.G2087C (p.R696P), c.C2128T (p.Q710X)
●	○	○	○	Familial Hyperinsulinism: Type 1: ABCC8 Related	10	♂ Genotyping | c.3989-9G>A, c.4159_4161delTTC (p.1387delF), c.C4258T (p.R1420C), c.C4477T (p.R1493W), c.G2147T (p.G716V), c.G4055C (p.R1352P), c.T560A (p.V187D), c.4516G>A (p.E1506K), c.C2506T (p.Q836X), c.579+2T>A
●	●	○	○	Fanconi Anemia: Type C	8	♂ Genotyping | c.456+4A>T, c.67delG, c.C37T (p.Q13X), c.C553T (p.R185X), c.T1661C (p.L554P), c.C1642T (p.R548X), c.G66A (p.W22X), c.G65A (p.W22X)
●	●	○	○	Gaucher Disease	6	♂ Genotyping | c.84_85insG, c.A1226G (p.N409S), c.A1343T (p.D448V), c.C1504T (p.R502C), c.G1297T (p.V433L), c.G1604A (p.R535H)
●	●	○	○	Glycogen Storage Disease: Type IA	13	♂ Genotyping | c.376_377insTA, c.79delC, c.979_981delTTC (p.327delF), c.C1039T (p.Q347X), c.C247T (p.R83C), c.C724T (p.Q242X), c.G248A (p.R83H), c.G562C (p.G188R), c.G648T, c.G809T (p.G270V), c.A113T (p.D38V), c.975delG (p.L326fs), c.724delC
●	○	○	○	Joubert Syndrome	1	♂ Genotyping | c.G35T (p.R12L)
●	●	○	○	Maple Syrup Urine Disease: Type 1B	6	♂ Genotyping | c.G1114T (p.E372X), c.G548C (p.R183P), c.G832A (p.G278S), c.C970T (p.R324X), c.G487T (p.E163X), c.C853T (p.R285X)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Maple Syrup Urine Disease: Type 3	8	♂ Genotyping | c.104_105insA, c.G685T (p.G229C), c.A214G (p.K72E), c.A1081G (p.M361V), c.G1123A (p.E375K), c.T1178C (p.I393T), c.C1463T (p.P488L), c.A1483G (p.R495G)
●	○	○	○	Mucolipidosis: Type IV	4	♂ Genotyping | c.406-2A>G, c.G1084T (p.D362Y), c.C304T (p.R102X), c.244delC (p.L82fsX)
●	○	○	○	Nemaline Myopathy: NEB Related	1	♂ Genotyping | c.7434_7536del2502bp
●	○	○	○	Niemann-Pick Disease: Type A	6	♂ Genotyping | c.996delC, c.G1493T (p.R498L), c.T911C (p.L304P), c.C1267T (p.H423Y), c.G1734C (p.K578N), c.1493G>A (p.R498H)
●	○	○	○	Spinal Muscular Atrophy: SMN1 Linked	19	♂ Genotyping | DEL EXON 7, c.22_23insA, c.43C>T (p.Q15X), c.91_92insT, c.305G>A (p.W102X), c.400G>A (p.E134K), c.439_443delGAAGT, c.558delA, c.585_586insT, c.683T>A (p.L228X), c.734C>T (p.P245L), c.768_778dupTGCTGATGCTT, c.815A>G (p.Y272C), c.821C>T (p.T274I), c.823G>A (p.G275S), c.834+2T>G, c.835-18_835-12delCCTTTAT, c.835G>T, c.836G>T
●	○	○	○	Tay-Sachs Disease	30	♂ Genotyping | c.1073+1G>A, c.1277_1278insTATC, c.1421+1G>C, c.805+1G>A, c.C532T (p.R178C), c.G533A (p.R178H), c.G805A (p.G269S), c.C1510T (p.R504C), c.G1496A (p.R499H), c.G509A (p.R170Q), c.A1003T (p.I335F), c.910_912delTTC (p.305delF), c.G749A (p.G250D), c.T632C (p.F211S), c.C629T (p.S210F), c.613delC, c.A611G (p.H204R), c.G598A (p.V200M), c.A590C (p.K197T), c.571-1G>T, c.C540G (p.Y180X), c.T538C (p.Y180H), c.G533T (p.R178L), c.C508T (p.R170W), c.C409T (p.R137X), c.T380G (p.L127R), c.346+1G>C, c.T116G (p.L39R), c.G78A (p.W26X), c.A1G (p.M1V)
●	○	○	○	Usher Syndrome: Type 1F	6	♂ Genotyping | c.C733T (p.R245X), c.2067C>A (p.Y684X), c.C7T (p.R3X), c.C1942T (p.R648X), c.2800C>T (p.R934X), c.4272delA (p.L1425fs)
●	○	○	○	Usher Syndrome: Type 3	4	♂ Genotyping | c.T144G (p.N48K), c.T359A (p.M120K), c.300T>G (p.Y176X), c.C634T (p.Q212X)
●	○	○	○	Walker-Warburg Syndrome	1	♂ Genotyping | c.1167insA (p.F390fs)