

Name: Dana Modani
MRN: _____
Date of visit: 8/18/15

New Patient Intake Form

Answer all questions as they apply to you. This form will be added to your medical record.

Please select your physician.

- ☐ Dr. Gelber ☐ Dr. Grunebaum ☐ Dr. Kalish ☐ Dr. Landres
☒ Dr. Lee ☐ Dr. Pri-Paz ☐ Dr. Sharma ☐ Dr. Wasden

Who referred you? _____
Age: 35

Primary Care Provider: _____
Reason for visit: Pregnancy

Menstrual History

Date of last menstrual period: 6/20/15 Age(yrs) at 1st period: 15 yrs. Age at Menopause: N/A
My period usually occurs every 28-29 days and lasts for 5 days.

Please circle No or Yes

Heavy Periods: No ☒ Yes Painful Periods: ☒ No Yes Irregular Bleeding: ☒ No Yes PMS (bloating, moody): No ☒ Yes

Genetic History

Ethnic Background: Eastern European Partner's Age & Ethnic Background: 28 Indian

Has anyone in your family or your partner's family had any of the following? If so, please include which family member(s).

Autism <u>None</u>	Mental Retardation <u>None</u>
Birth Defect _____	Muscular Dystrophy _____
Congenital Heart Disease _____	Open Spine (Bifida) _____
Cystic Fibrosis _____	Sickle Cell Anemia _____
Down's syndrome _____	Thalassemia _____
Hemophilia _____	Unexplained Fetal Loss _____
Huntington's disease _____	Other _____

Gynecologic History

Last Pap smear: 4/9/15 Abnormal Pap Smears: No ☒ Yes (Year & Treatment given) 2000 LEEP
Last Mammogram: N/A Abnormal Mammograms: No ☐ Yes (Year & Treatment given) _____

Have you ever had any of the following infections? (Please check all that apply).

Gonorrhea _____ Chlamydia _____ Herpes _____ Trichomonas _____ Genital Warts/HPV ☒ Syphilis _____ None _____
If so, when and how was it treated? 2000 LEEP to remove cells

Have you had any of the following conditions? (Please check all that apply).

Uterine Fibroids _____ Infertility _____ Ovarian Cysts _____ Breast disease/biopsy _____ Endometriosis _____ None ☒
If so, please detail year and how it was treated? _____

Contraception/Pregnancy History

Sexually active: No ☒ Yes Medical issues pertaining to sexual activity: _____
Current method of preventing pregnancy: condoms
Total number of: pregnancies 1 Vaginal deliveries 1 C-Section _____ Miscarriages _____ Abortions 1 Ectopic _____
Pregnancy Complications: _____

Would you accept a blood transfusion in a life threatening situation? NO ☐ YES ☒

Pregnancy History

Date	Delivery Type	Birth Weight	Gender/Name	Complication
<u>10/19/14</u>	<u>Vaginal</u>	<u>7lbs 9oz.</u>	<u>Male/ Camden</u>	

*** ALLERGIES: No ☒ Yes Please list allergies: Penicillin, Cefaclor

Weill Cornell Medical College
Department of Obstetrics and Gynecology
Division of Maternal Fetal Medicine

Name: Dana Moolani
MRN: _____
Date of visit: 8/18/15

Medications: (List names and dosages; include vitamins, herbs, and other supplements):

Medication	Dosage	How Often	Medication	Dosage	How Often
<u>Vitamin Pre Natal</u>	<u>2gummies</u>	<u>1/day</u>			

Past and Current Medical History (Please include year if diagnosis and treatment were given).

<u>Anemia (Blood Transfusion?)</u>	<u>Hemorrhoids</u>	<u>Neurologic Disorder</u>
<u>Anesthesia Complications</u>	<u>Hiatal Hernia</u>	<u>Psychiatric Disorder</u>
<u>Breast disease</u>	<u>2011 PCNL Surgery</u>	<u>Kidney Stones</u>
<u>Congenital Heart Problem</u>	<u>Lung Disease/Asthma</u>	<u>Sickle Cell/Carrier</u>
<u>Diabetes</u>	<u>Lupus</u>	<u>TB/ Positive PPD</u>
<u>Gastrointestinal/Gallstones</u>	<u>Migraine Headaches</u>	<u>Thrombotic Disorder (Blood Clots)</u>

Surgical History (Briefly include your surgical history). 2000 LEEP, 2011 PCNL Surgery (Kidney stone removal)
2013 Ureteroscopy

Family History

Mother: Living: Yes Deceased (Cause): _____ Father: Living: Yes Deceased (Cause): _____
Siblings Number Living: 1 Number deceased: _____ Cause: _____

Detail below if anyone in your immediate family had the diagnosis: (Please indicate Mother, Father, Sibling, Grandparents and which side).

Bleeding Disorder	<u>Paternal Grandmother</u>	<u>Father, Paternal grandfather</u>	Neurological Disease
Blood Clots	<u>Paternal Grandfather</u>	<u>Mother, Maternal grandmother</u>	Psychiatric Disease
Cancer: Breast/GYN	<u>Paternal Grandfather</u>		Thyroid Disease
Other			

Social History

Occupation: Accountant Marital Status: Married Do you smoke? NO If so, how many packs a day?

Do you drink alcohol? No Yes If so, how many drinks/week? Do you take drugs? NO If so, which ones?

Review of Systems: Are you experiencing any of the following symptoms? Please indicate all that apply or NO if they do not.

Constitutional	<u>No</u>	<u>Fatigue</u>	<u>Weight Loss</u>	<u>Weight Gain</u>	<u>Fever</u>
Eye Problems	<u>No</u>	<u>Vision Changes</u>	<u>Glasses/Contacts</u>		
Ear, Nose, Throat	<u>No</u>	<u>Headache</u>	<u>Sinusitis</u>	<u>Ringing in Ears</u>	<u>Nose Bleed</u>
Cardiovascular	<u>No</u>	<u>Shortness of Breathe</u>	<u>Chest Pain</u>	<u>Edema</u>	<u>Palpitations</u>
Respiratory	<u>No</u>	<u>Wheezing</u>	<u>Coughing Blood</u>	<u>Cough</u>	
Gastrointestinal	<u>No</u>	<u>Nausea/vomiting</u>	<u>Constipation</u>	<u>Diarrhea</u>	<u>Bloody Stool</u>
Genitourinary	<u>No</u>	<u>Bloody Urine</u>	<u>Painful Urination</u>	<u>Urgency</u>	<u>Frequency</u>
Musculoskeletal	<u>No</u>	<u>Muscle Weakness</u>	<u>Muscle Pain</u>		
Skin/Breast	<u>No</u>	<u>Breast Pain</u>	<u>Nipple Discharge</u>	<u>Breast Mass</u>	<u>Skin Rash</u>
Neurological	<u>No</u>	<u>Fainting</u>	<u>Seizures</u>	<u>Numbness</u>	<u>Trouble Walking</u>
Psychiatric	<u>No</u>	<u>Depression</u>	<u>Anxiety</u>		
Endocrine	<u>No</u>	<u>Dry skin</u>	<u>Abnormal Thirst</u>	<u>Hot Flashes</u>	
Blood/Lymph	<u>No</u>	<u>Easy Bruising</u>	<u>Abnormal Bleeding</u>	<u>Swollen Glands</u>	

Other _____

Reviewed By: _____, MD on _____

Date



Patient Information

Patient: DANA LEVY MOOLANI
DOB: 05/11/1980
Gender: Female
Ethnicity: JEWISH
Procedure ID: 5249
Report Date: 02/25/2014
Report Updated: 03/10/2014

Sample Information

Specimen Type: Blood
Specimen Number: 6348
Barcode: 20140114565122
Date Specimen Collected: 02/12/2014
Date Specimen Received: 02/14/2014
Date Specimen Analyzed: 02/14/2014

Ordering Practice

Practice Code: 34
New York University School of Medicine
660 First Ave at 38 St
New York, NY 10016
Physician: Frederick Licciardi

Summary of Genetic Testing Results

Disease	Disease Groups	Next Steps
Glycogen Storage Disease: Type II Gene: GAA Mutation: c.T-45G (1 abnormal copy)		Assess reproductive risk based on partner's test results.

Disease Groups

High Impact	Treatment Benefits	X-Linked	Moderate Impact
These diseases have a significant impact on life expectancy and quality of life.	Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.	These diseases are passed down by female carriers. Carriers may have symptoms.	These diseases typically do not affect life expectancy but can affect quality of life.

Summary of Testing Performed

Diseases Tested: 213	Mutations Tested: 1682	Genes Tested: 201
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All other mutations analyzed by Recombine were not detected. This reduces but does not eliminate your chance to be a carrier for the associated genetic diseases. Recombine does not screen for every possible genetic disease.

Learn More

A list of all the genetic diseases and mutations you were screened for is included in the latter pages of this report. For disease information, please visit www.recombine.com/our-test. To speak with a Genetic Counselor, call 855.OUR.GENES

Lab Technician: Bo Chu

Reviewed by: Pere Colls, PhD, HCLD

*Methods and Limitations: Recombine developed this genetic assay using the Illumina Infinium Custom HD Genotyping Assay. The test is intended for clinical preconception and/or prenatal screening purposes and is not validated for detection of homozygous mutations. False positive or negative results may occur for reasons that include: genetic variants, assay limitations, sample mix-up, sample contamination, and molecular and technical errors. Recombine tests for Spinal Muscular Atrophy 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).

Recombine
32 Union Square East #1008
New York, NY 10003
855.687.4363

Patient Name: Dana Levy Moolani

Date of Birth: 5/11/1980

Date of Phone Consultation: 3/5/2014

Name of Genetic Counselor: Shabnam Asgari

Dear Dr. Frederick Licciardi,

I spoke with Karim Moolani and Dana Levy Moolani today via phone consultation regarding their Recombine genetic testing results; specifically that he was not identified as a carrier for any of the conditions included on the NYUFC CarrierMap screening panel. Dana Levy Moolani was identified as a carrier of Glycogen Storage Disease: Type II.

Medical and Family History:

- Karim Moolani is a 27-year old male of Indian descent currently in good health. Dana Levy Moolani is a 33-year old female of Ashkenazi Jewish descent. Dana reported that she has Cystinuria, but is currently in good health. She reportedly had one pregnancy termination with a previous partner. Dana reported that she is currently pregnant, this is the couples first pregnancy.

Dana has one brother who is reportedly healthy, it is unknown if he has cystinuria. Dana reported that her brother is looking into getting testing.

Karim reported that one of his paternal uncles passed away in his late 20s, he reportedly had epilepsy, his exact cause of death was not known. Karim's paternal grandmother passed away at 50-years of age from breast cancer diagnosed in her late 40s. No other cancer was reported in the family history.

We discussed the natural history and etiology of cystinuria. We discussed that this disease is not part of the Recombine panel; therefore, the couple was not tested for this disease. We discussed that cystinuria is inherited in an autosomal recessive manner, meaning that an affected individual needs to have two non-working copies of their gene to be affected. We discussed that if Karim is found to be a carrier the couple would have a 1 in 2 (or 50%) chance to have an affected child. We discussed that carrier testing for this condition is available through Mayo Clinic and Mount Sinai School of Medicine. We discussed that Dana's

brother has a 1 in 4 (or 25%) chance to be affected as well, or a 2 in 3 (or 66%) chance to be a carrier and unaffected.

We discussed that seizures affect approximately 1% of the population and can be genetic or non-genetic (eg. illness and/or trauma). Seizures with a genetic origin may be isolated or may be part of a genetic syndrome. If a seizure is due to a genetic syndrome, the recurrence risk is based on that condition's inheritance pattern. Most seizure disorders are not caused by a genetic syndrome and are believed to result from multifactorial inheritance (genes and environmental interaction). The exact risk depends on the type of seizure and age of onset of the parent, the risk to offspring is higher when mothers rather than fathers have seizures [1]. Prenatal diagnosis for these types of seizures is not currently available. Non-genetic causes of recurrent seizures are due to environmental factors such as head trauma, tumors of the brain and infections and would not increase the risk of recurrence for other family members above the general population rate. The exact cause for his uncle's seizures was not known, we discussed that if they are non-genetic in etiology, then the couple's risk of having a child with a seizure disorder should not be increased over the general population background risk.

We discussed that most cancer is sporadic, only a small minority arise due to a cancer predisposition. We discussed that genetic testing for cancer is typically done in an affected individual and cancer screening begins 10-years earlier than the youngest cancer diagnosis in the family. We discussed that Karim should speak with his physician(s) regarding the family history and followup as recommended.

The reported family history is otherwise noncontributory for intellectual disabilities, learning disabilities, blindness, deafness, seizure disorders, birth defects, known genetic conditions, infant deaths, infertility, recurrent pregnancy loss and consanguinity.

Recombine Results:

- **Glycogen Storage Disease: Type II**

Dana was identified as a carrier for Glycogen Storage Disease Type II, otherwise known as Pompe disease. This condition is caused by a buildup of a complex sugar called glycogen in the body's cells, particularly damaging the muscles. In the classic form of infantile-onset Pompe, infants exhibit muscle weakness and heart defects within months after birth. If untreated, patients die from heart failure in their first year. The non-classic form of infantile-onset Pompe disease appears by age 1 and symptoms include delayed motor skills and muscle weakness leading to serious breathing problems. Affected children live only into early childhood. The late-onset type of Pompe disease appears in late childhood or adulthood and causes progressive muscle weakness in the legs, trunk, and muscles that control breathing. Affected patients die from respiratory failure in their 20-30s. Unfortunately, there is no cure for Pompe disease. Treatment

typically involves managing the symptoms of the condition, as well as nutritional and respiratory support.

c.T-45G: Dana was identified to carry the c.T-45G mutation in the GAA gene responsible for Pompe disease. This mutation is typically associated with the late-onset form of Pompe disease [2].

Reproductive Risk: Karim was not identified as a carrier for Pompe disease through the Recombine test. We discussed that this result reduces but does not eliminate the chance for this couple to have a child with Pompe disease. We discussed that the carrier frequency of this condition is not known for the general population or for individuals of Indian descent. We discussed that the Recombine detection rate for this condition in the general population is approximately 10%. We discussed that the couple's risk of having an affected child is difficult to quantify without a known carrier rate for this condition. We discussed the risks, benefits, and limitations of gene sequencing as an additional option to further refine the couple's chance to have a child with this condition. Gene sequencing is a more comprehensive test that can detect more disease-causing mutations; however, gene sequencing can also detect variants of uncertain significance (VUS) which are difficult to interpret clinically. Gene sequencing for Pompe disease is available through Prevention Genetics, GeneDx, and Emory Genetics Laboratory. Gene sequencing for Pompe disease is available through Prevention Genetics, GeneDx, and Emory Genetics Laboratory. Prevention Genetics offers the lowest cost at \$880. The couple indicated that they will discuss these results further.

- **Recombine Test**

We discussed that the Recombine test looked for mutations, or disease-causing changes, in genes associated with 191 recessive genetic diseases for Karim and 213 for Dana. Basic genetics including autosomal recessive inheritance were reviewed with the couple. The couple were not identified as carriers for any other conditions included on the NYUFC CarrierMap screening panel. We discussed that while the Recombine test significantly reduces the chance that someone is a carrier for the conditions tested, it does not completely eliminate that chance, which is less than 1%. We also discussed that the Recombine test does not test for every known genetic disease. This test does not look for chromosome problems, such as Down syndrome, and is not a substitute for routine prenatal care.

- The patient's questions and concerns were addressed.

Plan & Follow-Up

- Karim Moolani was not identified as a carrier for any of the conditions included on the NYUFC CarrierMap screening panel. Dana Levy Moolani was identified as a carrier of Glycogen Storage Disease: Type II. The couple indicated that they will discuss these results further and then decide if they want to pursue further genetic testing.

It was a pleasure speaking with this patient this day. Please do not hesitate to contact me with any other questions or concerns.



Shabnam Asgari, M.S., C.G.C.
Genetic Counselor

References:

- [1] Ottman et al., **Higher risk of seizures in offspring of mothers than of fathers with epilepsy.** American journal of human genetics, 1988. PubMed PMID: 3414683.
- [2] Hermans et al., **Twenty-two novel mutations in the lysosomal alpha-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II.** Human mutation, 2004. PubMed PMID: 14695532.

Diseases & Mutations Assayed

Groups	Disease	#	Mutations
● ● ● ●	17-Alpha-Hydroxylase Deficiency (CYP17A1)	17	c.T601A (p.Y201N). c.C1084T (p.R362C). c.T1216C (p.W406R). c.G51A (p.W17X). c.C1024A (p.P342T). c.G1073A (p.R358Q). c.C715T (p.R239X). c.157_159delTTC (p.53delF). c.C81A (p.Y27X). c.C286T (p.R96V). c.T985G (p.Y329D). c.G287A (p.R96Q). c.G1040A (p.R347H). c.T316C (p.S106P). c.T340G (p.F114V). c.A347T (p.D116V). c.C1039T (p.R347C)
● ● ● ●	17-Beta-Hydroxysteroid Dehydrogenase Type III Deficiency (HSD17B3)	8	c.G239A (p.R80Q). c.A703G (p.M235V). c.C695T (p.S232L). c.G166A (p.A56T). c.C608T (p.A203V). c.C238T (p.R80W). c.A389G (p.N130S). c.G803A (p.C268Y)
● ● ● ●	21-Hydroxylase-Deficient Congenital Classical Adrenal Hyperplasia (CYP21A2)	4	c.G1273A (p.G425S). c.293-13C>G. c.332_339delGAGACTAC. c.G877A (p.G293S)
● ● ● ●	21-Hydroxylase-Deficient Congenital Nonclassical Adrenal Hyperplasia (CYP21A2)	3	c.A188T (p.H63L). c.C1360T (p.P454S). c.G844C (p.V282L)
● ● ● ●	3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (HSD3B2)	8	c.742_747delGTCCGAinsAACTA (p.V248NfsR249X). c.A1119C (p.X373C). c.G512A (p.W171X). c.C745T (p.R249X). c.C29A (p.A10E). c.C1022T (p.P341L). c.C776T (p.T259M). c.G424A (p.E142K)
● ● ● ●	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC1 (MCCA) Related (MCCC1)	2	c.T1310C (p.L43T). c.A1155C (p.R385S)
● ● ● ●	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC2 (MCCB) Related (MCCC2)	8	c.A1309G (p.I437V). c.G803C (p.R268T). c.T499C (p.C167R). c.G295C (p.E99Q). c.G464A (p.R155Q). c.C929G (p.P310R). c.A569G (p.H190R). c.G838T (p.D280Y)
● ● ● ●	3-Methylglutaconic Aciduria: Type 3 (OPA3)	5	c.313C>G (p.Q105E). c.143-1G>C. c.320_337delAGCAGCGCCACAAAGGAGG (p.Q108_E113del). c.A15C>T (p.Q139X). c.277G>A (p.G93S)
● ● ● ●	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS)	6	c.259C>T (p.P87S). c.286G>A (p.D96N). c.155A>G (p.N52S). c.46C>T (p.R16C). c.347A>G (p.D116G). c.74G>A (p.R25Q)
● ● ● ●	Abetalipoproteinemia (MTTP)	2	c.G2593T (p.G865X). c.2212delT
● ● ● ●	Achromatopsia: CNGB3 Related (CNGB3)	6	c.G1208A (p.R403Q). c.991-3T>G. c.G1006T (p.E336X). c.1148delC (p.T383fs). c.817_824delCCAGACT. c.886_896delACTTCTACAAinsT
● ● ● ●	Acrodermatitis Enteropathica (SLC39A4)	10	c.C599T (p.P200L). c.968-971delAGTC. c.G1120C (p.G374R). c.C283T (p.R95C). c.G1576C (p.G526R). c.G909C (p.Q303H). c.G989A (p.G330D). c.1017ins53. c.1223-1227delCCGGG. c.C318A (p.N106K)
● ● ● ●	Acyl-CoA Oxidase I Deficiency (ACOX1)	5	c.C442T (p.R148X). c.372delCATGCCCGCTGGAACCTT. c.G532T (p.G178C). c.A832G (p.M278V). c.A926G (p.Q309R)
● ● ● ●	Adenosine Deaminase Deficiency (ADA)	22	c.43C>G (p.H15D). c.419G>A (p.G140E). c.302G>T (p.R101L). c.646G>A (p.G216R). c.220G>T (p.G74C). c.320T>C (p.L107P). c.301C>T (p.R101W). c.536C>A (p.A179D). c.445C>T (p.R149W). c.467G>A (p.R156H). c.596A>C (p.Q199P). c.631C>T (p.R211C). c.302G>A (p.R101Q). c.529G>A (p.V177M). c.454C>A (p.L152M). c.466C>T (p.R156C). c.385G>A (p.V129M). c.58G>A (p.G20R). c.248C>A (p.A83D). c.632G>A (p.R211H). c.872C>T (p.S291L). c.986C>T (p.A329V)
● ● ● ●	Adrenoleukodystrophy: X-Linked (ABCD1)	25	c.T520G (p.Y174D). c.C1817T (p.S606L). c.G421A (p.A141T). c.1791_1792delTA (p.A597AfsX599). c.871_873delGAG (291delE). c.G1202A (p.R401Q). c.G871A (p.E291K). c.G1429T (p.E477X). c.C1849T (p.R617C). c.1634+1G>A (IVS6+1G>A). c.1865+1G>A (IVS8+1G>A). c.1866-10G>A (IVS8-10G>A). c.G796C (p.G266R). c.1937delC. c.A443G (p.N148S). c.C1165G (p.R389G). c.C1390T (p.R464X). c.901-1G>A (IVS1-1G>A). c.C1252T (p.R418W). c.C1544T (p.S515F). c.G1850A (p.R617H). c.1635-2A>G (IVS6-2A>G). c.1415_1416delAG (1801delAG). c.C1552T (p.R518W). c.C1451G (p.P484R)
● ● ● ●	Alkaptonuria (HGD)	14	c.T899G (p.V300G). c.T360G (p.C120W). c.G808A (p.G270R). c.1111_1112insC. c.G990T (p.R330S). c.C688T (p.P230S). c.174delA. c.16-1G>A. c.457_458insG. c.C140T (p.S47L). c.342+1G>A. c.G481C (p.G161R). c.45_458insG. c.A1102G (p.M368V)
● ● ● ●	Alpha-1-Antitrypsin Deficiency (SERPINA1)	4	c.A1131T (p.L377F). c.G1096A (p.E366K). c.226_228delTTC (p.76delF). c.C187T (p.R63C)
● ● ● ●	Alpha-Mannosidosis (MAN2B1)	3	c.2248C>T (p.R750W). c.2426T>C (p.L809P). c.1830+1G>C (p.V549_E610del)
● ● ● ●	Alpha Thalassemia (HBA1)	18	Alpha3.7. c.427T>C (p.X143Qext32). c.207C>A (p.N69K). c.*+94A>G. c.207C>G (p.N69K). c.2T>C (p.M1T). c.94_95delAG (p.R32DfsX24). c.-5092_95+60del5247bp (Alpha 5.2). SEA deletion. THAI deletion. c.223G>C (p.D75G). HBA2c.-9111_HBA1c.*2791del16401bp (MED). Alpha4.2. 11.1kb deletion. c.377T>C (p.L126P). c.95+2_95+6delTGAGG. c.339C>G (p.H113Q). c.340_351delCTCCCCGCCGAG (p.L114_E117del)
● ● ● ●	Alport Syndrome: COL4A3 Related (COL4A3)	3	c.C4571G (p.S1524X). c.C4441T (p.R1481X). c.4415_4419delCTTTT
● ● ● ●	Alport Syndrome: COL4A4 Related (COL4A4)	5	c.C3713G (p.S1238X). c.G3601A (p.G1201S). c.C4129T (p.R1377X). c.C4923A (p.C1641X). c.C4715T (p.P1572L)
● ● ● ●	Alport Syndrome: X-linked (COL4A5)	3	c.T4946G (p.L1649R). c.G5030A (p.R1677Q). c.G4691C (p.C1564S)
● ● ● ●	Amegakaryocytic Thrombocytopenia (MPL)	3	c.C127T (p.R43X). c.79+2T>A. c.G305C (p.R102P)
● ● ● ●	Andermann Syndrome (SLC12A6)	5	c.2436delG (p.T813fsX813). c.901delA. c.C2023T (p.R675X). c.C3031T (p.R1011X). c.C619T (p.R207C)
● ● ● ●	Androgen Insensitivity Syndrome: Complete (AR)	18	c.C2323T (p.R ⁻ 5C). c.T1 ⁻ 48A (p.F583Y). c.A2650T (p.K884X). c.A2069C (p.H690P). c.A1 ⁻ 1T (p.K591X). c.C178T (p.Q60X). c.A2362G (p.M788V). c.T2123G (p.L708R). c.G2157A (p.W719X). c.G1739T (p.C580F). c.G2391A (p.W797X). c.G2231A (p.G744E). c.179insA. c.G2343T (p.M781I). c.G2324A (p.R775H). c.T2033C (p.L678P). c.180delGC. c.G2599A (p.V867M)
● ● ● ●	Argininosuccinate Lyase Deficiency (ASL)	6	c.A857G (p.Q286R). c.G532A (p.V178M). c.C283T (p.R95C). c.446+1G>A. c.C1153T (p.R385C). c.C1135T (p.R379C)
● ● ● ●	Aromatase Deficiency (CYP19A1)	11	c.C1303T (p.R435C). c.G628A (p.E210K). c.C1123T (p.R375C). c.629-3C>A. c.858+2T>C. c.1222_1224delC. c.G1310A (p.C437Y). c.G1094A (p.R365Q). c.G1310A (p.C437Y). c.468delC. c.451+1G>A

Groups	Disease	#	Mutations
● ● ● ●	ARSACS (SACS)	8	c.8844delT (p.L2949fs). c.T5836C (p.W1946R). c.6273delT. c.T9742C (p.W3248R). c.C4933T (p.R1645X). c.T3161C (p.F1054S). c.C12973T (p.R4325X). c.C7504T (p.R2502X)
● ● ● ●	Arts Syndrome (PRPS1)	2	c.T455C (p.L152P). c.A398C (p.Q133P)
● ● ● ●	Aspartylglycosaminuria (AGA)	8	c.T214C (p.S72P). c.199_200delGA. c.G904A (p.G302R). c.T916C (p.C306R). c.G482A (p.R161Q). c.G488C (p.C163S). c.C302T (p.A101V). c.G179A (p.G60D)
● ● ● ●	Ataxia-Telangiectasia (ATM)	19	c.A8030G (p.Y2677C). c.7517_7520delGAGA (p.R2506fs). c.C103T (p.R35X). c.T7271G (p.V2424G). c.C7327T (p.R2443X). c.G5932T (p.E1978X). c.G7876C (p.A2626P). c.5'62+1126A>G. c.630-2A>C. c.638_646delTAGAATTTC (p.R254_S2549delRIS). c.C5908T (p.Q1970X). c.3894insT. c.5712_5713insA (p.S1905fs). c.A7268G (p.E2423G). c.T7967C (p.L2656P). c.T8480G (p.F2827C). c.G3576A (p.K1192K). c.1564_1565delGA (p.E522fs). c.3245delATCinsTGAT (p.H1082fs)
● ● ● ●	Ataxia with Vitamin E Deficiency (TTPA)	5	c.487_488insTT (p.T163GfsX13). c.744delA. c.G575A (p.R192H). c.C400T (p.R134X). c.T303G (p.H101Q)
● ● ● ●	Autosomal Recessive Polycystic Kidney Disease (PKHD1)	17	c.9689delA. c.T10658C (p.I3553T). c.10174C>T (p.Q3392X). c.T10412G (p.V3471G). c.C1486T (p.R496X). c.6992T>A (p.I2331K). c.C101T (p.T36M). c.9530T>C (p.I311T). c.C8011T (p.R261X). c.3761_3762delCCinsG (p.A1254fs). c.5895insA. c.G5221A (p.V1741M). c.8870T>C (p.I2957T). c.C9053T (p.S3018F). c.C4991T (p.S1664F). c.2414C>T (p.P805L). c.664A>G (p.I222V)
● ● ● ●	Bardet-Biedl Syndrome: BBS10 Related (BBS10)	3	c.271_273ins1bp (p.C91fsX95). c.G101C (p.R34P). c.T931G (p.S311A)
● ● ● ●	Bardet-Biedl Syndrome: BBS12 Related (BBS12)	5	c.1063C>T (p.R355X). c.335_337delTAG. c.865G>C (p.A289P). c.1483_1484delGA (p.E495fsX498). c.1114_1115delTT (p.F372X)
● ● ● ●	Bardet-Biedl Syndrome: BBS1 Related (BBS1)	3	c.G1645T (p.E549X). c.T1169G (p.M390R). c.851delA
● ● ● ●	Bardet-Biedl Syndrome: BBS2 Related (BBS2)	3	c.C72G (p.Y24X). c.T224G (p.V75G). c.940delA
● ● ● ●	Bare Lymphocyte Syndrome: Type II (CLITA)	1	c.G1141T (p.E381X)
● ● ● ●	Bartter Syndrome: Type 4A (BSND)	6	c.G3A (p.M11). c.A1T (p.M1L). c.G28A (p.G10S). c.G23T (p.R8L). c.G139A (p.G47R). c.C22T (p.R8W)
● ● ● ●	Beta-Hexosaminidase Pseudodeficiency (HEXA)	2	c.C739T (p.R247W). c.C745T (p.R249W)
● ● ● ●	Beta-Ketothiolase Deficiency (ACAT1)	15	c.T99A (p.Y33X). c.1006-1G>C. c.G547A (p.G183R). c.G997C (p.A333P). c.T2A (p.M1K). c.1006-2A>C. c.G1138A (p.A380T). c.G1136T (p.G379V). c.C814T (p.Q272X). c.A278G (p.N93S). c.1033_1035delGAA (p.345delE). c.T935C (p.I312T). c.1083insA. c.826+1G>T. c.C433G (p.Q145E)
● ● ● ●	Beta Thalassemia (HBB)	91	c.c-142t. c.45_46insG (p.L16fs). c.a-79g. c.85_86insC (p.L29fs). c.250delG. c.T2C (p.M1T). c.2T>G (p.M1R). c.59A>G (p.N20S). c.g-29a. c.93-15T>G. c.G169C (p.G57R). c.c-136g. c.217_221delAGTGinsT (p.S73_D74delinsLfs). c.36delT (p.T13fs). c.271G>T (p.E91X). c.46delT (p.W16Gfs). c.92+2T>A. c.135delC (p.F46fs). c.316-1G>T. c.287_288insA (p.L97fs). c.90C>T (p.G30G). c.T75A (p.G25G). c.c-140t. c.17_18delCT. c.a-81g. c.203_204delITG (p.V68fs). c.316-197C>T. c.217insA. c.316-146T>G. c.A52T (p.K18X). c.G295A (p.V99M). c.315+2T>C. c.155delC (p.P52fs). c.316-2A>G. c.G34A (p.V12I). c.4delG. c.c-137g. c.92+5G>A. c.92+5G>T. c.c-138t. c.A-50C. c.383_385delIAGG (p.Q128_A129delQAlnsP). c.G415C (p.A139P). c.323_324insG (p.N109fs). c.444+110T>C. c.444+111A>G. c.c-151t. c.a-78g. c.G4A (p.W16X). c.316-106C>T. c.316-2A>C. c.G48A (p.W16X). c.C118T (p.Q40X). c.t-80a. c.92+2T>C. c.315+1G>A. c.316-3C>G. c.92+110G>A/c.93-21G>A. c.20delA. c.124_129TCTTT>TT. c.316-3C>A. c.315+705T>G. c.1A>G (p.M1V). c.316-1G>C. c.c-137t. c.51delC. c.93-1G>A. c.93-1G>T. c.230delC. c.223+702_444+342del620insAAGTAGA. c.316-1G>A. c.92+1G>C. c.G114A (p.W38X). c.112delT. c.93-1G>C. c.92+5G>C. c.G82T (p.A28S). c.315+745C>G. c.G113A (p.W38X). c.126delC. c.92+1G>A. c.25_26delAA. c.444+113A>G. c.225delC. c.315+654C>T. c.68_74delAAGTTGG. c.92+6T>C. c.G92C (p.R31T). c.92+1G>T. c.2_28insG. c.-17_92+25del293bp
● ● ● ●	Biotinidase Deficiency (BTD)	10	c.G511A (p.A171T). c.A1466C (p.N489T). c.G1330C (p.D444H). c.A1368C (p.Q456H). c.A755G (p.D252G). c.C1612T (p.R538C). c.C235T (p.R79C). c.G100A (p.G34S). c.T1207G (p.F403V). c.98_104delGCGGCTGinsTCC (p.C33FfsX68)
● ● ● ●	Bloom Syndrome (BLM)	9	c.2207delATCTGainsTAGATTC. c.G3107T (p.C1036F). c.2407insT. c.1933C>T (p.Q645X). c.1701G>A (p.W567X). c.C2528T (p.T843I). c.C2695T (p.R899X). c.1284G>A (p.W428X). c.557_559delCAA (p.S186X)
● ● ● ●	Canavan Disease (ASPA)	8	c.C693A (p.Y231X). c.A692G (p.Y231C). c.A854C (p.E285A). c.A71G (p.E24G). c.C693T (p.Y231Y). c.433-2A>G. c.C654A (p.C218X). c.C914A (p.A305E)
● ● ● ●	Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	7	c.A1079G (p.E360G). c.G2126A (p.G709E). c.G2129A (p.G710E). c.C1241T (p.A414V). c.A1493G (p.Y498C). c.A1361G (p.D454G). c.C1436T (p.P479L)
● ● ● ●	Carnitine Palmitoyltransferase II Deficiency (CPT2)	22	c.A1649G (p.Q550R). c.C680T (p.P227L). c.C370T (p.R124X). c.T1148A (p.F383Y). c.C1891T (p.R631C). c.G1646A (p.G549D). c.C338T (p.S113L). c.A1883C (p.Y628S). c.1923_1935delGAAGGCCCTTAGAA. c.C1810T (p.P604S). c.1737delC. c.533_556delTGAACCTGCAAAAAGTGACACTA. c.T1342C (p.F448L). c.G452A (p.R151Q). c.G520A (p.E114K). c.G1145A (p.R382K). c.A359G (p.Y120C). c.C149A (p.P50H). c.A983G (p.D328G). c.109_110insGC. c.1238_1239delAG. c.C1507T (p.R503C)
● ● ● ●	Cartilage-Hair Hypoplasia (RMRP)	2	c.A70G. c.G262T
● ● ● ●	Cerebrotendinous Xanthomatosis (CYP27A1)	13	c.G646C (p.A216P). c.C1016T (p.T339M). c.G434A (p.G145E). c.G1421A (p.R474Q). c.C379T (p.R127W). c.G1214A (p.R405Q). c.C1420T (p.R474W). c.C1435T (p.R479C). c.G583T (p.E195X). c.C1183T (p.R395C). c.1263+1G>A. c.844+1G>A. c.C819delT
● ● ● ●	Charcot-Marie-Tooth Disease with Deafness: X-Linked: GJB1 Related (GJB1)	23	c.T89A (p.I30N). c.A614G (p.N205S). c.C514T (p.P172S). c.C164T (p.T55I). c.C658T (p.R220X). c.C424T (p.R142V). c.T397C (p.W133R). c.225delG (p.R75fs). c.C223T (p.R75W). c.C254G (p.S85C). c.G283A (p.V95M). c.G304T (p.E102X). c.G181A (p.V63I). c.T145C (p.S49P). c.T66G (p.F235C). c.G37T (p.V13L). c.T467G (p.L156R). c.G415A (p.V139M). c.C43T (p.R15W). c.T408C (p.V136A). c.304delGAG (p.102delE). c.G123C (p.G41D). c.A194G (p.Y65C)

Groups	Disease	#	Mutations
● ● ● ●	Charcot-Marie-Tooth Disease with Deafness: X-Linked: PRPS1 Related (PRPS1)	2	c.A129C (p.E43D). c.T344C (p.M115T)
● ● ● ●	Cholesteryl Ester Storage Disease (LIPA)	4	c.883C>T (p.H295Y). c.652C>T (p.R218X). c.894G>A. c.1024G>A (p.G342R)
● ● ● ●	Choreoacanthocytosis (VPS13A)	1	c.6059delC
● ● ● ●	Choroideremia (CHM)	1	c.1609+2insT
● ● ● ●	Chronic Granulomatous Disease: X-Linked (CYBB)	14	c.C911G (p.P304R). c.C625T (p.H209Y). c.C217T (p.R73X). c.A302G (p.H101R). c.C676T (p.R226X). c.C1244A (p.P415H). c.G466A (p.A156T). c.C301T (p.H101Y). c.G1166C (p.G389A). c.G252A (p.A84A). c.45+6T>C. c.C90T (p.H303N). c.A1499G (p.D500G). c.252+5G>A
● ● ● ●	Citrullinemia: Type I (ASS1)	16	c.420+5G>A. c.G470A (p.R157H). c.G1085T (p.G362V). c.A928C (p.K310Q). c.G323T (p.R108L). c.G40A (p.G14S). c.C1087T (p.R363W). c.C256T (p.R86C). c.C53T (p.S18L). c.1168G>A (p.G390R). c.C835T (p.R279X). c.C910T (p.R304W). c.T535C (p.W179R). c.G970A (p.G324S). c.G539A (p.S180N). c.1194-1G>C
● ● ● ●	Classical Galactosemia (GALT)	17	c..1039_753del3162. c.C413T (p.T138M). c.134_138delCAGCT. c.T584C (p.L195P). c.253-2A>G. c.820+51_789del2294ins12. c.C404T (p.S135L). c.A563G (p.Q188R). c.A626G (p.Y209C). c.T512C (p.F111S). c.G855C (p.K285N). c.C99T (p.R333G). c.C505A (p.Q169K). c.G60T (p.E203K). c.T1138C (p.X380R). c.T221C (p.L74P). c.T425A (p.M142K)
● ● ● ●	Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)	3	c.G422A (p.R141H). c.C357A (p.F119L). c.G385A (p.V129M)
● ● ● ●	Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI)	1	c.G884A (p.R295H)
● ● ● ●	Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6)	4	c.T1432C (p.S478P). c.C998T (p.A333V). c.895_897delATA. c.257+5G>A
● ● ● ●	Congenital Lipoid Adrenal Hyperplasia (STAR)	11	c.178+3T>T. c.201_202delCT. c.64+1G>T. c.466-11T>A. c.G545T (p.R182L). c.C562T (p.R188C). c.G545A (p.R182H). c.C772T (p.Q258X). c.G559A (p.V187M). c.G749A (p.W250X). c.G650C (p.R217T)
● ● ● ●	Congenital Neutropenia: Recessive (HAX1)	6	c.C568T (p.Q190X). c.C256T (p.R86X). c.91delG. c.130insA. c.424insG. c.121_125insG
● ● ● ●	Corneal Dystrophy and Perceptive Deafness (SLC4A11)	7	c.2233_2240insTATGACAC. c.A2566G (p.M856V). c.1378delTACGinsA. c.473delGCTTCGCC. c.G1463A (p.R488K). c.T2528C (p.L843P). c.T637C (p.S213P)
● ● ● ●	Corticosterone Methyloxidase Deficiency (CYP11B2)	3	c.T1382C (p.L461P). c.A1492G (p.T498A). c.C541T (p.R181W)
● ● ● ●	Creatine Transporter Defect (SLC6A8)	8	c.778-2A>G. c.259G>A (p.G87R). c.950_951insA (p.Y317X). c.321_323delICTT (p.F107del). c.263-1G>C. c.1596+1G>A. c.1631C>T (p.P544L). c.1661C>T (p.P554L)
● ● ● ●	Crigler-Najjar Syndrome (UGT1A1)	11	c.A992G (p.Q331R). c.G923A (p.G308E). c.C991T (p.Q331X). c.C1124T (p.S375F). c.A1070G (p.Q357R). c.508_513delITTC (p.110delF). c.C1021T (p.R341X). c.C840A (p.C280X). c.A1198G (p.N400D). c.T524A (p.L175Q). c.T44G (p.L15R)
● ● ● ●	Cystic Fibrosis (CFTR)	124	c.3690delT. c.C1075A (p.Q359K). c.C1079A (p.T360K). c.2052delA (p.K684fs). c.1116+1305delT. c.3717+12191C>T. c.1116+1304_1305insT. c.3744delA. c.2052_2053insA (p.Q685fs). c.2737_2738insG (p.Y913X). c.3768insT. c.442delA. c.G3808A (p.D1270N). c.3535delACCA. c.3527delC. c.3038delC. c.3773_3774insT (p.L1258fs). c.G3454C (p.D1152H). c.G1438T (p.G480C). c.C1657T (p.R553X). c.2988+1G>A. c.261delTT. c.G350A (p.R117H). c.1521_1523delCTT (p.508delF). c.C3909G (p.N1303K). c.C1654T (p.Q552X). c.C1721A (p.P54H). c.1679+1634A>G. c.C3764G (p.S1255X). c.3659delC (p.T1220fs). c.C2668T (p.Q890X). c.3536_3539delCCAA (p.T1179fs). c.3067_3072delATAGTG (p.I1023_V1024delT). c.G3611A (p.W1204X). c.C2290T (p.R764X). c.G328C (p.D110H). c.C14T (p.P5L). c.325delITATinsG. c.313delA (p.I105fs). c.G171A (p.W57X). c.G1040A (p.R347H). c.G3752A (p.S1251N). c.G1040C (p.R347P). c.1976delA (p.N659fs). c.G3846A (p.W1282X). c.C1477T (p.Q493X). c.3140-26A>G. c.T1647G (p.S549R). c.3063delAGTGAT. c.G3848T (p.R1283M). c.1519_1521delATC (p.507delI). c.G532A (p.G178R). c.1155insTA. c.580-1G>T. c.1029delC. c.G988T (p.G330X). c.C223T (p.R75X). c.14delCA. c.T1090C (p.S364P). c.1766+1G>A. c.803delA (p.N268fs). c.164+12T>C. c.1545_1546delITA (p.Y155X). c.C2125T (p.R709X). c.T3302A (p.M1101K). c.T366G (p.Y122X). c.1585-1G>A. c.1543delITA. c.T617G (p.L206W). c.1680-1G>A. c.G1646A (p.S549N). c.G271A (p.G91R). c.G1652A (p.G551D). c.G1679C (p.R560T). c.G274T (p.E92X). c.C1000T (p.R334W). c.G1624T (p.G542X). c.C3472T (p.R1158X). c.G178T (p.E60X). c.G19T (p.E7X). c.G1558T (p.V520F). c.C3484T (p.R1162X). c.G1865A (p.G622D). c.G1055A (p.R352Q). c.C349T (p.R117C). c.A2128T (p.K710X). c.931delITTC (p.311delF). c.C1364A (p.A455E). c.946delIT. c.A1645C (p.S549R). c.1116+1305_1307delITTT. c.1923delCTCAAACTinsA. c.C1477T (p.Q493X). c.G1646T (p.S549I). c.1766+5G>T. c.1766+1G>T. c.1911delG. c.804delITA. c.1818delB4. c.802delA. c.2049insA. c.2049delA. c.579+5G>A. c.1973delGAAATTCATCTinsAGAAA. c.G1675A (p.A559T). c.579+1G>T. c.1116+1305_1306delITT. c.2051delAAinsG. c.531delIT. c.489+1G>T. c.2174insA. c.274-1G>A. c.2657+5G>A. c.C3196T (p.R1066C). c.G254A (p.G85E). c.C3276G (p.Y1092X). c.G3209A (p.R1070Q). c.273+3A>C. c.273+1G>A. c.C3587G (p.S1196X). c.C3712T (p.Q1238X). c.G3266A (p.W1089X). c.C1572A (p.C524X). c.C1013T (p.T338I)
● ● ● ●	Cystinosis (CTNS)	14	c.G124A (p.V42I). c.G613A (p.D205N). c.G283T (p.G95X). c.198delATTACTATCCTTGAGCTCCCC. c.18_21delGACT. c.G329T (p.G110V). c.G414A (p.W138X). c.C416T (p.S139F). c.G506A (p.G169D). c.G589A (p.G197R). c.C969G (p.N323K). c.T473C (p.L158P). c.G1015A (p.G339R). c.-39155_848del57119
● ● ● ●	D-Bifunctional Protein Deficiency (HSD17B4)	7	c.G46A (p.G16S). c.63G>T (p.L21F). c.422_423delAG. c.652G>T (p.V218L). c.317G>C (p.R106P). c.1369A>C (p.N457D). c.1369A>T (p.N457Y)
● ● ● ●	Diabetes: Recessive Permanent Neonatal (ABCC8)	2	c.G1144A (p.E382K). c.A215G (p.N72S)
● ● ● ●	Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	4	c.1905+1G>A. c.1897delC. c.295delTCAT. c.G2657A (p.R886H)
● ● ● ●	Du Pan Syndrome (GDF5)	5	c.T1315A (p.S439T). c.1309delITG. c.C1306A (p.P436T). c.G1133A (p.R378Q). c.T1322C (p.L441P)
● ● ● ●	Dystrophic Epidermolysis Bullosa: Recessive (COL7A1)	7	c.T8393A (p.M2798K). c.G4039C (p.G1347R). c.427-2A>G. c.7345-1G>A. c.C933A (p.Y311X). c.2470insG. c.5821-1G>A
● ● ● ●	Ehlers-Danlos Syndrome: Type VIIC (ADAMTS2)	1	c.C673T (p.Q225X)

Groups	Disease	#	Mutations
● ● ● ●	Ellis-van Creveld Syndrome (EVC2)	1	c.C3265T (p.Q1089X)
● ● ● ●	Emery-Dreifuss Myopathy: X-Linked (EMD)	3	c.C547A (p.P183T). c.C130T (p.Q44X). c.A1G (p.M1V)
● ● ● ●	Enhanced S-Cone (NR2E3)	1	c.G932A (p.R311Q)
● ● ● ●	Ethylmalonic Aciduria (ETHE1)	4	c.C487T (p.R163W). c.505+1G>T. c.G3T (p.M1I). c.221_222insA
● ● ● ●	Fabry's Disease (GLA)	23	c.G983C (p.G328A). c.T484C (p.W162R). c.C890T (p.S297F). c.T606G (p.C202W). c.G466A (p.A156T). c.C979A (p.Q327K). c.G194C (p.S65T). c.A791T (p.D264V). c.G1081C (p.G361R). c.T1095G (p.Y365X). c.T806C (p.V269A). c.G1025A (p.R342Q). c.A815G (p.N272S). c.T166G (p.C56G). c.A101G (p.N34S). c.A644G (p.N215S). c.C6 ⁻ 9T (p.R22 ⁻ X). c.A ⁻ 9 ⁻ T (p.D266V). c.G680A (p.R227Q). c.C436T (p.P146S). c.G427C (p.A143P). c.G888A (p.M296I). c.G982A (p.G328R)
● ● ● ●	Factor IX Deficiency (F9)	7	c.G217A (p.E73K). c.G677A (p.R226Q). c.G128A (p.R43Q). c.C223T (p.R75X). c.G316A (p.G106S). c.C1025T (p.T342M). c.T1328C (p.I443T)
● ● ● ●	Factor VIII Deficiency (F8)	34	c.C1750A (p.Q584K). c.C5122T (p.R1708C). c.C5143T (p.R1715X). c.C5422T (p.L1808F). c.C6413A (p.S2138Y). c.C6532T (p.R2178C). c.C6967T (p.R2323C). c.G121T (p.G41C). c.A940G (p.T314A). c.A5096T (p.Y1699F). c.A14 ⁻ 5G (p.Y492C). c.T935C (p.F312S). c.G2215A (p.E ⁻ 39K). c.G5305C (p.R1769R). c.A1226G (p.E409G). c.T1786C (p.S596P). c.G902T (p.R301L). c.T6360G (p.F2120L). c.G5123A (p.R1708H). c.G2167A (p.A723T). c.C43T (p.R15X). c.C1804T (p.R602X). c.G1293T (p.L431F). c.A1660G (p.S554G). c.G541A (p.V181M). c.G6545A (p.R2182H). c.G6506A (p.R2169H). c.T5372C (p.M1791T). c.A6278G (p.D2093G). c.A5822G (p.N1941S). c.G6683A (p.R2228Q). c.G6744T (p.V2248C). c.C1648T (p.R550C). c.G1957A (p.V653M)
● ● ● ●	Familial Dysautonomia (IKBKAP)	3	c.G2087C (p.R696P). c.C2741T (p.P914L). c.2204+6T>C
● ● ● ●	Familial Hyperinsulinism: Type 1: ABCC8 Related (ABCC8)	8	c.3989-9G>A. c.C4477T (p.R1493W). c.4516G>A (p.E1506K). c.G4055C (p.R1352P). c.G2147T (p.G ⁻ 16V). c.C4258T (p.R1420C). c.4159_4161delTTC (p.138 ⁻ delF). c.T560A (p.V18 ⁻ D)
● ● ● ●	Familial Hyperinsulinism: Type 2: KCNJ11 Related (KCNJ11)	6	c.G844A (p.E282K). c.C761T (p.P254L). c.G-134T. c.C36A (p.Y12X). c.T440C (p.L147P). c.A776G (p.H259R)
● ● ● ●	Familial Mediterranean Fever (MEFV)	11	c.C800T (p.T267I). c.A2080G (p.M694V). c.C1437G (p.F479L). c.G2082A (p.M694I). c.2076_2078delAAT (p.692delI). c.G2040A (p.M680I). c.G2230T (p.A744S). c.G2040C (p.M680I). c.T2177C (p.V726A). c.G2282A (p.R761H). c.G1958A (p.R653H)
● ● ● ●	Familial Mediterranean Fever: Mild Form (MEFV)	3	c.A2084G (p.K695R). c.G1223A (p.R408Q). c.C1105T (p.P369S)
● ● ● ●	Fanconi Anemia: Type C (FANCC)	8	c.456+4A>T. c.G65A (p.W22X). c.T1661C (p.L554P). c.C553T (p.R185X). c.C1642T (p.R548X). c.C37T (p.Q13X). c.G66A (p.W22X). c.66delG
● ● ● ●	Fragile X Syndrome (FMR1)	1	c.-129CGG(>40)
● ● ● ●	Fumarase Deficiency (FH)	1	c.1431_1433insAAA
● ● ● ●	Galactokinase Deficiency (GALK1)	6	c.1045G>A (p.G349S). c.238G>T (p.E80X). c.1144C>T (p.Q382X). c.1031C>T (p.T344M). c.94G>A (p.V32M). c.82C>A (p.P28T)
● ● ● ●	Gaucher Disease (GBA)	10	c.G1604A (p.R535H). c.T1448C (p.L483P). c.C1504T (p.R502C). c.G1342C (p.D448H). c.1263_1317del55. c.A1226G (p.N409S). c.84_85insG. c.A1343T (p.D448V). c.115+1G>A. c.G129 ⁻ T (p.V433L)
● ● ● ●	Gitelman Syndrome (SLC12A3)	9	c.C622T (p.R208W). c.1923-1G>T. c.2744+1G>T. c.C1043T (p.P348L). c.T1865C (p.L622P). c.C1760T (p.A587V). c.T1258C (p.C420R). c.G1961A (p.R654H). c.G1886T (p.G629V)
● ● ● ●	Globoid Cell Leukodystrophy (GALC)	10	c.C1586T (p.T529M). c.A2002C (p.T688P). c.G857A (p.G286D). c.1161+6555_9573del31670bp. c.G1153T (p.E385X). c.A1700C (p.Y567S). c.A913G (p.I305V). c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP). c.1472delA (p.K491fs). c.A246G (p.I82M)
● ● ● ●	Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD)	4	c.C653T (p.S218F). c.G1093A (p.A365T). c.G1466C (p.R489P). c.G1466T (p.R489L)
● ● ● ●	Glutaric Acidemia: Type I (GCDH)	2	c.C1262T (p.A421V). c.C1204T (p.R402W)
● ● ● ●	Glycine Encephalopathy: AMT Related (AMT)	6	c.826G>C (p.D276H). c.125A>G (p.H42R). c.878-1G>A. c.959G>A (p.R320H). c.574C>T (p.Q192X). c.139G>A (p.G47R)
● ● ● ●	Glycine Encephalopathy: GLDC Related (GLDC)	5	c.2T>C (p.M1T). c.1545G>C (p.R515S). c.2284G>A (p.G762R). c.2266_2268delTTC (p.756delF). c.1691G>T (p.S564I)
● ● ● ●	Glycogen Storage Disease: Type IA (G6PC)	11	c.A113T (p.D38V). c.C1039T (p.Q347X). c.79delC. c.377insTA. c.979_981delTTC (p.327delF). c.G562C (p.G188R). c.C724T (p.Q242X). c.C247T (p.R83C). c.G248A (p.R83H). c.G809T (p.G2 ⁻ 0V). c.G648T
● ● ● ●	Glycogen Storage Disease: Type IB (SLC37A4)	5	c.1042_1043delICT. c.G1015T (p.G339C). c.G1099A (p.A367T). c.G1016A (p.G339D). c.T352C (p.W118R)
● ● ● ●	Glycogen Storage Disease: Type II (GAA)	15	c.525delT (p.E176RfsX45). c.T-45G. c.G1561A (p.E521K). c.C710T (p.A237V). c.C2560T (p.R854X). c.T896G (p.L299R). c.C1935A (p.D645E). c.T953C (p.M318T). c.2481+110_2646+39del538 (p.G828_N882del55). c.C1634T (p.P545L). c.G1927A (p.G643R). c.1585_1586delTCT (p.S529V). c.C1935A (p.D645E). c.C2173T (p.R725V). c.2707_2709delIK (p.903delIK)
● ● ● ●	Glycogen Storage Disease: Type III (AGL)	14	c.G3980A (p.W1327X). c.4455delT. c.1384delG (p.V462X). c.C2590T (p.R864X). c.17_18delAG. c.3965delIT (p.V1322AfsX27). c.A3439G (p.R1147G). c.C16T (p.Q6X). c.C3682T (p.R1228X). c.C1222T (p.R408X). c.G2039A (p.W680X). c.G4342C (p.G1448R). c.2681+1G>A. c.4260-12A>G
● ● ● ●	Glycogen Storage Disease: Type IV (GBE1)	1	c.A986C (p.Y329S)
● ● ● ●	Glycogen Storage Disease: Type V (PYGM)	10	c.2128_2130delITTC (p.710delF). c.T2392C (p.W798R). c.G613A (p.G205S). c.T2392C (p.W798R). c.A1627T (p.K543X). c.G1827A (p.K609K). c.C255A (p.Y85X). c.A1628C (p.K543T). c.C148T (p.R50X). c.G1827A (p.K609K)
● ● ● ●	Glycogen Storage Disease: Type VII (PFKM)	3	c.C283T (p.R95X). c.G116T (p.R39L). c.593+1G>A

Groups	Disease	#	Mutations
● ● ● ● ●	GM1-Gangliosidosis (GLB1)	16	c.A1772G (p.Y591C), c.C145T (p.R49C), c.C1369T (p.R457X), c.A947G (p.Y316C), c.C202T (p.R68W), c.C245T (p.T82M), c.C601T (p.R201C), c.T152C (p.I51T), c.G1370A (p.R457Q), c.C622T (p.R208C), c.C1051T (p.R351X), c.G367A (p.G123R), c.G176A (p.R59H), c.T1771A (p.Y591N), c.75+2_75+3insT, c.1480-2A>G
● ● ● ● ●	GRACILE Syndrome (BCS1L)	12	c.G464C (p.R155P), c.A232G (p.S78G), c.G1057A (p.V353M), c.G103C (p.G35R), c.A148G (p.T50A), c.C246T (p.R45C), c.C166T (p.R56X), c.C296T (p.P99L), c.C547T (p.R183C), c.G548A (p.R183H), c.C550T (p.R184C), c.G830A (p.S277N)
● ● ● ● ●	Guanidinoacetate Methyltransferase Deficiency (GAMT)	5	c.309_310insCCGGGACTGGGCC (p.L99_A103fs), c.59G>C (p.Trp20Ser), c.148A>C (p.M50L), c.506G>A (p.C169Y), c.327G>A
● ● ● ● ●	Hemochromatosis: Type 1: HFE Related (HFE)	1	c.G845A (p.C282Y)
● ● ● ● ●	Hemochromatosis: Type 2A: HFE2 Related (HFE2)	1	c.G959T (p.G320V)
● ● ● ● ●	Hemochromatosis: Type 3: TFR2 Related (TFR2)	5	c.88_89insC (p.E60X), c.A2069C (p.Q690P), c.84insC, c.C750G (p.Y250X), c.T515A (p.M172K)
● ● ● ● ●	Hemoglobinopathy: Hb C (HBB)	1	c.G19A (p.E7K)
● ● ● ● ●	Hemoglobinopathy: Hb D (HBB)	1	c.G364C (p.E122Q)
● ● ● ● ●	Hemoglobinopathy: Hb E (HBB)	1	c.G ⁻ 9A (p.E2 ⁻ K)
● ● ● ● ●	Hemoglobinopathy: Hb O (HBB)	1	c.G364A (p.E122K)
● ● ● ● ●	Hereditary Fructose Intolerance (ALDOB)	10	c.T442C (p.V148R), c.C524A (p.A175D), c.865_867delCTT (p.289delL), c.C720A (p.C240X), c.C1005G (p.N335K), c.T612G (p.Y204X), c.G448C (p.A150P), c.357_360delAAAC, c.C10T (p.R4X), c.C178T (p.R60X)
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3)	1	c.C2116T (p.R706X)
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3)	5	c.C727T (p.Q243X), c.C1903T (p.R635X), c.C124T (p.R42X), c.C430T (p.R144X), c.3024delT
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2)	1	c.C283T (p.R95X)
● ● ● ● ●	Hermansky-Pudlak Syndrome: HPS3 Related (HPS3)	5	c.-3010_21 ⁺ +690del3.9kb, c.1691+2T>G, c.1691+2T>G, c.2589+1G>C, c.C1189T (p.R39 ⁻ W)
● ● ● ● ●	HMG-CoA Lyase Deficiency (HMGCL)	6	c.G122A (p.R41Q), c.561+1G>A, c.109G>T (p.E37X), c.G208C (p.V70L), c.914_915delTT, c.G835A (p.E279K)
● ● ● ● ●	Holocarboxylase Synthetase Deficiency (HLCS)	3	c.1795+5G>A, c.T710C (p.L237P), c.780delG
● ● ● ● ●	Homocystinuria Caused by CBS Deficiency (CBS)	7	c.572C>T (p.T191M), c.G919A (p.G307S), c.T833C (p.I278T), c.G797A (p.R266K), c.C341T (p.A114V), c.T959C (p.V320A), c.C1006T (p.R336C)
● ● ● ● ●	Hunter Syndrome (IDS)	7	c.T1264G (p.C422G), c.C998T (p.S333L), c.A404G (p.K135R), c.C1402T (p.R468W), c.C1327T (p.R443X), c.G1403T (p.R468L), c.G1403A (p.R468Q)
● ● ● ● ●	Hurler Syndrome (IDUA)	6	c.C208T (p.Q70X), c.G266A (p.R89Q), c.G1205A (p.W402X), c.T1960G (p.X654G), c.G979C (p.A327P), c.C1598G (p.P533R)
● ● ● ● ●	Hypohidrotic Ectodermal Dysplasia: X-Linked (EDA)	5	c.C1013T (p.T338M), c.G467A (p.R156H), c.C463T (p.R155C), c.C1072G (p.Q358E), c.C466T (p.R156C)
● ● ● ● ●	Hypophosphatasia (ALPL)	5	c.G571A (p.E191K), c.G1001A (p.G334D), c.A1133T (p.D378V), c.1559delT, c.T979C (p.F327L)
● ● ● ● ●	Inclusion Body Myopathy: Type 2 (GNE)	3	c.G131C (p.C44S), c.T2228C (p.M743T), c.G1807C (p.V603L)
● ● ● ● ●	Isovaleric Acidemia (IVD)	1	c.C941T (p.A314V)
● ● ● ● ●	Joubert Syndrome (TMEM216)	1	c.G35T (p.R12L)
● ● ● ● ●	Juvenile Retinoschisis: X-Linked (RS1)	3	c.G221T (p.G74V), c.G325C (p.G109R), c.G214A (p.E72K)
● ● ● ● ●	Laryngoonychocutaneous Syndrome (LAMA3)	1	c.151_152insG (p.V51GfsX3)
● ● ● ● ●	Leber Amaurosis (LCA5)	3	c.C835T (p.Q279X), c.1476_1477insA (p.P493TfsX1), c.1151delC
● ● ● ● ●	Leigh Syndrome: French-Canadian (LRPPRC)	1	c.C1061T (p.A354V)
● ● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2D (SGCA)	1	c.C229T (p.R77C)
● ● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2E (SGCB)	6	c.C452G (p.T151R), c.G272C (p.R91P), c.T323G (p.L108R), c.G272T (p.R91L), c.C341T (p.S114F), c.T299A (p.M100K)
● ● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2I (FKRP)	1	c.C826A (p.L276I)
● ● ● ● ●	Lipoprotein Lipase Deficiency (LPL)	1	c.G644A (p.G215E)
● ● ● ● ●	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)	2	c.C1132T (p.Q378X), c.G1528C (p.E510Q)
● ● ● ● ●	Luteinizing Hormone Resistance (Leydig Cell Hypoplasia) (LHCGR)	10	c.T1027A (p.C343S), c.T391C (p.C131R), c.C1635A (p.C545X), c.G1060A (p.E354K), c.1822_1827delCTGGTT (p.608_609delLV), c.G1777C (p.A593P), c.G430T (p.V144F), c.C1660T (p.R554X), c.T1627C (p.C543R), c.T1505C (p.L502P)
● ● ● ● ●	Maple Syrup Urine Disease: Type 1A (BCKDHA)	4	c.G730A (p.G244R), c.T1312A (p.Y438N), c.860_867delGAGGCCCC, c.117delC
● ● ● ● ●	Maple Syrup Urine Disease: Type 1B (BCKDHB)	3	c.G832A (p.G2 ⁻ 8S), c.G548C (p.R183P), c.G1114T (p.E3 ⁻ 2X)
● ● ● ● ●	Maple Syrup Urine Disease: Type 3 (DLD)	8	c.G1123A (p.E375K), c.G685T (p.G229C), c.T1178C (p.I393T), c.104_105insA, c.A1081G (p.M361V), c.C1463T (p.P488L), c.A1483G (p.R495G), c.A214G (p.K72E)
● ● ● ● ●	Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	8	c.T199C (p.Y67H), c.A985G (p.K329E), c.C616T (p.R206C), c.G617A (p.C206H), c.G583A (p.G195R), c.C362T (p.T121I), c.G799A (p.G267R), c.C250T (p.L84F)
● ● ● ● ●	Metachromatic Leukodystrophy (ARSA)	14	c.G302A (p.G99D), c.C641T (p.A214V), c.A856C (p.T286P), c.C1226T (p.T411I), c.1204+1G>A, c.G769C (p.D257H), c.C821T (p.T274M), c.459+1G>A, c.G733A (p.G245R), c.G257A (p.R86Q), c.C1130T (p.P3 ⁻ L), c.C1283T (p.P428L), c.T536G (p.I181S), c.C293T (p.S98F)
● ● ● ● ●	Methylmalonic Acidemia: MMAA Related (MMAA)	14	c.266T>C (p.L89P), c.1076G>A (p.R359Q), c.433C>T (p.R145X), c.358C>T (p.Q120X), c.64C>T (p.R22X), c.653G>A (p.G218E), c.988C>T (p.R330X), c.283C>T (p.Q95X), c.161G>A (p.W54X), c.650T>A (p.L217X), c.397C>T (p.Q133X), c.503delC (p.T168MfsX9), c.562G>C (p.G188R), c.733+1G>A

Groups	Disease	#	Mutations
● ● ● ●	Methylmalonic Acidemia: MMAB Related (MMAB)	11	c.403G>A (p.A135T). c.700C>T (p.Q234X). c.556C>T (p.R186W). c.291-1G>A. c.571C>T (p.R191W). c.572G>A (p.R191Q). c.656A>G (p.Y219C). c.568C>T (p.R190C). c.287T>C (p.I96T). c.197-1G>T. c.569G>A (p.R190H)
● ● ● ●	Methylmalonic Acidemia: MUT Related (MUT)	22	c.2150G>T (p.G717V). c.278G>A (p.R93H). c.655A>T (p.N219Y). c.1097A>G (p.N366S). c.281G>T (p.G94V). c.935G>T (p.G312V). c.607G>A (p.G203R). c.1105C>T (p.R369C). c.691T>A (p.Y231N). c.299A>G (p.Y100C). c.313T>C (p.W105R). c.643G>A (p.G215S). c.2099T>A (p.M700K). c.2080C>T (p.R694W). c.284C>G (p.P95R). c.1867G>A (p.G623R). c.322C>T (p.R108C). c.2054T>G (p.L685R). c.5-2C>A (p.A191E). c.1280G>A (p.G42-D). c.521T>C (p.F1-4S). c.1106G>A (p.R369H)
● ● ● ●	Methylmalonic Aciduria and Homocystinuria: Type cbIC (MMACHC)	5	c.482G>A (p.R161Q). c.609G>A (p.W203X). c.331C>T (p.R111X). c.271_273insA (p.R91KfsX14). c.394C>T (p.R132X)
● ● ● ●	MTHFR Deficiency: Severe (MTHFR)	7	c.652G>T (p.V218L). c.1166G>A (p.W389X). c.T1721G (p.V574G). c.T1304C (p.F435S). c.G1408T (p.E470X). c.523G>A (p.A175T). c.474A>T (p.G158G)
● ● ● ●	Mucopolipidosis: Type II/III (GNPTAB)	3	c.C3565T (p.R1189X). c.3503_3504delTC (p.L1168QfsX5). c.T1120C (p.F374L)
● ● ● ●	Mucopolipidosis: Type IV (MCOLN1)	4	c.-1015_78del6433. c.-1015_88del6433. c.G1084T (p.D362Y). c.406-2A>G
● ● ● ●	Muscle-Eye-Brain Disease (POMGNT1)	3	c.1539+1G>A. c.C1478G (p.P493R). c.C1324T (p.R442C)
● ● ● ●	Myotubular Myopathy: X-Linked (MTM1)	4	c.C205T (p.R69C). c.A566G (p.N189S). c.1261-10A>G. c.C721T (p.R241C)
● ● ● ●	Nemaline Myopathy: NEB Related (NEB)	1	c.7434_7536del2502bp
● ● ● ●	Nephrotic Syndrome: Type 1 (NPHS1)	3	c.121_122delCT. c.C3325T (p.R1109X). c.1481delC
● ● ● ●	Nephrotic Syndrome: Type 2 (NPHS2)	27	c.705_713delITCAGAGAG (p.L236_R238del). c.976_977insA (p.T326fsX345). c.412C>T (p.R138X). c.538G>A (p.V180M). c.964C>T (p.R322X). c.779T>A (p.V260E). c.851C>T (p.A284V). c.714G>T (p.R238S). c.353C>T (p.P118L). c.413G>A (p.R138Q). c.274G>T (p.G92C). c.871C>T (p.R291W). c.555delT (p.F185fsX186). c.622G>A (p.A208T). c.948delT (p.A31-L). c.868G>A (p.V290M). c.502C>A (p.R168S). c.503G>A (p.R168H). c.85G>A (p.A29T). c.862G>A (p.A288T). c.479A>G (p.D160G). c.502C>T (p.R168C). c.467_468insT (p.L156fsX166). c.467delT (p.L156fsX180). c.419delG (p.G140fsX180). c.855_856delAA (p.Q285fsX302). c.104_105insG (p.G35fsX69)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN3 Related (CLN3)	3	c.597C>A (p.Y199X). c.461-280_677+382del966 (p.G154AfsX29). c.883G>A (p.E295K)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5)	7	c.1175_1176delAT (p.Y392X). c.G377A (p.C126Y). c.A1121G (p.Y374C). c.G1054T (p.E352X). c.G225A (p.W-5X). c.G335A (p.R112H). c.G835A (p.D2-9N)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN6 Related (CLN6)	10	c.T200C (p.L67P). c.511_513delTAT (p.171delY). c.G308A (p.R103Q). c.C139T (p.L47F). c.460_462delATC (p.I154del). c.G17C (p.R6T). c.G368A (p.G123D). c.214G>T (p.E72X). c.C663G (p.Y221X). c.316_317insC (p.R106PfsX26)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8)	4	c.G88C (p.A30P). c.C70G (p.R24G). c.G789C (p.W263C). c.C610T (p.R204C)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: MFSD8 Related (MFSD8)	2	c.881C>A (p.T294K). c.754+2T>A
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1)	8	c.G322C (p.G108R). c.T29A (p.L10X). c.T656A (p.L219Q). c.C451T (p.R151X). c.G134A (p.C45Y). c.A236G (p.D79G). c.A364T (p.R122W). c.A223C (p.T75P)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: TPP1 Related (TPP1)	9	c.G1094A (p.C365Y). c.T1093C (p.C365R). c.C622T (p.R208X). c.523-1G>C. c.523-1G>A. c.G851T (p.G284V). c.G1340A (p.R477H). c.C616T (p.R206C). c.A857G (p.N286S)
● ● ● ●	Niemann-Pick Disease: Type A (SMPD1)	5	c.C1267T (p.H423Y). c.G1493T (p.R498L). c.G1734C (p.K578N). c.994delC. c.T911C (p.L304P)
● ● ● ●	Niemann-Pick Disease: Type B (SMPD1)	2	c.1828_1830delCGC (p.610delR). c.C880A (p.Q294K)
● ● ● ●	Niemann-Pick Disease: Type C1 (NPC1)	14	c.G2848A (p.V950M). c.T3182C (p.I1061T). c.G2974C (p.G992R). p.T1133C (p.V378A). c.C2932T (p.R978C). c.G2665A (p.V889M). c.G530A (c.C117Y). c.A3263G (p.Y1088C). c.T337C (p.C113R). c.A2783C (p.Q928P). c.A3467G (p.N1156S). c.A2324C (p.Q775P). c.C3107T (p.T1036M). c.G29-4T (p.G992W)
● ● ● ●	Niemann-Pick Disease: Type C2 (NPC2)	11	c.G58T (p.E20X). c.C358T (p.P120S). c.C133T (p.Q45X). c.C141A (p.C47X). c.332delA (p.N111fs). c.C436T (p.Q146X). c.G352T (p.E118X). c.190+5G>A. c.T295C (p.C99R). c.G115A (p.V39M). c.T199C (p.S67P)
● ● ● ●	Nijmegen Breakage Syndrome (NBN)	8	c.C643T (p.R215W). c.654_658delAAAAC (p.K219fs). c.698_701delAACA (p.K233SfsX4). c.C976T (p.Q326X). c.C1089A (p.Y363X). c.742insGG (p.E248GfsX5). c.835_838delCAGA (p.Q279PfsX1). c.1142delC (p.P381QfsX22)
● ● ● ●	Nonsyndromic Hearing Loss and Deafness: DFNB1 Related (GJB2)	14	c.G551C (p.R184P). c.358delGAG (p.120delE). c.G231A (p.W77X). c.C427T (p.R143W). c.G71A (p.W24X). c.T229C (p.W-TR). c.C3-0T (p.Q124X). c.T269C (p.L90P). c.T101C (p.M34T). c.16-delT. c.35delG. c.235delC. c.312_325delGAAGTTCATCAAGG. c.G109A (p.V37I)
● ● ● ●	Ornithine Transcarbamylase Deficiency (OTC)	3	c.C274T (p.R92X). c.C533T (p.T178M). c.G77A (p.R26Q)
● ● ● ●	Ornithine Translocase Deficiency (SLC25A15)	4	c.C95G (p.T32R). c.562_564delTTC (p.188delF). c.G824A (p.R275Q). c.C535T (p.R179X)
● ● ● ●	Pendred Syndrome (SLC26A4)	5	c.A1246C (p.T416P). c.T707C (p.L236P). c.A1151G (p.E384G). c.1001+1G>A. c.A2168G (p.H723R)
● ● ● ●	Persistent Mullerian Duct Syndrome: Type 1 (AMH)	2	c.C571T (p.R191X). c.G1144T (p.E382X)
● ● ● ●	Persistent Mullerian Duct Syndrome: Type II (AMHR2)	5	c.G1217A (p.R406Q). c.1330_1356delCTGGGCAATACCCCTACCTCTGATGAG. c.232+1G>T. c.G-42A (p.E248K). c.596delA
● ● ● ●	Phenylalanine Hydroxylase Deficiency (PAH)	17	c.C842T (p.P281L). c.T896G (p.F299C). c.C117G (p.F39L). c.G473A (p.R158Q). c.A1241G (p.Y414C). c.A1G (p.M1V). c.C1222T (p.R408W). c.G838A (p.E280K). c.C754T (p.R252W). c.G3A (p.M1I). c.G1223A (p.R408Q). c.1315+1G>A. c.1066-11G>A. c.T143C (p.L48S). c.G814T (p.G272X). c.G782A (p.R261Q). c.T194C (p.I65T)
● ● ● ●	Polyglandular Autoimmune Syndrome: Type I (AIRE)	5	c.1163_1164insA (p.Met388IlefsX36). c.C769T (p.R257X). c.C415T (p.R139X). c.A254G (p.Y85C). c.967_979delCTGCCCTCCGCG (p.Leu323SerfsX51)

Groups	Disease	#	Mutations
●●●●●	Primary Hyperoxaluria: Type 1 (AGXT)	12	c.G738A (p.W246X). c.G508A (p.G170R). c.G698A (p.R233H). c.33insC. c.G121A (p.G41R). c.C697T (p.R233C). c.T454A (p.F152I). c.T613C (p.S205P). c.T731C (p.I244T). c.G245A (p.G82E). c.C198G (p.Y66X). c.G466A (p.G156R)
●●●●●	Primary Hyperoxaluria: Type 2 (GRHPR)	3	c.C295T (p.R99X). c.103delG. c.404+3delAAGT
●●●●●	Primary Hyperoxaluria: Type 3 (HOGA1)	2	c.G860T (p.G287V). c.944_946delAGG (p.315delE)
●●●●●	Progressive Familial Intrahepatic Cholestasis: Type 2 (ABCB11)	5	c.C1723T (p.R575X). c.G1295C (p.R432T). c.C3169T (p.R1057X). c.A890G (p.E297G). c.3767_3768insC
●●●●●	Propionic Acidemia: PCCA Related (PCCA)	4	c.93~C>T (p.R313X). c.1685C>G (p.S562X). c.862A>G (p.R288G). c.1196G>A (p.R399Q)
●●●●●	Propionic Acidemia: PCCB Related (PCCB)	13	c.1495C>T (p.R499X). c.280G>T (p.G94X). c.1228C>T (p.R410W). c.502G>A (p.E168K). c.1606A>G (p.N536D). c.1556T>C (p.L519P). c.1539_1540insCCC (p.R514PfsX38). c.335G>A (p.G112D). c.457G>C (p.A153P). c.1304A>G (p.Y435C). c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA (p.G407fs). c.1283C>T (p.T428I). c.1534C>T (p.R512C)
●●●●●	Pseudocholinesterase Deficiency (BCHE)	1	c.A293G (p.D98G)
●●●●●	Pycnodysostosis (CTSK)	2	c.T926C (p.L309P). c.A990G (p.X330W)
●●●●●	Pyruvate Dehydrogenase Deficiency: Autosomal Recessive (PDHB)	2	c.C1030T (p.P344S). c.A395G (p.Y132C)
●●●●●	Pyruvate Dehydrogenase Deficiency: X-Linked (PDHA1)	4	c.A648C (p.L216F). c.1145_1146insATCA. c.C787G (p.R263G). c.G1133A (p.R378H)
●●●●●	Retinitis Pigmentosa: Autosomal Recessive: DHDDS Related (DHDDS)	1	c.A124G (p.K42E)
●●●●●	Rhizomelic Chondrodysplasia Punctata: Type I (PEX7)	8	c.T875A (p.L292X). c.12_18dupGTGCGGT (p.G7VfsX51). c.G649C (p.G217R). c.T345G (p.Y115X). c.903+1G>C. c.C120G (p.Y40X). c.C653T (p.A218V). c.A40C (p.T14P)
●●●●●	Salla Disease (SLC17A5)	5	c.802_816delTCATCATTAAGAAAT (p.Leu336fsX13). c.C1001G (p.P334R). c.A406G (p.K136E). c.A548G (p.H183R). c.C115T (p.R39C)
●●●●●	Sandhoff Disease (HEXB)	3	c.~6delA. c.445+1G>A. c.850C>T (p.R284X)
●●●●●	SCID: X-Linked (IL2RG)	12	c.G854A (p.R285Q). c.G341A (p.G114D). c.T878A (p.L293Q). c.C664T (p.R222C). c.C923A (p.S308X). c.T186A (p.C62X). c.C865T (p.R289X). c.T515C (p.L172P). c.T458A (p.I153N). c.454+1G>A. c.A292T (p.K98X). c.T343C (p.C115R)
●●●●●	Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	5	c.C1058T (p.S353L). c.C391T (p.R107C). c.C575T (p.A192V). c.C1138T (p.R380W). c.C1147T (p.R383C)
●●●●●	Sickle-Cell Anemia (HBB)	1	c.A20T (p.E7V)
●●●●●	Sjogren-Larsson Syndrome (ALDH3A2)	2	c.129~_1298delIGA (p.E433fs). c.C943T (p.P315S)
●●●●●	Smith-Lemli-Opitz Syndrome (DHCR7)	20	c.724C>T (p.R242C). c.964+1G>C. c.725G>A (p.R242H). c.A356T (p.H119L). c.506C>T (p.S169L). c.C1054T (p.R352W). c.906C>G (p.F302L). c.1228G>A (p.G410S). c.1342G>A (p.E448K). c.G1055A (p.R352Q). c.G1337A (p.R446Q). c.G744T (p.W248C). c.T470C (p.L157P). c.G453A (p.W151X). c.C1210T (p.R404C). c.T326C (p.L109P). c.C278T (p.T93M). c.G1139A (p.C380Y). c.G976T (p.V326L). c.G452A (p.W151X)
●●●●●	Spinal Muscular Atrophy: SMN1 Linked (SMN1)	23	DEL EXON 7. c.683T>A (p.L228X). c.43C>T (p.Q15X). c.734C>T (p.P245L). c.439_443delGAAGT. c.558delA. c.400G>A (p.E134K). c.815A>G (p.Y272C). c.836G>T. c.835G>T. c.823G>A (p.G275S). c.835-18_835-12delCCTTTAT. c.5C>G (p.A2G). c.~40_~41insC. c.~68_~8dupTGCTGATGCTT. c.509_510delGT. c.22_23insA. c.821C>T (p.T274I). c.305G>A (p.W102X). c.81_81+1insG. c.91_92insT. c.834+2T>G. c.585_586insT
●●●●●	Stuve-Wiedemann Syndrome (LIFR)	9	c.756_757insT (p.K253X). c.170delC. c.653_654insT. c.2434C>T (p.R812X). c.2472_2476delTATGT. c.1620_1621insA. c.1601+1G>A. c.1789C>T (p.R597X). c.2274_2275insT
●●●●●	Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2)	7	c.T1957A (p.C653S). c.C532T (p.R178X). c.G764A (p.G255E). c.C835T (p.R279W). c.1018_1020delIGTT (p.340delV). c.C398T (p.A133V). c.699+2T>C
●●●●●	Tay-Sachs Disease (HEXA)	32	c.~2564_253+5128del~945insG. c.12~8_12~9insTATC. c.10~4-1G>T. c.A1003T (p.I335F). c.1073+1G>A. c.913_915delTTC (p.305delF). c.T116G (p.L39R). c.T538C (p.Y180H). c.C532T (p.R178C). c.A611G (p.H204R). c.G509A (p.R170Q). c.C508T (p.R170W). c.C1510T (p.R504C). c.613delC. c.G1496A (p.R499H). c.G749A (p.G250D). c.C629T (p.S210F). c.G78A (p.W26X). c.G598A (p.V200M). c.T632C (p.F211S). c.G533T (p.R178L). c.571-1G>T. c.A590C (p.K197T). c.A1G (p.M1V). c.1421+1G>C. c.805+1G>A. c.T380G (p.L127R). c.G533A (p.R178H). c.G805A (p.G269S). c.346+1G>C. c.C540G (p.Y180X). c.C409T (p.R137X)
●●●●●	Tyrosine Hydroxylase Deficiency (TH)	1	c.G698A (p.R233H)
●●●●●	Tyrosinemia: Type I (FAH)	10	c.~0~1G>C. c.G1009A (p.G33~S). c.G1069T (p.E35~X). c.698A>T (p.D233V). c.554-1G>T. c.G786A (p.W262X). c.C782T (p.P261L). c.607-6T>G. c.707-1G>C. c.1062+5G>A
●●●●●	Usher Syndrome: Type 1B (MYO7A)	12	c.634C>T (p.R212C). c.3719G>A (p.R1240Q). c.1996C>T (p.R666X). c.C448T (p.R150X). c.635G>A (p.R212H). c.700C>T (p.Q234X). c.2476G>A (p.A826T). c.905G>A (p.R302H). c.1884C>A (p.C628X). c.1797G>A (p.M599I). c.6025delG (p.A2009fs). c.5581C>T (p.R1861X)
●●●●●	Usher Syndrome: Type 1C (USH1C)	5	c.91C>T (p.R31X). c.216G>A (p.V72fs). c.IVS5+1G>A. c.238_239insC. c.IVS1+1G>T
●●●●●	Usher Syndrome: Type 1D (CDH23)	14	c.4069C>T (p.Q1357X). c.4488G>C (p.Q1496H). c.3617C>G (p.P1206R). c.4504C>T (p.R1502X). c.3880C>T (p.Q1294X). c.849~C>G (p.R2833G). c.630~G>T (p.E2103X). c.523~G>A (p.R1~46Q). c.8230G>A (p.G2744S). c.7549A>G (p.S2517G). c.172C>T (p.Q58X). c.3367C>T (p.Q1123X). c.5985C>A (p.Y1995X). c.9524G>A (p.R3175H)
●●●●●	Usher Syndrome: Type 1F (PCDH15)	3	c.C2052A (p.Y684X). c.A5557C (p.M1853L). c.C733T (p.R245X)
●●●●●	Usher Syndrome: Type 2A (USH2A)	9	c.1000C>T (p.R334W). c.1256G>T (p.C419F). c.14020A>G (p.R4674G). c.923_924insGCCA (p.H308fs). c.239_240insGTAC. c.2299delG (p.E767SfsX21). c.12067-2A>G. c.2209C>T (p.R737X). c.2276G>T (p.C759F)
●●●●●	Usher Syndrome: Type 3A (CLRN1)	3	c.T528G (p.Y176X). c.T359A (p.M120K). c.T144G (p.N48K)

Groups	Disease	#	Mutations
● ● ● ●	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	8	c.1372T>C (p.F458L), c.779C>T (p.T260M), c.1405C>T (p.R469W), c.848T>C (p.V283A), c.1322G>A (p.G441D), c.1144A>C (p.K382Q), c.1226C>T (p.T409M), c.1837C>T (p.R613W)
● ● ● ●	Walker-Warburg Syndrome (FKTN)	1	c.1167insA (p.F390fs)
● ● ● ●	Wilson Disease (ATP7B)	7	c.G2337A (p.W779X), c.1340delAAAC, c.G2336A (p.W779X), c.G2333T (p.R778L), c.2304delC, c.C2332G (p.R778G), c.C3207A (p.H1069Q)
● ● ● ●	Wolman Disease (LIPA)	6	c.C964T (p.Q322X), c.G260T (p.G87V), c.966+1G>A, c.229+1G>A, c.419G>A (p.W140X), c.796G>T (p.G266X)
● ● ● ●	Zellweger Spectrum Disorders: PEX10 Related (PEX10)	2	c.~64_~65insA, c.8~4_8~5delCT
● ● ● ●	Zellweger Spectrum Disorders: PEX1 Related (PEX1)	3	c.G2528A (p.G843D), c.2097insT (p.I700fs), c.2916delA (p.G973fs)



Patient Information

Patient: KARIM MOOLANI
DOB: 12/18/1986
Gender: Male
Ethnicity: SOUTH ASIAN
Procedure ID: 5246
Report Date: 02/25/2014
Report Updated: 03/10/2014

Sample Information

Specimen Type: Blood
Specimen Number: 6345
Barcode: 20140114823560
Date Specimen Collected: 02/12/2014
Date Specimen Received: 02/14/2014
Date Specimen Analyzed: 02/14/2014

Ordering Practice

Practice Code: 34
New York University School of Medicine
660 First Ave at 38 St
New York, NY 10016
Physician: Frederick Licciardi

Summary of Genetic Testing Results

You did not test positive for any of the mutations assayed.

Disease Groups

High Impact	Treatment Benefits	X-Linked	Moderate Impact
These diseases have a significant impact on life expectancy and quality of life.	Treatment lessens disease symptoms. Newborn screening may be available for timely intervention.	These diseases are passed down by female carriers. Carriers may have symptoms.	These diseases typically do not affect life expectancy but can affect quality of life.

Summary of Testing Performed

Diseases Tested: 191	Mutations Tested: 1476	Genes Tested: 180
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All other mutations analyzed by Recombine were not detected. This reduces but does not eliminate your chance to be a carrier for the associated genetic diseases. Recombine does not screen for every possible genetic disease.

Learn More

A list of all the genetic diseases and mutations you were screened for is included in the latter pages of this report. For disease information, please visit www.recombine.com/our-test. To speak with a Genetic Counselor, call 855.OUR.GENES

Lab Technician: Bo Chu

Reviewed by: Pere Colls, PhD, HCLD

*Methods and Limitations: Recombine developed this genetic assay using the Illumina Infinium Custom HD Genotyping Assay. The test is intended for clinical preconception and/or prenatal screening purposes and is not validated for detection of homozygous mutations. False positive or negative results may occur for reasons that include: genetic variants, assay limitations, sample mix-up, sample contamination, and molecular and technical errors. Recombine tests for Spinal Muscular Atrophy 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).

Recombine
32 Union Square East #1008
New York, NY 10003
855.687.4363

Patient Name: Karim Moolani

Date of Birth: 12/18/1986

Date of Phone Consultation: 3/5/2014

Name of Genetic Counselor: Shabnam Asgari

Dear Dr. Frederick Licciardi,

I spoke with Karim Moolani and Dana Levy Moolani today via phone consultation regarding their Recombine genetic testing results; specifically that he was not identified as a carrier for any of the conditions included on the NYUFC CarrierMap screening panel. Dana Levy Moolani was identified as a carrier of Glycogen Storage Disease: Type II.

Medical and Family History:

- Karim Moolani is a 27-year old male of Indian descent currently in good health. Dana Levy Moolani is a 33-year old female of Ashkenazi Jewish descent. Dana reported that she has Cystinuria, but is currently in good health. She reportedly had one pregnancy termination with a previous partner. Dana reported that she is currently pregnant, this is the couples first pregnancy.

Dana has one brother who is reportedly healthy, it is unknown if he has cystinuria. Dana reported that her brother is looking into getting testing.

Karim reported that one of his paternal uncles passed away in his late 20s, he reportedly had epilepsy, his exact cause of death was not known. Karim's paternal grandmother passed away at 50-years of age from breast cancer diagnosed in her late 40s. No other cancer was reported in the family history.

We discussed the natural history and etiology of cystinuria. We discussed that this disease is not part of the Recombine panel; therefore, the couple was not tested for this disease. We discussed that cystinuria is inherited in an autosomal recessive manner, meaning that an affected individual needs to have two non-working copies of their gene to be affected. We discussed that if Karim is found to be a carrier the couple would have a 1 in 2 (or 50%) chance to have an affected child. We discussed that carrier testing for this condition is available through Mayo Clinic and Mount Sinai School of Medicine. We discussed that Dana's

brother has a 1 in 4 (or 25%) chance to be affected as well, or a 2 in 3 (or 66%) chance to be a carrier and unaffected.

We discussed that seizures affect approximately 1% of the population and can be genetic or non-genetic (eg. illness and/or trauma). Seizures with a genetic origin may be isolated or may be part of a genetic syndrome. If a seizure is due to a genetic syndrome, the recurrence risk is based on that condition's inheritance pattern. Most seizure disorders are not caused by a genetic syndrome and are believed to result from multifactorial inheritance (genes and environmental interaction). The exact risk depends on the type of seizure and age of onset of the parent, the risk to offspring is higher when mothers rather than fathers have seizures [1]. Prenatal diagnosis for these types of seizures is not currently available. Non-genetic causes of recurrent seizures are due to environmental factors such as head trauma, tumors of the brain and infections and would not increase the risk of recurrence for other family members above the general population rate. The exact cause for his uncle's seizures was not known, we discussed that if they are non-genetic in etiology, then the couple's risk of having a child with a seizure disorder should not be increased over the general population background risk.

We discussed that most cancer is sporadic, only a small minority arise due to a cancer predisposition. We discussed that genetic testing for cancer is typically done in an affected individual and cancer screening begins 10-years earlier than the youngest cancer diagnosis in the family. We discussed that Karim should speak with his physician(s) regarding the family history and followup as recommended.

The reported family history is otherwise noncontributory for intellectual disabilities, learning disabilities, blindness, deafness, seizure disorders, birth defects, known genetic conditions, infant deaths, infertility, recurrent pregnancy loss and consanguinity.

Recombine Results:

- **Glycogen Storage Disease: Type II**

Dana was identified as a carrier for Glycogen Storage Disease Type II, otherwise known as Pompe disease. This condition is caused by a buildup of a complex sugar called glycogen in the body's cells, particularly damaging the muscles. In the classic form of infantile-onset Pompe, infants exhibit muscle weakness and heart defects within months after birth. If untreated, patients die from heart failure in their first year. The non-classic form of infantile-onset Pompe disease appears by age 1 and symptoms include delayed motor skills and muscle weakness leading to serious breathing problems. Affected children live only into early childhood. The late-onset type of Pompe disease appears in late childhood or adulthood and causes progressive muscle weakness in the legs, trunk, and muscles that control breathing. Affected patients die from respiratory failure in their 20-30s. Unfortunately, there is no cure for Pompe disease. Treatment

typically involves managing the symptoms of the condition, as well as nutritional and respiratory support.

c.T-45G: Dana was identified to carry the c.T-45G mutation in the GAA gene responsible for Pompe disease. This mutation is typically associated with the late-onset form of Pompe disease [2].

Reproductive Risk: Karim was not identified as a carrier for Pompe disease through the Recombine test. We discussed that this result reduces but does not eliminate the chance for this couple to have a child with Pompe disease. We discussed that the carrier frequency of this condition is not known for the general population or for individuals of Indian descent. We discussed that the Recombine detection rate for this condition in the general population is approximately 10%. We discussed that the couple's risk of having an affected child is difficult to quantify without a known carrier rate for this condition. We discussed the risks, benefits, and limitations of gene sequencing as an additional option to further refine the couple's chance to have a child with this condition. Gene sequencing is a more comprehensive test that can detect more disease-causing mutations; however, gene sequencing can also detect variants of uncertain significance (VUS) which are difficult to interpret clinically. Gene sequencing for Pompe disease is available through Prevention Genetics, GeneDx, and Emory Genetics Laboratory. Gene sequencing for Pompe disease is available through Prevention Genetics, GeneDx, and Emory Genetics Laboratory. Prevention Genetics offers the lowest cost at \$880. The couple indicated that they will discuss these results further.

- **Recombine Test**

We discussed that the Recombine test looked for mutations, or disease-causing changes, in genes associated with 191 recessive genetic diseases for Karim and 213 for Dana. Basic genetics including autosomal recessive inheritance were reviewed with the couple. The couple were not identified as carriers for any other conditions included on the NYUFC CarrierMap screening panel. We discussed that while the Recombine test significantly reduces the chance that someone is a carrier for the conditions tested, it does not completely eliminate that chance, which is less than 1%. We also discussed that the Recombine test does not test for every known genetic disease. This test does not look for chromosome problems, such as Down syndrome, and is not a substitute for routine prenatal care.

- The patient's questions and concerns were addressed.

Plan & Follow-Up

- Karim Moolani was not identified as a carrier for any of the conditions included on the NYUFC CarrierMap screening panel. Dana Levy Moolani was identified as a carrier of Glycogen Storage Disease: Type II. The couple indicated that they will discuss these results further and then decide if they want to pursue further genetic testing.

It was a pleasure speaking with this patient this day. Please do not hesitate to contact me with any other questions or concerns.



Shabnam Asgari, M.S., C.G.C.
Genetic Counselor

References:

- [1] Ottman et al., **Higher risk of seizures in offspring of mothers than of fathers with epilepsy.** American journal of human genetics, 1988. PubMed PMID: 3414683.
- [2] Hermans et al., **Twenty-two novel mutations in the lysosomal alpha-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II.** Human mutation, 2004. PubMed PMID: 14695532.

Diseases & Mutations Assayed

Groups	Disease	#	Mutations
● ● ● ●	17-Alpha-Hydroxylase Deficiency (CYP17A1)	17	c.T340G (p.F114V), c.T985G (p.Y329D), c.T601A (p.Y201N), c.T1216C (p.W406R), c.157_159delITC (p.S3delF), c.C1039T (p.R347C), c.A347T (p.D116V), c.G51A (p.W17X), c.C1024A (p.P342T), c.C715T (p.R239X), c.G287A (p.R96Q), c.G1073A (p.R358Q), c.C286T (p.R96W), c.G1040A (p.R347H), c.C1084T (p.R362C), c.C81A (p.Y27X), c.T316C (p.S106P)
● ● ● ●	17-Beta-Hydroxysteroid Dehydrogenase Type III Deficiency (HSD17B3)	8	c.C238T (p.R80W), c.A389G (p.N130S), c.G166A (p.A56T), c.A703G (p.M235V), c.G803A (p.C268Y), c.G239A (p.R80Q), c.C608T (p.A203V), c.C695T (p.S232L)
● ● ● ●	21-Hydroxylase-Deficient Congenital Classical Adrenal Hyperplasia (CYP21A2)	4	c.293-13C>G, c.G877A (p.G293S), c.332_339delGAGACTAC, c.G1273A (p.G425S)
● ● ● ●	21-Hydroxylase-Deficient Congenital Nonclassical Adrenal Hyperplasia (CYP21A2)	3	c.A188T (p.H63L), c.G844C (p.V282L), c.C1360T (p.P454S)
● ● ● ●	3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency (HSD3B2)	8	c.G512A (p.W171X), c.C1022T (p.P341L), c.A1119C (p.X373C), c.G424A (p.E142K), c.C745T (p.R249X), c.742_747delGTCCGAGinsAACTA (p.V248NfsR249X), c.C29A (p.A10E), c.C776T (p.T259M)
● ● ● ●	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC1 (MCCA) Related (MCCC1)	2	c.T1310C (p.L43 ^{TP}), c.A1155C (p.R385S)
● ● ● ●	3-Methylcrotonyl-CoA Carboxylase Deficiency: MCCC2 (MCCB) Related (MCCC2)	8	c.A1309G (p.I437V), c.G803C (p.R268T), c.G295C (p.E99Q), c.G838T (p.D280Y), c.C929G (p.P310R), c.T499C (p.C167R), c.A569G (p.H190R), c.G464A (p.R155Q)
● ● ● ●	3-Methylglutaconic Aciduria: Type 3 (OPA3)	5	c.313C>G (p.Q105E), c.143-1G>C, c.320_337delAGCAGCGCCACAAAGGAGG (p.Q108_E113del), c.415C>T (p.Q139X), c.277G>A (p.G93S)
● ● ● ●	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS)	6	c.347A>G (p.D116G), c.155A>G (p.N52S), c.259C>T (p.P87S), c.286G>A (p.D96N), c.46C>T (p.R16C), c.74G>A (p.R25Q)
● ● ● ●	Abetalipoproteinemia (MTTP)	2	c.G2593T (p.G865X), c.2212delT
● ● ● ●	Achromatopsia: CNGB3 Related (CNGB3)	6	c.G1208A (p.R403Q), c.991-3T>G, c.1148delC (p.T383fs), c.G1006T (p.E336X), c.817_824delCCAGACT, c.886_896delACTTCTACAAinsT
● ● ● ●	Acrodermatitis Enteropathica (SLC39A4)	10	c.G1120C (p.G374R), c.968-971delAGTC, c.C283T (p.R95C), c.G909C (p.Q303H), c.G989A (p.G330D), c.G1576C (p.G526R), c.1017ins53, c.C318A (p.N106K), c.1223-1227delICGGGG, c.C599T (p.P200L)
● ● ● ●	Acyl-CoA Oxidase I Deficiency (ACOX1)	5	c.A832G (p.M278V), c.372delCATGCCCGCCTGGAACCTT, c.G532T (p.G178C), c.A926G (p.Q309R), c.C442T (p.R148X)
● ● ● ●	Adenosine Deaminase Deficiency (ADA)	22	c.986C>T (p.A329V), c.419G>A (p.G140E), c.454C>A (p.L152M), c.596A>C (p.Q199P), c.445C>T (p.R149W), c.646G>A (p.G216R), c.631C>T (p.R211C), c.536C>A (p.A179D), c.466C>T (p.R156C), c.632G>A (p.R211H), c.301C>T (p.R101W), c.385G>A (p.V129M), c.302G>A (p.R101Q), c.529G>A (p.V177M), c.467G>A (p.R156H), c.320T>C (p.L107P), c.872C>T (p.S291L), c.302G>T (p.R101L), c.248C>A (p.A83D), c.220G>T (p.G74C), c.58G>A (p.G20R), c.43C>G (p.H15D)
● ● ● ●	Alkaptonuria (HGD)	14	c.T899G (p.V300G), c.G808A (p.G270R), c.16-1G>A, c.G481C (p.G161R), c.457_458insG, c.174delA, c.1111_1112insC, c.A1102G (p.M368V), c.342+1G>A, c.C140T (p.S47L), c.C688T (p.P230S), c.G990T (p.R330S), c.T360G (p.C120V), c.457_458insG
● ● ● ●	Alpha-1-Antitrypsin Deficiency (SERPINA1)	4	c.G1096A (p.E366K), c.A1131T (p.L377F), c.226_228delITC (p.76delF), c.C187T (p.R63C)
● ● ● ●	Alpha-Mannosidosis (MAN2B1)	3	c.2248C>T (p.R750W), c.2426T>C (p.L809P), c.1830+1G>C (p.V549_E610del)
● ● ● ●	Alpha Thalassemia (HBA1)	18	11.1kb deletion, c.-5092_95+60del5247bp (Alpha 5.2), Alpha3.7, SEA deletion, c.207C>G (p.N69K), c.427T>C (p.X143Qext32), c.2T>C (p.M1T), Alpha4.2, THAI deletion, HBA2c-, 9111_HBA1c, 2791del16401bp (MED), c.339C>G (p.H113Q), c.377T>C (p.L126P), c.*+94A>G, c.207C>A (p.N69K), c.223G>C (p.D75G), c.94_95delAG (p.R32DfsX24), c.340_351delICTCCCGCCGAG (p.L114_E11 ^{TP} del), c.95+2_95+6delITGAGG
● ● ● ●	Alport Syndrome: COL4A3 Related (COL4A3)	3	c.4415_4419delCTTTT, c.C4571G (p.S1524X), c.C4441T (p.R1481X)
● ● ● ●	Alport Syndrome: COL4A4 Related (COL4A4)	5	c.C3713G (p.S1238X), c.C4715T (p.P1572L), c.C4923A (p.C1641X), c.C4129T (p.R1377X), c.G3601A (p.G1201S)
● ● ● ●	Amegakaryocytic Thrombocytopenia (MPL)	3	c.C127T (p.R43X), c.79+2T>A, c.G305C (p.R102P)
● ● ● ●	Andermann Syndrome (SLC12A6)	5	c.2436delG (p.T813fsX813), c.901delA, c.C2023T (p.R675X), c.C3031T (p.R1011X), c.C619T (p.R207C)
● ● ● ●	Argininosuccinate Lyase Deficiency (ASL)	6	c.A857G (p.Q286R), c.C1135T (p.R379C), c.C283T (p.R95C), c.C1153T (p.R385C), c.G532A (p.V178M), c.446+1G>A
● ● ● ●	Aromatase Deficiency (CYP19A1)	11	c.C1303T (p.R435C), c.G1310A (p.C437Y), c.C1123T (p.R375C), c.G1310A (p.C437Y), c.1222_1224delC, c.G628A (p.E210K), c.451+1G>A, c.468delC, c.G1094A (p.R365Q), c.629-3C>A, c.858+2T>C
● ● ● ●	ARSACS (SACS)	8	c.6273delT, c.8844delT (p.I2949fs), c.T5836C (p.W1946R), c.T9742C (p.W3248R), c.C4933T (p.R1645X), c.T3161C (p.F1054S), c.C12973T (p.R4325X), c.C7504T (p.R2502X)
● ● ● ●	Aspartylglycosaminuria (AGA)	8	c.G488C (p.C163S), c.199_200delGA, c.T214C (p.S72P), c.G482A (p.R161Q), c.T916C (p.C306R), c.G904A (p.G302R), c.C302T (p.A101V), c.G1 ^{TP} 9A (p.G60D)
● ● ● ●	Ataxia-Telangiectasia (ATM)	19	c.5762+1126A>G, c.C103T (p.R35X), c.C7327T (p.R2443X), c.G3576A (p.K1192K), c.3245delATCinsTGAT (p.H1082fs), c.A8030G (p.Y2677C), c.3894insT, c.5712_5713insA (p.S1905fs), c.T7271G (p.V2424G), c.G5932T (p.E1978X), c.C5908T (p.Q1970X), c.A7268G (p.E2423G), c.T8480G (p.F2827C), c.1564_1565delGA (p.E522fs), c.7517_7520delGAGA (p.R2506fs), c.7638_7646delTAGAATTC (p.R2547_S2549delRIS), c.7630-2A>C, c.G7876C (p.A2626P), c.T7967C (p.L2656P)
● ● ● ●	Ataxia with Vitamin E Deficiency (TTPA)	5	c.C400T (p.R134X), c.744delA, c.487_488insTT (p.T163GfsX13), c.G575A (p.R192H), c.T303G (p.H101Q)

Groups	Disease	#	Mutations
● ● ● ● ●	Autosomal Recessive Polycystic Kidney Disease (PKHD1)	17	c.5895insA. c.C1486T (p.R496X). c.T10658C (p.I3553T). c.9689delA. c.3761_3762delCCinsG (p.A1254fs). c.C4991T (p.S1664F). c.T10412G (p.V3471G). c.8870T>C (p.I2957T). c.C1077T (p.T36M). c.C8011T (p.R2671X). c.10174C>T (p.Q3392X). c.9530T>C (p.I3177T). c.6992T>A (p.I2331K). c.G5221A (p.V1741M). c.2414C>T (p.P805L). c.664A>G (p.I222V). c.C9053T (p.S3018F)
● ● ● ● ●	Bardet-Biedl Syndrome: BBS10 Related (BBS10)	3	c.G101C (p.R34P). c.271_273ins1bp (p.C91fsX95). c.T931G (p.S311A)
● ● ● ● ●	Bardet-Biedl Syndrome: BBS12 Related (BBS12)	5	c.1483_1484delGA (p.E495fsX498). c.335_337delTAG. c.1063C>T (p.R355X). c.865G>C (p.A289P). c.1114_1115delITT (p.F3 ⁻ 2X)
● ● ● ● ●	Bardet-Biedl Syndrome: BBS1 Related (BBS1)	3	c.G1645T (p.E549X). c.T1169G (p.M390R). c.851delA
● ● ● ● ●	Bardet-Biedl Syndrome: BBS2 Related (BBS2)	3	c.940delA. c.C72G (p.Y24X). c.T224G (p.V75G)
● ● ● ● ●	Bare Lymphocyte Syndrome: Type II (CLITA)	1	c.G1141T (p.E381X)
● ● ● ● ●	Bartter Syndrome: Type 4A (BSND)	6	c.G139A (p.G47R). c.G3A (p.M1I). c.A1T (p.M1L). c.C22T (p.R8W). c.G23T (p.R8L). c.G28A (p.G10S)
● ● ● ● ●	Beta-Hexosaminidase Pseudodeficiency (HEXA)	2	c.C745T (p.R249W). c.C739T (p.R247W)
● ● ● ● ●	Beta-Ketothiolase Deficiency (ACAT1)	15	c.T2A (p.M1K). c.1006-1G>C. c.G99 ⁻ C (p.A333P). c.1006-2A>C. c.G1138A (p.A380T). c.G1136T (p.G379V). c.A278G (p.N93S). c.G547A (p.G183R). c.C433G (p.Q145E). c.1033_1035delGAA (p.345delE). c.1083insA. c.826+1G>T. c.T99A (p.Y33X). c.T935C (p.I312T). c.C814T (p.Q272X)
● ● ● ● ●	Beta Thalassemia (HBB)	91	c.93-1G>A. c.36delT (p.T13fs). c.17_18delCT. c.155delC (p.P52fs). c.a-79g. c.316-3C>G. c.323_324insG (p.N109fs). c.316-146T>G. c.G169C (p.G57R). c.2T>G (p.M1R). c.T2C (p.M1T). c.46delT (p.W16Gfs). c.316-3C>A. c.316-1G>T. c.316-2A>G. c.a-81g. c.316-2A>C. c.316-106C>T. c.316-1G>C. c.T75A (p.G25G). c.217insA. c.316-1G>A. c.315+745C>G. c.315+2T>C. c.315+705T>G. c.316-197C>T. c.287_288insA (p.L97fs). c.A52T (p.K18X). c.G295A (p.V99M). c.59A>G (p.N20S). c.315+654C>T. c.315+1G>A. c.c-13 ⁻ T. c.c-136g. c.4delG. c.25_26delAA. c.217_221delAGTGinsT (p.S73_D74delinsLfs). c.230delC. c.G34A (p.V12I). c.271G>T (p.E91X). c.223+702_444+342del620insAAGTAGA. c.92+5G>A. c.203_204delTG (p.V68fs). c.45_46insG (p.L16fs). c.c-137g. c.20delA. c.92+5G>T. c.124_129TTCTTT>TT. c.93-1G>T. c.A-50C. c.85_86insC (p.L29fs). c.c-138I. c.92+2T>C. c.135delC (p.F46fs). c.51delC. c.G415C (p.A139P). c.92+110G>A/c.93-21G>A. c.92+2T>A. c.444+110T>C. c.G47A (p.W16X). c.92+1G>A. c.444+111A>G. c.90C>T (p.G30G). c.a-78g. c.g-29a. c.93-15T>G. c.250delG. c.c-151t. c.G48A (p.W16X). c.92+5G>C. c.92+6T>C. c.C118T (p.Q40X). c.1A>G (p.M1V). c.t-80a. c.c-140t. c.c-142t. c.383_385delAGG (p.Q128_A129delQAinsP). c.G114A (p.W38X). c.92+1G>T. c.G113A (p.W38X). c.G92C (p.R31T). c.444+113A>G. c.27_28insG. c.93-1G>C. c.92+1G>C. c.126delC. c.225delC. c.-176_92+25del1293bp. c.68_74delAAGTTGG. c.G82T (p.A28S). c.112delT
● ● ● ● ●	Biotinidase Deficiency (BTD)	10	c.G511A (p.A171T). c.G1330C (p.D444H). c.A1466C (p.N489T). c.A1368C (p.Q456H). c.A755G (p.D252G). c.C1612T (p.R538C). c.C235T (p.R79C). c.G100A (p.G34S). c.T1207G (p.F403V). c.98_104delGCGGCTGinsTCC (p.C33FfsX68)
● ● ● ● ●	Bloom Syndrome (BLM)	9	c.2407insT. c.2207delATCTGAinsTAGATT. c.557_559delCAA (p.S186X). c.1933C>T (p.Q645X). c.1 ⁻ 01G>A (p.W56 ⁻ X). c.C2695T (p.R899X). c.1284G>A (p.W428X). c.C2528T (p.T843I). c.G310 ⁻ T (p.C1036F)
● ● ● ● ●	Canavan Disease (ASPA)	8	c.433-2A>G. c.C693A (p.Y231X). c.C654A (p.C218X). c.C693T (p.Y231Y). c.A692G (p.Y231C). c.C914A (p.A305E). c.A854C (p.E285A). c.A71G (p.E24G)
● ● ● ● ●	Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	7	c.A1079G (p.E360G). c.A1493G (p.Y498C). c.G2129A (p.G710E). c.A1361G (p.D454G). c.C1241T (p.A414V). c.G2126A (p.G709E). c.C1436T (p.P479L)
● ● ● ● ●	Carnitine Palmitoyltransferase II Deficiency (CPT2)	22	c.T1342C (p.F448L). c.T1148A (p.F383Y). c.G1145A (p.R382K). c.A359G (p.Y120C). c.533_556delITGAACCTGCAAAAAGTGACACTA. c.1923_1935delGAAGGCCCTTAGAA. c.G1646A (p.G549D). c.C1810T (p.P604S). c.A1649G (p.Q550R). c.C370T (p.R124X). c.C680T (p.P227L). c.G452A (p.R151Q). c.G520A (p.E174K). c.C1891T (p.R631C). c.C338T (p.S113L). c.A1883C (p.Y628S). c.C149A (p.P50H). c.A983G (p.D328G). c.109_110insGC. c.1238_1239delAG. c.1737delC. c.C1507T (p.R503C)
● ● ● ● ●	Cartilage-Hair Hypoplasia (RMRP)	2	c.G262T. c.A70G
● ● ● ● ●	Cerebrotendinous Xanthomatosis (CYP27A1)	13	c.C1435T (p.R479C). c.G646C (p.A216P). c.C1183T (p.R395C). c.C1420T (p.R474W). c.G434A (p.G145E). c.G583T (p.E195X). c.C379T (p.R127V). c.C819delT. c.G1214A (p.R405Q). c.G1421A (p.R4 ⁻ 4Q). c.C1016T (p.T339M). c.844+1G>A. c.1263+1G>A
● ● ● ● ●	Cholesteryl Ester Storage Disease (LIPA)	4	c.652C>T (p.R218X). c.883C>T (p.H295Y). c.894G>A. c.1024G>A (p.G342R)
● ● ● ● ●	Choreoacanthocytosis (VPS13A)	1	c.6059delC
● ● ● ● ●	Citrullinemia: Type I (ASS1)	16	c.C53T (p.S18L). c.1168G>A (p.G390R). c.C910T (p.R304W). c.C835T (p.R279X). c.G470A (p.R157H). c.G1085T (p.G362V). c.G539A (p.S180N). c.G970A (p.G324S). c.T535C (p.W179R). c.C256T (p.R86C). c.G323T (p.R108L). c.G40A (p.G14S). c.1194-1G>C. c.420+5G>A. c.A928C (p.K310Q). c.C1087T (p.R363V)
● ● ● ● ●	Classical Galactosemia (GALT)	1 ⁻	c.C413T (p.T138M). c.-1039_53del3162. c.134_138delCAGCT. c.820+51_89del2294ins12. c.253-2A>G. c.T584C (p.L195P). c.A626G (p.Y209C). c.A563G (p.Q188R). c.C404T (p.S135L). c.C505A (p.Q169K). c.G855C (p.K285N). c.G607A (p.E203K). c.C997G (p.R333G). c.T1138C (p.X380R). c.T221C (p.L74P). c.T425A (p.M142K). c.T512C (p.F171S)
● ● ● ● ●	Congenital Disorder of Glycosylation: Type 1A: PMM2 Related (PMM2)	3	c.G422A (p.R141H). c.C357A (p.F119L). c.G385A (p.V129M)
● ● ● ● ●	Congenital Disorder of Glycosylation: Type 1B: MPI Related (MPI)	1	c.G884A (p.R295H)
● ● ● ● ●	Congenital Disorder of Glycosylation: Type 1C: ALG6 Related (ALG6)	4	c.T1432C (p.S478P). c.257+5G>A. c.895_897delIATA. c.C998T (p.A333V)
● ● ● ● ●	Congenital Lipoid Adrenal Hyperplasia (STAR)	11	c.178+3T>T. c.C772T (p.Q258X). c.64+1G>T. c.G545A (p.R182H). c.G559A (p.V187M). c.G545T (p.R182L). c.G650C (p.R21 ⁻ T). c.C562T (p.R188C). c.G ⁻ 49A (p.W250X). c.201_202delCT. c.466-11T>A
● ● ● ● ●	Congenital Neutropenia: Recessive (HAX1)	6	c.C568T (p.Q190X). c.121_125insG. c.130insA. c.424insG. c.91delG. c.C256T (p.R86X)

Groups	Disease	#	Mutations
● ● ● ● ●	Corneal Dystrophy and Perceptive Deafness (SLC4A11)	7	c.T2528C (p.L843P). c.A2566G (p.M856V). c.473delGCTTCGCC. c.2233_2240insTATGACAC. c.1378delTACGinsA. c.G1463A (p.R488K). c.T637C (p.S213P)
● ● ● ● ●	Corticosterone Methyloxidase Deficiency (CYP11B2)	3	c.C541T (p.R181W). c.T1382C (p.L461P). c.A1492G (p.T498A)
● ● ● ● ●	Crigler-Najjar Syndrome (UGT1A1)	11	c.C1021T (p.R341X). c.A992G (p.Q331R). c.A1070G (p.Q357R). c.508_513delITTC (p.170delF). c.C991T (p.Q331X). c.C1124T (p.S375F). c.T44G (p.L15R). c.C840A (p.C280X). c.G923A (p.G308E). c.A1198G (p.N400D). c.T524A (p.L175Q)
● ● ● ● ●	Cystic Fibrosis (CFTR)	124	c.1116+1305delIT. c.3690delIT. c.G3454C (p.D1152H). c.C1079A (p.T360K). c.G1558T (p.V520F). c.3 ⁻ 1 ⁻ +12191C>T. c.1116+1304_1305insT. c.T3302A (p.M1101K). c.G350A (p.R11 ⁻ H). c.2052delA (p.K684fs). c.C1075A (p.Q359K). c.579+1G>T. c.3744delA. c.1543delITA. c.2052_2053insA (p.Q685fs). c.2737_2738insG (p.Y913X). c.3768insT. c.274-1G>A. c.G3611A (p.W1204X). c.C349T (p.R117C). c.442delA. c.1973delGAAATTCATCCTinsAGAAA. c.1521_1523delICTT (p.508delF). c.G3752A (p.S1251N). c.C1654T (p.Q552X). c.1519_1521delATC (p.507delI). c.C3196T (p.R1066C). c.C1721A (p.P574H). c.C1657T (p.R553X). c.3535delACCA. c.3527delIC. c.325delITATinsG. c.G1040C (p.R347P). c.C3909G (p.N1303K). c.G3846A (p.W1282X). c.G3808A (p.D12 ⁻ ON). c.261delIT. c.19 ⁻ 6delA (p.N659fs). c.G1040A (p.R34 ⁻ H). c.C14T (p.P5L). c.3536_3539delICCAA (p.T1179fs). c.313delA (p.I105fs). c.G3848T (p.R1283M). c.C2290T (p.R764X). c.G171A (p.W57X). c.3659delIC (p.T1220fs). c.G328C (p.D110H). c.G532A (p.G178R). c.G1055A (p.R352Q). c.3140-26A>G. c.3063delAGTGAT. c.3038delIC. c.G988T (p.G330X). c.2988+1G>A. c.T1090C (p.S364P). c.C2668T (p.Q890X). c.580-1G>T. c.1155insTA. c.1029delIC. c.C1477T (p.Q493X). c.1477delICA. c.803delA (p.N268fs). c.G19T (p.E7X). c.1116+1305_1306delIT. c.T366G (p.Y122X). c.2174insA. c.1766+1G>A. c.T617G (p.L206WV). c.164+12T>C. c.C2125T (p.R709X). c.3067_3072delATAGTG (p.I1023_V1024delIT). c.G3209A (p.R10 ⁻ Q). c.3 ⁻ 1 ⁻ 3 ⁻ 3 ⁻ 4insT (p.L1258fs). c.G2 ⁻ 1A (p.G91R). c.G16 ⁻ 9C (p.R560T). c.G2 ⁻ 4T (p.E92X). c.G1646A (p.S549N). c.1585-1G>A. c.G1624T (p.G542X). c.C1013T (p.T338I). c.G1438T (p.G480C). c.A1645C (p.S549R). c.946delIT. c.931delITTC (p.311delF). c.A2128T (p.K710X). c.1680-1G>A. c.1679+1634A>G. c.C223T (p.R75X). c.C1000T (p.R334WV). c.C1477T (p.Q493X). c.C3472T (p.R1158X). c.C3484T (p.R1162X). c.G1652A (p.G551D). c.G1646T (p.S549I). c.1911delIG. c.804delITA. c.802delA. c.G1675A (p.A559T). c.1766+1G>T. c.579+5G>A. c.1766+5G>T. c.1116+1305_1307delITTT. c.1818del84. c.G1865A (p.G622D). c.531delIT. c.G178T (p.E60X). c.1923delICTCAAACinsA. c.489+1G>T. c.2049insA. c.2049delA. c.1545_1546delITA (p.Y1515X). c.G254A (p.G85E). c.2051delAAinsG. c.T1647G (p.S549R). c.2657+5G>A. c.C3276G (p.Y1092X). c.C1364A (p.A455E). c.273+3A>C. c.273+1G>A. c.C3587G (p.S1196X). c.C3764G (p.S1255X). c.C3712T (p.Q1238X). c.C1572A (p.C524X). c.G3266A (p.W1089X)
● ● ● ● ●	Cystinosis (CTNS)	14	c.G329T (p.G110V). c.G124A (p.V42I). c.G506A (p.G169D). c.G283T (p.G95X). c.C416T (p.S139F). c.198delATTACTATCCTTGGCTCCCC. c.18_21delGACT. c.G589A (p.G197R). c.G414A (p.W138X). c.G613A (p.D205N). c.C969G (p.N323K). c.G1015A (p.G339R). c.T473C (p.L158P). c.-39155_848del57119
● ● ● ● ●	D-Bifunctional Protein Deficiency (HSD1 ⁻ B4)	-	c.63G>T (p.L21F). c.1369A>G (p.N45 ⁻ D). c.G46A (p.G16S). c.31 ⁻ G>C (p.R106P). c.1369A>T (p.N457Y). c.652G>T (p.V218L). c.422_423delIAG
● ● ● ● ●	Diabetes: Recessive Permanent Neonatal (ABCC8)	2	c.G1144A (p.E382K). c.A215G (p.N72S)
● ● ● ● ●	Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	4	c.1905+1G>A. c.1897delIC. c.G2657A (p.R886H). c.295delITCAT
● ● ● ● ●	Du Pan Syndrome (GDF5)	5	c.T1315A (p.S439T). c.1309delTTG. c.T1322C (p.L441P). c.C1306A (p.P436T). c.G1133A (p.R378Q)
● ● ● ● ●	Dystrophic Epidermolysis Bullosa: Recessive (COL7A1)	7	c.T8393A (p.M2798K). c.5821-1G>A. c.G4039C (p.G1347R). c.C933A (p.Y311X). c.7345-1G>A. c.24 ⁻ 0insG. c.42 ⁻ 2A>G
● ● ● ● ●	Ehlers-Danlos Syndrome: Type VIIC (ADAMTS2)	1	c.C673T (p.Q225X)
● ● ● ● ●	Ellis-van Creveld Syndrome (EVC2)	1	c.C3265T (p.Q1089X)
● ● ● ● ●	Enhanced S-Cone (NR2E3)	1	c.G932A (p.R311Q)
● ● ● ● ●	Ethylmalonic Aciduria (ETHE1)	4	c.G3T (p.M1I). c.505+1G>T. c.C487T (p.R163W). c.221_222insA
● ● ● ● ●	Familial Dysautonomia (IKBKAP)	3	c.G2087C (p.R696P). c.C2741T (p.P914L). c.2204+6T>C
● ● ● ● ●	Familial Hyperinsulinism: Type 1: ABCC8 Related (ABCC8)	8	c.4516G>A (p.E1506K). c.C4477T (p.R1493W). c.C4258T (p.R1420C). c.G2147T (p.G716V). c.G4055C (p.R1352P). c.3989-9G>A. c.T560A (p.V187D). c.4159_4161delITTC (p.1387delF)
● ● ● ● ●	Familial Hyperinsulinism: Type 2: KCNJ11 Related (KCNJ11)	6	c.T440C (p.L14 ⁻ P). c.C36A (p.Y12X). c.G844A (p.E282K). c.C ⁻ 61T (p.P254L). c.G-134T. c.A ⁻ 6G (p.H259R)
● ● ● ● ●	Familial Mediterranean Fever (MEFV)	11	c.G2082A (p.M694I). c.G2282A (p.R761H). c.C1437G (p.F479L). c.G2040C (p.M680I). c.G2040A (p.M680I). c.C800T (p.T267I). c.G2230T (p.A744S). c.T2177C (p.V726A). c.A2080G (p.M694V). c.2076_2078delAAT (p.692delI). c.G1958A (p.R653H)
● ● ● ● ●	Familial Mediterranean Fever: Mild Form (MEFV)	3	c.G1223A (p.R408Q). c.A2084G (p.K695R). c.C1105T (p.P369S)
● ● ● ● ●	Fanconi Anemia: Type C (FANCC)	8	c.456+4A>T. c.C553T (p.R185X). c.G65A (p.W22X). c.66delIG. c.T1661C (p.L554P). c.G66A (p.W22X). c.C3 ⁻ T (p.Q13X). c.C1642T (p.R548X)
● ● ● ● ●	Fumarase Deficiency (FH)	1	c.1431_1433insAAA
● ● ● ● ●	Galactokinase Deficiency (GALK1)	6	c.1045G>A (p.G349S). c.1144C>T (p.Q382X). c.238G>T (p.E80X). c.1031C>T (p.T344M). c.82C>A (p.P28T). c.94G>A (p.V32M)
● ● ● ● ●	Gaucher Disease (GBA)	10	c.G1604A (p.R535H). c.T1448C (p.L483P). c.A1343T (p.D448V). c.A1226G (p.N409S). c.G1342C (p.D448H). c.84_85insG. c.1263_1317del55. c.115+1G>A. c.G1297T (p.V433L). c.C1504T (p.R502C)
● ● ● ● ●	Gitelman Syndrome (SLC12A3)	9	c.C622T (p.R208W). c.T1865C (p.L622P). c.G1961A (p.R654H). c.T1258C (p.C420R). c.G1886T (p.G629V). c.1923-1G>T. c.2 ⁻ 44+1G>T. c.C1043T (p.P348L). c.C1 ⁻ 60T (p.A58 ⁻ V)

Groups	Disease	#	Mutations
● ● ● ● ●	Globoid Cell Leukodystrophy (GALC)	10	c.A913G (p.I305V). c.A2002C (p.T688P). c.683_694delATCTCTGGGAGTinsCTC (p.N228_S232del5insTP). c.1472delA (p.K491fs). c.1161+6555_9573del31670bp. c.G857A (p.G286D). c.G1153T (p.E385X). c.C1586T (p.T529M). c.A246G (p.I82M). c.A1700C (p.Y567S)
● ● ● ● ●	Glutaric Acidemia: Type I (GCDH)	2	c.C1204T (p.R402W). c.C1262T (p.A421V)
● ● ● ● ●	Glycine Encephalopathy: AMT Related (AMT)	6	c.826G>C (p.D276H). c.878-1G>A. c.574C>T (p.Q192X). c.959G>A (p.R320H). c.139G>A (p.G47R). c.125A>G (p.H42R)
● ● ● ● ●	Glycine Encephalopathy: GLDC Related (GLDC)	5	c.2266_2268delTTC (p.756delF). c.1545G>C (p.R515S). c.2T>C (p.M1T). c.2284G>A (p.G762R). c.1691G>T (p.S564I)
● ● ● ● ●	Glycogen Storage Disease: Type IA (G6PC)	11	c.377insTA. c.A113T (p.D38V). c.C1039T (p.Q347X). c.79delC. c.979_981delTTC (p.327delF). c.G248A (p.R83H). c.C724T (p.Q242X). c.C247T (p.R83C). c.G809T (p.G270V). c.G562C (p.G188R). c.G648T
● ● ● ● ●	Glycogen Storage Disease: Type IB (SLC37A4)	5	c.1042_1043delICT. c.G1015T (p.G339C). c.G1099A (p.A367T). c.G1016A (p.G339D). c.T352C (p.W118R)
● ● ● ● ●	Glycogen Storage Disease: Type II (GAA)	15	c.2481+110_2646+39del538 (p.G828_N882del55). c.C1634T (p.P545L). c.T-45G. c.C2560T (p.R854X). c.G192A (p.G643R). c.G1561A (p.E521K). c.T953C (p.M318T). c.C1935A (p.D645E). c.C1935A (p.D645E). c.1585_1586delTCT (p.S529V). c.C2173T (p.R725W). c.2707_2709delIK (p.903delIK). c.C710T (p.A237V). c.525delIT (p.E176RfsX45). c.T896G (p.L299R)
● ● ● ● ●	Glycogen Storage Disease: Type III (AGL)	14	c.G2039A (p.W680X). c.C1222T (p.R408X). c.A3439G (p.R1147G). c.1384delG (p.V462X). c.2681+1G>A. c.G3980A (p.W1327X). c.4455delIT. c.3965delIT (p.V1322AfsX27). c.17_18delAG. c.C2590T (p.R864X). c.C16T (p.Q6X). c.C3682T (p.R1228X). c.G4342C (p.G1448R). c.4260-12A>G
● ● ● ● ●	Glycogen Storage Disease: Type IV (GBE1)	1	c.A986C (p.Y329S)
● ● ● ● ●	Glycogen Storage Disease: Type V (PYGM)	10	c.G1827A (p.K609K). c.T2392C (p.W798R). c.2128_2130delTTC (p.710delF). c.G613A (p.G205S). c.T2392C (p.W798R). c.A162T (p.K543X). c.G182A (p.K609K). c.A1628C (p.K543T). c.C148T (p.R50X). c.C255A (p.Y85X)
● ● ● ● ●	Glycogen Storage Disease: Type VII (PFKM)	3	c.C283T (p.R95X). c.G116T (p.R39L). c.593+1G>A
● ● ● ● ●	GM1-Gangliosidosis (GLB1)	16	c.T1771A (p.Y591N). c.G176A (p.R59H). c.G1370A (p.R457Q). c.C245T (p.T82M). c.C1051T (p.R351X). c.A947G (p.Y316C). c.C601T (p.R201C). c.C622T (p.R208C). c.A1772G (p.Y591C). c.T152C (p.I51T). c.G367A (p.G123R). c.C202T (p.R68W). c.C1369T (p.R457X). c.1480-2A>G. c.75+2_75+3insT. c.C145T (p.R49C)
● ● ● ● ●	GRACILE Syndrome (BCS1L)	12	c.G464C (p.R155P). c.A232G (p.S78G). c.C166T (p.R56X). c.G548A (p.R183H). c.C54T (p.R183C). c.G830A (p.S277N). c.G1057A (p.V353M). c.G103C (p.G35R). c.C550T (p.R184C). c.A148G (p.T50A). c.C296T (p.P99L). c.C246T (p.R45C)
● ● ● ● ●	Guanidinoacetate Methyltransferase Deficiency (GAMT)	5	c.309_310insCCGGAGCTGGGCC (p.L99_A103fs). c.148A>C (p.M50L). c.506G>A (p.C169Y). c.59G>C (p.Trp20Ser). c.327G>A
● ● ● ● ●	Hemochromatosis: Type 1: HFE Related (HFE)	1	c.G845A (p.C282Y)
● ● ● ● ●	Hemochromatosis: Type 2A: HFE2 Related (HFE2)	1	c.G959T (p.G320V)
● ● ● ● ●	Hemochromatosis: Type 3: TFR2 Related (TFR2)	5	c.88_89insC (p.E60X). c.84insC. c.T515A (p.M172K). c.C750G (p.Y250X). c.A2069C (p.Q690P)
● ● ● ● ●	Hemoglobinopathy: Hb C (HBB)	1	c.G19A (p.E7K)
● ● ● ● ●	Hemoglobinopathy: Hb D (HBB)	1	c.G364C (p.E122Q)
● ● ● ● ●	Hemoglobinopathy: Hb E (HBB)	1	c.G79A (p.E27K)
● ● ● ● ●	Hemoglobinopathy: Hb O (HBB)	1	c.G364A (p.E122K)
● ● ● ● ●	Hereditary Fructose Intolerance (ALDOB)	10	c.C10T (p.R4X). c.T442C (p.W148R). c.C178T (p.R60X). c.C524A (p.A175D). c.C720A (p.C240X). c.C1005G (p.N335K). c.357_360delAAAC. c.G448C (p.A150P). c.865_867delCTT (p.289delL). c.T612G (p.Y204X)
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMA3 Related (LAMA3)	1	c.C2116T (p.R706X)
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMB3 Related (LAMB3)	5	c.C727T (p.Q243X). c.C1903T (p.R635X). c.C124T (p.R42X). c.3024delIT. c.C430T (p.R144X)
● ● ● ● ●	Herlitz Junctional Epidermolysis Bullosa: LAMC2 Related (LAMC2)	1	c.C283T (p.R95X)
● ● ● ● ●	Hermansky-Pudlak Syndrome: HPS3 Related (HPS3)	5	c.-3010_217+690del3.9kb. c.C1189T (p.R397W). c.1691+2T>G. c.2589+1G>C. c.1691+2T>G
● ● ● ● ●	HMG-CoA Lyase Deficiency (HMGCL)	6	c.914_915delITT. c.G122A (p.R41Q). c.109G>T (p.E37X). c.561+1G>A. c.G208C (p.V70L). c.G835A (p.E279K)
● ● ● ● ●	Holocarboxylase Synthetase Deficiency (HLCS)	3	c.780delG. c.1795+5G>A. c.T710C (p.L237P)
● ● ● ● ●	Homocystinuria Caused by CBS Deficiency (CBS)	7	c.G919A (p.G307S). c.572C>T (p.T191M). c.C341T (p.A114V). c.T833C (p.I278T). c.G797A (p.R266K). c.C1006T (p.R336C). c.T959C (p.V320A)
● ● ● ● ●	Hurler Syndrome (IDUA)	6	c.G266A (p.R89Q). c.G1205A (p.W402X). c.G979C (p.A327P). c.C1598G (p.P533R). c.C208T (p.Q70X). c.T1960G (p.X654G)
● ● ● ● ●	Hypophosphatasia (ALPL)	5	c.1559delIT. c.G1001A (p.G334D). c.A1133T (p.D378V). c.G571A (p.E191K). c.T979C (p.F327L)
● ● ● ● ●	Inclusion Body Myopathy: Type 2 (GNE)	3	c.G131C (p.C44S). c.T2228C (p.M743T). c.G1807C (p.V603L)
● ● ● ● ●	Isovaleric Acidemia (IVD)	1	c.C941T (p.A314V)
● ● ● ● ●	Joubert Syndrome (TMEM216)	1	c.G35T (p.R12L)
● ● ● ● ●	Laryngoonychocutaneous Syndrome (LAMA3)	1	c.151_152insG (p.V51GfsX3)
● ● ● ● ●	Leber Amaurosis (LCA5)	3	c.C835T (p.Q279X). c.1476_1477insA (p.P493TfsX1). c.1151delC
● ● ● ● ●	Leigh Syndrome: French-Canadian (LRPPRC)	1	c.C1061T (p.A354V)
● ● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2D (SGCA)	1	c.C229T (p.R77C)
● ● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2E (SGCB)	6	c.C452G (p.T151R). c.G272T (p.R91L). c.T323G (p.L108R). c.C341T (p.S114F). c.G272C (p.R91P). c.T299A (p.M100K)

Groups	Disease	#	Mutations
● ● ● ●	Limb-Girdle Muscular Dystrophy: Type 2I (FKRP)	1	c.C826A (p.L276I)
● ● ● ●	Lipoprotein Lipase Deficiency (LPL)	1	c.G644A (p.G215E)
● ● ● ●	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)	2	c.C1132T (p.Q378X). c.G1528C (p.E510Q)
● ● ● ●	Luteinizing Hormone Resistance (Leydig Cell Hypoplasia) (LHCGR)	10	c.T1505C (p.L502P). c.G1777C (p.A593P). c.T1627C (p.C543R). c.T391C (p.C131R). c.T1027A (p.C343S). c.G430T (p.V144F). c.C1660T (p.R554X). c.C1635A (p.C545X). c.G1060A (p.E354K). c.1822_1827delCTGGTT (p.608_609delLV)
● ● ● ●	Maple Syrup Urine Disease: Type 1A (BCKDHA)	4	c.117delC. c.G730A (p.G244R). c.T1312A (p.Y438N). c.860_867delGAGGCCCC
● ● ● ●	Maple Syrup Urine Disease: Type 1B (BCKDHB)	3	c.G1114T (p.E3~2X). c.G548C (p.R183P). c.G832A (p.G2~8S)
● ● ● ●	Maple Syrup Urine Disease: Type 3 (DLD)	8	c.G1123A (p.E375K). c.104_105insA. c.C1463T (p.P488L). c.G685T (p.G229C). c.A1081G (p.M361V). c.A214G (p.K72E). c.A1483G (p.R495G). c.T1178C (p.I393T)
● ● ● ●	Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	8	c.T199C (p.Y67H). c.C362T (p.T121I). c.G799A (p.G267R). c.G617A (p.C206H). c.C250T (p.L84F). c.G583A (p.G195R). c.A985G (p.K329E). c.C616T (p.R206C)
● ● ● ●	Metachromatic Leukodystrophy (ARSA)	14	c.C1136T (p.P377L). c.C827T (p.T276M). c.A459+1G>A. c.A856C (p.T288P). c.C1283T (p.P426L). c.C635T (p.A212V). c.G251A (p.R86Q). c.1204+1G>A. c.T536G (p.I181S). c.C1232T (p.T409I). c.C28~T (p.S96F). c.G302A (p.G99D). c.G~39A (p.G245R). c.G~63C (p.D25~H)
● ● ● ●	Methylmalonic Acidemia: MMAA Related (MMAA)	14	c.266T>C (p.L89P). c.650T>A (p.L217X). c.562G>C (p.G188R). c.653G>A (p.G218E). c.733+1G>A. c.64C>T (p.R22X). c.1076G>A (p.R359Q). c.988C>T (p.R330X). c.503delC (p.T168MfsX9). c.161G>A (p.W54X). c.283C>T (p.Q95X). c.358C>T (p.Q120X). c.397C>T (p.Q133X). c.433C>T (p.R145X)
● ● ● ●	Methylmalonic Acidemia: MMAB Related (MMAB)	11	c.569G>A (p.R190H). c.700C>T (p.Q234X). c.656A>G (p.Y219C). c.556C>T (p.R186W). c.403G>A (p.A135T). c.291-1G>A. c.571C>T (p.R191W). c.572G>A (p.R191Q). c.287T>C (p.I96T). c.197-1G>T. c.568C>T (p.R190C)
● ● ● ●	Methylmalonic Acidemia: MUT Related (MUT)	22	c.691T>A (p.Y231N). c.655A>T (p.N219Y). c.2150G>T (p.G~1~V). c.109~A>G (p.N366S). c.935G>T (p.G312V). c.643G>A (p.G215S). c.1105C>T (p.R369C). c.2099T>A (p.M700K). c.607G>A (p.G203R). c.1867G>A (p.G623R). c.1106G>A (p.R369H). c.1280G>A (p.G427D). c.2054T>G (p.L685R). c.2080C>T (p.R694W). c.278G>A (p.R93H). c.322C>T (p.R108C). c.299A>G (p.Y100C). c.284C>G (p.P95R). c.572C>A (p.A191E). c.521T>C (p.F174S). c.313T>C (p.W105R). c.281G>T (p.G94V)
● ● ● ●	Methylmalonic Aciduria and Homocystinuria: Type cblC (MMACHC)	5	c.482G>A (p.R161Q). c.271_273insA (p.R91KfsX14). c.394C>T (p.R132X). c.331C>T (p.R111X). c.609G>A (p.W203X)
● ● ● ●	MTHFR Deficiency: Severe (MTHFR)	7	c.1166G>A (p.W389X). c.T1304C (p.F435S). c.G1408T (p.E470X). c.474A>T (p.G158G). c.523G>A (p.A175T). c.652G>T (p.V218L). c.T1721G (p.V574G)
● ● ● ●	Mucopolidosis: Type II/III (GNPTAB)	3	c.3503_3504delITC (p.L1168QfsX5). c.T1120C (p.F374L). c.C3565T (p.R1189X)
● ● ● ●	Mucopolidosis: Type IV (MCOLN1)	4	c.-1015_788delG6433. c.406-2A>G. c.G1084T (p.D362Y). c.-1015_788delG6433
● ● ● ●	Muscle-Eye-Brain Disease (POMGNT1)	3	c.1539+1G>A. c.C1478G (p.P493R). c.C1324T (p.R442C)
● ● ● ●	Nemaline Myopathy: NEB Related (NEB)	1	c.7434_7536del12502bp
● ● ● ●	Nephrotic Syndrome: Type 1 (NPHS1)	3	c.121_122delICT. c.C3325T (p.R1109X). c.1481delC
● ● ● ●	Nephrotic Syndrome: Type 2 (NPHS2)	2~	c.419delG (p.G140fsX180). c.~9T>A (p.V260E). c.502C>A (p.R168S). c.9~6_9~insA (p.T326fsX345). c.413G>A (p.R138Q). c.412C>T (p.R138X). c.353C>T (p.P118L). c.274G>T (p.G92C). c.104_105insG (p.G35fsX69). c.85G>A (p.A29T). c.503G>A (p.R168H). c.555delT (p.F185fsX186). c.538G>A (p.V180M). c.467_468insT (p.L156fsX166). c.479A>G (p.D160G). c.964C>T (p.R322X). c.851C>T (p.A284V). c.705_713delICTAGAGAG (p.L236_R238del). c.502C>T (p.R168C). c.467delT (p.L156fsX180). c.871C>T (p.R291W). c.948delT (p.A317L). c.862G>A (p.A288T). c.868G>A (p.V290M). c.855_856delAA (p.Q285fsX302). c.714G>T (p.R238S). c.622G>A (p.A208T)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN3 Related (CLN3)	3	c.461-280_677+382del966 (p.G154AfsX29). c.883G>A (p.E295K). c.597C>A (p.Y199X)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN5 Related (CLN5)	7	c.G377A (p.C126Y). c.1175_1176delAT (p.Y392X). c.A1121G (p.Y374C). c.G1054T (p.E352X). c.G225A (p.W75X). c.G835A (p.D279N). c.G335A (p.R112H)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN6 Related (CLN6)	10	c.G17C (p.R6T). c.T200C (p.L67P). c.511_513delITAT (p.L171delY). c.C663G (p.Y221X). c.G368A (p.G123D). c.C139T (p.L47F). c.214G>T (p.E72X). c.316_317insC (p.R106PfsX26). c.460_462delATC (p.I154del). c.G308A (p.R103Q)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: CLN8 Related (CLN8)	4	c.G88C (p.A30P). c.C70G (p.R24G). c.C610T (p.R204C). c.G789C (p.W263C)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: MFSD8 Related (MFSD8)	2	c.~54+2T>A. c.881C>A (p.T294K)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: PPT1 Related (PPT1)	8	c.G322C (p.G108R). c.C451T (p.R151X). c.T29A (p.L10X). c.T656A (p.L219Q). c.A236G (p.D79G). c.A364T (p.R122W). c.G134A (p.C45Y). c.A223C (p.T75P)
● ● ● ●	Neuronal Ceroid-Lipofuscinosis: TPP1 Related (TPP1)	9	c.523-1G>C. c.A857G (p.N286S). c.G851T (p.G284V). c.T1093C (p.C365R). c.G1094A (p.C365Y). c.523-1G>A. c.C622T (p.R208X). c.C616T (p.R206C). c.G1340A (p.R477H)
● ● ● ●	Niemann-Pick Disease: Type A (SMPD1)	5	c.994delC. c.G1734C (p.K578N). c.G1493T (p.R498L). c.C1267T (p.H423Y). c.T911C (p.L304P)
● ● ● ●	Niemann-Pick Disease: Type B (SMPD1)	2	c.1828_1830delICG (p.610delR). c.C880A (p.Q294K)
● ● ● ●	Niemann-Pick Disease: Type C1 (NPC1)	14	c.G2848A (p.V950M). c.A346~G (p.N1156S). c.A3263G (p.Y1088C). c.T3182C (p.I1061T). p.T1133C (p.V378A). c.C3107T (p.T1036M). c.A2783C (p.Q928P). c.G2665A (p.V898M). c.T337C (p.C113R). c.A2324C (p.Q775P). c.G2974C (p.G992R). c.C2932T (p.R978C). c.G530A (c.C117Y). c.G2974T (p.G992W)
● ● ● ●	Niemann-Pick Disease: Type C2 (NPC2)	11	c.G58T (p.E20X). c.G115A (p.V39M). c.G352T (p.E118X). c.T199C (p.S67P). c.C133T (p.Q45X). c.C358T (p.P120S). c.C436T (p.Q146X). c.T295C (p.C99R). c.332delA (p.N111fs). c.C141A (p.C47X). c.190+5G>A

Groups	Disease	#	Mutations
● ● ● ●	Nijmegen Breakage Syndrome (NBN)	8	c.654_658delAAAAA (p.K219fs), c.742insGG (p.E248GfsX5), c.698_701delAACA (p.K233SfsX4), c.C976T (p.Q326X), c.1142delC (p.P381QfsX22), c.835_838delCAGA (p.Q279PfsX1), c.C643T (p.R215W), c.C1089A (p.Y363X)
● ● ● ●	Nonsyndromic Hearing Loss and Deafness: DFNB1 Related (GJB2)	14	c.C427T (p.R143W), c.358delGAG (p.120delE), c.G231A (p.W77X), c.G71A (p.W24X), c.T269C (p.L90P), c.C370T (p.Q124X), c.G109A (p.V37I), c.T229C (p.W77R), c.167delIT (c.T101C (p.M34T), c.35delG, c.312_325delGAAGTTCATCAAGG, c.235delC, c.G551C (p.R184P)
● ● ● ●	Ornithine Translocase Deficiency (SLC25A15)	4	c.C535T (p.R179X), c.562_564delTTC (p.188delF), c.C95G (p.T32R), c.G824A (p.R275Q)
● ● ● ●	Pendred Syndrome (SLC26A4)	5	c.T ⁻ 0 ⁻ C (p.L236P), c.A1151G (p.E384G), c.A1246C (p.T416P), c.A2168G (p.H ⁻ 23R), c.1001+1G>A
● ● ● ●	Persistent Mullerian Duct Syndrome: Type 1 (AMH)	2	c.C571T (p.R191X), c.G1144T (p.E382X)
● ● ● ●	Persistent Mullerian Duct Syndrome: Type II (AMHR2)	5	c.G742A (p.E248K), c.232+1G>T, c.G1217A (p.R406Q), c.1330_1356delCTGGGCAATACCCCTACCTCTGATGAG, c.596delA
● ● ● ●	Phenylalanine Hydroxylase Deficiency (PAH)	17	c.C842T (p.P281L), c.C117G (p.F39L), c.T896G (p.F299C), c.G838A (p.E280K), c.1315+1G>A, c.A1G (p.M1V), c.A1241G (p.Y414C), c.G1223A (p.R408Q), c.C1222T (p.R408W), c.C754T (p.R252W), c.G3A (p.M1I), c.1066-11G>A, c.G473A (p.R158Q), c.T143C (p.L48S), c.T194C (p.I65T), c.G814T (p.G2 ⁻ 2X), c.G ⁻ 82A (p.R261Q)
● ● ● ●	Polyglandular Autoimmune Syndrome: Type I (AIRE)	5	c.C769T (p.R257X), c.A254G (p.Y85C), c.967_979delCTGTCCCTCCCGC (p.Leu323SerfsX51), c.C415T (p.R139X), c.1163_1164insA (p.Met388IlefsX36)
● ● ● ●	Primary Hyperoxaluria: Type 1 (AGXT)	12	c.T454A (p.F152I), c.T613C (p.S205P), c.C697T (p.R233C), c.G508A (p.G170R), c.C198G (p.Y66X), c.G245A (p.G82E), c.G466A (p.G156R), c.33insC, c.G738A (p.W246X), c.T731C (p.I244T), c.G698A (p.R233H), c.G121A (p.G41R)
● ● ● ●	Primary Hyperoxaluria: Type 2 (GRHPR)	3	c.C295T (p.R99X), c.103delG, c.404+3delAAGT
● ● ● ●	Primary Hyperoxaluria: Type 3 (HOGA1)	2	c.G860T (p.G287V), c.944_946delIAGG (p.315delE)
● ● ● ●	Progressive Familial Intrahepatic Cholestasis: Type 2 (ABCB11)	5	c.C1 ⁻ 23T (p.R5 ⁻ 5X), c.3 ⁻ 6 ⁻ 3 ⁻ 68insC, c.A890G (p.E29 ⁻ G), c.C3169T (p.R105 ⁻ X), c.G1295C (p.R432T)
● ● ● ●	Propionic Acidemia: PCCA Related (PCCA)	4	c.1685C>G (p.S562X), c.1196G>A (p.R399Q), c.937C>T (p.R313X), c.862A>G (p.R288G)
● ● ● ●	Propionic Acidemia: PCCB Related (PCCB)	13	c.1495C>T (p.R499X), c.1606A>G (p.N536D), c.1534C>T (p.R512C), c.1539_1540insCCC (p.R514PfsX38), c.1556T>C (p.L519P), c.1304A>G (p.Y435C), c.1228C>T (p.R410W), c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA (p.G407fs), c.502G>A (p.E168K), c.335G>A (p.G112D), c.280G>T (p.G94X), c.1283C>T (p.T428I), c.457G>C (p.A153P)
● ● ● ●	Pseudocholinesterase Deficiency (BCHE)	1	c.A293G (p.D98G)
● ● ● ●	Pycnodysostosis (CTSK)	2	c.A990G (p.X330W), c.T926C (p.L309P)
● ● ● ●	Pyruvate Dehydrogenase Deficiency: Autosomal Recessive (PDHB)	2	c.C1030T (p.P344S), c.A395G (p.Y132C)
● ● ● ●	Retinitis Pigmentosa: Autosomal Recessive: DHDDS Related (DHDDS)	1	c.A124G (p.K42E)
● ● ● ●	Rhizomelic Chondrodysplasia Punctata: Type I (PEX7)	8	c.C120G (p.Y40X), c.903+1G>C, c.A40C (p.T14P), c.G649C (p.G217R), c.T875A (p.L292X), c.T345G (p.Y115X), c.C653T (p.A218V), c.12_18dupGTGCGGT (p.G7VfsX51)
● ● ● ●	Salla Disease (SLC17A5)	5	c.A406G (p.K136E), c.A548G (p.H183R), c.802_816delTCATCATTAAGAAAT (p.Leu336fsX13), c.C1001G (p.P334R), c.C115T (p.R39C)
● ● ● ●	Sandhoff Disease (HEXB)	3	c.850C>T (p.R284X), c.445+1G>A, c. ⁻ 6delA
● ● ● ●	Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	5	c.C391T (p.R107C), c.C1147T (p.R383C), c.C1138T (p.R380W), c.C575T (p.A192V), c.C1058T (p.S353L)
● ● ● ●	Sickle-Cell Anemia (HBB)	1	c.A20T (p.E7V)
● ● ● ●	Sjogren-Larsson Syndrome (ALDH3A2)	2	c.1297_1298delIGA (p.E433fs), c.C943T (p.P315S)
● ● ● ●	Smith-Lemli-Opitz Syndrome (DHCR7)	20	c.G452A (p.W151X), c.1342G>A (p.E448K), c.A356T (p.H119L), c.G1337A (p.R446Q), c.G453A (p.W151X), c.C1054T (p.R352W), c.T470C (p.L157P), c.G744T (p.W248C), c.C1210T (p.R404C), c.G9 ⁻ 6T (p.V326L), c.1228G>A (p.G410S), c.C2 ⁻ 8T (p.T93M), c.T326C (p.L109P), c.964-1G>C, c.906C>G (p.F302L), c.725G>A (p.R242H), c.724C>T (p.R242C), c.G1139A (p.C380Y), c.506C>T (p.S169L), c.G1055A (p.R352Q)
● ● ● ●	Spinal Muscular Atrophy: SMN1 Linked (SMN1)	23	c.740_741insC, DEL EXON 7, c.43C>T (p.Q15X), c.815A>G (p.Y272C), c.836G>T, c.821C>T (p.T274I), c.768_778dupTGCTGATGCTT, c.835-18_835-12delCCTTTAT, c.585_586insT, c.823G>A (p.G275S), c.558delA, c.22_23insA, c.835G>T, c.834+2T>G, c.683T>A (p.L228X), c.734C>T (p.P245L), c.439_443delGAAGT, c.305G>A (p.W102X), c.91_92insT, c.400G>A (p.E134K), c.5C>G (p.A2G), c.509_510delGT, c.81_81+1insG
● ● ● ●	Stuve-Wiedemann Syndrome (LIFR)	9	c. ⁻ 56 ⁻ 5 ⁻ insT (p.K253X), c.1 ⁻ 0delC, c.653_654insT, c.1 ⁻ 89C>T (p.R59 ⁻ X), c.2434C>T (p.R812X), c.2472_2476delTATGT, c.2274_2275insT, c.1620_1621insA, c.1601-1G>A
● ● ● ●	Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2)	7	c.C835T (p.R279W), c.G764A (p.G255E), c.C398T (p.A133V), c.C532T (p.R178X), c.699+2T>C, c.T1957A (p.C653S), c.1018_1020delGTT (p.340delV)
● ● ● ●	Tay-Sachs Disease (HEXA)	32	c.-2564_253+5128del7945insG, c.1278_1279insTATC, c.C508T (p.R170W), c.C532T (p.R178C), c.1073+1G>A, c.G509A (p.R170Q), c.G533A (p.R178H), c.T632C (p.F211S), c.G749A (p.G250D), c.A590C (p.K197T), c.805+1G>A, c.1074-1G>T, c.1421+1G>C, c.913_915delITC (p.305delF), c.A1003T (p.I335F), c.G805A (p.G269S), c.G598A (p.V200M), c.T380G (p.L12 ⁻ R), c.C540G (p.Y180X), c.571-1G>T, c.T538C (p.Y180H), c.G533T (p.R178L), c.C409T (p.R137X), c.C629T (p.S210F), c.346+1G>C, c.613delC, c.T116G (p.L39R), c.G1496A (p.R499H), c.C1510T (p.R504C), c.G78A (p.W26X), c.A1G (p.M1V), c.A611G (p.H204R)
● ● ● ●	Tyrosine Hydroxylase Deficiency (TH)	1	c.G698A (p.R233H)
● ● ● ●	Tyrosinemia: Type I (FAH)	10	c.707-1G>C, c.698A>T (p.D233V), c.G786A (p.W262X), c.G1009A (p.G337S), c.G1069T (p.E357X), c.1062+5G>A, c.707-1G>C, c.C782T (p.P261L), c.554-1G>T, c.607-6T>G

Groups	Disease	#	Mutations
● ● ● ● ●	Usher Syndrome: Type 1B (MYO7A)	12	c.3719G>A (p.R1240Q), c.700C>T (p.Q234X), c.635G>A (p.R212H), c.5581C>T (p.R1861X), c.C448T (p.R150X), c.6025delG (p.A2009fs), c.2476G>A (p.A826T), c.1996C>T (p.R666X), c.1797G>A (p.M599I), c.905G>A (p.R302H), c.634C>T (p.R212C), c.1884C>A (p.C628X)
● ● ● ● ●	Usher Syndrome: Type 1C (USH1C)	5	c.IVS5+1G>A, c.216G>A (p.V72fs), c.91C>T (p.R31X), c.238_239insC, c.IVS1+1G>T
● ● ● ● ●	Usher Syndrome: Type 1D (CDH23)	14	c.8230G>A (p.G2744S), c.172C>T (p.Q58X), c.4504C>T (p.R1502X), c.5237G>A (p.R1746Q), c.5985C>A (p.Y1995X), c.6307G>T (p.E2103X), c.3880C>T (p.Q1294X), c.3617C>G (p.P1206R), c.7549A>G (p.S2517G), c.4488G>C (p.Q1496H), c.3367C>T (p.Q1123X), c.4069C>T (p.Q1357X), c.9524G>A (p.R31 ⁵ H), c.849 ⁻ C>G (p.R2833G)
● ● ● ● ●	Usher Syndrome: Type 1F (PCDH15)	3	c.A5557C (p.M1853L), c.C2052A (p.Y684X), c.C733T (p.R245X)
● ● ● ● ●	Usher Syndrome: Type 2A (USH2A)	9	c.1000C>T (p.R334W), c.14020A>G (p.R4674G), c.923_924insGCCA (p.H308fs), c.12067-2A>G, c.2299delG (p.E767SfsX21), c.2276G>T (p.C759F), c.2209C>T (p.R737X), c.1256G>T (p.C419F), c.239_240insGTAC
● ● ● ● ●	Usher Syndrome: Type 3A (CLRN1)	3	c.T528G (p.Y176X), c.T359A (p.M120K), c.T144G (p.N48K)
● ● ● ● ●	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	8	c.1372T>C (p.F458L), c.1322G>A (p.G441D), c.1144A>C (p.K382Q), c.1405C>T (p.R469W), c.1226C>T (p.T409M), c.183 ⁻ C>T (p.R613V), c.848T>C (p.V283A), c. ⁻ 9C>T (p.T260M)
● ● ● ● ●	Walker-Warburg Syndrome (FKTN)	1	c.1167insA (p.F390fs)
● ● ● ● ●	Wilson Disease (ATP7B)	7	c.2304delC, c.G2333T (p.R778L), c.1340delAAAC, c.G2336A (p.W779X), c.C2332G (p.R778G), c.C3207A (p.H1069Q), c.G2337A (p.W779X)
● ● ● ● ●	Wolman Disease (LIPA)	6	c.419G>A (p.W140X), c.C964T (p.Q322X), c.796G>T (p.G266X), c.966+1G>A, c.229+1G>A, c.G260T (p.G87V)
● ● ● ● ●	Zellweger Spectrum Disorders: PEX10 Related (PEX10)	2	c.764_765insA, c.874_875delCT
● ● ● ● ●	Zellweger Spectrum Disorders: PEX1 Related (PEX1)	3	c.2916delA (p.G973fs), c.G2528A (p.G843D), c.2097insT (p.I700fs)