**Patient Results: [**#**] [Known/novel] Heterozygous [Pathogenic/Likely Pathogenic] Variant Detected**

**DNA Variants:**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Inheritance** | **Disease** | **Prevalence** | **Variant** | **Classification** |
|  | [**mode**] |   |  | c.#A>T(p.aa#aa) | [**path/likely path**] |

**Interpretation:**

A sample from this individual (**subject ID**) was referred to our laboratory for analysis of Next-Generation Genome Sequencing (NGS) and Sanger confirmation of variants identified in carrier screening for: [**insert list of categories selected by participant**].

[**#**] [**known/novel**] heterozygous [**type**] variant, c.#A>T (p.aa#aa) (**ID**), was detected in exon [**#**] of the *[****gene******name****]* gene of this individual by NGS. This study indicated that this individual is a carrier (i.e. not affected) of a [**pathogenic/likely pathogenic**] variant in the *[****gene name****]* gene. The *[****gene name****]* gene encodes for [**description**] (OMIM#: **#**). When mutated, the *[****gene name****]* gene is associated with [**mode**] [**condition name**], [**description**] (MIM#: **#**). [**describe related symptoms**] (**citation**). This condition is [**mode**] and is considered a [**category**] condition. We have confirmed this finding in our laboratory using Sanger sequencing.

It is important to understand that next generation genome sequencing is a screening test. This individual could carry a variant not detected by this test, or in genes that are not analyzed (see test limitations). In addition, only known pathogenic and likely pathogenic variants are reported.

**Recommendations:**

In general we recommend that the reproductive partner of an individual who is a carrier be tested. [**testing status of partner: tested/not tested**]. Genetic counseling is recommended.

**Evidence for Variant Interpretations:**

c.#A>T (p.aa#aa) **in Exon** [**#**] **of the *[gene name]* gene (NM ID, chr [#]:** **[position]) is interpreted as [pathogenic/likely pathogenic].**

The c.#A>T (p.aa#aa) [**type**] variant in the *[****gene name****]* gene has been previously reported in [**describe**] with [**mode**] [**condition name**] and is predicted to [**describe**] in exon [**#**] (out of a total of [**#**] exons in the coding sequence). [**describe**] (**citation**). [**type**] variants have been described in the *[****gene name****]* gene in several affected individuals (**citation**) and are, therefore, a common mechanism of disease. This [**variant**] [**has/has not**] been reported in the three control population databases (Exome Sequencing Project [ESP], 1000 Genomes, and ExAc) and displayed a [**high/low**] CADD score (**#**). Therefore, this collective evidence supports the classification of the c.#A>T (p.aa#aa) as a recessive [**pathogenic/likely pathogenic**] variant for [**condition**].

**Method:**

Next-generation genome sequencing was performed in the Illumina CLIA laboratory. Genomic DNA was prepped with TruSeq DNA LT and then sequenced on a HiSeq 2000 or 2500 (Illumina, version 3 chemistry) with 100bp paired-end reads. Resulting sequences were aligned to the human genome reference (hg19) using the Burrows-Wheeler Aligner (BWA) and variants identified with the Genome Analysis Toolkit (GATK) at the University of Washington (UW). A modified version of the SeattleSeq tool was used to annotate variants found within a defined set of colon cancer and actionable genes. OHSU laboratory analyzed the annotated variant list to identify pathogenic variants in the attached gene list for evaluation of carrier status. For confirmation studies, genomic DNA was extracted in our laboratory using the Puregene extraction method, and pathogenic variants were confirmed by custom designed Sanger sequencing. The sequence was assembled and analyzed in comparison to the published reference sequence for each gene in which a pathogenic variant was identified. Only known pathogenic and likely pathogenic variants were confirmed and reported.

The NGS data was also assessed for the average depth of coverage and data quality threshold values:

|  |  |
| --- | --- |
| Mean Depth of Coverage1 | **#** |
| Quality threshold2 | **#**%  |

1Mean depth of coverage refers to the sequence mean read depth across the genome.

2The quality threshold refers to the percentage of the genome where read depth was at least 30X coverage to permit high quality variant base calling, annotation and evaluation. Average quality threshold is **#**% at >=30X of the genome, indicating that a small portion of the target region may not be covered with sufficient depth or quality to call variant positions confidently.

**List of Carrier Status Genes Covered at Less Than 10X Read Depth:**

[**insert list**]

**Limitations:**

1. This assay has limited ability to detect large deletions or duplications as well as small insertions and deletions.
2. This test also has limited ability to detect mosaicism.
3. This test does not provide complete coverage of all coding exons.
4. Noncoding regions may have limited information and limited ability to interpret.
5. The assay does not detect variants located:
	1. in regions of insufficient coverage,
	2. in regions containing paralogous genes or pseudogenes,
	3. in regions where the reference genome is inaccurate or contains gaps and insertions,
	4. in regions of high GC content
6. All identified variants of uncertain significance are not reported.
7. Genes not associated clinically with Mendelian disorders at the time this test was performed were not analyzed.
8. Genes not analyzed.

**References:**

1. Exome Variant Server (ESP 6500) (http://evs.gs.washington.edu/EVS/)
2. Richards *et al.* (2015) Genet Med; 17(5):405-24.
3. Rehm *et al.* (2013) Genet Med; 15(9):733-47.
4. Dorschner *et al.* (2013) Am J Hum Genet; 93:631-640.

**Diseases and Associated Genes:**

**Cardiomyopathy, Dilated, 3b, Cmd3b –** DMD; **Arterial calcification, Generalized, Of Infancy, Gaci -** ENPP1; **Glycogen Storage Disease Ii (Pompe) -** GAA; **Jervell and Lange Nielsen Syndrome1, Jlns1 -** KCNQ1; **Danon Disease (Lysosomal Glycogen Storage Disease Without Acid Maltase) -** LAMP2; **Barth Syndrome, Bths -** TAZ; **Cardiomyopathy, Dilated 3a, Cmd3a -** TAZ; **Myopathy, Early Onset, With Fatal Cardiomyopathy -** TTN; **Ichthyosis Congenita, Harlequin Fetus -** ABCA12; **Chanarin - Dorfman Syndrome (Ichthyotic Neutral Lipid Storage Disease) -** ABHD5; **Ichthyosis, Congenital, Autosomal Recessive 2, Arci2 -** ALOX12B; **Fibromatosis, Juvenile Hyaline -** ANTXR2; **Cutis Laxa, Ar, Ii -** ATP6V0A2; **Ichthyosis, Leukocyte Vacuoles , Alopecia, And Sclerosing Cholangitis -** CLDN1; **Epidermolysis Bullosa, Junctional, Nonherlitz -** COL17A1; **Ehlers Danlos Syndrome, Ar, Cardiac Valvular -** COL1A2; **Epidermolysis Bullosa Dystrophica, Ar, Rdeb -** COL7A1; **17α-hydroxylase/17,20-lyase Deficiency -** CYP17A1;**Xeroderma Pigmentosum, Complementation Group E -** DDB2; **Skin Fragility Woolly Hair Syndrome -** DSP; **Epidermolysis Bullosa, Lethal Acantholytic -** DSP; **Ectodermal Dysplasia, Hypohidrotic, Xlr, Xhed -** EDA; **Abcd Syndrome -** EDNRB; **Cutis Laxa, Ar, I -** EFEMP2; **Xeroderma Pigmentosum, Complementation Group D, Xpd -** ERCC2; **Trichothiodystrophy, Photosensitive, Ttdp -** ERCC2; **Xeroderma Pigmentosum, Complementation Group B, Xpb -** ERCC3; **Xeroderma Pigmentosum, Complementation Group F, Xpf -** ERCC4; **Xeroderma Pigmentosum, Complementation Group G, Xpg -** ERCC5; **De Sanctis Cacchione Syndrome -** ERCC6; **Cutis Laxa, Ar, I -** FBLN5; **Fibromatosis, Juvenile Hyaline -** FECH; **Trichothiodystrophy, Photosensitive, Ttdp -** GTF2H5; **Hermansky - Pudlak Syndrome 1, Hps1 (Heterogenous) -** HPS1; **Incontinentia Pigmenti, Ip -** IKBKG; **Epidermolysis Bullosa Junctionalis With Pyloric Atresia -** ITGA6; **Epidermolysis Bullosa Junctionalis With Pyloric Atresia -** ITGB4; **Epidermolysis Bullosa, Junctional, Non Herlitz -** ITGB4; **Laryngo Onychocutaneous Syndrome, Locs -** LAMA3; **Epidermolysis Bullosa, J Unctional, Nonherlitz -** LAMA3; **Epidermolysis Bullosa, Junctional, Nonherlitz -** LAMB3; **Epidermolysis Bullosa, Junctional, Nonherlitz -** LAMC2; **Tight Skin Contracture Syndrome, Lethal -** LMNA; **Epidermolysis Bullosa Simplex With Muscular Dystrophy -** PLEC1; **3-Hydroxy-3-methylglutaryl CoA lysase deficiency –** PTS; **Yemenite Deaf Blind Hypopigmentation Syndrome -** SOX10; **Ichthyosis, X - Linked, Xli (Steroid Sulfatase Deficiency) -** STS; **Ichthyosis, Lamellar, 1, Li1 -** TGM1; **Porphyria, Congenital Erythropoietic -** UROS; **Odontoonychodermal Dysplasia, Oodd -** WNT10A; **De Sanctis Cacchione Syndrome -** XPA; **Xeroderma Pigmentosum, Complementation Groupa, Xpa -** XPA; **Tight Skin Contracture Syndrome, Lethal -** ZMPSTE24; **Brittle Cornea Syndrome, Bcs -** ZNF469; **Deafness, Neurosensory, Ar2, Dfnb2 -** MYO7A; **Fragile X Fraxe -** AFF2; **Sjogren - Larsson Syndrome, Sls -** ALDH3A2; **Chondrodysplasia Punctata1, Xlr Recessive, Cdpx1 -** ARSE; **Lissencephaly, Xlr, 2lisx2 -** ARX; **Multiple Pterygium Syndrome, Lethal -** CHRNA1; **Multiple Pterygium Syndrome, Lethal -** CHRND; **Multiple Pterygium Syndrome, Lethal -** CHRNG; **Multiple Pterygium Syndrome, Escobar -** CHRNG; **>272430 Crisponi Syndrome (Cold - Induced Sweating Synd) -** CRLF1; **Desmosterolosis -** DHCR24; **ICOS Deficiency (Lethal Congenital Contracture Synd) , LCCS2 -** ERBB3; **Cerebrooculofacioskeletal Syndrome1, Cofs1 -** ERCC6; **Roberts Syndrome, Rbs -** ESCO2; **Vacterl Association With Hydrocephalus, Xlr -** FANCB; **Aarskog - Scott Syndrome, Aas -** FGD1; **Fragile X -** FMR1; **Fraser Syndrome -** FRAS1; **Fraser Syndrome -** FREM2; **Down Syndrome (?) -** GATA1; **Lethal Congenital Contracture Syndrome1, Lccs1 -** GLE1; **Acrocallosal Syndrome, Acls -** KIF7; **Simpson - Golabi - Behmel Syndrome, Type 1, Sgbs1 -** GPC3; **Hydrolethalus Syndrome1 -** HYLS1l; **Corpus Callosum, Agenesis Of, With Mental Retardation, Ocularcoloboma -** IGBP1; **Hydrocephalus Due To Congenital Stenosis Of Aqueduct Of Sylvius -** L1CAM; **Masa Syndrome -** L1CAM; **Donnai - Barrow Syndrome -** LRP2; **Opitz - Kaveggia Syndrome, Oks (Fg Syndrome) -** MED12; **Meckel Syndrome, 1, Mks1 - MKS1**; **Norrie Disease, Nd -** NDP; **Dosage Sensitive Sex Reversal, Dss -** NR0B1; **Lowe Oculocerebro Renal Syndrome, Ocrl -** OCRL; **Simpson Golabi Behmel Syndrome, 2 -** OFD1; **Orofaciodigital Syndrome I, Ofd1 -** OFD1; **>225400 Nevo Syndrome (Edsvi) -** PLOD1; **Renpenning Syndrome1, Rens1 -** PQBP1; **Carpenter Syndrome -** RAB23; **Warburg Micro Syndrome, Warbm -** RAB3GAP1; **Fetal Akinesia Deformation Sequence –** RAPSN, DOK7; **Lissencephaly2, Flis2 -** RELN; **Meckel Syndrome, 5, Mks5 -** RPGRIP1L (8 genes); **Coffin - Lowry Syndrome, Cls -** RPS6KA3; **Andermann Syndrome -** SLC12A6; **Diastrophic Dysplasia (Sulfate Transporter - Related Osterchondrodysplasia) -** SLC26A2; **Atelosteogenesis, Ii, Aoii -** SLC26A2; **Microphthalmia, Syndromic 9, Mcops9 -** STRA6; **Opticoacoustic Nerve Atrophy With Dementia -** TIMM8A; **Mulibrey Nanism -** TRIM37; **Johanson Blizzard Syndrome, Jbs -** UBR1; **Arthrogryposis, Renal Dysfunction, And Cholestasis -** VPS33B; **Cohen Syndrome -** VPS13B; **Tetra Amelia, Ar -** WNT3; **Heterotaxy, Visceral, 1, Xlr, Htx1 -** ZIC3; **Mandibuloacral Dysplasia With Type B Lipodystrophy, MADB -** ZMPSTE24; **Fanconi Anemia Complementation Group A -** FANCA; **Fanconi Anemia Complementation Group C -** FANCC; **Fanconi Anemia D1, Brca2 -** FANCD1; **Fanconi Anemia B -** FAAP95; **Bloom Syndrome -** RECQL3; **Achalasia - Addisonianism - Alacrima - Syndrome, Aaa -** AAAS; **Familial Hyperinsulinemia -** ABCC8; **(>312300) Infertile Male Syndrome -** AR; **Androgen Insensitivity Syndrome, Ais -** AR; **Lipoid Congenital Adrenal Hyperplasia -** CYP11A1; **Adrenal Hyperplasia, Congenital, Due to 21 hydroxylase deficiency -** CYP11A1; **Vitamin D Dependent Rickets, I -** CYP27B1; **Adrenal Hypoplasia, Congenital, Ahc -** DAX1; **Pituitary Dwarfism Iii -** HESX1; **Insulin Like Growth Factor I, Resistance To -** IGF1; **Donohue Syndrome -** INSR; **Kallmann Syndrome -** KAL1; **Pituitary Dwarfismiii -** LHX3; **Pituitary Hormone Deficiency, Combined, 2, Cphd2 -** PROP1; **Pituitary Dwarfismiii -** POU1F1; **Lipoid Congenital Adrenal Hyperplasia -** STAR; **Hypothyroidism, Congenital, Nongoitrous, 4, Chng4 -** TSHB; **Intestinal Pseudo Obstruction, Neuronal, Chronicidiopathic, Xlr -** FLNA; **Colorectal Adenomatous Polyposis, Ar -** MUTYH; **Diarrhea 4, Malabsorptive, Congenital -** NEUROG3; **Thrombotic Thrombocytopenic Purpura, Congenital, Ttp -** ADAMTS13; **Thalassemia/Mental Retardation Syndrome, Nondeletion, Xlratrx -** ATRX; **Afibrinogenemia, Congenital -** FGA; **Afibrinogenemia, Congenital -** FGB; **Afibrinogenemia, Congenital -** FGG; **Thrombophilia Due To Thrombin Defect, Thph1 (Prothrombin) -** F2; **Factor V Leiden -** F5; **Hemophilia A (Factor Viii) -** F8; **Factor Xi - F11**; **G6pd (Glucose - 6 - Phosphate Dehydrogenase) -** G6PD; **Glutathione Synthetase Deficiency -** GSS; **Hemochromatosis, Juvenile, Jh -** HAMP; **Alpha Thalassemia** - HBA1/HBA2; **Beta Thalassemia -** HBB; **Sickle Cell Anemia -** HBB; **Hemochromatosis, Type 1 -** HFE; **Hemochromatosis, Juvenile, Jh -** HFE2; **Amegakaryocytic Thrombocytopenia, Congenital, Camt -** MPL; **Pyruvate Kinase Deficiency Of Red Cells -** PKLR; **Plasminogen Deficiency, I -** PLG; **Thrombophilia, Hereditary, Due To Protein C Deficiency, Autosomal -** PROC; **Shwachman Diamond Syndrome, Sds -** SBDS; **Hemochromatosis, 3 -** TFR2; **Cirrhosis, Familial (Multifactoral) -** KRT18; **Cirrhosis, Familial (Multifactoral) -** KRT8; **Gilbert Syndrome -** UGT1A1; **Severe Combined Immunodeficiency, Ar, T Cell - Negative -** ADA; **Autoimmune Polyendocrine Syndrome, I, Aps1 (Polyglandular) -** AIRE; **Agammaglobulinemia, Xlr xla -** BTK; **Immunodeficiency With Hyperigm, 1, HIGM1 -** CD40LG; **Properdin Deficiency, Xlr -** CFP; **Omenn Syndrome -** DCLRE1C; **Hoyeraal Hreidarsson Syndrome, Hhs -** DKC1; **Immunodeficiency Centromeric Instability Facial Anomalies syndrome -** DNMT3B; **Tcell Immunodeficiency, Congenital Alopecia, And Nail Dystrophy -** FOXN1; **Immunodysregulation, Polyendocrinopathy, And Enteropathy, Xlr -** FOXP3; **Ectodermal Dysplasia, Hypohidrotic, With Immune Deficiency -** IKBKG; **Osteopetrosis, Lymphedema, Ectodermal dysplasia, Anhidrosis, Immunodeficiency, Oledaid -** IKBKG; **Severe Combined Immunodeficiency, Xlr, Scidx1 -** IL2RG; **Combined Immunodeficiency, Xlr, Cidx -** IL2RG; **Severe Combined Immunodeficiency, Ar, Tcellnegative, Bcellpositive, Nkcellnegative -** JAK3; **Chediak Higashi Syndrome, Chs -** LYST; **Nijmegen Breakage Syndrome -** NBS1; **Hemophagocytic Lymphohistiocytosis, Familial, 2, (Heterogenous) -** PRF1; **Griscelli Syndrome, 2, Gs2 -** RAB27A; **Severe Combined Immunodeficiency, Ar, TCell Negative -** RAG1; **Omenn Syndrome -** RAG1; **Severe Combined Immunodeficiency, Ar, Tcell negative -** RAG2; **Omenn Syndrome -** RAG2; **Lympho proliferative Syndrome, Xlr, 1, Xlp1 -** SH2D1A; **Hepatic Venoocclusive Disease With Immunodeficiency, Vodi -** SP110; **Wiskott Aldrich Syndrome, Was -** WAS; **Cholestasis, Progressive Familial Intrahepatic2, Pfic2 -** ABCB11; **Acylcoa Dehydrogenase Family, Member9, Deficiency Of -** ACAD9; **Acylcoa Dehydrogenase, Long Chain, Deficiency Of -** ACADL; **Acyl - Coa Dehydrogenase, Medium - Chain, Deficiency Of (Mcad) -** ACADM; **Acyl - Coa Dehydrogenase, Short - Chain, Deficiency Of, Acadsd -** ACADS; **2 Methylbutyrylcoa Dehydrogenase Deficiency - ACADSB**; **Acyl - Coa Dehydrogenase, Very Long - Chain, Deficiency Of (Vlcad) -** ACADVL; **Peroxisomal Acyl - Coa Oxidase Deficiency -** ACOX1; **Aspartylglycosaminuria -** AGA; **Glycogen Storage Disease Iii -** AGL; **Rhizomelic Chondrodysplasia Punctata, 3, Rcdp3 -** AGPS; **Succinic Semi Aldehyde Dehydrogenase Deficiency -** ALDH5A1; **Fructose Intolerance, Hereditary -** ALDOB; **Congenital Disorder Of Glycosylation, Ik, Cdg1k -** ALG1; **Congenital Disorder Of Glycosylation, Ic, Cdg1c -** ALG6; **Hypophosphatasia, Infantile -** ALPL; **Bile Acid Synthesis Defect, Congenital, 4 -** AMACR; **Glycine Encephalopathy, Gce -** AMT; **Metachromatic Leukodystrophy -** ARSA; **Mucopolysaccharidosis Vi -** ARSB; **Argininosuccinic Aciduria -** ASL; **Canavan -** ASPA; **Citrullinemia Type 1 -** ASS1; **Aicar Transylase/Imp Cyclohydrolase, Deficiency Of -** ATIC; **Menkes -** ATP7A; **Wilson Disease -** ATP7B; **Cholestasis, Progressive Familial Intrahepatic1, Pfic1 -** ATP8B1; **3 Methylglutaconic Aciduria, I -** AUH; **Congenital Disorder Of Glycosylation, Iid, Cdg2d -** B4GALT1; **Maple Syrup Urine Disease Ia -** BCKDHA; **Maple Syrup Urine Disease, Classic, Ib -** BCKDHB; **Mitochondrialcomplex Iii Deficiency -** BCS1L; **Gracile Syndrome -** BCS1L; **Biotinidase Deficiency -** BTD; **Homocystinuria Due To Cystathionine Beta - Synthase Deficiency -** CBS; **Carbamoyl Phosphate Synthetasei Deficiency, Hyperammonemia -** CPS1; **Carnitine Palmitoyltransferase Deficiency 1a (Cpt1) -** CPT1A; **Carnitine Palmitoyltransferase Deficiency Ii (Cpt2) -** CPT2; **Cystinosis, Nephropathic, Ctns -** CTNS; **Cerebrotendinous Xanthomatosis -** CYP27A1; **D2hydroxyglutaric Aciduria -** D2HGDH; **Aromatic L - Amino Acid Decarboxylase Deficiency -** DDC; **Smith - Lemli - Opitz Syndrome, Slos -** DHCR7; **Dihydrolipoamide Dehydrogenase Deficiency (Lipoamide Dehydrogenase) Dld -** DLD; **Maple Syrup Urine Disease Iii -** DLD; **3 - Methylglutaconic Aciduria, V -** DNAJC19; **Congenital Disorder Of Glycosylation, Im, Cdg1m -** DOLK; **Congenital Disorder O Fglycosylation, Ij, Cdg1j -** DPAGT1; **Congenita Ldisorder Of Glycosylation, Ie, Cdg1e -** DPM1; **Dihydropyrimidine Dehydrogenase, Dpyd (Hereditary Thymine - Uraciluria) -** DPYD; **Multiple Acylcoa Dehydrogenase Deficiency, Madd -** ETFA; **Multiple Acylcoa Dehydrogenase Deficiency, Madd -** ETFB; **Multiple Acylcoa Dehydrogenase Deficiency, Madd -** ETFDH; **Encephalopathy, Ethylmalonic -** ETHE1; **Tyrosinemia -** FAH; **Fumarase Deficiency -** FH; **Fucosidosis -** FUCA1; **Glycogen Storage Disease I -** G6PC3; **Glycogen Storage Disease Ii -** GAA; **Krabbe -** GALC; **Galactosemia -** GALT; **Gaucher -** GBA; **Glycogen Storage Disease Iv -** GBE1; **Glutaric Acidemia I -** GCDH; **Congenital Disorder Of Glycosylation, Type Iib, Cdg2b -** GCS1; **Glycine Encephalopathy, Gce -** GCSH; **Combined Oxidative Phosphorylation Deficiency1, Coxpd1 -** GFM1; **Fabry (Alpha - Galactosidase A Deficiency) -** GLA; **Gm1 Gangliosidosis, Ii -** GLB1; **Glycine Encephalopathy, Gce -** GLDC; **Mucolipidosisii Alpha/Beta -** GNPTAB; **Mucolipidosisiii Alpha/Beta -** GNPTAB; **(601492) Mucopolysaccharidosis Viii -** GNS; **Mucopolysaccharidosis Vii -** GUSB; **3 - Hydroxyacyl - Coa Dehydrogenase Deficiency -** HADH; **Trifunctional Protein Deficiency -** HADHA; **Long - Chain 3 - Hydroxyacyl - Coa Dehydrogenase Deficiency (Lchad) -** HADHA; **Trifunctional Protein Deficiency -** HADHB; **Tay - Sachs -** HEXA; **Alkaptonuria -** HGD; **Mucopolysaccharidosis Iiic -** HGSNAT; **Beta - Hydroxyisobutyryl Coa Deacylase, Deficiency Of -** HIBCH; **3 - Hydroxyhydroxy - 3 - Methylglutaryl - Coa Lyase Deficiency -** HMGCL; **Lesch - Nyhan Syndrome, Lns -** HPRT1; **D - Bifunctional Protein Deficiency -** HSD17B4; **Hunter Syndrome (Mps Ii) -** IDA; **Hurler Syndrome (Mps1h) -** IDUA; **Isovaleric Acidemia, Iva -** IVD; **Leigh Syndrome, French Canadian, Lsfc -** LRPPRC; **Alpha Mannosidosis -** MAN2B1; **3 - Methylcrotonyl - Coa Carboxylase2 Deficiency -** MCCC2; **Mucolipidosis Type Iv -** MCOLN1; **Congenital Disorder Of Glycosylation, Iia, Cdg2a -** MGAT2; **Methylmalonicaciduria, Cblb -** MMAB; **Methyl Malonic Aciduria And Homocystinuria, Cblc -** MMACHC; **Congenital Disorder O Fglycosylation, Iib, Cdg2b -** MOGS; **Congenital Disorder Of Glycosylation, Ib, Cdg1b -** MPI; **Combined Oxidative Phosphorylation Deficiency2, Coxpd2 -** MRPS16; **Combined Oxidative Phosphorylation Deficiency5, Coxpd5 -** MRPS22; **Homocystinuria due to deficiency of n(5, 10) Methylenetetrahydrofolate -** MTHFR; **Abetalipoproteinemia, Abl -** MTTP; **Methyl Malonic Aciduria Due To Methyl Malonyl Coa Mutase Def -** MUT; **Mevalonic Aciduria - MVK**; **Schindler Disease, I -** NAGA; **Nacetyl Glutamate Synthase Deficiency -** NAGS; **Neuraminidase Deficiency (Salodosis) -** NEU1; **Niemann - Pick Type C (Also Type D) -** NPC1; **Peroxisome Biogenesis Disorder 1a (Zellweger), Pbd1a -** PEX1; **Phenylketonuria, Pku -** PAH; **Niemann - Pick Type A -** SMPD1; **3 - Methylglutaconic Aciduria, Iii (Costeff Optic Atrophy) -** OPA3; **Ornithine Transcarbamylase Deficiency, Hyperammonemia Due To -** OTC; **Pyruvate Carboxylase Deficiency -** PC; **Pyruvate Dehydrogenase E3 Bindingprotein Deficiency -** PDHX; **Pyruvatede Hydrogenase Phosphatase Deficiency -** PDP1; **Congenital Disorder Of Glycosylation, Type Ia, Cdg1a -** PMM2; **Pyridoxamine 5 Prime Phosphateoxidase Deficiency -** PNPO; **Glycogen Storage Disease Of Heart, Lethalcongenital -** PRKAG2; **Combined Saposin Deficiency -** PSAP; **Phosphoserine Aminotransferase Deficiency -** PSAT1; **Glycogen Storage Disease V -** PYGM; **Lathosterolosis -** SC5DL; **Cardioencephalomyopathy, Fatal Infantile, Due To Cytochrome C Oxidase -** SCO2; **Surfactant Metabolism Dysfunction, Pulmonary, 1, Smdp1 -** SFTPB; **Mucopolysaccharidosis Iiia -** SGSH; **Sialuria, Finnish (Salla Disease) -** SLC17A5; **Infantile Sialic Acid Storage Disorder -** SLC17A5; **Hyperornithinemia Hyperammonemia Homocitrullinuria Syndrome -** SLC25A15; **Congenital Disorder Of Glycosylation, Iif, Cdg2f -** SLC35A1; **Congenita Ldisorder O Fglycosylation, Iic, Cdg2c -** SLC35C1; **Niemann - Pick Type A -** SMPD1; **Lactic Acidosis, Fata Linfantile -** SUCLG1; **Sulfocysteinuria -** SUOX; **Combined Oxidative Phosphorylation Deficiency3, Coxpd3 -** TSFM; **Mitochondrial Complex Iii Deficiency -** UQCRB; **Mitochondrialcomplex Iii Deficiency -** UQCRQ; **ALSTROM SYNDROME, ALMS Stopped Review 10/29/13 -** ALMS1; **Multiple System Atrophy 1, Susceptibility To, Msa1 -** COQ2; **Neurodegeneration With Brain Iron Accumulation 1, Nbia1 –** ATP13A2, CP, DCAF17,PANK2, PLA2G6, FTL ETC; **Choreoacanthocytosis, Chac -** VPS13A; **Adrenoleukodystrophy, Ald -** ABCD1; **Ehlers Danlos Syndrome, Vii, Ar -** ADAMTS2; **Joubert Syndrome3, Jbts3 -** AHI1; **Amyotrophic Latera Lsclerosis2, Juvenile, Als2 -** ALS2; **Primary Lateral Sclerosis, Juvenile, Plsj -** ALS2; **Ataxia, Early Onset, With Oculomotor Apraxia And Hypoalbuminemia, -** APTX; **Coenzyme Q10 Deficiency -** APTX; **Epileptic Encephalopathy, Early Infantile, 1 -** ARX; **Corpus Callosum Agenesis Of With Abnormalgenitalia -** ARX; **Ataxia Telangiectasia -** ATM; **Familial Neurohypophyseal Diabetes Insipidus –** AVP; **Mitochondrial Dna Depletion Syndrome, Hepatocerebral -** C10ORF2(DGUOK); **Infantileonsetspinocerebellarataxia, Iosca (Mito Dna Depletion 7) -** C10ORF2; **Coenzyme Q10 Deficiency -** CABC1; **Epileptic Encephalopathy, Early Infantile, 2 -** CDKL5; **Joubert Syndrome 5, Jbts5 -** CEP290; **Batten (Ceroid Lipofuscinosis, Neuronal3 -** CLN3; **Ceroid Lipofuscinosis, Neuronal, 5, Cln5 -** CLN5; **Ceroid Lipofuscinosis, Neuronal, 6, Cln6 -** CLN6; **Ceroid Lipofuscinosis, Neuronal, 8, Northern Epilepsy Variant -** CLN8; **Coenzyme Q10 Deficiency -** COQ2; **Myoclonic Epilepsy of Unverricht And Lundborg -** CSTB; **Ceroidlipofuscinosis, Neuronal, 10, Cln10 -** CTSD; **Lissencephaly, Xlr, 1, Lisx1 -** DCX; **Mitochondrial DNA Depletion Syndrome, Hepatocerebral -** DGUOK; **Hypertrophic Neuropathy Of Dejerine Sottas Cmt3, Cmt4f -** EGR2 (AD vs AR); **Neuropathy, Hypomyelinating/Charcot - Marie - Tooth - Disease, 4e -** EGR2; **Charcot - Marie - Tooth - Disease, 4d –** NDRG1; **Myoclonic Epilepsy Of Lafora -** EPM2A; **Cockayne Syndrome, B, Csb -** ERCC6; **Cockayne Syndrome, A, Csa -** ERCC8; **Leukodystrophy, Hypomyelinating, 5 -** FAM126A; **Charcot - Marie - Tooth - Disease, 4h, Cmt4h -** FGD4; **Fukuyama - Congenital - Muscular - Dystrophy, Fcmd -** ;; **Friedreich Ataxia 1, Frda -** FXN; **Charcot - Marie - Tooth Disease, X - Linked Dominant, 1, Cmtx1 -** GJB1; **Leukodystrophy, Hypomyelinating, 2 -** GJC2; **Gm1 Gangliosidosis, I -** GLB1; **Bernard-Soulier Syndrome Type A1** - GP1BA; **Bernard-Soulier Syndrome Type C** - GP9**; Bilateral frontoparietal polymicrogyria** - GPR56; **Mental Retardation, Xlr, Syndromic10, Mrxs10 -** HSD17B10; **Spinal Muscular Atrophy, Distal, Ar, 1, Dsma1 -** IGHMBP2; **Neuropathy, Hereditary Sensory Autonomic, Iii, Hsan3 (Familial Dysautonomia) -** IKBKAP; **Epilepsy, Progressive Myoclonic 3, Epm3 -** KCTD7; **Muscular Dystrophy, Congenital Merosin Deficient, 1a, Mdc1a -** LAMA2; **Familial Hypercholesterolemia –** LDLR, LDLRAP1; **Lujan Fryns Syndrome -** MED12; **Ceroid Lipofuscinosis , Neuronal, 7, Cln7 -** MFSD8; **Megalencephalic Leukoencephalopathy With Subcorticalcysts, Mlc -** MLC1; **Molybdenum Cofactor Deficiency -** MOCS1; **Molybdenum Cofactor Deficiency -** MOCS2; **Navajo Neurohepatopathy, Nn -** MPV17; **Mitochondrial DNA Depletion Syndrome, Hepatocerebral -** MPV17; **Hypertrophic Neuropathy Of Dejerine Sottas.Cmt3, Cmt4f –** MPZ, PMP22; **Neuropathy, Hypomyelinating/Charcot - Marie - Tooth - Disease, 4e -** MPZ; **Myotubular Myopathy1, Mtm1 -** MTM1; **Griscelli Syndrome, 1, Gs1 -** MYO5A; **Elejalde Disease - MYO5A**; **Nemaline Myopathy2, Nem2 -** NEB; **Nemaline Myopathy 5, -** NEM5; **Nemaline Myopathy 8, -** KLHL40; **Myoclonic Epilepsy Of Lafora -** NHLRC1; **Joubert Syndrome 4, Jbts4 -** NPHP1; **Insensitivity To Pain, Congenital, With Anhidrosis, Cipa -** NTRK1; **Striatonigral Degeneration, Infantile, Sndi -** NUP62; **Leigh Syndrome, Xlr -** PDHA1; **Leigh Syndrome –** AARS2, ABCB7, ACO2, AFG3L2, AGK, AIFM1, ALAS2, ALDH1B1, ATP5E, ATPAF2, BOLA3, C12orf65, CISD2, COA5, COQ6, COQ9, COX10, COX14, COX15, COX20, COX6B1, DARS2, DLAT, DNM1L, EARS2, FARS2, FASTKD2, FBP1, FOXRED1, G6PC, GFER, GFM2, GYS2, HARS2, HLCS, HSPD1, ISCU, LARS, LIAS, MARS2, MFN2, MPC1, MRPL44, MTFMT, MTO1, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF6, NDUFAF7, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OPA1, PDHB, PNPT1, POLG2, PUS1, RARS2, REEP1, RMND1, RRM2B, SARS2, SCO1, SDHA, SDHAF1, SERAC1, SLC19A3, SLC25A3, SLC25A4, SLC37A4, 125 SOD1, 126 SPG7, SUCLA2, SURF1, TACO1, TIMM44, TMEM70, TPK1, TRMU, TTC19, TUFM, WFS1, YARS2; **Meckel Syndrome -** B9D1, B9D2, C5orf42, CC2D2A, CEP41, NPP5E, TCTN1, TCTN2, TCTN3, TMEM126A, TMEM138, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423; **Coenzyme Q10 Deficiency -** PDSS1; **Coenzyme Q10 Deficiency -** PDSS2; **Neuroaxonal Dystrophy, Infantile, Inad1 -** PLA2G6; **Spinal Muscular Atrophy, Distal, Ar, 4, Dsma4 -** PLEKHG5; **Spastic Paraplegia2 , Xlr, Spg2 (Heterogenous) -** PLP1; **Pelizaeus - Merzbacher Disease, Pmd -** PLP1; **Hypertrophic Neuropathy Of Dejerine Sottas.Cmt3, Cmt4f** - PMP22 (AD/AR); **Alpers Diffuse Degeneration Of Cerebral Gray Matter With Hepatic Cirrhosis -** POLG; **Ceroid Lipofuscinosis, Neuronal, 1, Cln1 -** PPT1; **Arts Syndrome, Arts -** PRPS1; **Hypertrophic Neuropathy Of Dejerine Sottas.Cmt3, Cmt4f -** PRX; **Metachromatic Leukodystrophy Due To Saposinb Deficiency -** PSAP; **Krabbe Disease, Atypical, Due To Saposina Deficiency -** PSAP; **Martsolf Syndrome -** RAB3GAP2; **Arsacs -** SACS; **Rigid Spine Muscular Dystrophy1, Rsmd1 -** SEPN1; **Marinesco Sjogren Syndrome -** SIL1; **Allan - Herndon - Dudley Syndromeahds -** SLC16A2; **Epileptic Encephalopathy, Early Infantile, 3 -** SLC25A22; **Creatine Deficiency Syndrome, Xlr -** SLC6A8; **Mental Retardation, Xlr, Syndromic, Christianson -** SLC9A6; **Spinal Muscular Atrophy -** SMN1; **Cerebral Dysgenesis, Neuropathy, Ichthyosis, Palmoplantar Keratoderma -** SNAP29; **Amish Infantile Epilepsy Syndrome -** ST3GAL5; **Hypoparathyroidism Retardation Dysmorphism Syndrome, Hrd -** TBCE; **Segawa Syndrome, Autosomal Recessive -** TH; **Mitochondrial Dna Depletion Syndrome, Myopathic -** TK2; **Joubert Syndrome Type 2 -** TMEM216; **Joubert Syndrome 6 -** TNNT1; **Joubert Syndrome –** ARL13B; **Ceroid Lipofuscinosis, Neuronal, 2, Cln2 (Tpp1 - Related) -** TPP1; **Aicardi Goutieres Syndrome1, Ags1 -** TREX1; **Pontocerebellar Hypoplasia4, Pch4 -** TSEN54; **Pontocerebellar Hypoplasia2a, Pch2a -** TSEN54; **Spinal Muscular Atrophy, X - Linked 2, Smax2 -** UBA1; **Cerebellar Hypoplasia And Mentalretardation With/Without Quadrupedal -** VLDLR; **Myoadenylate Deaminase Deficiency, Myopathy Due To -** AMPD1; **Muscular Dystrophy, Congenital, Megaconial Type, Mdcmc -** CHKB; **Ullrich Congenital Muscular Dystrophy, Ucmd -** UCMD; **Muscular Dystrophy, Duchenne Type, Dmd -** DMD; **Citrin Deficiency -** SLC25A13; **Compton North Congenital Myopathy** - CNTN1; **Congenital Myasthenic syndrome** – CHRNE; **D-2-Hydroxyglutaric Aciduria** - IDH2; **Emery - Dreifuss Muscular Dystrophy 1, X - Linked, Edmd1 -** EMD; **Inclusion Body Myopathy 2, Autosomal Recessive, Ibm2 -** GNE; **Muscular Dystrophy, Congenital, 1d -** LARGE; **Walker - Warburg (Muscle - Eye - Brain) -** POMT1; **Muscular Dystrophy, Limb - Girdle, Type 2d, Lgmd2d -** SGCA; **Muscular Dystrophy, Limb - Girdle, Type 2e, Lgmd2e -** SGCB; **Carnitine Deficiency, Systemic Primary, Cdsp -** SLC22A5; **Myopathy, Early - Onset, With Fatal Cardiomyopathy Ttn -** TTN; **Ataxia With Vitamin E Deficiency (Ved) -** TTPA; **Mitochondrial Dna Depletion Syndrome 1 (Mngie Type), Mtdps1 -** TYMP, TK2, DGUOK ETC; **Renaltubular Dysgenesis, Rtd -** ACE; **Renaltubular Dysgenesis, Rtd -** AGT; **Hyperoxaluria, Primary, Type I, Hp1 -** AGXT; **Renaltubular Dysgenesis, Rtd -** AGTR1; **Hypomagnesemia, Renal, With Ocular Involvement -** CLDN19; **Alport Syndrome, Arcol4a3renal -** COL4A4 (COL4A3); **Alport Syndrome, X - Linked, Ats -** COL4A5; **Polycystic Kidney Disease (Arpkd) -** FCYT; **Hyperoxaluria, Primary, Type Ii, Hp2 -** GRHPR; **Nephronophthisis 2, Nphp2 -** INVS; **Senior Loken Syndrome5, Slsn5 -** IQCB1; **Bartter Syndrome, Antenatal, 2 -** KCNJ1; **Pierson Syndrome -** LAMB2; **Familial Mediterranean Fever, Fmf -** MEFV; **Nephronophth Isis 1, Nphp1 -** NPHP1; **Senior Loken Syndrome1, Slsn1 -** NPHP1; **Renal Hepatic Pancreatic Dysplasia, Rhpd -** NPHP3; **Nephronophthisis4, Nphp4 -** NPHP4; **Nephrotic Syndrome, Type 1, Nphs1 -** NPHS1; **Nephrotic Syndrome, Type 2 (Steroid - Resistant) -** PDCN; **Nephrotic Syndrome, 3, Nphs3 -** PLCE1; **Hypotonia Cystinuria Syndrome -** PREPL; **Renaltubular Dysgenesis, Rtd -** REN; **Pseudohypoaldosteronism, I, Ar, Pha1 -** SCNN1A; **Pseudohypoaldosteronism, I, Ar, Pha1 -** SCNN1B; **Pseudohypoaldosteronism, I, Ar, Pha1 -** SCNN1G; **Bartter Syndrome, Antenatal, 1 -** SLC12A1; **Hypotonia Cystinuria Syndrome -** SLC3A1; **Nephrotic Syndrome, Early Onset, With Diffuse Mesangial Sclerosis -** WT1; **Cystic Fibrosis -** CFTR; **Alpha 1 - Antitrypsin Deficiency -** SERPINA1; **Respiratory Distress Syndrome In Premature Infants -** SFTPA1; **Respiratory Distress Syndrome In Premature Infants -** SFTPB; **Respiratory Distress Syndrome In Premature Infants -** SFTPC; **Pulmonary Alveolar Microlithiasis -** SLC34A2; **Sudden Infant Death With Dysgenesis Of The Testes Syndrome, Siddt -** TSPYL1; **Geleophysic Dysplasia -** ADAMTSL2; **Hyalinosis, Infantile Systemic -** ANTXR2; **Seckel Syndrome1 -** ATR; **Osteopetrosis, Ar3, Optb3 -** CA2; **Otospondylomegaepiphyseal Dysplasia, Osmed -** COL11A2; **Osteogenesis Imperfecta, Iib -** CRTAP; **Pycnodysostosis -** CTSK; **Spondylocostal Dysostosis, Ar1, Scdo1 -** DLL3; **Hypophosphatemic Rickets, Ar -** DMP1; **Epiphyseal Dysplasia, Multiple, With Early Onset Diabetes Mellitus -** EIF2AK3; **Ellis - Van Creveld Syndrome, Evc -** EVC; **Raine Syndrome, Rns -** FAM20C; **Antley Bixler Syndrome, Abs -** FGFR2; **Antley-Bixler syndrome with disordered steroidogenesis -** POR**; Dyssegmental Dysplasia, Silverman - Handmaker, Ddsh -** HSPG2; **Asphyxiating Thoracic Dystrophy 2, Atd2 (Jeune) -** IFT80; **Hydrops - Ectopic Calcification - Moth - Eaten Skeletal Dysplasia (Greenberg) -** LBR; **Osteogenesis Imperfecta, Viii -** LEPRE1; **Stuve - Wiedemann Syndrome -** LIFR; **Ellis - Van Creveld Syndrome, Evc -** LBN; **Osteoporosis Pseudoglioma Syndrome, Oppg -** LRP5; **Osteopetrosis, Ar5, Optb5 -** OSTM1; **Chondrodysplasia, Blomstrand, Bocd -** PTH1R; **Rhizomelic Chondrodysplasia Punctata, Type 1, Rcdp1 -** PEX7; **Baller - Gerold Syndrome, Bgs -** RECQL4; **Anauxetic Dysplasia -** RMRP; **Cartilage - Hair Hypoplasia -** RMRP; **Achondrogenesis, Ib, Acg1b -** SLC26A2; **Schneckenbecken Dysplasia -** SLC35D1; **Osteopetrosis, Ar1, Optb1 -** TCIRG1; **Paget Disease, Juvenile -** TNFRSF11B; **Vitamin D Dependent Rickets, Ii -** VDR; **Fibular Aplasia Or Hypoplasia, Femoral Bowing And Poly, Syn, And -** WNT7A; **Ulna And Fibula, Absence Of, With Severe Limb Deficiency -** WNT7A; **Stargardt Disease -** ABCA4; **Usher Syndrome, Id, Ush1d -** CDH23; **Choroideremia -** CHM; **Achromatopsia –** CNGA3,CNGB3, GNAT2, PDE6C, PDE6H; **Usher Syndrome, Iii, Ush3 -** CLRN1; **Usher Syndrome, Iic, Ush2 -** CGPR98; **Albinism, Ocular, Type I, Oa1 -** GPR143; **Usher Syndrome, I -** MYO7A; **Usher Syndrome, If, Ush1f -** PCDH15; **Retinoschisis 1, X - Linked, Juvenile, Rs1 -** RS1; **Corneal Dystrophy And Perceptive Deafness -** SLC4A11; **Albinism, Oculocutaneous, Type Ib, Oca1b -** TYR; **Usher Syndrome, Ic, Ush1c -** USH1C; **Usher Syndrome, Ig, Ush1g -** USH1G; **Usher Syndrome, Iia, Ush2a -** USH2A.