Fairfax Cryobank
The Trusted Choice for Donor Sperm

Donor 5316

Genetic Testing Summary

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

Last Updated: 05/06/19

Donor Reported Ancestry: Russian, English 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 130 mutations in the CFTR gene	1/350
Spinal Muscular Atrophy (SMA) carrier screening	Negative for deletions of exon 7 in the SMN1 gene	1/632
Additional testing attached- 229 diseases by genotyping	Carrier: Joubert Syndrome (TMEM216) Carrier: Nonsyndromic Hearing Loss and Deafness: GJB2 Related Negative for other mutations tested	Partner testing is recommended before using this donor.

*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.

Recombine

CarrierMap™

Ordering Practice:

Practice Code: [redacted]
Fairfax Cryobank - [redacted]
[redacted]
Physician: [redacted]
Report Generated: 2015-10-12

Donor 5316

DOB: [redacted]
Gender: Male
Ethnicity: European
Procedure ID: 31313
Kit Barcode: [redacted]
Method: Genotyping
Specimen: Blood, #32857
Specimen Collection: 2015-09-25
Specimen Received: 2015-09-28
Specimen Analyzed: 2015-10-06

Partner Not Tested

SUMMARY OF RESULTS

MUTATION(S) IDENTIFIED

Disease	Donor 5316	Partner Not Tested
Joubert Syndrome High Impact	Carrier (1 abnormal copy) Mutation: c.G35T (p.R12L) Gene: TMEM216 Method: Genotyping Reproductive risk detected. Consider partner testing.	
Nonsyndromic Hearing Loss and Deafness: GJB2 Related High Impact Treatment Benefits	Carrier (1 abnormal copy) Mutation: c.35delG Gene: GJB2 Method: Genotyping Reproductive risk detected. Consider partner testing.	

All other 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: CarrierMap Expanded , Diseases Tested: 229, Mutations Tested: 1741, Genes Tested: 221, Null Calls: 0

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

Reviewed by Pere Colls, PhD, HCLD, Lab Director

Joubert Syndrome

Joubert syndrome causes brain abnormalities. In this disease, the TMEM216 gene responsible for forming structures important for chemical signaling during development is defective. As a result, certain tissues do not develop correctly. Patients affected exhibit weak muscle tone in infancy, which progresses in early childhood into difficulty with movement. Affected individuals have delayed development and may exhibit intellectual disability from moderate severe with prominent speech and behavioral problems. Patients with the TMEM216 related form of this disease often develop kidney disease and in some cases have extra fingers or toes. Heart abnormalities and encephalocele, a type of neural tube defect are also sometimes seen in this condition.

Clinical Information

Physical Impairment	●
Cognitive Impairment	●
Shortened Lifespan	●
Effective Treatment	

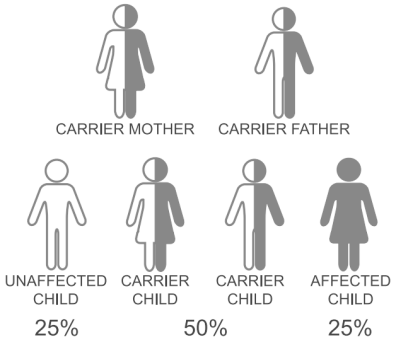
● High Impact

Status

Donor 5316:

Carrier (1 abnormal copy)
Mutation: c.G35T (p.R12L)
Gene: TMEM216
Method: Genotyping

Inheritance



Treatment

Treatment is management orientated. No treatment that reverses or slows the natural history of Joubert syndrome is available. Care should be taken in managing respiratory and feeding problems related to either breathing abnormalities or hypotonia. Rehabilitation strategies must be planned for cognitive and behavioral difficulties and specific manifestations such as the visual impairment.

Prognosis

Prognosis is generally unfavorable. Early psychomotor development is invariably delayed, and the cognitive functioning of all the patients eventually progresses to the mildly to severely retarded range. Oral motor difficulties are invariably present from birth, resulting in swallowing and chewing difficulties and, in most patients, failure to thrive. Some patients suffer from end-stage renal insufficiency by mid-adolescence

To learn more visit http://recombine.com/diseases/joubert-syndrome

Carrier Risk Assessment

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Ashkenazi Jewish	>99%	1/92	1/9200

Nonsyndromic Hearing Loss and Deafness: GJB2 Related

Nonsyndromic hearing loss and deafness is an inherited form of hearing loss that is not associated with other medical findings. There are many genetic causes of nonsyndromic hearing loss and deafness, with mutations in the GJB2 gene accounting for a large proportion of cases. This gene is responsible for a protein called connexin 26, which is normally involved in maintaining the proper balance of nutrients within the inner ear. Affected individuals typically have hearing loss from birth. The degree of hearing impairment is similar in both ears, may be mild to profound, and generally remains stable over time.

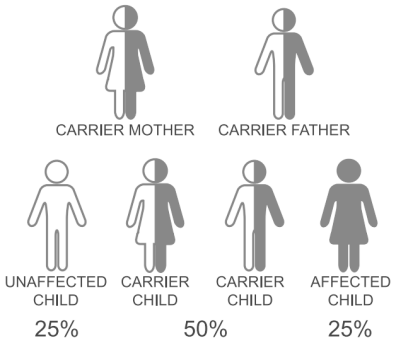
High Impact
Treatment Benefits

Status

Donor 5316:

Carrier (1 abnormal copy)
Mutation: c.35delG
Gene: GJB2
Method: Genotyping

Inheritance



Clinical Information

Physical Impairment	●
Cognitive Impairment	
Shortened Lifespan	
Effective Treatment	

Treatment

The degree of hearing loss should be assessed in infancy by auditory brain response audiometry. Hearing aids, including cochlear implants for those with profound deafness, can be considered. An appropriate educational program is necessary to prevent learning delays.

Prognosis

Prognosis is good if hearing loss is detected by age 2. The hearing impairment is typically present at birth and the degree of impairment can be mild to profound and varies greatly between affected individuals, even among siblings. The degree of hearing loss typically remains stable over time.

Carrier Risk Assessment

Ethnicity	Detection Rate	Pre-Test Risk	Post-Test Risk
Ashkenazi Jewish	95.83%	1/20	1/480
Chinese	82.26%	1/100	1/564
European	77.73%	1/53	1/238
Indian	66.98%	Unknown	Unknown
Israeli	93.10%	1/16	1/232
Japanese	69.44%	1/75	1/245
Roma	92.86%	Unknown	Unknown
United States	46.50%	1/34	1/64

To learn more visit http://recombine.com/diseases/nonsyndromic-hearing-loss-and-deafness-gjb2-related

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
●	●	○	○	11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia	1	♂ Genotyping | c.G1343A (p.R448H)
●	●	○	○	17-Alpha-Hydroxylase Deficiency	20	♂ Genotyping | c.157_159delTTC (p.53delF), c.T316C (p.S106P), c.C715T (p.R239X), c.C1024A (p.P342T), c.C286T (p.R96W), c.G1040A (p.R347H), c.G1073A (p.R358Q), c.G51A (p.W17X), c.T340G (p.F114V), c.A347T (p.D116V), c.C1039T (p.R347C), c.C1084T (p.R362C), c.T1216C (p.W406R), c.T985G (p.Y329D), c.T601A (p.Y201N), c.C81A (p.Y27X), c.G287A (p.R96Q), c.C1226G (p.P409R), c.T1250G (p.F417C), c.T278G (p.F93C)
●	●	○	○	17-Beta-Hydroxysteroid Dehydrogenase Type III Deficiency	8	♂ Genotyping | c.C695T (p.S232L), c.A703G (p.M235V), c.G239A (p.R80Q), c.C608T (p.A203V), c.C238T (p.R80W), c.G166A (p.A56T), c.A389G (p.N130S), c.G803A (p.C268Y)
●	●	○	○	21-Hydroxylase-Deficient Congenital Classical Adrenal Hyperplasia	3	♂ Genotyping | c.293-13C>G, c.332_339delGAGACTAC, c.G1273A (p.G425S)
○	●	○	●	21-Hydroxylase-Deficient Congenital Nonclassical Adrenal Hyperplasia	2	♂ Genotyping | c.C1360T (p.P454S), c.G844C (p.V282L)
●	●	○	○	3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	6	♂ Genotyping | c.G512A (p.W171X), c.742_747delGTCCGAinsAACTA (p.V248NfsR249X), c.C745T (p.R249X), c.C29A (p.A10E), c.G424A (p.E142K), c.C664A (p.P222T)
●	●	○	○	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC1 Related	2	♂ Genotyping | c.A1155C (p.R385S), c.T1310C (p.L437P)
●	●	○	○	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC2 Related	8	♂ Genotyping | c.G295C (p.E99Q), c.T499C (p.C167R), c.G464A (p.R155Q), c.A569G (p.H190R), c.G803C (p.R268T), c.G838T (p.D280Y), c.C929G (p.P310R), c.A1309G (p.I437V)
●	○	○	○	3-Methylglutaconic Aciduria: Type 3	4	♂ Genotyping | c.415C>T (p.Q139X), c.313C>G (p.Q105E), c.277G>A (p.G93S), c.143-1G>C
●	●	○	○	3-Phosphoglycerate Dehydrogenase Deficiency	7	♂ Genotyping | c.G1468A (p.V490M), c.C403T (p.R135W), c.712delG (p.G238fsX), c.G1273A (p.V425M), c.G1117A (p.A373T), c.G781A (p.V261M), c.G1129A (p.G377S)
○	○	○	●	5-Alpha Reductase Deficiency	10	♂ Genotyping | c.C736T (p.R246W), c.T164A (p.L55Q), c.G344A (p.G115D), c.G547A (p.G183S), c.C679T (p.R227X), c.G682A (p.A228T), c.G586A (p.G196S), c.A692G (p.H231R), c.C635G (p.P212R), c.G591T (p.E197D)
●	●	○	○	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency	6	♂ Genotyping | c.46C>T (p.R16C), c.74G>A (p.R25Q), c.155A>G (p.N52S), c.259C>T (p.P87S), c.286G>A (p.D96N), c.347A>G (p.D116G)
●	○	○	○	ARSACS	6	♂ Genotyping | c.C12973T (p.R4325X), c.C7504T (p.R2502X), c.T9742C (p.W3248R), c.8844delT (p.I2949fs), c.T5836C (p.W1946R), c.T3161C (p.F1054S)
●	●	○	○	Abetalipoproteinemia	2	♂ Genotyping | c.G2593T (p.G865X), c.2211delT
●	●	○	○	Acrodermatitis Enteropathica	9	♂ Genotyping | c.1223-1227delCCGGG, c.968-971delAGTC, c.C283T (p.R95C), c.C318A (p.N106K), c.C599T (p.P200L), c.G1120A (p.G374R), c.G1576C (p.G526R), c.G909C (p.Q303H), c.G989A (p.G330D)
●	○	○	○	Acyl-CoA Oxidase I Deficiency	5	♂ Genotyping | c.372delCATGCCCGCCTGGAACTT, c.A832G (p.M278V), c.A926G (p.Q309R), c.C442T (p.R148X), c.G532T (p.G178C)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Adenosine Deaminase Deficiency	22	♂ Genotyping | c.986C>T (p.A329V), c.872C>T (p.S291L), c.646G>A (p.G216R), c.632G>A (p.R211H), c.631C>T (p.R211C), c.596A>C (p.Q199P), c.536C>A (p.A179D), c.529G>A (p.V177M), c.467G>A (p.R156H), c.466C>T (p.R156C), c.454C>A (p.L152M), c.445C>T (p.R149W), c.419G>A (p.G140E), c.385G>A (p.V129M), c.320T>C (p.L107P), c.302G>A (p.R101Q), c.302G>T (p.R101L), c.301C>T (p.R101W), c.248C>A (p.A83D), c.220G>T (p.G74C), c.58G>A (p.G20R), c.43C>G (p.H15D)
○	○	○	●	Alkaptonuria	14	♂ Genotyping | c.1111_1112insC, c.16-1G>A, c.174delA, c.342+1G>A, c.A1102G (p.M368V), c.C140T (p.S47L), c.C688T (p.P230S), c.G481A (p.G161R), c.G808A (p.G270R), c.T899G (p.V300G), c.G990T (p.R330S), c.457_458insG, c.T360G (p.C120W), c.1112A>G (p.H371R)
●	○	○	○	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
○	○	○	●	Alpha-1-Antitrypsin Deficiency	4	♂ Genotyping | c.226_228delTTC (p.76delF), c.A1131T (p.L377F), c.C187T (p.R63C), c.G1096A (p.E366K)
●	○	○	○	Alpha-Mannosidosis	3	♂ Genotyping | c.2426T>C (p.L809P), c.2248C>T (p.R750W), c.1830+1G>C (p.V549_E610del)
●	○	○	○	Alport Syndrome: COL4A3 Related	3	♂ Genotyping | c.4420_4423delCTTTT, c.C4441T (p.R1481X), c.C4571G (p.S1524X)
●	○	○	○	Alport Syndrome: COL4A4 Related	5	♂ Genotyping | c.C3713G (p.S1238X), c.C4129T (p.R1377X), c.C4715T (p.P1572L), c.C4923A (p.C1641X), c.G3601A (p.G1201S)
●	○	○	○	Amegakaryocytic Thrombocytopenia	3	♂ Genotyping | c.79+2T>A, c.C127T (p.R43X), c.G305C (p.R102P)
●	○	○	○	Andermann Syndrome	5	♂ Genotyping | c.2436delG (p.T813fsX813), c.901delA, c.C2023T (p.R675X), c.C3031T (p.R1011X), c.C619T (p.R207C)
●	○	○	○	Antley-Bixler Syndrome	4	♂ Genotyping | c.G859C (p.A287P), c.G1615A (p.G539R), c.T1475A (p.V492E), c.G1370A (p.R457H)
●	●	○	○	Argininosuccinate Lyase Deficiency	7	♂ Genotyping | c.446+1G>A, c.A857G (p.Q286R), c.C1135T (p.R379C), c.C1153T (p.R385C), c.C283T (p.R95C), c.G532A (p.V178M), c.C1060T (p.Q354X)
●	●	○	○	Aromatase Deficiency	8	♂ Genotyping | c.1222delC, c.468delC, c.629-3C>A, c.C1123T (p.R375C), c.C1303T (p.R435C), c.G1094A (p.R365Q), c.G1310A (p.C437Y), c.G628A (p.E210K)
●	○	○	○	Arthrogryposis, Mental Retardation, & Seizures	2	♂ Genotyping | c.1012A>G (p.S338G), c.C514T (p.Q172X)
●	○	○	○	Aspartylglycosaminuria	8	♂ Genotyping | c.200_201delAG, c.G482A (p.R161Q), c.G488C (p.C163S), c.T214C (p.S72P), c.T916C (p.C306R), c.G904A (p.G302R), c.C302T (p.A101V), c.G179A (p.G60D)
●	●	○	○	Ataxia with Vitamin E Deficiency	4	♂ Genotyping | c.744delA, c.G575A (p.R192H), c.C400T (p.R134X), c.T303G (p.H101Q)
●	○	○	○	Ataxia-Telangiectasia	19	♂ Genotyping | c.C103T (p.R35X), c.1564_1565delGA (p.E522fs), c.3245delATCinsTGAT (p.H1082fs), c.G3576A (p.K1192K), c.3894insT, c.5712_5713insA (p.S1905fs), c.5762+1126A>G, c.C5908T (p.Q1970X), c.G5932T (p.E1978X), c.A7268G (p.E2423G), c.T7271G (p.V2424G), c.C7327T (p.R2443X), c.7517_7520delGAGA (p.R2506fs), c.7630-2A>C, c.7638_7646delTAGAATTTC (p.R2547_S2549delRIS), c.G7876C (p.A2626P), c.T7967C (p.L2656P), c.A8030G (p.Y2677C), c.T8480G (p.F2827C)

H	T	X	M	Disease	#	Mutations
●	○	○	○	Autosomal Recessive Polycystic Kidney Disease	17	♂ Genotyping | c.5895insA (p.L1966fsX1969), c.9689delA (p.D3230fs), c.C107T (p.T36M), c.C1486T (p.R496X), c.T10412G (p.V3471G), c.T10658C (p.I3553T), c.10174C>T (p.Q3392X), c.9530T>C (p.I3177T), c.C9053T (p.S3018F), c.8870T>C (p.I2957T), c.C8011T (p.R2671X), c.6992T>A (p.I2331K), c.G5221A (p.V1741M), c.C4991T (p.S1664F), c.3761_3762delCCinsG (p.A1254fs), c.2414C>T (p.P805L), c.664A>G (p.I222V)
●	○	○	○	Bardet-Biedl Syndrome: BBS1 Related	3	♂ Genotyping | c.851delA, c.G1645T (p.E549X), c.T1169G (p.M390R)
●	○	○	○	Bardet-Biedl Syndrome: BBS10 Related	3	♂ Genotyping | c.271_273ins1bp (p.C91fsX95), c.G101C (p.R34P), c.T931G (p.S311A)
●	○	○	○	Bardet-Biedl Syndrome: BBS11 Related	1	♂ Genotyping | c.C388T (p.P130S)
●	○	○	○	Bardet-Biedl Syndrome: BBS12 Related	5	♂ Genotyping | c.335_337delTAG, c.865G>C (p.A289P), c.1063C>T (p.R355X), c.1114_1115delTT (p.F372X), c.1483_1484delGA (p.E495fsX498)
●	○	○	○	Bardet-Biedl Syndrome: BBS2 Related	5	♂ Genotyping | c.940delA, c.C72G (p.Y24X), c.T224G (p.V75G), c.A311C (p.D104A), c.G1895C (p.R632P)
●	○	○	○	Bare Lymphocyte Syndrome: Type II	1	♂ Genotyping | c.G1141T (p.E381X)
●	●	○	○	Bartter Syndrome: Type 4A	6	♂ Genotyping | c.A1T (p.M1L), c.C22T (p.R8W), c.G139A (p.G47R), c.G23T (p.R8L), c.G28A (p.G10S), c.G3A (p.M1I)
●	●	○	○	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
●	○	○	○	Beta-Hexosaminidase Pseudodeficiency	2	♂ Genotyping | c.C739T (p.R247W), c.C745T (p.R249W)
●	●	○	○	Beta-Ketothiolase Deficiency	14	♂ Genotyping | c.1006-1G>C, c.1006-2A>C, c.1083insA, c.826+1G>T, c.A278G (p.N93S), c.C433G (p.Q145E), c.C814T (p.Q272X), c.G1136T (p.G379V), c.G1138A (p.A380T), c.G547A (p.G183R), c.G997C (p.A333P), c.T2A (p.M1K), c.T935C (p.I312T), c.T99A (p.Y33X)
●	●	○	○	Biotinidase Deficiency	10	♂ Genotyping | c.98_104delGCGGCTGinsTCC (p.C33FfsX68), c.A1368C (p.Q456H), c.A755G (p.D252G), c.C1612T (p.R538C), c.C235T (p.R79C), c.G100A (p.G34S), c.G1330C (p.D444H), c.G511A (p.A171T), c.T1207G (p.F403V), c.A1466C (p.N489T)
●	○	○	○	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)

H	T	X	M	Disease	#	Mutations
●	○	○	○	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)
●	●	○	○	Carnitine Palmitoyltransferase IA Deficiency	7	♂ Genotyping | c.A1079G (p.E360G), c.A1361G (p.D454G), c.C1241T (p.A414V), c.C1436T (p.P479L), c.G2126A (p.G709E), c.G2129A (p.G710E), c.A1493G (p.Y498C)
●	●	○	○	Carnitine Palmitoyltransferase II Deficiency	21	♂ Genotyping | c.109_110insGC, c.1238_1239delAG, c.1737delC, c.1923_1935delGAAGGCCTTAGAA, c.A1649G (p.Q550R), c.A1883C (p.Y628S), c.A359G (p.Y120C), c.A983G (p.D328G), c.C149A (p.P50H), c.C1507T (p.R503C), c.C1810T (p.P604S), c.C1891T (p.R631C), c.C338T (p.S113L), c.C370T (p.R124X), c.C680T (p.P227L), c.G1145A (p.R382K), c.G1646A (p.G549D), c.G452A (p.R151Q), c.G520A (p.E174K), c.T1148A (p.F383Y), c.T1342C (p.F448L)
●	○	○	○	Carpenter Syndrome	2	♂ Genotyping | c.T434A (p.L145X), c.408_409insT (p.136fsX)
●	○	○	○	Cartilage-Hair Hypoplasia	2	♂ Genotyping | c.A71G, c.-624C>A
●	○	○	○	Cerebrotendinous Xanthomatosis	13	♂ Genotyping | c.1263+1G>A, c.844+1G>A, c.C1016T (p.T339M), c.C1183T (p.R395C), c.C1420T (p.R474W), c.C1435T (p.R479C), c.C379T (p.R127W), c.819delT, c.G1214A (p.R405Q), c.G1421A (p.R474Q), c.G434A (p.G145E), c.G583T (p.E195X), c.G646C (p.A216P)
●	○	○	○	Choreoacanthocytosis	1	♂ Genotyping | c.6058delC (p.P2020fs)
●	●	○	○	Citrullinemia: Type I	9	♂ Genotyping | c.1194-1G>C, c.A928C (p.K310Q), c.C835T (p.R279X), c.G1085T (p.G362V), c.G470A (p.R157H), c.G539A (p.S180N), c.G970A (p.G324S), c.T535C (p.W179R), c.1168G>A (p.G390R)
●	●	○	○	Classical Galactosemia	17	♂ Genotyping | c.253-2A>G, c.A563G (p.Q188R), c.A626G (p.Y209C), c.C404T (p.S135L), c.C413T (p.T138M), c.C505A (p.Q169K), c.C997G (p.R333G), c.G607A (p.E203K), c.G855T (p.K285N), c.T1138C (p.X380R), c.T221C (p.L74P), c.T425A (p.M142K), c.T512C (p.F171S), c.T584C (p.L195P), c.134_138delCAGCT, c.-1039_753del3162, c.820+51_*789del2294ins12
●	○	○	○	Cohen Syndrome	8	♂ Genotyping | c.T6578G (p.L2193R), c.C7051T (p.R2351X), c.G4471T (p.E1491X), c.C2911T (p.R971X), c.G7934A (p.G2645D), c.C10888T (p.Q3630X), c.9259_9260insT (p.L3087fs), c.3348_3349delCT (p.C1117fx)
●	●	○	○	Combined Pituitary Hormone Deficiency: PROP1 Related	11	♂ Genotyping | c.G218A (p.R73H), c.150delA (p.G50fsX), c.C358T (p.R120C), c.112_124delTCGAGTGCTCCAC (p.S38fsX), c.T2C (p.M1T), c.157delA (p.R53fsX), c.G212A (p.R71H), c.C217T (p.R73C), c.G582A (p.W194X), c.109+1G>T, c.301delAG (p.S101fsX)
●	○	○	○	Congenital Disorder of Glycosylation: Type 1A: PMM2 Related	5	♂ Genotyping | c.C357A (p.F119L), c.G422A (p.R141H), c.C338T (p.P113L), c.G691A (p.V231M), c.T470C (p.F157S)
●	●	○	○	Congenital Disorder of Glycosylation: Type 1B: MPI Related	1	♂ Genotyping | c.G884A (p.R295H)
●	○	○	○	Congenital Disorder of Glycosylation: Type 1C: ALG6 Related	4	♂ Genotyping | c.257+5G>A, c.895_897delATA, c.C998T (p.A333V), c.T1432C (p.S478P)
●	○	○	○	Congenital Lipoid Adrenal Hyperplasia	10	♂ Genotyping | c.201_202delCT, c.466-11T>A, c.64+1G>T, c.C562T (p.R188C), c.C772T (p.Q258X), c.G545A (p.R182H), c.G545T (p.R182L), c.G559A (p.V187M), c.G650C (p.R217T), c.G749A (p.W250X)
●	○	○	○	Congenital Neutropenia: Recessive	5	♂ Genotyping | c.121_125insG, c.130_131insA, c.91delG, c.C256T (p.R86X), c.C568T (p.Q190X)
●	○	○	○	Corneal Dystrophy and Perceptive Deafness	8	♂ Genotyping | c.1378delTACGinsA, c.2233_2240insTATGACAC, c.473delGCTTCGCC, c.A2566G (p.M856V), c.G1463A (p.R488K), c.T2528C (p.L843P), c.T637C (p.S213P), c.2321+1G>A

H	T	X	M	Disease	#	Mutations
●	●	○	○	Corticosterone Methyloxidase Deficiency	3	♂ Genotyping | c.A1492G (p.T498A), c.C541T (p.R181W), c.T1382C (p.L461P)
●	○	○	○	Crigler-Najjar Syndrome	11	♂ Genotyping | c.508_513delTTC (p.170delF), c.A1070G (p.Q357R), c.C1021T (p.R341X), c.C1124T (p.S375F), c.C840A (p.C280X), c.C991T (p.Q331X), c.G923A (p.G308E), c.A1198G (p.N400D), c.A992G (p.Q331R), c.T44G (p.L15R), c.T524A (p.L175Q)
●	●	○	○	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)
●	●	○	○	Cystinosis	12	♂ Genotyping | c.18_21delGACT, c.198_218delTATTACTATCCTTGAGCTCCC , c.G283T (p.G95X), c.G414A (p.W138X), c.G506A (p.G169D), c.G613A (p.D205N), c.T473C (p.L158P), c.G329T (p.G110V), c.C416T (p.S139F), c.G589A (p.G197R), c.C969G (p.N323K), c.G1015A (p.G339R)
●	●	○	○	Cystinuria: Non-Type I	15	♂ Genotyping | c.G508A (p.V170M), c.G313A (p.G105R), c.G583A (p.G195R), c.G775A (p.G259R), c.C997T (p.R333W), c.T131C (p.I44T), c.C782T (p.P261L), c.A695G (p.Y232C), c.G544A (p.A182T), c.C368T (p.T123M), c.520insT (p.F112fs), c.614_615insA (p.K205fs), c.789+2T>C, c.605-3C>A (IVS5-3C>A), c.C1445T (p.P482L)
●	●	○	○	Cystinuria: Type I	10	♂ Genotyping | c.T1400C (p.M467T), c.T2033C (p.L678P), c.G542A (p.R181Q), c.C1955G (p.T652R), c.C1843A (p.P615T), c.G1085A (p.R362H), c.T1597A (p.Y533N), c.C647T (p.T216M), c.C808T (p.R270X), c.A452G (p.Y151C)
●	○	○	○	D-Bifunctional Protein Deficiency	6	♂ Genotyping | c.G46A (p.G16S), c.63G>T (p.L21F), c.422_423delAG, c.652G>T (p.V218L), c.1369A>T (p.N457Y), c.1369A>G (p.N457D)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Diabetes: Recessive Permanent Neonatal	2	♂ Genotyping | c.A215G (p.N72S), c.G1144A (p.E382K)
●	○	○	○	Du Pan Syndrome	5	♂ Genotyping | c.1309delTTG, c.C1306A (p.P436T), c.G1133A (p.R378Q), c.T1315A (p.S439T), c.T1322C (p.L441P)
●	●	○	○	Dyskeratosis Congenita: RTEL1 Related	5	♂ Genotyping | c.C2869T (p.R981W), c.C2920T (p.R998X), c.G1548T (p.M516I), c.G2216T (p.G763V), c.G3791A (p.R1264H)
●	○	○	○	Dystrophic Epidermolysis Bullosa: Recessive	10	♂ Genotyping | c.2470_2471insG, c.C933A (p.Y311X), c.G4039C (p.G1347R), c.T8393A (p.M2798K), c.A425G (p.K142R), ,C.8441-14_8435delGCTCTTGGCTCCAGGACCCCT, c.4783-1G>A, c.G7344A (p.V2448X), c.G4991C (p.G1664A), c.497_498insA (p.V168GfsX179)
●	○	○	○	Ehlers-Danlos Syndrome: Type VIIC	2	♂ Genotyping | c.C673T (p.Q225X), c.G2384A (p.W795X)
●	○	○	○	Ellis-van Creveld Syndrome: EVC Related	8	♂ Genotyping | c.T919C (p.S307P), c.1694delC (p.A565VfsX23), c.734delT (p.L245fs), c.910-911insA (p.R304fs), c.C2635T (p.Q879X), c.1886+5G>T, c.1098+1G>A, c.C1018T (p.R340X)
●	○	○	○	Ellis-van Creveld Syndrome: EVC2 Related	1	♂ Genotyping | c.C3025T (p.Q1009X)
●	○	○	○	Enhanced S-Cone	1	♂ Genotyping | c.G932A (p.R311Q)
●	●	○	○	Ethylmalonic Aciduria	3	♂ Genotyping | c.505+1G>T, c.C487T (p.R163W), c.G3T (p.M1I)
●	○	○	○	Familial Chloride Diarrhea	6	♂ Genotyping | c.344delT (p.I115I), c.G559T (p.G187X), c.951delGGT (p.V318del), c.G1386A (p.W462X), c.A371T (p.H124L), c.2023_2025dupATC (p.I675L)
●	○	○	○	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
●	○	○	○	Familial Hyperinsulinism: Type 2: KCNJ11 Related	6	♂ Genotyping | c.A776G (p.H259R), c.C36A (p.Y12X), C.C761T (p.P254L), c.G-134T, c.G844A (p.E282K), c.T440C (p.L147P)
●	●	○	○	Familial Mediterranean Fever	12	♂ Genotyping | c.2076_2078delAAT (p.692delI), c.A2080G (p.M694V), c.A2084G (p.K695R), c.C1437G (p.F479L), c.C800T (p.T267I), c.G1958A (p.R653H), c.G2040A (p.M680I), c.G2040C (p.M680I), c.G2082A (p.M694I), c.G2230T (p.A744S), c.G2282A (p.R761H), c.T2177C (p.V726A)
●	●	○	○	Fanconi Anemia: Type A	1	♂ Genotyping | c.C295T (p.Q99X)
●	●	○	○	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)
●	●	○	○	Fanconi Anemia: Type G	5	♂ Genotyping | c.1480+1G>C, c.307+1G>C, c.1794_1803delCTGGATCCGT (p.W599Pfs), c.637_643delTACCGCC (p.Y213K+4X), c.925-2A>G
●	●	○	○	Fanconi Anemia: Type J	1	♂ Genotyping | c.2392C>T (p.R798X)
●	○	○	○	Fumarase Deficiency	1	♂ Genotyping | c.1431_1433insAAA
●	○	○	○	GM1-Gangliosidoses	16	♂ Genotyping | c.1480-2A>G, c.75+2_75+3insT, c.A1772G (p.Y591C), c.A947G (p.Y316C), c.C1051T (p.R351X), c.C1369T (p.R457X), c.C145T (p.R49C), c.C202T (p.R68W), c.C245T (p.T82M), c.C601T (p.R201C), c.C622T (p.R208C), c.G1370A (p.R457Q), c.G176A (p.R59H), c.G367A (p.G123R), c.T152C (p.I51T), c.T1771A (p.Y591N)

H	T	X	M	Disease	#	Mutations
●	○	○	○	GRACILE Syndrome	11	♂ Genotyping | c.A232G (p.S78G), c.G103C (p.G35R), c.A148G (p.T50A), c.C166T (p.R56X), c.C296T (p.P99L), c.G464C (p.R155P), c.C547T (p.R183C), c.G548A (p.R183H), c.C550T (p.R184C), c.G830A (p.S277N), c.G1057A (p.V353M)
○	●	○	●	Galactokinase Deficiency	7	♂ Genotyping | c.1144C>T (p.Q382X), c.1045G>A (p.G349S), c.1031C>T (p.T344M), c.238G>T (p.E80X), c.94G>A (p.V32M), c.82C>A (p.P28T), c.C593T (p.A198V)
●	●	○	○	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)
○	●	○	●	Gitelman Syndrome	7	♂ Genotyping | c.1926-1G>T, c.C1043T (p.P348L), c.C1760T (p.A587V), c.C622T (p.R208W), c.G1886T (p.G629V), c.T1258C (p.C420R), c.T1865C (p.L622P)
●	○	○	○	Globoid Cell Leukodystrophy	10	♂ Genotyping | c.G1153T (p.E385X), c.G857A (p.G286D), c.A2002C (p.T668P), c.A1700C (p.Y567S), c.C1586T (p.T529M), c.1472delA (p.K491fs), c.A913G (p.I305V), c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP), c.A246G (p.I82M), c.1161+6555_*9573del31670bp
●	●	○	○	Glutaric Acidemia: Type I	8	♂ Genotyping | c.C1204T (p.R402W), c.C1262T (p.A421V), c.C743T (p.P248L), c.G1093A (p.E365K), c.G877A (p.A293T), c.1083-2A>C (IVS10-2A>C), c.G680C (p.R227P), c.G1198A (p.V400M)
●	○	○	○	Glycine Encephalopathy: AMT Related	6	♂ Genotyping | c.959G>A (p.R320H), c.878-1G>A, c.826G>C (p.D276H), c.574C>T (p.Q192X), c.139G>A (p.G47R), c.125A>G (p.H42R)
●	○	○	○	Glycine Encephalopathy: GLDC Related	5	♂ Genotyping | c.2284G>A (p.G762R), c.2266_2268delTTC (p.756delF), c.1691G>T (p.S564I), c.1545G>C (p.R515S), c.2T>C (p.M1T)
●	●	○	○	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
●	●	○	○	Glycogen Storage Disease: Type IB	5	♂ Genotyping | c.1042_1043delCT, c.G1015T (p.G339C), c.G1016A (p.G339D), c.G1099A (p.A367T), c.T352C (p.W118R)
●	●	○	○	Glycogen Storage Disease: Type II	12	♂ Genotyping | c.C1935A (p.D645E), c.C2560T (p.R854X), c.-32-13T>G, c.525delT (p.E176Rfs), c.C710T (p.A237V), c.T896G (p.L299R), c.T953C (p.M318T), c.G1561A (p.E521K), c.C1634T (p.P545L), c.G1927A (p.G643R), c.C2173T (p.R725W), c.2707_2709delK (p.903delK)
●	●	○	○	Glycogen Storage Disease: Type III	14	♂ Genotyping | c.17_18delAG, c.4455delT, c.C1222T (p.R408X), c.C16T (p.Q6X), c.1384delG (p.V462X), c.G2039A (p.W680X), c.C2590T (p.R864X), c.2681+1G>A, c.A3439G (p.R1147G), c.C3682T (p.R1228X), c.3965delT (p.V1322AfsX27), c.G3980A (p.W1327X), c.4260-12A>G, c.G4342C (p.G1448R)
●	○	○	○	Glycogen Storage Disease: Type IV	2	♂ Genotyping | c.A986C (p.Y329S), c.986A>G
○	○	○	●	Glycogen Storage Disease: Type V	8	♂ Genotyping | c.2128_2130delTTC (p.710delF), c.A1627T (p.K543X), c.A1628C (p.K543T), c.C148T (p.R50X), c.C255A (p.Y85X), c.G613A (p.G205S), c.T2392C (p.W798R), c.G1827A (p.K609K)
○	○	○	●	Glycogen Storage Disease: Type VII	2	♂ Genotyping | c.G116T (p.R39L), c.C283T (p.R95X)
●	●	○	○	Guanidinoacetate Methyltransferase Deficiency	4	♂ Genotyping | c.506G>A (p.C169Y), c.327G>A, c.309_310insCCGGGACTGGGCC (p.L99_A103fs), c.148A>C (p.M50L)
●	●	○	○	HMG-CoA Lyase Deficiency	6	♂ Genotyping | c.914_915delTT, c.G122A (p.R41Q), c.G208C (p.V70L), c.G835A (p.E279K), c.561+1G>A, c.109G>T (p.E37X)
●	●	○	○	Hemochromatosis: Type 2A: HFE2 Related	1	♂ Genotyping | c.G959T (p.G320V)

H	T	X	M	Disease	#	Mutations
○	●	○	●	Hemochromatosis: Type 3: TFR2 Related	4	♂ Genotyping | c.A2069C (p.Q690P), c.C750G (p.Y250X), c.T515A (p.M172K), c.88_89insC (p.E60X)
○	●	○	●	Hemoglobinopathy: Hb C	1	♂ Genotyping | c.G19A (p.E7K)
○	●	○	●	Hemoglobinopathy: Hb D	1	♂ Genotyping | c.G364C (p.E122Q)
○	●	○	●	Hemoglobinopathy: Hb E	1	♂ Genotyping | c.G79A (p.E27K)
○	●	○	●	Hemoglobinopathy: Hb O	1	♂ Genotyping | c.G364A (p.E122K)
○	●	○	●	Hereditary Fructose Intolerance	10	♂ Genotyping | c.357_360delAAAC, c.C1005G (p.N335K), c.C524A (p.A175D), c.G448C (p.A150P), c.T612G (p.Y204X), c.865_867delCTT (p.289delL), c.C720A (p.C240X), c.T442C (p.W148R), c.C178T (p.R60X), c.C10T (p.R4X)
●	○	○	○	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related	6	♂ Genotyping | c.3024delT, c.C124T (p.R42X), c.C1903T (p.R635X), c.C430T (p.R144X), c.C727T (p.Q243X), c.C3247T (p.Q1083X)
●	○	○	○	Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related	1	♂ Genotyping | c.C283T (p.R95X)
●	○	○	○	Hermansky-Pudlak Syndrome: Type 1	1	♂ Genotyping | c.1470_1486dup16 (p.H497Qfs)
●	○	○	○	Hermansky-Pudlak Syndrome: Type 3	5	♂ Genotyping | c.C1189T (p.R397W), c.1691+2T>G, c.2589+1G>C, c.2482-2A>G, c.1163+1G>A
●	●	○	○	Holocarboxylase Synthetase Deficiency	7	♂ Genotyping | c.1795+5G>A, c.780delG, c.T710C (p.L237P), c.C1522T (p.R508W), c.G1648A (p.V550M), c.G1513C (p.G505R), c.772_781delACAAGCAAGG (p.T258fs)
●	●	○	○	Homocystinuria Caused by CBS Deficiency	7	♂ Genotyping | c.G919A (p.G307S), c.T833C (p.I278T), c.C1006T (p.R336C), c.T959C (p.V320A), c.G797A (p.R266K), c.572C>T (p.T191M), c.C341T (p.A114V)
●	●	○	○	Hurler Syndrome	8	♂ Genotyping | c.C1598G (p.P533R), c.C208T (p.Q70X), c.G1205A (p.W402X), c.G979C (p.A327P), c.G266A (p.R89Q), c.T1960G (p.X654G), c.G152A (p.G51D), c.T1037G (p.L346R)
●	○	○	○	Hypophosphatasia	5	♂ Genotyping | c.1559delT, c.A1133T (p.D378V), c.G1001A (p.G334D), c.G571A (p.E191K), c.T979C (p.F327L)
●	○	○	○	Inclusion Body Myopathy: Type 2	3	♂ Genotyping | c.T2228C (p.M743T), c.G1807C (p.V603L), c.G131C (p.C44S)
●	●	○	○	Isovaleric Acidemia	1	♂ Genotyping | c.C941T (p.A314V)
●	○	○	○	Joubert Syndrome	1	♂ Genotyping | c.G35T (p.R12L)
●	●	○	○	Lamellar Ichthyosis: Type 1	1	♂ Genotyping | c.877-2A>G (IVS5-2A>G)
●	○	○	○	Laryngoonychocutaneous Syndrome	1	♂ Genotyping | c.151_152insG (p.V51GfsX3)
●	○	○	○	Leber Amaurosis: CEP290 Related	1	♂ Genotyping | c.2991+1655A>G (p.C998X)
●	○	○	○	Leber Amaurosis: GUCY2D Related	2	♂ Genotyping | c.T1694C (p.F565S), c.2943delG (p.G982V)
●	○	○	○	Leber Amaurosis: LCA5 Related	3	♂ Genotyping | c.C835T (p.Q279X), c.1476_1477insA (p.P493TfsX1), c.1151delC
●	○	○	○	Leber Amaurosis: RDH12 Related	6	♂ Genotyping | c.C565T (p.Q189X), c.C184T (p.R62X), c.C464T (p.T155I), c.A677G (p.Y226C), c.C146T (p.T49M), c.C295A (p.L99I)
●	○	○	○	Leigh Syndrome: French-Canadian	1	♂ Genotyping | c.C1061T (p.A354V)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Leydig Cell Hypoplasia (Luteinizing Hormone Resistance)	13	♂ Genotyping | c.1822_1827delCTGGTT (p.608_609delLV), c.G1777C (p.A593P), c.C1660T (p.R554X), c.G1060A (p.E354K), c.C1635A (p.C545X), c.T391C (p.C131R), c.T1027A (p.C343S), c.T1627C (p.C543R), c.T1505C (p.L502P), c.G430T (p.V144F), c.C1847A (p.S616Y), c.T455C (p.I152T), c.537-3C>A
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2A	6	♂ Genotyping | c.G1715A (p.R572Q), c.G1469A (p.R490Q), c.550delA (p.T184fs), c.G2306A (p.R769Q), c.2362_2363delAGinsTCATCT,c.2362AG>TCATCT (p.R788Sfs), c.G1525T (p.V509F)
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2C	4	♂ Genotyping | c.G848A (p.C283Y), c.G787A (p.E263K), c.525delT (p.F175fsX), c.87dupT (p.Y29fsX)
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2D	1	♂ Genotyping | c.C229T (p.R77C)
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2E	6	♂ Genotyping | c.C341T (p.S114F), c.C452G (p.T151R), c.G272C (p.R91P), c.G272T (p.R91L), c.T299A (p.M100K), c.T323G (p.L108R)
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2F	5	♂ Genotyping | c.C493T (p.R165X), c.G89A (p.W30X), c.G784A (p.E262K), c.G391C (p.A131P), c.653delC (p.A218fs)
●	○	○	○	Limb-Girdle Muscular Dystrophy: Type 2I	1	♂ Genotyping | c.C826A (p.L276I)
○	●	○	●	Lipoprotein Lipase Deficiency	1	♂ Genotyping | c.G644A (p.G215E)
●	●	○	○	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	2	♂ Genotyping | c.C1132T (p.Q378X), c.G1528C (p.E510Q)
●	●	○	○	Lysinuric Protein Intolerance	4	♂ Genotyping | c.C1228T (p.R410X), c.G726A (p.W242X), c.1384_1385insATCA (p.R462fs), c.895-2A>T
●	●	○	○	MTHFR Deficiency: Severe	6	♂ Genotyping | c.T1721G (p.V574G), c.G1408T (p.E470X), c.1166G>A (p.W389X), c.652G> T (p.V218L), c.523G>A (p.A175T), c.474A>T (p.G158G)
●	●	○	○	Maple Syrup Urine Disease: Type 1A	3	♂ Genotyping | c.860_867delGAGGCCCC, c.T1312A (p.Y438N), c.288+1G>A
●	●	○	○	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)
●	●	○	○	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)
●	○	○	○	Meckel Syndrome: Type 1	5	♂ Genotyping | c.1408-35_1408-7del29 (p.G470fs), c.80+2T>C (IVS1+2T>C), c.1024+1G>A (IVS11+1G>A), c.G417A (p.E139X), c.50insCCGGG (p.D19AfsX)
●	●	○	○	Medium Chain Acyl-CoA Dehydrogenase Deficiency	8	♂ Genotyping | c.A985G (p.K329E), c.C362T (p.T121I), c.G583A (p.G195R), c.G799A (p.G267R), c.T199C (p.Y67H), c.C250T (p.L84F), c.C616T (p.R206C), c.G617A (p.C206H)
●	○	○	○	Megalencephalic Leukoencephalopathy	5	♂ Genotyping | c.G176A (p.G59E), c.C278T (p.S93L), c.135_136insC (p.P45fsX), c.908_918delTGCTGCTGCTGinsGCA (p.Val303GlyfsX96), c.C880T (p.P294S)
●	○	○	○	Metachromatic Leukodystrophy	16	♂ Genotyping | c.1204+1G>A, c.459+1G>A, c.A862C (p.T288P), c.C1136T (p.P379L), c.C1283T (p.P428L), c.C827T (p.T276M), c.T542G (p.I181S), c.C1232T (p.T411I), c.G769C (p.D257H), c.G739A (p.G247R), c.C641T (p.A214V), c.G302A (p.G101D), c.C293T (p.S98F), c.G257A (p.R86Q), c.G263A (p.G86D), c.C1114T (p.R372W)
●	●	○	○	Methylmalonic Acidemia: MMAA Related	14	♂ Genotyping | c.64C>T (p.R22X), c.161G>A (p.W54X), c.266T>C (p.L89P), c.283C>T (p.Q95X), c.358C>T (p.Q120X), c.397C>T (p.Q133X), c.433C>T (p.R145X), c.503delC (p.T168MfsX9), c.562G>C (p.G188R), c.650T>A (p.L217X), c.653G>A (p.G218E), c.733+1G>A, c.988C>T (p.R330X), c.1076G>A (p.R359Q)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Methylmalonic Acidemia: MMAB Related	11	♂ Genotyping | c.700C>T (p.Q234X), c.656A>G (p.Y219C), c.572G>A (p.R191Q), c.571C>T (p.R191W), c.569G>A (p.R190H), c.568C>T (p.R190C), c.556C>T (p.R186W), c.403G>A (p.A135T), c.291-1G>A, c.287T>C (p.I96T), c.197-1G>T
●	●	○	○	Methylmalonic Acidemia: MUT Related	22	♂ Genotyping | c.2150G>T (p.G717V), c.2099T>A (p.M700K), c.2080C>T (p.R694W), c.2054T>G (p.L685R), c.1867G>A (p.G623R), c.1280G>A (p.G427D), c.1106G>A (p.R369H), c.1105C>T (p.R369C), c.1097A>G (p.N366S), c.935G>T (p.G312V), c.691T>A (p.Y231N), c.655A>T (p.N219Y), c.643G>A (p.G215S), c.607G>A (p.G203R), c.572C>A (p.A191E), c.521T>C (p.F174S), c.322C>T (p.R108C), c.313T>C (p.W105R), c.299A>G (p.Y100C), c.284C>G (p.P95R), c.281G>T (p.G94V), c.278G>A (p.R93H)
●	●	○	○	Methylmalonic Aciduria and Homocystinuria: Type cblC	5	♂ Genotyping | c.271_273insA (p.R91KfsX14), c.331C>T (p.R111X), c.394C>T (p.R132X), c.482G>A (p.R161Q), c.609G>A (p.W203X)
●	○	○	○	Mucolipidosis: Type II	3	♂ Genotyping | c.3503_3504delTC (p.L1168QfsX5), c.C3565T (p.R1189X), c.T1120C (p.F374L)
●	○	○	○	Mucolipidosis: Type IV	4	♂ Genotyping | c.406-2A>G, c.G1084T (p.D362Y), c.C304T (p.R102X), c.244delC (p.L82fsX)
●	○	○	○	Multiple Pterygium Syndrome	6	♂ Genotyping | c.C715T (p.R239C), c.C13T (p.Q5X), c.T320G (p.V107G), c.401_402delCT (p.P134fs), c.C1408T (p.R470X), c.C136T (p.R46X)
●	○	○	○	Multiple Sulfatase Deficiency	1	♂ Genotyping | c.T463C (p.S155P)
●	○	○	○	Muscle-Eye-Brain Disease	3	♂ Genotyping | c.1539+1G>A , c.C1324T (p.R442C), c.C1478G (p.P493R)
●	○	○	○	Navajo Neurohepatopathy	1	♂ Genotyping | c.G149A (p.R50Q)
●	○	○	○	Nemaline Myopathy: NEB Related	1	♂ Genotyping | c.7434_7536del2502bp
●	○	○	○	Nephrotic Syndrome: Type 1	5	♂ Genotyping | c.121_122delCT (p.L41Dfs), c.1481delC, c.C3325T (p.R1109X), c.C3478T (p.R1160X), c.2335-1G>A
●	○	○	○	Nephrotic Syndrome: Type 2	25	♂ Genotyping | c.976_977insA (p.T326fsX345), c.964C>T (p.R322X), c.948delT (p.A317L), c.871C>T (p.R291W), c.868G>A (p.V290M), c.862G>A (p.A288T), c.855_856delAA (p.Q285fsX302), c.851C>T (p.A284V), c.779T>A (p.V260E), c.714G>T (p.R238S), c.622G>A (p.A208T), c.555delT (p.F185fsX186), c.538G>A (p.V180M), c.503G>A (p.R168H), c.502C>A (p.R168S), c.502C>T (p.R168C), c.479A>G (p.D160G), c.467delT (p.L156fsX180), c.467_468insT (p.L156fsX166), c.419delG (p.G140fsX180), c.413G>A (p.R138Q), c.412C>T (p.R138X), c.353C>T (p.P118L), c.274G>T (p.G92C), c.85G>A (p.A29T)
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: CLN5 Related	7	♂ Genotyping | c.1175_1176delAT (p.Y392X), c.G225A (p.W75X), c.G835A (p.D279N), c.G335A (p.R112H), c.G377A (p.C126Y), c.G1054T (p.E352X), c.A1121G (p.Y374C)
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: CLN6 Related	9	♂ Genotyping | c.C663G (p.Y221X), c.511_513delTAT (p.171delY), c.460_462delATC (p.I154del), c.G368A (p.G123D), c.G308A (p.R103Q), c.214G>T (p.E72X), c.T200C (p.L67P), c.C139T (p.L47F), c.G17C (p.R6T)
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: CLN8 Related	4	♂ Genotyping | c.C70G (p.R24G), c.G789C (p.W263C), c.G88C (p.A30P), c.C610T (p.R204C)
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: MFSD8 Related	2	♂ Genotyping | c.881C>A (p.T294K), c.754+2T>A
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: PPT1 Related	8	♂ Genotyping | c.A223C (p.T75P), c.A364T (p.R122W), c.C451T (p.R151X), c.T29A (p.L10X), c.T656A (p.L219Q), c.G322C (p.G108R), c.A236G (p.D79G), c.G134A (p.C45Y)
●	○	○	○	Neuronal Ceroid-Lipofuscinosis: TPP1 Related	9	♂ Genotyping | c.523-1G>A, c.509-1G>C, c.C622T (p.R208X), c.G851T (p.G284V), c.G1340A (p.R477H), c.G1094A (p.C365Y), c.T1093C (p.C365R), c.A857G (p.N286S), c.C616T (p.R206C)

H	T	X	M	Disease	#	Mutations
●	○	○	○	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)
●	○	○	○	Niemann-Pick Disease: Type B	3	♂ Genotyping | c.1828_1830delCGC (p.610delR), c.C880A (p.Q294K), c.A1280G (p.H427R)
●	○	○	○	Niemann-Pick Disease: Type C1	14	♂ Genotyping | c.A2783C (p.Q928P), c.A3263G (p.Y1088C), c.A3467G (p.N1156S), c.C3107T (p.T1036M), c.T3182C (p.I1061T), c.G2974C (p.G992R), c.C2932T (p.R978C), c.G2848A (p.V950M), c.G2665A (p.V889M), c.A2324C (p.Q775P), p.T1133C (p.V378A), c.G530A (c.C117Y), c.T337C (p.C113R), c.G2974T (p.G992W)
●	○	○	○	Niemann-Pick Disease: Type C2	11	♂ Genotyping | c.G58T (p.E20X), c.C436T (p.Q146X), c.C358T (p.P120S), c.G352T (p.E118X), c.332delA (p.N111Ifs), c.T295C (p.C99R), c.T199C (p.S67P), c.190+5G>A, c.C141A (p.C47X), c.C133T (p.Q45X), c.G115A (p.V39M)
●	○	○	○	Nonsyndromic Hearing Loss and Deafness: DFNB3 Related	9	♂ Genotyping | c.453_455delCGAinsTGGACGCCTGGTCGGGCAGTGG (p.E152GfsX81), c.A7801T (p.K2601X), c.A6337T (p.I2113F), c.3866+1G>T, c.G3313T (p.E1105X), c.3334delG (p.G1112fs), c.G8148T (p.Q2716H), c.A6331T (p.N2111Y), c.C3685T (p.Q1229X)
●	●	○	○	Nonsyndromic Hearing Loss and Deafness: GJB2 Related	15	♂ Genotyping | c.167delT, c.235delC, c.312_325delGAAGTTCATCAAGG, c.358delGAG (p.120delE), c.35delG, c.C370T (p.Q124X), c.C427T (p.R143W), c.G109A (p.V37I), c.G231A (p.W77X), c.G551C (p.R184P), c.G71A (p.W24X), c.T101C (p.M34T), c.T229C (p.W77R), c.T269C (p.L90P), c.35G>T
●	○	○	○	Oculocutaneous Albinism: Type 1	9	♂ Genotyping | c.G272A (p.C91Y), c.C242T (p.P81L), c.T265C (p.C89R), c.A1G (p.M1V), c.G140A (p.G47D), c.G325A (p.G109R), c.568delG (p.G191Dfs), c.G707A (p.W236X), c.C832T (p.R278X)
●	○	○	○	Oculocutaneous Albinism: Type 4	2	♂ Genotyping | c.G469A (p.D157N), c.G563T (p.G188V)
●	●	○	○	Omenn Syndrome	1	♂ Genotyping | c.C597A (p.Y199X)
●	●	○	○	Ornithine Translocase Deficiency	3	♂ Genotyping | c.562_564delTTC (p.188delF), c.C95G (p.T32R), c.C535T (p.R179X)
●	○	○	○	POLG Related Disorders: Autosomal Recessive	16	♂ Genotyping | c.G695A (p.R232H), c.C752T (p.T251I), c.G1399A (p.A467T), c.C1760T (p.P587L), c.G2243C (p.W748S), c.G2542A (p.G848S), c.T3488G (p.M1163R), c.T911G (p.L304R), c.G8C (p.R3P), c.G2617T (p.E873X), c.C2794T (p.H932Y), c.G3151C (p.G1051R), c.A2591G (p.N864S), c.G1491C (p.Q497H), c.C679T (p.R227W), c.C3218T (p.P1073L)
●	●	○	○	Pendred Syndrome	7	♂ Genotyping | c.1001+1G>A, c.A1151G (p.E384G), c.A1246C (p.T416P), c.A2168G (p.H723R), c.T707C (p.L236P), c.T716A (p.V239D), c.919-2A>G
●	●	○	○	Persistent Mullerian Duct Syndrome: Type 1	5	♂ Genotyping | c.G1144T (p.E382X), c.C571T (p.R191X), c. C1518G (p.H506Q), c. G1574A (p.C525Y), c.C283T (p.R95X)
●	●	○	○	Persistent Mullerian Duct Syndrome: Type 2	14	♂ Genotyping | c.232+1G>A, c.1330_1356delCTGGGCAATACCCCTACCTCTGATGAG, c.596delA, c.G1217A (p.R406Q), c.G742A (p.E248K), c. A1277G (p.D426G), c. T846G (p.H282Q), c. T1373C (p.V458A), c. G1471C (p.D491H), c. C1510T (p.R504C), c.118G>T (p.G40X), c.289C>T (p.R97X), c.160C>T (p.R54C), c.425G>T (p.G142V)
●	●	○	○	Phenylalanine Hydroxylase Deficiency	17	♂ Genotyping | c.1066-11G>A, c.1315+1G>A, c.A1241G (p.Y414C), c.C1222T (p.R408W), c.C754T (p.R252W), c.G1223A (p.R408Q), c.G473A (p.R158Q), c.G782A (p.R261Q), c.G814T (p.G272X), c.T143C (p.L48S), c.T194C (p.I65T), c.T896G (p.F299C), c.C842T (p.P281L), c.G838A (p.E280K), c.C117G (p.F39L), c.G3A (p.M1I), c.A1G (p.M1V)
○	○	○	●	Polyglandular Autoimmune Syndrome: Type I	5	♂ Genotyping | c.C769T (p.R257X), c.A254G (p.Y85C), c.1163_1164insA (p.Met388IlefsX36), c.967_979delCTGTCCCCTCCGC (p.Leu323SerfsX51), c.C415T (p.R139X)

H	T	X	M	Disease	#	Mutations
●	●	○	○	Primary Carnitine Deficiency	12	♂ Genotyping | c.G506A (p.R169Q), c.G396A (p.W132X), c.C1195T (p.R399W), c.C1433T (p.P478L), c.G43T (p.G15W), c.1324_1325delGCinsAT (p.A442I), c.A632G (p.Y211C), c.1202_1203insA (p.Y401fsX), c.C844T (p.R282X), c.C505T (p.R169W), c.G1196A (p.R399Q), c.A95G (p.N32S)
●	●	○	○	Primary Hyperoxaluria: Type 1	11	♂ Genotyping | c.G508A (p.G170R), c.T454A (p.F152I), c.T731C (p.I244T), c.G121A (p.G41R), c.C198G (p.Y66X), c.G245A (p.G82E), c.G466A (p.G156R), c.T613C (p.S205P), c.C697T (p.R233C), c.G698A (p.R233H), c.G738A (p.W246X)
●	○	○	○	Primary Hyperoxaluria: Type 2	3	♂ Genotyping | c.103delG, c.404+3delAAGT, c.C295T (p.R99X)
●	●	○	○	Primary Hyperoxaluria: Type 3	2	♂ Genotyping | c.944_946delAGG (p.315delE), c.G860T (p.G287V)
●	○	○	○	Progressive Cerebello Cerebral Atrophy: Type 2	2	♂ Genotyping | c.A2084G (p.Q695R), c.1556+5G>A
●	○	○	○	Progressive Familial Intrahepatic Cholestasis: Type 2	5	♂ Genotyping | c.3767_3768insC, c.A890G (p.E297G), c.C1723T (p.R575X), c.C3169T (p.R1057X), c.G1295C (p.R432T)
●	●	○	○	Propionic Acidemia: PCCA Related	4	♂ Genotyping | c.862A>G (p.R288G), c.937C>T (p.R313X), c.1196G>A (p.R399Q), c.1685C>G (p.S562X)
●	●	○	○	Propionic Acidemia: PCCB Related	13	♂ Genotyping | c.280G>T (p.G94X), c.335G>A (p.G112D), c.457G>C (p.A153P), c.502G>A (p.E168K), c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA (p.G407fs), c.1228C>T (p.R410W), c.1283C>T (p.T428I), c.1304A>G (p.Y435C), c.1495C>T (p.R499X), c.1534C>T (p.R512C), c.1539_1540insCCC (p.R514PfsX38), c.1556T>C (p.L519P), c.1606A>G (p.N536D)
○	●	○	●	Pseudocholinesterase Deficiency	1	♂ Genotyping | c.A293G (p.D98G)
●	○	○	○	Pycnodysostosis	2	♂ Genotyping | c.A990G (p.X330W), c.T926C (p.L309P)
●	○	○	○	Pyruvate Dehydrogenase Deficiency: Autosomal Recessive	2	♂ Genotyping | c.A395G (p.Y132C), c.C1030T (p.P344S)
●	○	○	○	Retinal Dystrophies: RLBP1 Related	1	♂ Genotyping | c.C700T (p.R234W)
●	○	○	○	Retinitis Pigmentosa: Autosomal Recessive: DHDDS Related	1	♂ Genotyping | c.A124G (p.K42E)
●	○	○	○	Rhizomelic Chondrodysplasia Punctata: Type I	7	♂ Genotyping | c.903+1G>C, c.G649A (p.G217R), c.T875A (p.L292X), c.45_52insGGGACGCC (p.H18RfsX35), c.C120G (p.Y40X), c.T345G (p.Y115X), c.C653T (p.A218V)
●	○	○	○	Salla Disease	5	♂ Genotyping | c.802_816delTCATCATTAAGAAAT (p.Leu336fsX13), c.A406G (p.K136E), c.C115T (p.R39C), c.A548G (p.H183R), c.C1001G (p.P334R)
●	○	○	○	Sandhoff Disease	3	♂ Genotyping | c.76delA, c.445+1G>A, c.850C>T (p.R284X)
●	○	○	○	Sanfilippo Syndrome: Type A	11	♂ Genotyping | c.G734A (p.R245H), c.C220T (p.R74C), c.C197G (p.S66W), c.G449A (p.R150Q), c.G1339A (p.E447K), c.G1105A (p.E369K), c.G1298A (p.R433Q), c.C383T (p.P128L), c.G617C (p.R206P), c.T892C (p.S298P), c.1080delC (p.T360fsX)
●	○	○	○	Sanfilippo Syndrome: Type B	10	♂ Genotyping | c.G2021A (p.R674H), c.C889T (p.R297X), c.G1928A (p.R643H), c.C1927T (p.R643C), c.C1562T (p.P521L), c.C1444T (p.R482W), c.C1693T (p.R565W), c.G1694C (p.R565P), c.C700T (p.R234C), c.C1876T (p.R626X)
●	○	○	○	Sanfilippo Syndrome: Type C	13	♂ Genotyping | c.C848T (p.P311L), c.T962G (p.L321X), c.T1529A (p.M510K), c.C1030T (p.R344C), c.C1553T (p.S518F), c.C1150T (p.R384X), c.493+1G>A (IVS4+1G>A), c.372-2A>G (IVS3-2A>G), c.C1622T (p.S541L), c.852-1G>A, c.525_526insT (p.A175fsX), c.1345insG (p.D449fsX), c.234+1G>A (IVS2+1G>A)

H	T	X	M	Disease	#	Mutations
●	○	○	○	Sanfilippo Syndrome: Type D	5	♂ Genotyping | c.C1063T (p.R355X), c.C1168T (p.Q390X), c.1226insG (p.R409fsX), c.1138insGTCCT (p.D380fsX), c.1169delA (p.Q390fsX)
○	●	○	●	Short Chain Acyl-CoA Dehydrogenase Deficiency	5	♂ Genotyping | c.C1058T (p.S353L), c.C1138T (p.R380W), c.C1147T (p.R383C), c.C319T (p.R107C), c.C575T (p.A192V)
●	●	○	○	Sickle-Cell Anemia	1	♂ Genotyping | c.A20T (p.E7V)
●	○	○	○	Sjogren-Larsson Syndrome	2	♂ Genotyping | c.C943T (p.P315S), c.1297_1298delGA (p.E433fs)
●	○	○	○	Smith-Lemli-Opitz Syndrome	21	♂ Genotyping | c.964-1G>C, c.A356T (p.H119L), c.C1054T (p.R352W), c.C1210T (p.R404C), c.C278T (p.T93M), c.G1055A (p.R352Q), c.G1139A (p.C380Y), c.G1337A (p.R446Q), c.G452A (p.W151X), c.G453A (p.W151X), c.G744T (p.W248C), c.G976T (p.V326L), c.T326C (p.L109P), c.T470C (p.L157P), c.1342G>A (p.E448K), c.1228G>A (p.G410S), c.906C>G (p.F302L), c.725G>A (p.R242H), c.724C>T (p.R242C), c.506C>T (p.S169L), c.A1G (p.M1V)
●	○	○	○	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
●	○	○	○	Stargardt Disease	18	♂ Genotyping | c.C3083T (p.A1028V), c.C52T (p.R18W), c.C5338G (p.P1780A), c.G2791A (p.V931M), c.T1018G (p.Y340D), c.G1715A (p.R572Q), c.T2461A (p.W821R), c.G2565A (p.W855X), c.G3106A (p.E1036K), c.3210_3211insGT (p.S1071Vfs), c.C634T (p.R212C), c.C3113T (p.A1038V), c.T1622C (p.L541P), c.G3364A (p.E1122K), c.C6079T (p.L2027F), c.G2588C (p.G863A), c.1938-1G>A, c.571-2A>G
●	○	○	○	Stuve-Wiedemann Syndrome	8	♂ Genotyping | c.2472_2476delTATGT, c.2434C>T (p.R812X), c.2274_2275insT, c.1789C>T (pR597X), c.1620_1621insA, c.756_757insT (p.K253X), c.653_654insT, c.170delC
●	○	○	○	Sulfate Transporter-Related Osteochondrodysplasia	6	♂ Genotyping | c.1018_1020delGTT (p.340delV), c.C532T (p.R178X), c.C835T (p.R279W), c.T1957A (p.C653S), c.C398T (p.A133V), c.G764A (p.G255E)
●	○	○	○	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)
●	●	○	○	Tyrosine Hydroxylase Deficiency	1	♂ Genotyping | c.G698A (p.R233H)
●	●	○	○	Tyrosinemia: Type I	10	♂ Genotyping | c.1062+5>G>A, c.554-1G>T, c.607-6T>G, c.707-1G>C, c.C782T (p.P261L), c.G1069T (p.E357X), c.G786A (p.W262X), c.698A>T (p.D233V), c.G1009A (p.G337S), c.G192T (p.Q64H)
●	○	○	○	Usher Syndrome: Type 1B	12	♂ Genotyping | c.93C>A (p.C31X), c.C448T (p.R150X), c.634C>T (p.R212C), c.635G>A (p.R212H), c.700C>T (p.Q234X), c.905G>A (p.R302H), c.1797G>A (p.M599I), c.1996C>T (p.R666X), c.2476G>A (p.A826T), c.3719G>A (p.R1240Q), c.5581C>T (p.R1861X), c.6025delG (p.A2009fs)
●	○	○	○	Usher Syndrome: Type 1C	4	♂ Genotyping | c.IVS5+1G>A, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.36+1G>T,c.IVS1+1G>T

H	T	X	M	Disease	#	Mutations
●	○	○	○	Usher Syndrome: Type 1D	15	♂ Genotyping | c.172C>T (p.Q58X), c.3367C>T (p.Q1123X), c.3617C>G (p.P1206R), c.3713_3714delCT (p.S1238fs), c.3880C>T (p.Q1294X), c.4069C>T (p.Q1357X), c.4488G>C (p.Q1496H), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.7549A>G (p.S2517G), c.8230G>A (p.G2744S), c.8497C>G (p.R2833G), c.9524G>A (p.R3175H)
●	○	○	○	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 2A	21	♂ Genotyping | c.14020A>G (p.R4674G), c.12067-2A>G, c.4338_4339delCT (p.C1447fs), c.2299delG (p.E767SfsX21), c.2276G>T (p.C759F), c.2209C>T (p.R737X), c.1256G>T (p.C419F), c.1000C>T (p.R334W), c.923_924insGCCA (p.H308fs), c.T12708A (p.C4236X), c.C13576T (p.R4526X), c.1840+1G>A, c.T11328G (p.Y3776X), c.C5329T (p.R1777W), c.9165_9168delCTAT (p.I3055MfsX2), c.C9469T (p.Q3157X), c.C1876T (p.R626X), c.7123delG (p.G2375fs), c.A6235T (p.K2079X), c.C14403G (p.Y4801X), c.G3788A (p.W1263X)
●	○	○	○	Usher Syndrome: Type 3	4	♂ Genotyping | c.T144G (p.N48K), c.T359A (p.M120K), c.300T>G (p.Y176X), c.C634T (p.Q212X)
●	●	○	○	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	9	♂ Genotyping | c.779C>T (p.T260M), c.848T>C (p.V283A), c.1144A>C (p.K382Q), c.1226C>T (p.T409M), c.1322G>A (p.G441D), c.1372T>C (p.F458L), c.1405C>T (p.R469W), c.1837C>T (p.R613W), c.A739C (p.K247Q)
●	○	○	○	Walker-Warburg Syndrome	1	♂ Genotyping | c.1167insA (p.F390fs)
●	○	○	○	Werner Syndrome	6	♂ Genotyping | c.3139-1G>C (IVS25-1G>C), c.C3913T (p.R1305X), c.C3493T (p.Q1165X), c.A1730T (p.K577M), c.C1336T (p.R368X), c.2089-3024A>G
●	●	○	○	Wilson Disease	14	♂ Genotyping | c.1340delAAAC, c.2304delC (p.M769Cfs), c.C2332G (p.R778G), c.C3207A (p.H1069Q), c.G2333T (p.R778L), c.G2336A (p.W779X), c.G2337A (p.W779X), c.G2906A (p.R969Q), c.T1934G (p.M645R), c.T2123C (p.L708P), c.A3191C (p.E1064A), c.C3817T (p.P1273S), c.G3683C (p.R1228T), c.A3809G (p.N1270S)
●	○	○	○	Zellweger Spectrum Disorders: PEX1 Related	3	♂ Genotyping | c.G2528A (p.G843D), c.2916delA (p.G973fs), c.2097insT (p.I700fs)
●	○	○	○	Zellweger Spectrum Disorders: PEX10 Related	2	♂ Genotyping | c.764_765insA, c.874_875delCT
●	○	○	○	Zellweger Spectrum Disorders: PEX2 Related	1	♂ Genotyping | c.C355T (p.R119X)
●	○	○	○	Zellweger Spectrum Disorders: PEX6 Related	7	♂ Genotyping | c.1130+1G>A (IVS3+1G>A), c.1688+1G>A (IVS7+1G>A), c.1962-1G>A (p.L655fsX3), c.1301delC (p.S434Ffs), c.T1601C (p.L534P), c.511insT (p.G171Wfs), c.802_815delGACGGACTGGCGCT ([p.Val207_Gln294del, Val76_Gln294del])