

15 Crawford St., STE 100
Needham, MA 02494
(p) 626-350-0537
(f) 626-454-1667
Lab Director: Arash Radfar M.D.
CLIA: 22D0957540



Patient Information:

14644, DONOR

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8759403

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: SSB Genetics, Dept

San Diego Sperm Bank

4915 25th Avenue NE, Ste 204W

Seattle, WA 98105

Phone: (206) 588-1484

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Lawrence M. Weiss, MD

Report Date: **Jun 26, 2024**

Accession:

FT-7032182

Test#: FT-TS14866159

Specimen Type: Blood (EDTA)

Collected: Jun 11, 2024

Accession:

N/A

FINAL RESULTS



Carrier for **ONE** genetic condition
Genetic counseling is
recommended.

TEST PERFORMED

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders)

(515 Gene Panel; gene sequencing with deletion and
duplication analysis)

Condition and Gene	Inheritance	14644, DONOR	Partner
Phenylalanine Hydroxylase deficiency (Phenylketonuria) <i>PAH</i>	AR	⊕ Carrier c.1066-11G>A (p.?)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these results, this individual is positive for a carrier mutation in 1 gene. Carrier screening for the reproductive partner is recommended to accurately assess the risk for any autosomal recessive conditions. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- Testing for copy number changes in the SMN1 gene was performed to screen for the carrier status of Spinal Muscular Atrophy. The results for this individual are within the normal range for non-carriers. See Limitations section for more information.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- X-linked genes are not routinely analyzed for male carrier screening tests. Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)

Patient: 14644, DONOR; Sex: M;
DOB: [REDACTED] **MR#:** [REDACTED]

Accession#: FT-7032182; FD Patient#: FT-PT8759403;
DocID: FT-TS14866159AA; **PAGE 1 of 5**



PHENYLALANINE HYDROXYLASE DEFICIENCY (PHENYLKETONURIA)

Patient	14644, DONOR	Partner
Result	⊕ Carrier	N/A
Variant Details	PAH (NM_000277.3) c.1066-11G>A (p.?)	N/A

What is Phenylalanine Hydroxylase deficiency (Phenylketonuria)?

Phenylalanine hydroxylase deficiency (phenylketonuria, or "PKU") is an inherited disorder that increases the levels of a substance called phenylalanine in the blood. Without treatment, phenylalanine builds up in the body, and affected children develop permanent intellectual disabilities. Seizures, delayed development, behavioral problems, and psychiatric disorders are also common. Less severe forms of this condition, sometimes called variant PKU and non-PKU hyperphenylalaninemia, have a smaller risk of brain damage.

What is my risk of having an affected child?

PKU is inherited in an autosomal recessive manner. This means that when both parents are carriers, there is a 25% (1 in 4) risk of having an affected child.

What kind of medical management is available?

PKU is screened for via newborn screening in all 50 states. There is currently no cure for PKU, but with a restricted diet and lifestyle modification, the prognosis is good. However, a restricted diet can be difficult. The support of physicians, nutritionists, social workers, nurses, and family is necessary to maintain an appropriate diet. The US Food and Drug Administration (FDA) has approved a drug (sapropterin hydrochloride) that is effective in allowing a less restricted diet without negative symptoms in some individuals with PKU.

What mutation was detected?

The detected heterozygous variant was NM_000277.3:c.1066-11G>A (p.?). This variant is predicted to create a cryptic splice acceptor site in intron 10 and disrupt the canonical splice acceptor site of exon 10. This alteration (also referred to as IVS10+546 G>A) is a common pathogenic variant in association with PKU and has been demonstrated to significantly reduce the enzyme activity (PubMed: [1769645](#), [23500595](#), [25596310](#), [12655546](#), [8990013](#)). This variant has been reviewed and classified as "Pathogenic" by ClinGen PAH variant curation expert panel in ClinVar, with multiple submitters in agreement (ClinVar: 607). The laboratory classifies this variant as pathogenic.



GENES TESTED:

Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) - 515 Genes

This analysis was run using the Beacon Preconception Carrier Screening - 515 Genes (without X-linked Disorders) gene list. 515 genes were tested with 99.5% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCC2, ABCC8, ACAD9, ACADM, ACADVL, ACAT1, ACOX1, ACSF3, ADA, ADAMTS2, ADAMTSL4, ADGRG1, ADGRV1, AGA, AGL, AGPS, AGXT, AHI1, AIPL1, AIRE, ALDH3A2, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, AMN, AMT, ANO10, AP1S1, AQP2, ARG1, ARL6, ARSA, ARSB, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7B, ATP8B1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCKDHA, BCKDHB, BCS1L, BLM, BLOC1S3, BLOC1S6, BMP1, BRIP1, BSND, CAD, CANT1, CAPN3, CASQ2, CBS, CC2D1A, CC2D2A, CCDC103, CCDC39, CCDC88C, CD3D, CD3E, CD40, CD59, CDH23, CEP152, CEP290, CERKL, CFTR, CHAT, CHRNE, CHRNA, CIITA, CLCN1, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL27A1, COL4A3, COL4A4, COL7A1, COX15, CPS1, CPT1A, CPT2, CRB1, CRTAP, CRYL1, CTNS, CTSA, CTSC, CTSD, CTSK, CYBA, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, CYP7B1, DBT, DCAF17, DCLRE1C, DDX11, DGAT1, DGUOK, DHCR7, DHDDS, DLD, DLL3, DNAH11, DNAH5, DNAI1, DNAI2, DNMT3B, DOK7, DUOX2, DYNCH2H1, DYSF, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELP1, EPG5, ERCC2, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, FAH, FAM161A, FANCA, FANCC, FANCD2, FANCE, FANCG, FANCI, FANCL, FBP1, FBXO7, FH, FKBP10, FKBP, FKTN, FMO3, FOXN1, FOXRED1, FRAS1, FREM2, FUCA1, G6PC, G6PC3, GAA, GALT, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GDF5, GFM1, GHR, GJB2, GJB6, GJB1, GLDC, GLE1, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GRHRP, GRIP1, GSS, GUCY2D, GUSB, HADH, HADHA, HADHB, HAMP, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HJV, HLCS, HMGCL, HMOX1, HOGA1, HPD, HPS1, HPS3, HPS4, HPS5, HPS6, HSD17B3, HSD17B4, HSD3B2, HYAL1, HYLS1, IDUA, IGHMBP2, IKBK, IL7R, INVS, ITGA6, ITGB3, ITGB4, IVD, JAK3, KCNJ1, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LARGE1, LCA5, LDLR, LDLRAP1, LHX3, LIFR, LIG4, LIPA, LMBRD1, LOXHD1, LPL, LRAT, LRP2, LRPPRC, LYST, MAK, MAN2B1, MANBA, MCEE, MCOLN1, MCPH1, MECP, MED17, MESF2, MFSDB, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MPI, MPL, MPV17, MRE11, MTHFR, MTR, MTRR, MTPP, MUSK, MUT, MVK, MYO15A, MYO7A, NAGA, NAGLU, NAGS, NBN, NCF2, NDRG1, NDUFAF2, NDUFAF5, NDUFS4, NDUFS6, NDUFS7, NDUFV1, NEB, NEU1, NGLY1, NPC1, NPC2, NPHP1, NPHS1, NPHS2, NR2E3, NSMCE3, NTRK1, OAT, OCA2, OPA3, OSTM1, OTOA, OTOF, P3H1, PAH, PANK2, PC, PCBD1, PCCA, PCCB, PCDH15, PCNT, PDHB, PEPD, PET100, PEX1, PEX10, PEX12, PEX13, PEX16, PEX2, PEX26, PEX5, PEX6, PEX7, PFKM, PGM3, PHGDH, PHKB, PHKG2, PHYH, PIGN, PJKV, PKHD1, PLA2G6, PLEKHG5, PLOD1, PMM2, PNPO, POLG, POLH, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PRCD, PRDM5, PRF1, PROP1, PSAP, PTPRC, PTS, PUS1, PYGM, QDPR, RAB23, RAG1, RAG2, RARSN, RARS2, RDH12, RLBP1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPE65, RPGRIP1L, RTEL1, RXYLT1, RYR1, SACS, SAMD9, SAMHD1, SCQ2, SEC23B, SEPSecs, SGCA, SGCB, SGCD, SGCG, SGSH, SKIV2L, SLC12A1, SLC12A3, SLC12A6, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC26A2, SLC26A3, SLC26A4, SLC27A4, SLC35A3, SLC37A4, SLC38A8, SLC39A4, SLC45A2, SLC4A11, SLC5A5, SLC7A7, SMARCA1, SMN1, SMPD1, SNAP29, SPG11, SPR, SRD5A2, ST3GAL5, STAR, STX11, STXBP2, SUMF1, SUOX, SURF1, SYNE4, TANGO2, TAT, TBCD, TBCE, TCIRG1, TCN2, TECPR2, TERT, TF, TFR2, TG, TGM1, TH, TK2, TMC1, TMEM216, TMEM67, TMPPRS3, TPO, TPP1, TREX1, TRIM32, TRIM37, TRMU, TSEN54, TSFM, TSHB, TSHR, TTC37, TTPA, TULP1, TYMP, TYR, TYRP1, UBR1, UNC13D, USH1C, USH2A, VDR, VLDLR, VPS11, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, WISP3, WNT10A, WRN, XPA, XPC, ZBTB24, ZFYVE26, ZNF469

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 99.57% and 99.53% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The Fulgent Germline v2019.2 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or



otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.

Gene Specific Notes and Limitations

ALG1: Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 6-13 of the ALG1 gene (NM_019109.4). **CEP290:** Copy number analysis for exons 8-13 and exons 39-42 may have reduced sensitivity in the CEP290 gene. Confirmation of these exons are limited to individuals with a positive personal history of CEP290-related conditions and/or individuals carrying a pathogenic/likely pathogenic sequence variant. **CFTR:** Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. Analysis of the intron 8 polymorphic region (e.g. IVS8-5T allele) is only performed if the p.Arg117His (R117H) mutation is detected. Single exon deletion/duplication analysis is limited to deletions of previously reported exons: 1, 2, 3, 11, 19, 20, 21. CFTR variants primarily associated with CFTR-related isolated congenital bilateral absence of the vas deferens and CFTR-related pancreatitis are not included in this analysis. CFTR variants with insufficient evidence of being cystic fibrosis mutations will not be reported either. **CRYL1:** As mutations in the CRYL1 gene are not known to be associated with any clinical condition, sequence variants in this gene are not analyzed. However, to increase copy number detection sensitivity for large deletions including this gene and a neighboring gene on the panel (GJB6, also known as connexin 30), this gene was evaluated for copy number variation. **CYP11B1:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP11B2:** The current testing method is not able to reliably detect certain pathogenic variants in this gene due to the interference by highly homologous regions. This analysis is not designed to detect or rule-out copy-neutral chimeric CYP11B1/CYP11B2 gene. **CYP21A2:** Significant pseudogene interference and/or reciprocal exchanges between the CYP21A2 gene and its pseudogene, CYP21A1P, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of the clinical findings, biochemical profile, and family history of each patient. LR-PCR is not routinely ordered for NM_000500.9:c.955C>T (p.Gln319Ter). Individuals with c.955C>T (p.Gln319Ter) will be reported as a Possible Carrier indicating that the precise nature of the variant has not been determined by LR-PCR and that the variant may occur in the CYP21A2 wild-type gene or in the CYP21A1P pseudogene. The confirmation test is recommended if the second reproductive partner is tested positive for variants associated with classic CAH. **DDX11:** Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in the DDX11 gene. **DUOX2:** The current testing method is not able to reliably detect variants in exons 6-8 of the DUOX2 gene (NM_014080.5) due to significant interference by the highly homologous gene, DUOX1. **FANCD2:** Due to pseudogene interference, copy-number-variants within exon 14-17 of the FANCD2 gene (NM_033084.4) are not evaluated and detection of single-nucleotide variants and small insertions/deletions in this region is not guaranteed. **GALT:** In general, the D2 "Duarte" allele is not reported if detected, but can be reported upon request. While this allele can cause positive newborn screening results, it is not known to cause clinical symptoms in any state. See GeneReviews for more information: <https://www.ncbi.nlm.nih.gov/books/NBK1518/> **GBA:** Significant pseudogene interference and/or reciprocal exchanges between the GBA gene and its pseudogene, GBAP1, have been known to occur and may impact results. As such, the relevance of variants reported in this gene must be interpreted clinically in the context of this individual's clinical findings, biochemical profile, and family history. The current testing method cannot detect copy-neutral rearrangements between the pseudogene and the functional gene, which have been reported in very rare cases of Gaucher disease (PubMed: 21704274). **HBA1:** Significant interference



from highly homologous regions in exons 1-2 of the HBA1 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. HBA2: Significant interference from highly homologous regions in exons 1-2 of the HBA2 gene has been recognized to occur, potentially impeding the assay's technical capability to detect pathogenic alterations during sequencing analyses. HSD17B4: Copy number analysis for exons 4-6 may have reduced sensitivity in the HSD17B4 gene. Confirmation of these exons are limited to individuals with a positive personal history of D-bifunctional protein deficiency and Perrault syndrome and/or individuals carrying a pathogenic/likely pathogenic sequence variant. LMBRD1: Copy number analysis for exons 9-12 may have reduced sensitivity in the LMBRD1 gene. Confirmation of these exons are limited to individuals with a positive personal history of combined methylmalonic aciduria and homocystinuria and/or individuals carrying a pathogenic/likely pathogenic sequence variant. MTHFR: As recommended by ACMG, the two common polymorphisms in the MTHFR gene - c.1286A>C (p.Glu429Ala, also known as c.1298A>C) and c.665C>T (p.Ala222Val, also known as c.677C>T) - are not reported in this test due to lack of sufficient clinical utility to merit testing (PubMed: [23288205](#)). NEB: This gene contains a 32-kb triplicate region (exons 82-105) which is not amenable to sequencing and deletion/duplication analysis. NPHS2: If detected, the variant NM_014625.3:c.686G>A (p.Arg229Gln) will not be reported as this variant is not significantly associated with disease when homozygous or in the compound heterozygous state with variants in exons 1-6 of NPHS2. OTOA: Due to pseudogene interference, our current testing method is not able to reliably detect variants in exons 20-28 (NM_144672.3) in the OTOA gene. SMN1: The current testing method detects sequencing variants in exon 7 and copy number variations in exons 7-8 of the SMN1 gene (NM_022874.2). Sequencing and deletion/duplication analysis are not performed on any other region in this gene. About 5%-8% of the population have two copies of SMN1 on a single chromosome and a deletion on the other chromosome, known as a [2+0] configuration (PubMed: [20301526](#)). The current testing method cannot directly detect carriers with a [2+0] SMN1 configuration but can detect linkage between the silent carrier allele and certain population-specific single nucleotide changes. As a result, a negative result for carrier testing greatly reduces but does not eliminate the chance that a person is a carrier. Only abnormal results will be reported. TERT: The TERT promoter region is analyzed for both sequencing and copy number variants. TYR: Due to the interference by highly homologous regions, our current testing method has less sensitivity to detect variants in exons 4-5 of the TYR gene (NM_000372.5). VPS45: LoF is not a known disease mechanism. WRN: Due to the interference by highly homologous regions within the WRN gene, our current testing method has less sensitivity to detect variants in exons 10-11 of WRN (NM_000553.6).

SIGNATURE:



Yan Meng, Ph.D., CGMB, FACMG on 6/26/2024
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed and its performance characteristics determined by Fulgent Therapeutics LLC CAP #8042697 CLIA #05D2043189; 4399 Santa Anita Ave., El Monte, CA, 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at [626-350-0537](tel:626-350-0537) or by email at info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes on this test please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)



CGT plus v5.4.8

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	SSB04	Sample type:	Saliva	Clinic:	Seattle Sperm Bank
Patient name:	DONOR 14644	Date of draw:		Doctor:	James Kuan
Patient DOB:		Date of receipt:	07/26/2026		
Ethnic group:	European	Report date/time:	08/11/2026		
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)

Gene :	PEX1	Allele:	Het
DNA Change:	NM_000466.3:c.1810G>A	Inheritance:	AR
Protein change:	p.Gly604Arg	OMIM phenotype:	234580; 214100; 601539
Variant classification:	Likely Pathogenic		

Hypermethioninemia due to adenosine kinase deficiency

Gene :	ADK	Allele:	Het
DNA Change:	NM_006721.4:c.829C>T	Inheritance:	AR
Protein change:	p.Arg277*	OMIM phenotype:	614300
Variant classification:	Likely Pathogenic		

Phenylketonuria

Gene :	PAH	Allele:	Het
DNA Change:	NM_000277.3:c.1066-11G>A	Inheritance:	AR
Protein change:	-	OMIM phenotype:	261600
Variant classification:	Pathogenic		



INTERPRETATION OF TEST RESULTS

Typically, a positive result does not have direct clinical consequences for the carrier individual. There is another normal gene copy for all positive autosomal recessive (AR) genes indicated in the table which provides normal biological information. The likelihood of transmission of the variant(s) to offspring is 50%, independent for each variant. If the partner, or gamete donor, screens negative for the pathogenic or likely pathogenic variants in the gene(s) included in the table for this patient, the reproductive risk would be reduced. Please note that family members may also carry the variant(s) reported here, and this information may be significant for them and their offspring.

If a patient and partner, or gamete donor, are both carriers of variants in the same gene associated with AR inheritance, there is a 25% chance that any child they have together would be affected. If a female patient is a carrier for an X-linked condition, there is a 50% chance that each of the reproductive couple's children would also be a carrier. Males would typically express symptoms of the condition, and females are typically unaffected or may display milder symptoms.

For genes with a negative test result, the risk of having children affected by the associated disorders decreases significantly compared to the general population. This also the case for a negative personal result when a reproductive partner or a gamete donor is a carrier for a pathogenic or likely pathogenic variant in one or more of the tested genes. However, due to test limitations associated with any genetic test, this low risk is not zero (see limitations section and informed consent form)

TEST DESCRIPTION

The Carrier Genetic Test (CGT) is a preconception DNA screening test that aims to identify individuals and couples at increased risk of conceiving children affected by a monogenic disease. Knowledge of this risk may influence a couple's decision to conceive or encourage the couple to adopt preventive measures, including preimplantation genetic testing for the at risk disease (PGT-M) prenatal genetic testing, or to use donated gametes. The multigene CGT interrogates thousands of DNA variants using a high-throughput technology (Next Generation Sequencing, NGS).

COMMENTS

None

TEST METHODOLOGY

DNA is isolated from the sample, usually blood or saliva, and analyzed by whole exome sequencing by NGS. This includes capture and sequence of all human exons and other gene regions of interest where known disease-causing variants are located. Sequencing raw data is then analyzed using bioinformatics (bioinformatic pipeline v3.0), which includes sequence alignment against the GRCh37 human genome reference, variant calling, annotation, and real-time interpretation of variants. QC parameters include, all reported samples that will have a minimum of 7Gb of data, with minimal mean coverage greater than 75x, and a specific depth analysis for more than 68,000 DNA positions where known pathogenic variants are located. In addition, complementary tests (non-NGS techniques) are performed for the following genes, if included, CFTR gene intronic variant/s; SMN1 gene exon 7-deletion; CYP21A2 gene frequent mutations; HBA1 and HBA2 genes frequent deletions; FXN gene GAA repeat sizing; FMR1 gene CGG repeat sizing (females only); DMD gene frequent deletions/duplications; F8 gene intron 22 inversion (females only). When requested, CNV analysis by MLPA is performed for CFTR, HBB and HBA1/HBA2. Based on our validations studies, reported samples will have analytical detection rate for SNV variants as per the control sample NA12878 (Control positive); PASS value: NA12878 Sensitivity SNV ≥ 0.97000 .



TEST LIMITATIONS

In the general population, there is a 3-5% risk for birth defects caused by genetic and/or non-genetic factors not detected by this type of test.

Analytically, the CGT test does not cover all known monogenic diseases nor all disease-causing variants for each tested gene. The test does not include the analysis of conditions associated with mitochondrial DNA nor multifactorial nor digenic inheritance. The test does not detect large rearrangements (inversions, deletions and duplications more than 15 nucleotides), variants located in regulatory regions or intronic regions outside the +/-3bp cut off (except if otherwise indicated), or in low sequence coverage areas (<7x). DNA changes caused by trinucleotide repeat expansions are not detected, except those indicated in the methodology section. For copy number variation analysis, when a normal result is obtained (2 copies detected), it is not possible to confirm that one copy is present in each of the two alleles (non-carrier) or if both copies are present in cis on the same allele, with no copies in the other allele (silent carrier). Clinical sensitivity varies among conditions. In particular, the sensitivity for SMN1 is approximately 96% because it is not possible to identify silent carriers among patients with 2 SMN1 copies detected and because point mutations or small indels are not analyzed. The CYP21A2 gene analysis presents unique challenges due to its high sequence homology (~98%) with the pseudogene CYP21A1P, which leads to frequent gene rearrangements and complex mutations. These challenges can cause difficulties in distinguishing CYP21A2 from CYP21A1P, increasing the risk of misdiagnosis. Different testing methods have specific limitations, requiring a combination of techniques such as long-range PCR, Sanger sequencing, next-generation sequencing (NGS), MLPA, and qPCR to achieve accurate results. These challenges and limitations may lead to false or inconclusive results. Therefore, genetic counselling is strongly recommended to evaluate the findings, discuss potential implications, and determine whether additional testing (such as MLPA) is necessary for an accurate diagnosis. In summary, sensitivity to detect pathogenic variants, if they result from complex gene conversion/gene rearrangements events, may be reduced. For the HEXB gene, the common 16 kb deletion that causes disease in 30% of affected patients is not included in CGT analysis. Furthermore, this test does not evaluate the HFE gene.

Then, a negative CGT result significantly reduces but does not completely exclude the possibility of being a carrier of a variant associated with single gene disorders (see residual risk table). The presence of pseudogenes and/or rare polymorphisms and/or homopolymers may lead to false negative or false positive results. In addition, a negative result for the CGT variants does not exclude the possibility of a de novo variant occurring in the offspring. Germline mosaicism or low-level somatic mosaicism cannot be detected. As with any laboratory test, there is a small chance that this result may be inaccurate for a procedural reason such as an error during sample collection, labelling, processing, data collection or interpretation. Please note that the clinical classification of variants can change over time. To check whether there have been any changes to the classification of reported variants, please contact IGENOMIX.

LEGAL/QUALITY

Igenomix USA, Inc will only release the report once a completed test requisition form is received. The clinic/clinician/certified health professional requesting the test is responsible for obtaining and taking custody of "Informed Consent" from the patient as depicted by national guidelines and/or legislation. This test was developed, and its performance characteristics determined by IGENOMIX SPAIN LAB, SLU. It has not been cleared or approved by the US Food and Drug Administration. The test is used as a laboratory developed test for clinical purposes.

Part of this test has been outsourced to a reference laboratory whose Quality Management System is based on high Quality Standards, periodically monitored by Igenomix SPAIN* and audited by independent external groups.

*IGENOMIX SPAIN holds CLIA Certificate of Compliance: #99D2146167.



EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by Igenomix USA, Inc are exclusively intended to be interpreted by qualified/certified health professionals. The Carrier screening test (CGT) for USA samples is currently being performed outside of Igenomix USA.

The result obtained by this test and the information that could be derived from it, cannot be considered in any case as substitute of genetic counselling or medical treatment by a trained professional neither represent itself a medical enquiry. We recommend that you consult your physician for genetic testing & counselling upon reception of your results.

Any result should be interpreted in the context of all available clinical findings, within the general context of a medical investigation, which must be conducted by clinically trained professionals. Igenomix USA, Inc is not responsible for any decisions made or actions undertaken by the contracting party based on the results provided by Igenomix USA, Inc or otherwise., nor the harmful temporary consequences diverted by its use, making specific discretion of taking appropriate legal measures assuming an improper use of those mentioned studies and analysis.

SIGNED

COUNTERSIGNED

Arantxa Hervas PhD
3025-CV
General Supervisor

Diana Valbuena

This test or part of this test has been outsourced to a referral Laboratory. Lab CLIA No.: 99D2146167

Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)

What is Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)?

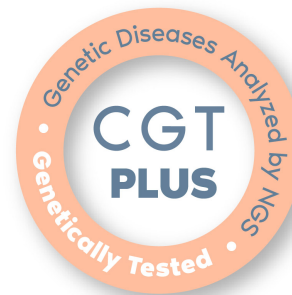
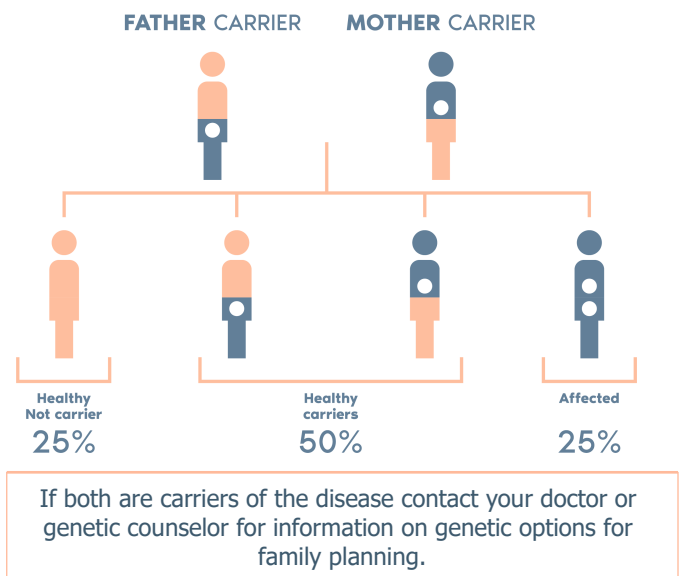
Heimler syndrome-1 (HMLR1), which represents the mildest end of the peroxisomal biogenesis disorder spectrum, is a rare autosomal recessive disorder characterized by sensorineural hearing loss, enamel hypoplasia of the secondary dentition, and nail abnormalities (Ratbi et al., 2015). Zellweger syndrome is an autosomal recessive systemic disorder characterized clinically by severe neurologic dysfunction, craniofacial abnormalities, and liver dysfunction, and biochemically by the absence of peroxisomes. Most severely affected individuals with classic Zellweger syndrome phenotypes die within the first year of life (summary by Wanders, 2004). Peroxisome biogenesis disorder-1B (PBD1B) is characterized by the overlapping phenotypes of neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), which represent the milder manifestations of the Zellweger syndrome spectrum (ZSS) of peroxisome biogenesis disorders (PBDs). Initial presentation and natural history varies, with many children presenting as newborns, whereas others do not come to attention until later. Most affected children have hypotonia, but unlike Zellweger syndrome, there is a degree of psychomotor development, and some patients achieve head control, sit unsupported, and may even walk independently. Many can communicate, and although the language is rare, there have been children who have near-normal language for their age. Craniofacial anomalies are similar to but less pronounced than Zellweger syndrome. In some individuals leukodystrophy develops, with degeneration of myelin, loss of previously acquired skills, and development of spasticity; this may stabilize, or progress and be fatal. In PBD1B, the most common manifestations that are less apparent in ZS are sensorineural hearing loss and retinitis pigmentosa (summary by Steinberg et al., 2006). While Zellweger syndrome usually results in death in the first year of life, children with the NALD presentation may reach their teens, and those with the IRD presentation may reach adulthood (summary by Waterham and Ebberink, 2012).

What is the next step if I am a carrier of Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)?

If you are a carrier of Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD) it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

If your partner tests negative for Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD), the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.



What if both parents are carriers of Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)?

When both parents are carriers of Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD), the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

In this case it is advisable to use the same assay (CGT) to test candidate donors and choose one that is negative for the same condition.

Hypermethioninemia due to adenosine kinase deficiency

What is Hypermethioninemia due to adenosine kinase deficiency?

Hypermethioninemia due to adenosine kinase deficiency is an autosomal recessive inborn error of metabolism characterized by global developmental delay, early-onset seizures, mild dysmorphic features, and characteristic biochemical anomalies, including persistent hypermethioninemia with increased levels of S-adenosylmethionine (AdoMet) and S-adenosylhomocysteine (AdoHcy); homocysteine is typically normal (Bjursell et al., 2011).

What is the next step if I am a carrier of Hypermethioninemia due to adenosine kinase deficiency?

If you are a carrier of Hypermethioninemia due to adenosine kinase deficiency it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

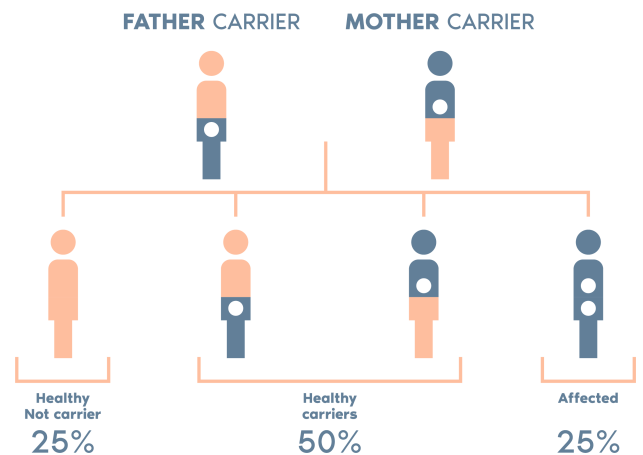
If your partner tests negative for Hypermethioninemia due to adenosine kinase deficiency, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Hypermethioninemia due to adenosine kinase deficiency?

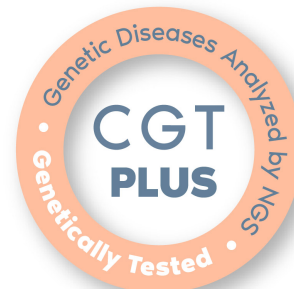
When both parents are carriers of Hypermethioninemia due to adenosine kinase deficiency, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

In this case it is advisable to use the same assay (CGT) to test candidate donors and choose one that is negative for the same condition.



If both are carriers of the disease contact your doctor or genetic counselor for information on genetic options for family planning.



Phenylketonuria

What is Phenylketonuria?

Phenylketonuria follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the PAH gene located on chromosomal region 12q23.2. The age of onset is neonatal. This disease is characterized by gradual developmental delay, stunted growth, microcephaly, seizures, tremors, eczema, vomiting, and musty odor. Untreated patients subsequently develop intellectual disability, behavioral disorders (hyperactivity) and motor disorders. The prevalence is 1:2,600-1:200,000.

What is the next step if I am a carrier of Phenylketonuria?

If you are a carrier of Phenylketonuria it is important that your partner (or gamete donor) is tested to determine if she/he is also a carrier of this condition.

What if my partner isn't a carrier?

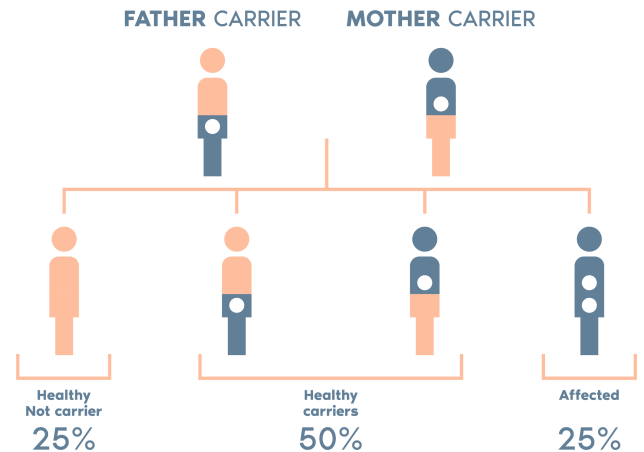
If your partner tests negative for Phenylketonuria, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Phenylketonuria?

When both parents are carriers of Phenylketonuria, the probability of having a child with the disease is 25% in each pregnancy. (See graph)

What if I am going to use gamete donation?

In this case it is advisable to use the same assay (CGT) to test candidate donors and choose one that is negative for the same condition.



If both are carriers of the disease contact your doctor or genetic counselor for information on genetic options for family planning.



LIST OF ANALYZED GENES

Gene mean coverage >100x CHRNA, CHRNA, ADK, NAGA, SLC3A1, SERPINA1, AQP2, CYP19A1, ASNS, CAPN3, CASQ2, TYR1, CTSD, CLCN1, CYP11A1, COL4A3, COL7A1, COL4A4, GJB2, CNGA1, POR, CYP11B2, PCBD1, ERCC2, SLC26A3, ETVB, TYMP, ERCC3, ERCC5, FMO3, FH, MPV17, GNRHR, GHRHR, HBA1, HBA2, HBB, HSPG2, VSX2, HADHB, LAMC2, LAMB3, LIFR, LHCGR, PLOD1, LAMA2, MTR, MTTP, MPL, NDUFV1, NEB, PRF1, PEX2, ALPL, POU1F1, POLG, PRDX1, PSAP, F2, BCHE, PDHB, RAG1, RAG2, RPE65, PDE6A, AHCY, SAG, SURF1, TG, TSHB, TGM1, TH, NTRK1, DPAGT1, UGT1A1, ATP6V1B1, BBS1, ETVF1, PCCA, PCCB, GLDC, AMT, GCSH, DLD, DBT, BCKDHB, MVK, IDUA, F11, MYO7A, LRP2, SGCA, JAK3, GUCY2D, GCH1, ACADSB, PEX5, TTPA, IGHMBP2, ABCC8, AUH, STAR, CPT2, PPT1, CNGB1, LAMA3, DDB2, HADHA, SLC12A3, GSS, NPC2, CTSK, MOGS, SGCD, PEX6, PROP1, RAPS, HADH, ABCA3, ABCA4, PEX7, PEX12, CYP11B1, PMM2, SLC5A5, HSD17B4, PEX1, VRK1, TRIM32, ATP8B1, CFTR, MTRR, MYO15A, NBN, SLC37A4, NDUFS4, NPHS1, SPG7, ADGRV1, DHCR7, FANCG, DYSF, COLQ, AGPS, ABCB11, ABCD4, PNPO, DYNC2H1, DNAH5, GLE1, TSHR, SLC22A5, CCN6, ASS1, DPM1, MTMR2, LARGE1, SLC7A7, PLA2G6, BCS1L, OTOF, ELP1, GNE, NDUFS6, SLC25A13, SLC25A15, SLC19A2, EIF2B5, EIF2AK3, EDAR, ADGRG1, SLC7A9, CRB1, SCO2, GRHR, GALK1, SLC17A5, DNAI1, AIPL1, GJB6, NR2E3, SACS, IDH3B, ADAMTS2, ALG6, TCIRG1, GRIP1, BLM, WRN, TFR2, TSFM, NPHS2, ACAD8, EVC, SLC12A6, HPS1, TRIM37, CNGB3, USH1C, MCOLN1, NDRG1, SGSH, AAAS, DNAI2, CRTAP, TMPRSS3, PCDH15, CDH23, HSD17B3, SLC35A1, SLC26A4, LDLRAP1, MLC1, VPS13A, HAX1, DNAJC12, HPS3, RAB23, BBS2, WNT10A, CTNS, CLRN1, GBA1, NOP10, EXOSC3, CYP27A1, GDAP1, TREX1, SMARCA1, GNMT, GFM1, GP1BA, PKHD1, SLC26A2, CLN6, SAMHD1, DUOX2, TPO, GAA, FTCD, PRODH, ALDH4A1, POMGNT1, ALMS1, HEXB, PHGDH, ATP7B, ACADS, GALC, LYST, TYR, LDLR, GALE, GALT, ACADM, IVD, SLC39A4, NPHP1, FANCA, EVC2, POMT1, POMT2, FKTN, SBDs, HGD, MMAA, LRPPRC, MMAB, ATM, SMPD1, NPC1, OSTM1, CTH, GNS, USH1G, ABCA12, ACAT1, VPS13B, FRAS1, GBE1, GNPTAB, WHRN, SUMF1, TPP1, ETVF, MEV, DHDDS, SH3TC2, NAGS, CPS1, ASL, ARG1, BCKDHA, HJV, CERKL, USH2A, MCEE, ETHE1, PYGM, COL27A1, CYBA, NCF2, TSEN54, PC, GCDH, RDH12, RTEL1, AHI1, SGCG, UNC13D, ADA, MCCC1, MCCC2, HLCS, MMUT, CYP17A1, ESCO2, MAN2B1, MANBA, CYP27B1, ALDH3A2, ACADVL, NDUFAF2, NLRP7, HPD, LPL, ACOX1, MKS1, TMEM67, VPS45, POLR1C, CEP290, BBS10, SLC4A11, TRMU, RNASEH2B, RNASEH2C, P3H1, HGSNAT, MAT1A, CYP11B1, PFKM, BBS12, DOLK, SPG11, AGL, RPRIP1L, G6PC3, ACAD9, MFSD8, XPA, CCDC88C, LCA5, OCA2, GLB1, CA2, GUSB, RARS2, ARSB, SLC46A1, MMADHC, ZFYVE26, CC2D2A, GALNS, FUCA1, F5, PAH, EYS, LMBRD1, QDPR, PTS, ALDOB, DUOX2A, DPYD, SEPS2, TAT, LOXHD1, CTSA, XPC, AGA, TMEM216, OAT, CBS, LIPA, FAM161A, HOGA1, FOXRED1, SLC25A20, ANO10, G6PC1, CYP21A2, FAH, HSD3B2, HMGCL, FANCC, SKIC3

Gene mean coverage 50x-100x CHAT, B4GALT1, HMOX1, MPI, RMRP, GP9, CPT1A, LHX3, KCNJ11, TNXB, GDF5, GAMT, UBE3A, NDUFS7, RAX, CTSC, PEX10, TRDN, COX15, MED17, AGXT, ST3GAL5, MESP2, CHST6, SLC35C1, ALG1, DCLRE1C, SLC19A3, SLC45A2, NHP2, OPA3, HEXA, CLN3, HYAL1, MTHFR, MCPH1, SRD5A2, AIRE, LIPH, ARSA, GNPTG, ASPA, CLN5, PUS1, NEU1, NCF1, SLC6A19, ARL13B, ERCC8, ERCC6, NAGLU, MMACHC, DOK7, HYL1, FAM20C, IYD, NDUFAF5, ACF3, TECPR2, NADK2, VPS53

Gene mean coverage < 50x GP1BB, TF, CIITA, SMN1, SLC35A3, PANK2, BSND, FKRP, MLYCD, FXN, CLN8, PEX26, BTG, SLC35D1, PPM1K

GLOSSARY

TYPES OF INHERITANCE:

- AR: Autosomal recessive**
Inherited conditions that require two pathogenic variants (one from each parent) in a given gene to display symptoms.
- XR: X-linked recessive**
The gene is located on the X chromosome. Men with a pathogenic variant have the disease. Women with a pathogenic variant are carriers and generally asymptomatic or may mild symptoms.
- Digenic inheritance**
In some diseases, the symptoms could be explained by the coexistence of pathogenic variants in two different genes related with the disease instead of two pathogenic variants in the same gene.

ALLELES:

Pathogenic variants present in the two copies of a gene.

- Homozygous pathogenic variant (Hom.):**
Each copy of the gene has the same pathogenic variant. Generally, this is associated with clinical symptoms.
- Compound heterozygous (Het.):**
Each copy of the gene has a different pathogenic variant. Generally, this is associated with clinical symptoms. This situation is referred as having variants "in trans".

Pathogenic variant present in one copy of a gene.

- Heterozygous pathogenic variant (Het.):**
Only one copy of a gene has a pathogenic variant. There is another normal gene copy.

Note: Sometimes an individual has two pathogenic variants in the same gene copy. This situation is referred as having variants in cis and it is considered as a single pathogenic variant.

CNV:

Refers to copy number variation (deletion or duplication), i.e., the number of copies of a particular gene (or gene region) is different from the usual two copies.

LARGE GENE CONVERSION:

Refers to pathogenic variants caused by gene sequence exchange or replacement between a normal functional gene and a quasi-identical non-functional gene (pseudogene).



Autosomal recessive conditions				
Chrom	Gene	Disease/ Condition	Carrier Rate	Residual Risk
12	AAAS	Triple-A syndrome (achalasia-addisonianism-alacrimia)	1 in 436	1 in 8266
2	ABCA12	Ichthyosis, congenital, autosomal recessive, type 4A; ICAR, type 4B (harlequin)	1 in 194	1 in 715
16	ABCA3	Surfactant metabolism dysfunction, pulmonary, type 3	1 in 500	1 in 7143
1	ABCA4	Stargardt disease 1; Retinitis pigmentosa 19; Cone-rod dystrophy 3	1 in 62	1 in 3100
2	ABCB11	Cholestasis, benign recurrent intrahepatic, type 2; Cholestasis, progressive familial intrahepatic, type 2	1 in 276	1 in 3450
11	ABCC8	Hyperinsulinemic hypoglycemia, type 1 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 192	1 in 1920
14	ABCD4	Methylmalonic aciduria and homocystinuria, cbJ type	1 in 496	1 in 49501
11	ACAD8	Isobutyryl-CoA dehydrogenase deficiency	< 1 in 500	Reduced
3	ACAD9	Acyl-CoA dehydrogenase 9 deficiency (mitochondrial complex I deficiency, nuclear, type 20)	1 in 309	1 in 3090
1	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	1 in 60	1 in 600
12	ACADS	Short-chain acyl-CoA dehydrogenase deficiency	1 in 102	1 in 10200
10	ACADSB	Short/branched-chain acyl-CoA dehydrogenase deficiency	1 in 500	1 in 1125
17	ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	1 in 112	1 in 1120
11	ACAT1	Alpha-methylacetoacetic aciduria (3-ketothiolase deficiency)	1 in 300	1 in 3750
17	ACOX1	Peroxisomal acyl-CoA oxidase deficiency	< 1 in 500	Reduced
16	ACSF3	Combined malonic and methylmalonic aciduria	1 in 90	1 in 900
20	ADA	Severe combined immunodeficiency due to adenosine deaminase deficiency (ADA)	1 in 390	1 in 2600
5	ADAMTS2	Ehlers-Danlos syndrome, dermatosparaxis type	< 1 in 500	Reduced
16	ADGRG1	Polymicrogyria, bilateral frontoparietal	< 1 in 500	Reduced
5	ADGRV1	Usher syndrome, type 2C	1 in 80	1 in 800
10	ADK	Hypermethioninemia due to adenosine kinase deficiency	1 in 500	1 in 1498
4	AGA	Aspartylglucosaminuria (glycosylasparaginase deficiency)	< 1 in 500	Reduced
1	AGL	Glycogen storage disease, type 3	1 in 200	1 in 2000
2	AGPS	Rhizomelic chondrodysplasia punctata, type 3	< 1 in 500	Reduced
2	AGXT	Hyperoxaluria, primary, type 1	1 in 174	1 in 2486
20	AHCY	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	< 1 in 500	Reduced
6	AHI1	Joubert syndrome, type 3	1 in 334	1 in 706
17	AIP1	Leber congenital amaurosis, type 4	1 in 400	1 in 1060
21	AIRE	Autoimmune polyendocrinopathy syndrome, type 1	1 in 310	1 in 4429
17	ALDH3A2	Sjogren-Larsson syndrome	< 1 in 500	Reduced
1	ALDH4A1	Hyperprolinemia, type 2	1 in 500	1 in 49951
9	ALDOB	Fructose intolerance, hereditary	1 in 80	1 in 3300
16	ALG1	Congenital disorder of glycosylation, type 1K	1 in 87	1 in 1740
1	ALG6	Congenital disorder of glycosylation, type 1C	1 in 500	1 in 5000
2	ALMS1	Alström syndrome	1 in 250	1 in 1667
1	ALPL	ALPL-related conditions	1 in 274	1 in 2740
3	AMT	Glycine encephalopathy	1 in 310	1 in 6200
3	ANO10	Spinocerebellar ataxia, autosomal recessive, type 10	1 in 224	1 in 2236
12	AQP2	Diabetes insipidus, nephrogenic, type 2	< 1 in 500	Reduced
6	ARG1	Argininemia (arginase deficiency)	1 in 418	1 in 13933
3	ARL13B	Joubert syndrome type 8	< 1 in 500	Reduced
22	ARSA	Metachromatic leukodystrophy	1 in 192	1 in 1920
5	ARSB	Mucopolysaccharidosis, type 6 (Maroteaux-Lamy syndrome)	1 in 314	1 in 3925
7	ASL	Argininosuccinic aciduria	1 in 116	1 in 1170
7	ASNS	Asparagine synthetase deficiency	< 1 in 500	Reduced
17	ASPA	Canavan disease	1 in 416	1 in 13867
9	ASS1	Citrullinemia, type 1	1 in 300	1 in 3750
11	ATM	ATM-related conditions	1 in 150	1 in 1000
2	ATP6V1B1	Renal tubular acidosis with deafness	< 1 in 500	Reduced
13	ATP7B	Wilson disease	1 in 90	1 in 3000
18	ATP8B1	Cholestasis, progressive familial intrahepatic, type 1; Cholestasis, benign recurrent intrahepatic, type 1	1 in 738	1 in 1803
9	AUH	3-methylglutaconic aciduria, type 1	< 1 in 500	Reduced
9	B4GALT1	Congenital disorder of glycosylation, type 2D	< 1 in 500	<1 in 50000
11	BBS1	Bardet-Biedl syndrome, type 1	1 in 152	1 in 1520
12	BBS10	Bardet-Biedl syndrome, type 10	1 in 237	1 in 4740
4	BBS12	Bardet-Biedl syndrome, type 12	1 in 500	1 in 10000
16	BBS2	Bardet-Biedl syndrome, type 2	1 in 200	1 in 4000
3	BCHE	Butyrylcholinesterase deficiency	1 in 30	1 in 200
19	BCKDHA	Maple syrup urine disease, type 1A	1 in 320	1 in 3200
6	BCKDHB	Maple syrup urine disease, type 1B	1 in 365	1 in 2808
2	BCS1L	Mitochondrial complex III deficiency nuclear type 1; GRACILE syndrome; Bjornstad syndrome	1 in 320	1 in 2133
15	BLM	Bloom syndrome	1 in 320	1 in 3200
1	BSND	Bartter syndrome, type 4A	< 1 in 500	Reduced
3	BTD	Biotinidase deficiency	1 in 120	1 in 6000
8	CA2	Osteopetrosis with renal tubular acidosis (osteopetrosis, autosomal recessive, type 3)	< 1 in 500	<1 in 1000
15	CAPN3	Limb-girdle muscular dystrophy, type 1 (LGMD R1)	1 in 103	1 in 686
1	CASQ2	Ventricular tachycardia, catecholaminergic polymorphic, type 2	1 in 168	1 in 8354

21	CBS	Homocystinuria due to cystathionine beta-synthase	1 in 274	1 in 2740
4	CC2D2A	Joubert syndrome, type 9; Meckel syndrome, type 6; COACH syndrome, 2	1 in 196	1 in 2800
14	CCDC88C	Hydrocephalus, congenital, type 1	1 in 500	1 in 7143
6	CCN6	Progressive pseudorheumatoid dysplasia	< 1 in 500	Reduced
10	CDH23	Deafness, autosomal recessive, type 12; Usher syndrome, type 1D	1 in 216	1 in 1080
12	CEP290	Meckel syndrome, type 4; Joubert syndrome, type 5; Leber congenital amaurosis, type 10	1 in 150	1 in 3750
2	CERKL	Retinitis pigmentosa, type 26	1 in 250	1 in 1667
7	CFTR	Cystic fibrosis	1 in 25	1 in 833
10	CHAT	Myasthenic syndrome, congenital, type 6, presynaptic	1 in 600	1 in 30000
17	CHRNAE	Myasthenic syndrome, congenital, type 4B, fast-channel; Myasthenic syndrome, congenital, type 4C, associated with acetylcholine receptor deficiency	1 in 244	1 in 2440
2	CHRNAE	Multiple pterygium syndrome (MPS), Escobar type; MPS, lethal type	1 in 400	1 in 1597
16	CHST6	Macular corneal dystrophy	1 in 80	1 in 2666
16	CIITA	Bare lymphocyte syndrome, type 2, complementation group A	< 1 in 500	Reduced
7	CLCN1	Myotonia congenita, recessive	1 in 159	1 in 319
16	CLN3	Ceroid lipofuscinosis, neuronal, type 3	1 in 242	1 in 367
13	CLN5	Ceroid lipofuscinosis, neuronal, type 5	1 in 400	1 in 8000
15	CLN6	Ceroid lipofuscinosis, neuronal, type 6	< 1 in 500	Reduced
8	CLN8	Ceroid lipofuscinosis, neuronal, type 8	< 1 in 500	Reduced
3	CLRN1	Usher syndrome, type 3A	1 in 250	1 in 1667
4	CNGA1	Retinitis pigmentosa type 49	1 in 625	1 in 1171
16	CNGB1	Retinitis pigmentosa type 45	1 in 200	1 in 4000
8	CNGB3	Achromatopsia, type 3	1 in 125	1 in 1250
9	COL27A1	Steel syndrome	1 in 500	1 in 2500
2	COL4A3	Alport syndrome, autosomal recessive, type 3B	1 in 300	1 in 1500
2	COL4A4	Alport syndrome, autosomal recessive, type 2	1 in 425	1 in 4250
3	COL7A1	Dystrophic epidermolysis bullosa (DEB), Hallopeau-Siemens (HS) type and non-HS type; DEB pruriginosa; DEB pretibial	1 in 150	1 in 1000
3	COLQ	Myasthenic syndrome, congenital, type 5	1 in 805	1 in 1420
10	COX15	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 2; Leigh syndrome due to cytochrome c oxidase deficiency	1 in 480	1 in 9600
2	CPS1	Carbamoylphosphate synthetase 1 deficiency	1 in 500	1 in 2500
11	CPT1A	Carnitine palmitoyltransferase type 1A deficiency, hepatic	< 1 in 500	Reduced
1	CPT2	Carnitine palmitoyltransferase type 2 deficiency, lethal neonatal; Carnitine palmitoyltransferase type 2 deficiency, infantile	1 in 100	1 in 667
1	CRB1	Retinitis pigmentosa, type 12; Leber congenital amaurosis, type 8	1 in 158	1 in 1580
3	CRTAP	Osteogenesis imperfecta, type 7	1 in 1416	1 in 3539
1	CTH	Cystathioninuria	1 in 160	1 in 3200
17	CTNS	Nephropathic cystinosis	1 in 200	1 in 400
20	CTSA	Galactosialidosis	1 in 557	1 in 11140
11	CTSC	Haim-Munk syndrome; Papillon-Lefevre syndrome	1 in 500	1 in 2496
11	CTSD	Ceroid lipofuscinosis, neuronal, type 10	< 1 in 500	Reduced
1	CTSK	Pycnodysostosis	< 1 in 500	Reduced
16	CYBA	Chronic granulomatous disease, type 4	< 1 in 500	Reduced
15	CYP11A1	46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency	1 in 500	1 in 7143
8	CYP11B1	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	1 in 300	1 in 2000
8	CYP11B2	Hypoadosteronism, congenital, due to CMO I deficiency	< 1 in 500	Reduced
10	CYP17A1	17 alpha(a)-hydroxylase/17,20-lyase deficiency	< 1 in 500	Reduced
15	CYP19A1	Aromatase deficiency	< 1 in 500	Reduced
2	CYP18A1	Glaucoma, primary congenital, type 3A	1 in 196	1 in 1960
6	CYP21A2	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	1 in 62	1 in 1240
2	CYP27A1	Cerebrotendinous xanthomatosis	1 in 275	1 in 5500
12	CYP27B1	Vitamin D-dependent rickets, type 1	< 1 in 500	Reduced
1	DBT	Maple syrup urine disease, type 2	1 in 410	1 in 2733
10	DCLRE1C	Omenn syndrome; Severe combined immunodeficiency, Athabaskan type	< 1 in 500	Reduced
11	DDB2	Xeroderma pigmentosum, complementation group E	< 1 in 500	Reduced
11	DHCR7	Smith-Lemli-Opitz syndrome	1 in 100	1 in 1000
1	DHDDS	Retinitis pigmentosa, type 59	< 1 in 500	Reduced
7	DLD	Dihydrolipoamide dehydrogenase deficiency	< 1 in 500	Reduced
5	DNAH5	Ciliary dyskinesia, primary, type 3, with or without situs inversus	1 in 130	1 in 520
9	DNAI1	Ciliary dyskinesia, primary, type 1, with or without situs inversus	1 in 323	1 in 1615
17	DNAI2	Ciliary dyskinesia, primary, type 9, with or without situs inversus	< 1 in 500	Reduced
10	DNAJC12	Hyperphenylalaninemia, mild, non-BH4-deficient	1 in 500	Reduced
4	DOK7	Fetal akinesia deformation sequence, type 3; Myasthenic syndrome, congenital, type 10	1 in 262	1 in 2620
9	DOLK	Congenital disorder of glycosylation, type 1M	< 1 in 500	Reduced
11	DPAGT1	Congenital disorder of glycosylation, type 1J; Myasthenic syndrome, congenital, type 13	< 1 in 500	Reduced
20	DPM1	Congenital disorder of glycosylation, type 1E	< 1 in 500	<1 in 1750
1	DPYD	Dihydropyrimidine dehydrogenase deficiency	1 in 558	1 in 55701
15	DUOX2	Thyroid dysmorphogenesis, type 6	1 in 80	1 in 2600
15	DUOX2	Thyroid dysmorphogenesis, type 5	1 in 80	1 in 800
11	DYNC2H1	Short-rib thoracic dysplasia, type 3, with or without polydactyly	1 in 50	1 in 500
2	DYSF	Miyoshi muscular dystrophy, type 1; Limb-girdle muscular dystrophy, type 2 (LGMD R2)	1 in 300	1 in 3000

2	EDAR	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type	1 in 746	1 in 14920
2	EIF2AK3	Wolcott-Rallison syndrome	< 1 in 500	<1 in 2500
3	EIF2B5	Leukoencephalopathy with vanishing white matter (VWM)	1 in 400	1 in 8000
9	ELP1	Familial dysautonomia	1 in 200	1 in 2000
19	ERCC2	Trichothiodystrophy, type 1; Xeroderma pigmentosum, group D	1 in 500	1 in 10000
2	ERCC3	Trichothiodystrophy, type 2	1 in 436	1 in 14533
13	ERCC5	Cerebrooculofacioskeletal syndrome 3; Xeroderma pigmentosum, group G;Xeroderma pigmentosum, group G/Cockayne syndrome	< 1 in 500	Reduced
10	ERCC6	Cockayne syndrome, type B; Cerebrooculofacioskeletal syndrome, type 1	1 in 300	1 in 2000
5	ERCC8	Cockayne syndrome, type A	1 in 514	1 in 3960
8	ESCO2	Roberts syndrome	< 1 in 500	Reduced
15	ETFA	Glutaric acidemia, type 2A	< 1 in 500	Reduced
19	ETFB	Glutaric acidemia, type 2B	< 1 in 500	Reduced
4	ETFDH	Glutaric acidemia, type 2C	1 in 300	1 in 2000
19	ETHE1	Ethylmalonic encephalopathy	< 1 in 500	Reduced
4	EVC	Ellis-van Creveld syndrome	1 in 240	1 in 960
4	EVC2	Ellis-van Creveld syndrome	1 in 300	1 in 2000
9	EXOSC3	Pontocerebellar hypoplasia, type 1B	1 in 139	1 in 4633
6	EYS	Retinitis pigmentosa, type 25	1 in 100	1 in 1000
4	F11	Factor XI deficiency	1 in 200	1 in 2500
11	F2	Prothrombin deficiency	1 in 300	1 in 1500
1	F5	Factor V deficiency	1 in 500	1 in 10000
15	FAH	Tyrosinemia, type 1	1 in 200	1 in 2000
2	FAM161A	Retinitis pigmentosa, type 28	1 in 350	1 in 3500
7	FAM20C	Raine syndrome	< 1 in 500	<1 in 1000
16	FANCA	Fanconi anemia, complementation group A	1 in 200	1 in 400
9	FANCC	Fanconi anemia, complementation group C	1 in 480	1 in 2400
9	FANCG	Fanconi anemia, complementation group G	< 1 in 500	Reduced
1	FH	Fumarase deficiency	1 in 500	1 in 3333
19	FKRP	Muscular dystrophy-dystroglycanopathy, type 5A (Walker-Warburg syndrome); Type 5B; Type 5C (limb-girdle muscular dystrophy, type 9 [LGMDR9])	1 in 176	1 in 2514
9	FKTN	Muscular dystrophy-dystroglycanopathy, type 4A (Walker-Warburg syndrome); Type 4B; Type 4C (limb-girdle muscular dystrophy, type 13 [LGMD R13])	< 1 in 500	Reduced
1	FMO3	Trimethylaminuria	1 in 100	1 in 1000
11	FOXRED1	Mitochondrial complex I deficiency, nuclear type 19	1 in 439	1 in 21950
4	FRAS1	Fraser syndrome, type 1	1 in 300	1 in 3000
21	FTCD	Glutamate formiminotransferase deficiency	1 in 355	1 in 7000
1	FUCA1	Fucosidosis	1 in 1149	1 in 4880
9	FXN	Friedreich ataxia	1 in 91	1 in 1014
17	G6PC1	Glycogen storage disease, type 1A	1 in 300	1 in 3000
17	G6PC3	Dursun syndrome	< 1 in 500	<1 in 1170
17	GAA	Glycogen storage disease, type 2	1 in 100	1 in 666
14	GALC	Krabbe disease	1 in 120	1 in 218
1	GALE	Galactose epimerase deficiency	< 1 in 500	Reduced
17	GALK1	Galactokinase deficiency with cataracts	1 in 200	1 in 2500
16	GALNS	Mucopolysaccharidosis, type 4A	1 in 311	1 in 5180
9	GALT	Galactosemia	1 in 109	1 in 727
19	GAMT	Cerebral creatine deficiency syndrome, type 2	1 in 500	1 in 10000
1	GBA1	Gaucher Disease, type I-III; GD IIIC; GD, perinatal lethal	1 in 125	1 in 1563
3	GBE1	Glycogen storage disease, type 4	1 in 192	1 in 960
19	GCDH	Glutaricaciduria, type 1	1 in 200	1 in 4000
14	GCH1	Hyperphenylalaninemia, BH4-deficient, type B	1 in 250	1 in 1600
16	GCSH	Multiple mitochondrial dysfunctions syndrome 7	< 1 in 500	Reduced
8	GDAP1	Charcot-Marie-Tooth disease, recessive intermediate, type A	1 in 180	1 in 9000
20	GDF5	Chondrodysplasia, Grebe type	< 1 in 500	Reduced
3	GFM1	Combined oxidative phosphorylation deficiency, type 1	1 in 450	1 in 1500
7	GHRHR	Growth hormone deficiency, isolated, type 1B	1 in 692	1 in 34560
13	GJB2	Deafness, autosomal recessive, type 1A; Deafness, digenic, GJB2/GJB6	1 in 40	1 in 4000
13	GJB6	Deafness, autosomal recessive, type 1B; Deafness, digenic GJB2/GJB6	1 in 421	1 in 4210
3	GLB1	GM1-gangliosidosis, types 1-3; Mucopolysaccharidosis, type 4B (Morquio)	1 in 277	1 in 2770
9	GLDC	Glycine encephalopathy	1 in 180	1 in 720
9	GLE1	Lethal congenital contracture syndrome, type 1; Congenital arthrogryposis with anterior horn cell disease	1 in 350	1 in 3500
9	GNE	Inclusion body myopathy, type 2 (Nonaka myopathy)	1 in 203	1 in 4060
6	GNMT	Glycine N-methyltransferase deficiency	1 in 500	1 in 24951
12	GNPTAB	Mucopolipidosis 2 alpha/beta; Mucopolipidosis 3 alpha/beta	1 in 176	1 in 17522
16	GNPTG	Mucopolipidosis III gamma	< 1 in 500	Reduced
4	GNRHR	Hypogonadotropic hypogonadism, type 7, without anosmia	1 in 500	1 in 2097
12	GNS	Mucopolysaccharidosis, type 3D (Sanfilippo syndrome D)	< 1 in 500	Reduced
17	GP1BA	Bernard-Soulier syndrome, type A1	< 1 in 500	Reduced
22	GP1BB	Bernard-Soulier syndrome, type B	1 in 500	1 in 1666
3	GP9	Bernard-Soulier syndrome, type C	1 in 451	1 in 4510

9	GRHPR	Hyperoxaluria, primary, type 2	1 in 433	1 in 21650
12	GRIP1	Fraser syndrome 3	1 in 1218	1 in 12180
20	GSS	Glutathione synthetase deficiency	< 1 in 500	Reduced
17	GUCY2D	Leber congenital amaurosis, type 1	1 in 248	1 in 8260
7	GUSB	Mucopolysaccharidosis, type 7	1 in 552	1 in 16531
4	HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	1 in 415	1 in 622
2	HADHA	Long-chain 3-hydroxy-CoA dehydrogenase (LCHAD) deficiency; Mitochondrial trifunctional protein deficiency	1 in 250	1 in 5000
2	HADHB	Mitochondrial trifunctional protein deficiency	< 1 in 500	Reduced
1	HAX1	Neutropenia, severe congenital, type 3, autosomal recessive	1 in 219	1 in 2190
16	HBA1	Alpha thalassemia	1 in 30	1 in 200
16	HBA2	Alpha thalassemia	1 in 30	1 in 200
11	HBB	HBB-related hemoglobinopathies	1 in 67	1 in 6700
15	HEXA	Tay-Sachs disease	1 in 250	1 in 1250
5	HEXB	Sandhoff disease, infantile, juvenile, and adult forms	1 in 202	1 in 1347
3	HGD	Alkaptonuria	1 in 250	1 in 2500
8	HGSNAT	Mucopolysaccharidosis type 3C (Sanfilippo syndrome C)	1 in 345	1 in 4313
1	HJV	Hemochromatosis, type 2A	< 1 in 500	Reduced
21	HLCS	Holocarboxylase synthetase deficiency	1 in 300	1 in 3000
1	HMGCL	HMG-CoA lyase deficiency	< 1 in 500	Reduced
22	HMOX1	Heme oxygenase-1 deficiency	1 in 500	Reduced
10	HOGA1	Hyperoxaluria, primary, type 3	1 in 169	1 in 8400
12	HPD	Tyrosinemia, type 3	< 1 in 500	1 in 16666
10	HPS1	Hermansky-Pudlak syndrome, type 1	1 in 493	1 in 4930
3	HPS3	Hermansky-Pudlak syndrome, type 3	1 in 300	1 in 1500
9	HSD17B3	46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency	< 1 in 500	<1 in 2750
5	HSD17B4	D-bifunctional protein deficiency	1 in 534	1 in 13350
1	HSD3B2	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	1 in 862	1 in 8620
1	HSPG2	Schwartz-Jampel syndrome, type 1; Dyssegmental dysplasia, Silverman-Handmaker type	< 1 in 500	<1 in 1625
3	HYAL1	?Mucopolysaccharidosis, type 9	< 1 in 500	Reduced
11	HYLS1	Hydrolethalus syndrome	1 in 500	1 in 714
20	IDH3B	Retinitis pigmentosa, type 46	1 in 500	1 in 999
4	IDUA	Mucopolysaccharidosis type 1	1 in 153	1 in 3825
11	IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S	< 1 in 500	<1 in 4000
15	IVD	Isovaleric acidemia	1 in 115	1 in 1917
6	IYD	Thyroid dysmorphogenesis, type 4	1 in 159	1 in 633
19	JAK3	Severe Combined Immunodeficiency, autosomal recessive, T-negative/B-positive type	1 in 475	1 in 2375
11	KCNJ11	Hyperinsulinemic hypoglycemia, type 2 (congenital hyperinsulinism); Permanent neonatal diabetes mellitus (PNDM)	1 in 232	1 in 4640
6	LAMA2	LAMA2-related muscular dystrophy	1 in 125	1 in 625
18	LAMA3	Epidermolysis bullosa, junctional 2A, intermediate;Epidermolysis bullosa, junctional 2B, severe;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous	< 1 in 500	Reduced
1	LAMB3	Junctional epidermolysis bullosa (JEB) Herlitz type; JEB non-Herlitz type	1 in 222	1 in 11100
1	LAMC2	Epidermolysis bullosa, junctional 3A, intermediate;Epidermolysis bullosa, junctional 3B, severe	< 1 in 500	Reduced
22	LARGE1	Muscular dystrophy-dystroglycanopathy, type 6A and 6B	1 in 123	1 in 410
6	LCA5	Leber congenital amaurosis, type 5	< 1 in 500	Reduced
19	LDLR	Hypercholesterolemia, familial, type 1	1 in 118	1 in 280
1	LDLRAP1	Hypercholesterolemia, familial, autosomal recessive	< 1 in 500	Reduced
2	LHCGR	Leydig cell hypoplasia	< 1 in 500	<1 in 1060
9	LHX3	Pituitary hormone deficiency, combined, type 3	1 in 1398	1 in 13980
5	LIFR	Stuve-Wiedemann syndrome / Schwartz-Jampel type 2 syndrome	< 1 in 500	Reduced
10	LIPA	Lysosomal acid lipase deficiency	1 in 112	1 in 2240
3	LIPH	Hypotrichosis, type 7 or woolly hair, autosomal recessive, type 2, with or without hypotrichosis	1 in 500	1 in 2995
6	LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	1 in 414	1 in 13800
18	LOXHD1	Deafness, autosomal recessive, type 77	1 in 150	1 in 1500
8	LPL	Lipoprotein lipase deficiency	1 in 374	1 in 3740
2	LRP2	Donnai-Barrow syndrome	< 1 in 500	Reduced
2	LRPPRC	Leigh syndrome, French-Canadian type	< 1 in 500	Reduced
1	LYST	Chediak-Higashi syndrome	< 1 in 500	Reduced
19	MAN2B1	Alpha-mannosidosis	1 in 274	1 in 5480
4	MANBA	Mannosidosis, beta	< 1 in 500	Reduced
10	MAT1A	Methionine adenosyltransferase deficiency, autosomal recessive	< 1 in 500	Reduced
3	MCCC1	3-Methylcrotonyl-CoA carboxylase deficiency, type 1	1 in 353	1 in 7060
5	MCCC2	3-Methylcrotonyl-CoA carboxylase deficiency, type 2	1 in 204	1 in 4080
2	MCEE	Methylmalonyl-CoA epimerase deficiency	1 in 500	1 in 50000
19	MCOLN1	Mucopolidosis type 4	1 in 1166	1 in 4850
8	MCPH1	Microcephaly type 1, primary, autosomal recessive	1 in 500	1 in 8333
11	MED17	Microcephaly, postnatal progressive, with seizures and brain atrophy	< 1 in 500	Reduced
16	MEFV	Familial Mediterranean fever	1 in 40	1 in 160
15	MESP2	Spondylocostal dysostosis, type 2, autosomal recessive	1 in 500	1 in 50000
4	MFSD8	Ceroid lipofuscinosis, neuronal, type 7	1 in 300	1 in 3000
17	MKS1	Bardet-Biedl syndrome type 13; Meckel syndrome, type 1; Joubert syndrome, type 28	1 in 246	1 in 2460

22	MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	< 1 in 500	Reduced
16	MLYCD	Malonyl-CoA decarboxylase deficiency	1 in 500	1 in 1000
4	MMAA	Methylmalonic aciduria, vitamin B12-responsive	1 in 677	1 in 4513
12	MMAB	Methylmalonic aciduria, vitamin B12-responsive, type cblB	< 1 in 500	Reduced
1	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type	1 in 170	1 in 2429
2	MMADHC	Homocystinuria, cblD type, variant 1	< 1 in 500	Reduced
6	MMUT	Methylmalonic aciduria, mut(0) type	1 in 135	1 in 3375
2	MOGS	Congenital disorder of glycosylation, type 2B	< 1 in 500	Reduced
15	MPI	Congenital disorder of glycosylation, type 1B	1 in 473	1 in 11825
1	MPL	Thrombocytopenia, congenital amegakaryocytic	1 in 241	1 in 2410
2	MPV17	Mitochondrial DNA depletion syndrome type 6 (hepatocerebral); Charcot-Marie-Tooth disease, axonal, type 2EE	1 in 612	1 in 7650
1	MTHFR	Homocystinuria due to MTHFR deficiency	1 in 1119	1 in 11190
11	MTMR2	Charcot-Marie-Tooth disease, type 4B1	1 in 500	1 in 1000
1	MTR	Homocystinuria-megaloblastic anemia, cblG complementation type	< 1 in 500	1 in 5000
5	MTRR	Homocystinuria-megaloblastic anemia, cbl E type	< 1 in 500	Reduced
4	MTTP	Abetalipoproteinemia	< 1 in 500	Reduced
12	MVK	Mevalonic aciduria	1 in 286	1 in 2261
17	MYO15A	Deafness, autosomal recessive, type 3	1 in 1000	1 in 10000
11	MYO7A	Usher syndrome, type 1B; Deafness, autosomal recessive, type 2	1 in 129	1 in 2580
2	NADK2	2,4-dienoyl-CoA reductase deficiency	1 in 500	Reduced
22	NAGA	Schindler disease, type I; Schindler disease, type III; Kanzaki disease	1 in 500	1 in 5000
17	NAGLU	Mucopolysaccharidosis, type 3B (Sanfilippo B)	1 in 346	1 in 1384
17	NAGS	N-acetylglutamate synthase deficiency	< 1 in 500	Reduced
8	NBN	Nijmegen breakage syndrome	1 in 525	1 in 17500
7	NCF1	Chronic granulomatous disease, type 1	1 in 343	1 in 1027
1	NCF2	Chronic granulomatous disease, type 2	< 1 in 500	Reduced
8	NDRG1	Charcot-Marie-Tooth disease, type 4D	< 1 in 500	Reduced
5	NDUFAF2	Mitochondrial complex I deficiency, nuclear type 10	< 1 in 500	Reduced
20	NDUFAF5	Mitochondrial complex I deficiency, nuclear type 16	1 in 982	1 in 4350
5	NDUFS4	Mitochondrial complex I deficiency, nuclear type 1	< 1 in 500	Reduced
5	NDUFS6	Mitochondrial complex I deficiency, nuclear type 9	< 1 in 500	Reduced
19	NDUFS7	Mitochondrial complex I deficiency, nuclear type 3	< 1 in 500	Reduced
11	NDUFV1	Mitochondrial complex I deficiency, nuclear type 4	1 in 469	1 in 736
2	NEB	Nemaline myopathy type 2	1 in 175	1 in 2188
6	NEU1	Sialidosis, type 1 and type 2	< 1 in 500	Reduced
5	NHP2	Dyskeratosis congenita, autosomal recessive type 2	1 in 250	1 in 24964
19	NLRP7	Hydatidiform mole, recurrent, type 1	1 in 500	1 in 722
15	NOP10	Dyskeratosis congenita, autosomal recessive type 1	< 1 in 500	Reduced
18	NPC1	Niemann-Pick disease, type C1	1 in 163	1 in 652
14	NPC2	Niemann-pick disease, type C2	< 1 in 500	Reduced
2	NPHP1	Joubert syndrome type 4	1 in 418	1 in 1393
19	NPHS1	Nephrotic syndrome, type 1	1 in 112	1 in 1400
1	NPHS2	Nephrotic syndrome, type 2	1 in 226	1 in 11300
15	NR2E3	Enhanced S-cone syndrome (Goldmann-Favre); Retinitis pigmentosa, type 37	1 in 278	1 in 5560
1	NTRK1	Insensitivity to pain, congenital, with anhidrosis	1 in 1122	1 in 11220
10	OAT	Gyrate atrophy of choroid and retina	< 1 in 500	Reduced
15	OCA2	Oculocutaneous albinism type 2	1 in 101	1 in 1010
19	OPA3	3-methylglutaconic aciduria, type 3	< 1 in 500	Reduced
6	OSTM1	Osteopetrosis, autosomal recessive type 5	< 1 in 500	Reduced
2	OTOF	Deafness, autosomal recessive, type 9	1 in 228	1 in 2270
1	P3H1	Osteogenesis imperfecta, type 8	1 in 567	1 in 1447
12	PAH	Phenylketonuria	1 in 60	1 in 857
20	PANK2	Neurodegeneration with brain iron accumulation type 1	1 in 400	1 in 5000
11	PC	Pyruvate carboxylase deficiency	1 in 251	1 in 3150
10	PCBD1	Hyperphenylalaninemia, BH4-deficient, type D	< 1 in 500	Reduced
13	PCCA	Propionic acidemia	1 in 636	1 in 2544
3	PCCB	Propionic acidemia	1 in 635	1 in 7938
10	PCDH15	Deafness, autosomal recessive, type 23; Usher syndrome, type 1D/F digenic	1 in 497	1 in 1657
5	PDE6A	Retinitis pigmentosa type 43	1 in 500	1 in 863
3	PDHB	Pyruvate dehydrogenase E1-beta deficiency	< 1 in 500	Reduced
7	PEX1	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)	1 in 191	1 in 3820
1	PEX10	Peroxisome biogenesis disorder, type 6A (Zellweger syndrome); Peroxisome biogenesis disorder, type 6B	1 in 1287	1 in 16088
17	PEX12	Peroxisome biogenesis disorder type 3A (Zellweger)	< 1 in 500	Reduced
8	PEX2	Peroxisome biogenesis disorder type 5A (Zellweger)	< 1 in 500	Reduced
22	PEX26	Peroxisome biogenesis disorder type 7A (Zellweger)	< 1 in 500	Reduced
12	PEX5	Peroxisome biogenesis disorder type 2A (Zellweger)	< 1 in 500	Reduced
6	PEX6	Peroxisome biogenesis disorder, type 4A (Zellweger syndrome); Peroxisome biogenesis disorder, type 4B; Heimler syndrome 2	1 in 83	1 in 2766
6	PEX7	Rhizomelic chondrodysplasia punctata, type 1	1 in 371	1 in 7420
12	PFKM	Glycogen storage disease, type 7	< 1 in 500	Reduced

1	PHGDH	Neu-Laxova syndrome, type 1; Phosphoglycerate dehydrogenase deficiency	1 in 631	1 in 4207
6	PKHD1	Polycystic kidney disease type 4	1 in 66	1 in 1100
22	PLA2G6	Infantile neuroaxonal dystrophy type 1	1 in 343	1 in 856
1	PLOD1	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	1 in 159	1 in 450
16	PMM2	Congenital disorder of glycosylation, type 1A	1 in 71	1 in 3550
17	PNPO	Pyridoxamine 5'-phosphate oxidase deficiency	1 in 1107	1 in 3983
15	POLG	POLG-related disorders	1 in 194	1 in 3800
6	POLR1C	Leukodystrophy, hypomyelinating, type 11; Treacher Collins syndrome 3	1 in 1021	1 in 1659
1	POMGNT1	Muscular dystrophy-dystroglycanopathy, type 3A (Walker-Warburg syndrome); Type 3B; Type 3C (limb-girdle muscular dystrophy, type 15 [LGMDR15])	1 in 315	1 in 15750
9	POMT1	Muscular dystrophy-dystroglycanopathy, type 1A (Walker-Warburg syndrome); Type 1B; Type 1C (limb-girdle muscular dystrophy, type 11 [LGMD R11])	1 in 372	1 in 3720
14	POMT2	Muscular dystrophy-dystroglycanopathy, type 2A (Walker-Warburg syndrome); Type 2B; Type 2C (limb-girdle muscular dystrophy, type 14 [LGMD R14])	< 1 in 500	Reduced
7	POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	1 in 268	1 in 1069
3	POU1F1	Pituitary hormone deficiency, combined, type 1	1 in 1230	Reduced
4	PPM1K	?Maple syrup urine disease, mild variant	1 in 200	1 in 4000
1	PPT1	Ceroid lipofuscinosis, neuronal, type 1	1 in 488	1 in 16200
1	PRDX1	Methylmalonic aciduria and homocystinuria, cblC type, digenic	< 1 in 500	Reduced
10	PRF1	Hemophagocytic lymphohistiocytosis, familial, type 2	1 in 308	1 in 538
22	PRODH	Hyperprolinemia, type 1	1 in 500	1 in 3333
5	PROP1	Pituitary hormone deficiency, combined, type 2	1 in 591	1 in 29495
10	PSAP	Combined SAP deficiency	< 1 in 500	Reduced
11	PTS	Hyperphenylalaninemia, BH4-deficient, type A	1 in 478	1 in 1593
12	PUG1	Myopathy, lactic acidosis, and sideroblastic anemia, type 1	< 1 in 500	Reduced
11	PYGM	McArdle disease	1 in 206	1 in 2060
4	QDPR	Hyperphenylalaninemia, BH4-deficient, type C	< 1 in 500	Reduced
6	RAB23	Carpenter syndrome	< 1 in 500	Reduced
11	RAG1	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 344	1 in 614
11	RAG2	Omenn syndrome; Severe combined immunodeficiency, B cell-negative	1 in 1925	1 in 19250
11	RAPSN	Fetal akinesia deformation sequence, type 2; Myasthenic syndrome, congenital, type 11, associated with AChR deficiency	1 in 165	1 in 1650
6	RARS2	Pontocerebellar hypoplasia, type 6	1 in 269	1 in 3363
18	RAX	Isolated microphthalmia, type 3	< 1 in 500	Reduced
14	RDH12	Leber congenital amaurosis, type 13	1 in 456	1 in 4560
9	RMRP	Anauxetic dysplasia, type 1	1 in 500	1 in 6250
13	RNASEH2B	Aicardi-Goutieres syndrome, type 2	1 in 440	1 in 7333
11	RNASEH2C	Aicardi-Goutieres syndrome, type 3	< 1 in 500	Reduced
1	RPE65	RPE65-related Leber congenital amaurosis/early-onset severe retinal dystrophy	1 in 366	1 in 18300
16	RPGRIP1L	Joubert syndrome, type 7; Meckel syndrome, type 5; COACH syndrome	1 in 319	1 in 860
20	RTEL1	Dyskeratosis congenita, autosomal recessive type 5	< 1 in 500	Reduced
13	SACS	Spastic ataxia, Charlevoix-Saguenay, type	1 in 100	1 in 1000
2	SAG	Oguchi disease, type 1	< 1 in 500	Reduced
20	SAMHD1	Aicardi-Goutieres syndrome, type 5	1 in 610	1 in 2033
7	SBD5	Shwachman-Diamond syndrome	1 in 224	1 in 804
22	SCO2	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, type 1	1 in 500	1 in 8333
4	SEPSECS	Pontocerebellar hypoplasia, type 2D	< 1 in 500	Reduced
14	SERPINA1	Alpha-1 antitrypsin deficiency	1 in 24	1 in 2400
17	SGCA	Limb-girdle muscular dystrophy, type 3 (LGMD R3)	1 in 288	1 in 1920
4	SGCB	Limb-girdle muscular dystrophy, type 4 (LGMD R4)	1 in 628	1 in 2093
5	SGCD	Limb-girdle muscular dystrophy, type 6 (LGMD R6)	< 1 in 500	Reduced
13	SGCG	Limb-girdle muscular dystrophy, type 5 (LGMD R5)	1 in 1132	1 in 5468
17	SGSH	Mucopolysaccharidosis, type 3A (Sanfilippo A)	1 in 253	1 in 5060
5	SH3TC2	Charcot-Marie-Tooth disease, type 4C	1 in 130	1 in 1300
5	SKIC3	Trichohepatoenteric syndrome, type 1 (diarrhea, syndromic)	1 in 640	1 in 64000
16	SLC12A3	Gitelman syndrome	1 in 73	1 in 2400
15	SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	< 1 in 500	Reduced
6	SLC17A5	Salla disease	1 in 328	1 in 2187
1	SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	1 in 500	1 in 888
2	SLC19A3	Thiamine metabolism dysfunction syndrome, type 2 (biotin- or thiamine-responsive encephalopathy type)	1 in 232	1 in 1785
5	SLC22A5	Carnitine deficiency, systemic primary	1 in 251	1 in 717
7	SLC25A13	Citrullinemia, type 2, neonatal-onset; Citrullinemia, type 2, adult-onset	1 in 619	1 in 2063
13	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	< 1 in 500	Reduced
3	SLC25A20	Carnitine-acylcarnitine translocase deficiency	< 1 in 500	Reduced
5	SLC26A2	Achondrogenesis Ib; Atelosteogenesis, type II; De la Chapelle dysplasia; Diastrophic dysplasia; Diastrophic dysplasia, broad bone-platyspondylic variant; Epiphyseal dysplasia, multiple, 4	1 in 129	1 in 4300
7	SLC26A3	Diarrhea 1, secretory chloride, congenital	< 1 in 500	Reduced
7	SLC26A4	Deafness, autosomal recessive, type 4; Pendred syndrome	1 in 88	1 in 587
6	SLC35A1	Congenital disorder of glycosylation, type 2F	< 1 in 500	<1 in 1500
1	SLC35A3	Arthrogryposis, impaired intellectual development, and seizures	< 1 in 500	Reduced
11	SLC35C1	Congenital disorder of glycosylation, type 2C	< 1 in 500	Reduced
1	SLC35D1	Schneckenbecken dysplasia	< 1 in 500	Reduced

11	SLC37A4	Glycogen storage disease, type 1B	1 in 500	1 in 7143
8	SLC39A4	Acrodermatitis enteropathica	1 in 316	1 in 1580
2	SLC3A1	Cystinuria	1 in 42	1 in 1050
5	SLC45A2	Albinism, oculocutaneous, type 4	< 1 in 500	<1 in 1600
17	SLC46A1	Folate malabsorption, hereditary	< 1 in 500	Reduced
20	SLC4A11	Corneal endothelial dystrophy, autosomal recessive	1 in 806	1 in 26800
19	SLC5A5	Thyroid dysmorphogenesis, type 1	< 1 in 500	Reduced
5	SLC6A19	Hartnup disorder	1 in 87	1 in 2900
14	SLC7A7	Lysinuric protein intolerance	< 1 in 500	Reduced
19	SLC7A9	Cystinuria	1 in 42	1 in 2100
2	SMARCAL1	Schimke immunoosseous dysplasia	1 in 451	1 in 3007
5	SMN1	Spinal muscular atrophy	1 in 50	1 in 588
11	SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B	1 in 350	1 in 3500
15	SPG11	Amyotrophic lateral sclerosis 5, juvenile; Charcot-Marie-Tooth disease, axonal, type 2X; Spastic paraplegia 11	1 in 192	1 in 900
16	SPG7	Spastic paraplegia, type 7, autosomal recessive	1 in 80	1 in 1600
2	SRD5A2	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency (pseudovaginal perineoscrotal hypospadias)	1 in 400	1 in 4000
2	ST3GAL5	Salt and pepper developmental regression syndrome	< 1 in 500	Reduced
8	STAR	Lipoid adrenal hyperplasia	1 in 1147	1 in 14338
3	SUMF1	Multiple sulfatase deficiency	< 1 in 500	Reduced
9	SURF1	Mitochondrial complex IV deficiency, nuclear type 1; Charcot-Marie-Tooth disease, type 4K	1 in 191	1 in 9550
16	TAT	Tyrosinemia, type 2	< 1 in 500	Reduced
11	TCIRG1	Osteopetrosis, autosomal recessive, type 1	1 in 399	1 in 7980
14	TECPR2	Spastic paraplegia, type 49, autosomal recessive	1 in 1946	1 in 12973
3	TF	Atransferrinemia	1 in 500	1 in 7143
7	TFR2	Hemochromatosis, type 3	< 1 in 500	Reduced
8	TG	Thyroid dysmorphogenesis, type 3	1 in 159	1 in 5300
14	TGM1	Ichthyosis, congenital, autosomal recessive, type 1	1 in 186	1 in 1860
11	TH	Segawa syndrome, recessive	< 1 in 500	Reduced
11	TMEM216	Joubert syndrome, type 2; Meckel syndrome, type 2	< 1 in 500	Reduced
8	TMEM67	Meckel syndrome 3; COACH syndrome 1; Joubert syndrome 6; Nephronophthisis 11	1 in 147	1 in 2940
21	TMPRSS3	Deafness, autosomal recessive, type 8/10	1 in 135	1 in 2700
6	TNXB	Ehlers-Danlos syndrome, classic-like	1 in 335	1 in 1675
2	TPO	Thyroid dysmorphogenesis, type 2A	1 in 72	1 in 2400
11	TPP1	Ceroid lipofuscinosis, neuronal, type 2; Spinocerebellar ataxia, autosomal recessive, type 7	1 in 266	1 in 1773
6	TRDN	Ventricular tachycardia, catecholaminergic polymorphic, type 5, with or without muscle weakness	1 in 287	Reduced
3	TREX1	Aicardi-Goutieres syndrome, type 1	1 in 500	1 in 10000
9	TRIM32	Limb-girdle muscular dystrophy, type 8 (LGMD R8)	1 in 1273	1 in 12730
17	TRIM37	Mulibrey nanism	< 1 in 500	Reduced
22	TRMU	Liver failure, transient infantile	< 1 in 500	Reduced
17	TSEN54	Pontocerebellar hypoplasia, type 2A; Pontocerebellar hypoplasia, type 4	1 in 223	1 in 3997
12	TSMF	Combined oxidative phosphorylation deficiency, type 3	< 1 in 500	Reduced
1	TSHB	Hypothyroidism, congenital, nongoitrous, type 4	< 1 in 500	Reduced
14	TSHR	Hypothyroidism, congenital, nongoitrous, type 1	1 in 325	1 in 3250
8	TTPA	Ataxia with isolated vitamin E deficiency	< 1 in 500	Reduced
22	TYMP	Mitochondrial DNA depletion syndrome, type 1 (MNGIE type)	1 in 425	1 in 10625
11	TYR	Oculocutaneous albinism (OCA) type 1A; OCA type 1B	1 in 92	1 in 1840
9	TYRP1	Albinism, oculocutaneous, type 3	< 1 in 500	<1 in 1400
15	UBE3A	Angelman syndrome	N/A due to imprinting mechanism	Reduced
2	UGT1A1	Crigler-Najjar syndrome, type 1; Crigler-Najjar syndrome, type 2	1 in 500	1 in 5496
17	UNC13D	Hemophagocytic lymphohistiocytosis, familial, type 3	1 in 108	1 in 1080
11	USH1C	Usher syndrome, type 1C; Deafness, autosomal recessive, type 18A	1 in 257	1 in 3671
17	USH1G	Usher syndrome, type 1G	< 1 in 500	Reduced
1	USH2A	Usher syndrome, type 2A; Retinitis pigmentosa 39	1 in 60	1 in 600
9	VPS13A	Choreoacanthocytosis	1 in 341	1 in 974
8	VPS13B	Cohen syndrome	1 in 224	1 in 747
1	VPS45	Neutropenia, severe congenital, type 5	< 1 in 500	Reduced
17	VPS53	Pontocerebellar hypoplasia, type 2E	1 in 1506	Reduced
14	VRK1	Pontocerebellar hypoplasia, type 1A	< 1 in 500	1 in 10000
14	VXK2	Microphthalmia with coloboma 3; Isolated microphthalmia 2	1 in 1337	1 in 8913
9	WHRN	Usher syndrome, type 2D; Deafness, autosomal recessive, type 31	1 in 1000	Reduced
2	WNT10A	WNT10A-related conditions	1 in 238	1 in 2975
8	WRN	Werner syndrome	1 in 474	1 in 681
9	XPA	Xeroderma pigmentosum, group A	< 1 in 500	Reduced
3	XPC	Xeroderma pigmentosum, group C	< 1 in 500	Reduced
14	ZFYVE26	Spastic paraplegia, type 15, autosomal recessive	< 1 in 500	Reduced

N/A: no data prevalence unknown



Patient Information:

14644, DONOR

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8759403

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: SSB Genetics, Dept

San Diego Sperm Bank

4915 25th Avenue NE, Ste 204W

Seattle, WA 98105

Phone: (206) 588-1484

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: **Oct 10, 2025**

Accession:

FT-7032182

Test#: FT-TS15363289

Specimen Type: Blood (EDTA)

Collected: Jun 11, 2024

Accession:

N/A

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Custom Beacon Preconception Carrier Screening Panel

(2 Gene Panel: *BTBD* and *MCCC2*; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)



GENES TESTED:

Custom Beacon Preconception Carrier Screening Panel - 2 Genes

This analysis was run using the Custom Beacon Preconception Carrier Screening Panel gene list. 2 genes were tested with 100.0% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

BTD, MCCC2

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions ($\pm$ 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.



Gene Specific Notes and Limitations

No gene specific limitations apply to the genes on the tested panel.

SIGNATURE:

A handwritten signature in black ink, appearing to read "Yan Meng".

Yan Meng, Ph.D., CGMB, FACMG on 10/10/2025
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed, performed, and its performance characteristics determined by **Fulgent Therapeutics LLC** (CAP# 8042697, CLIA# 05D2043189), 4399 Santa Anita Ave., El Monte, CA 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **(626) 350-0537** or info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

4399 Santa Anita Ave.
El Monte, CA, 91731
(p) 626-350-0537 (f) 626-454-1667
info@fulgentgenetics.com
www.fulgentgenetics.com



To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes tested on any Beacon panel, please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)



Patient Information

Patient Name: Donor 14644

Date Of Birth: [REDACTED]

Gender: Male

Patient ID: N/A

Medical Record #: N/A

Collection Kit: 48298984-2-C

Accession ID: N/A

Case File ID: 18136052

Ethnicity: Northern European
Caucasian**Test Information**

Ordering Physician: Dr. James Kuan

Clinic Information: Seattle Sperm Bank

Phone: (206) 588-1484

Report Date: 09/16/2025

Sample Collected: 09/05/2025

Sample Received: 09/08/2025

Sample Type: Blood

**CARRIER SCREENING REPORT**

ABOUT THIS SCREEN: Horizon™ is a carrier screen for specific autosomal recessive and X-linked diseases. This information can help patients learn their risk of having a child with specific genetic conditions.

ORDER SELECTED: The Horizon Custom panel was ordered for this patient. Males are not screened for X-linked diseases

FINAL RESULTS SUMMARY:**Negative for 1 out of 1 diseases**

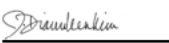
No Pathogenic variants were detected in the genes that were screened. The patient's remaining carrier risk after the negative screening results is listed for each disease/gene on the Horizon website at <https://www.natera.com/panel-option/h-all/>. Please see the following pages of this report for a comprehensive list of all conditions included on this individual's screen.

Carrier screening is not diagnostic and may not detect all possible pathogenic variants in a given gene.

RECOMMENDATIONS

Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting naterasession.com. Clinicians with questions may contact Natera at 650-249-9090 or email support@natera.com.


Christine M. Eng, M.D.
Medical Director, Baylor Genetics


J. Dianne Koen-Kim, Ph.D., FACMG
Senior Laboratory Director, Natera


Linyan Meng, Ph.D.
Laboratory Director, Baylor Genetics


Yang Wang, Ph.D., FACMG
Laboratory Director, Natera

Patient Information

Patient Name: Donor 14644

Date Of Birth: [REDACTED]

Case File ID: 18136052

Test Information

Ordering Physician: Dr. James Kuan

Clinic Information: Seattle Sperm Bank

Report Date: 09/16/2025

**DISEASES SCREENED**

Below is a list of all diseases screened and the result. Certain conditions have unique patient-specific numerical values, therefore, results for those conditions are formatted differently.

Autosomal Recessive**P**PSEUDOXANTHOMA ELASTICUM (ABCC6) **negative**

Patient Information

Patient Name: Donor 14644

Date Of Birth:

Case File ID: 18136052

Test Information

Ordering Physician: Dr. James Kuan

Clinic Information: Seattle Sperm Bank

Report Date: 09/16/2025

**Testing Methodology, Limitations, and Comments:****Next-generation sequencing (NGS)**

Sequencing library prepared from genomic DNA isolated from a patient sample is enriched for targets of interest using standard hybridization capture protocols and PCR amplification (for targets specified below). NGS is then performed to achieve the standards of quality control metrics, including a minimum coverage of 99% of targeted regions at 20X sequencing depth. Sequencing data is aligned to human reference sequence, followed by deduplication, metric collection and variant calling (coding region +/- 20bp). Variants are then classified according to ACMGG/AMP standards of interpretation using publicly available databases including but not limited to ENSEMBL, HGMD Pro, ClinGen, ClinVar, 1000G, ESP and gnomAD. Variants predicted to be pathogenic or likely pathogenic for the specified diseases are reported. It should be noted that the data interpretation is based on our current understanding of the genes and variants at the time of reporting. Putative positive sequencing variants that do not meet internal quality standards or are within highly homologous regions are confirmed by Sanger sequencing or gene-specific long-range PCR as needed prior to reporting.

Copy Number Variant (CNV) analysis is limited to deletions involving two or more exons for all genes on the panel, in addition to specific known recurrent single-exon deletions. CNVs of small size may have reduced detection rate. This method does not detect gene inversions, single-exonic and sub-exonic deletions (unless otherwise specified), and duplications of all sizes (unless otherwise specified). Additionally, this method does not define the exact breakpoints of detected CNV events. Confirmation testing for copy number variation is performed by specific PCR, Multiplex Ligation-dependent Probe Amplification (MLPA), next generation sequencing, or other methodology.

This test may not detect certain variants due to local sequence characteristics, high/low genomic complexity, homologous sequence, or allele dropout (PCR-based assays). Variants within noncoding regions (promoter, 5'UTR, 3'UTR, deep intronic regions, unless otherwise specified), small deletions or insertions larger than 25bp, low-level mosaic variants, structural variants such as inversions, and/or balanced translocations may not be detected with this technology.

SPECIAL NOTES

For ABCC6, sequencing variants in exons 1-7 are not detected due to the presence of regions of high homology.

Variant Classification

Only pathogenic or likely pathogenic variants are reported. Other variants including benign variants, likely benign variants, variants of uncertain significance, or inconclusive variants identified during this analysis may be reported in certain circumstances. Our laboratory's variant classification criteria are based on the ACMG and internal guidelines and our current understanding of the specific genes. This interpretation may change over time as more information about a gene and/or variant becomes available. Natera and its lab partner(s) may reclassify variants at certain intervals but may not release updated reports without a specific request made to Natera by the ordering provider. Natera may disclose incidental findings if deemed clinically pertinent to the test performed.

Negative Results

A negative carrier screening result reduces the risk for a patient to be a carrier of a specific disease but does not completely rule out carrier status. Please visit <https://www.natera.com/panel-option/h-all/> for a table of carrier rates, detection rates, residual risks and promised variants/exons per gene. Carrier rates before and after testing vary by ethnicity and assume a negative family history for each disease screened and the absence of clinical symptoms in the patient. Any patient with a family history for a specific genetic disease will have a higher carrier risk prior to testing and, if the disease-causing mutation in their family is not included on the test, their carrier risk would remain unchanged. Genetic counseling is recommended for patients with a family history of genetic disease so that risk figures based on actual family history can be determined and discussed along with potential implications for reproduction. Horizon carrier screening has been developed to identify the reproductive risks for monogenic inherited conditions. Even when one or both members of a couple screen negative for pathogenic variants in a specific gene, the disease risk for their offspring is not zero. There is still a low risk for the condition in their offspring due to a number of different mechanisms that are not detected by Horizon including, but not limited to, pathogenic variant(s) in the tested gene or in a different gene not included on Horizon, pathogenic variant(s) in an upstream regulator, uniparental disomy, de novo mutation(s), or digenic or polygenic inheritance.

Additional Comments

These analyses generally provide highly accurate information regarding the patient's carrier status. Despite this high level of accuracy, it should be kept in mind that there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

15 Crawford St., STE 100
Needham, MA 02494
(p) 626-350-0537
(f) 626-454-1667
Lab Director: Arash Radfar M.D.
CLIA: 22D0957540



Patient Information:

14644, DONOR

DOB: [REDACTED]

Sex: M

MR#:

Patient#: FT-PT8759403

Partner Information:

Not Tested

Physician:

Kuan, James

ATTN: SSB Genetics, Dept

San Diego Sperm Bank

4915 25th Avenue NE, Ste 204W

Seattle, WA 98105

Phone: (206) 588-1484

Laboratory:

Fulgent Therapeutics LLC

CAP#: 8042697

CLIA#: 05D2043189

Laboratory Director:

Dr. Amar Jariwala

Report Date: **Apr 11, 2025**

Accession:

FT-7032182

Test#: FT-TS15150531

Specimen Type: Blood (EDTA)

Collected: Jun 11, 2024

Accession:

N/A

FINAL RESULTS



No carrier mutations identified

TEST PERFORMED

Single Gene Carrier Screening: FTCD

(1 Gene Panel: *FTCD*; gene sequencing with deletion and duplication analysis)

INTERPRETATION:

Notes and Recommendations:

- No carrier mutations were identified in the submitted specimen. A negative result does not rule out the possibility of a genetic predisposition nor does it rule out any pathogenic mutations in areas not assessed by this test or in regions that were covered at a level too low to reliably assess. Also, it does not rule out mutations that are of the sort not queried by this test; see Methods and Limitations for more information. A negative result reduces, but does not eliminate, the chance to be a carrier for any condition included in this screen. Please see the supplemental table for details.
- This carrier screening test does not screen for all possible genetic conditions, nor for all possible mutations in every gene tested. This report does not include variants of uncertain significance; only variants classified as pathogenic or likely pathogenic at the time of testing, and considered relevant for reproductive carrier screening, are reported. Please see the gene specific notes for details. Please note that the classification of variants can change over time.
- Patients may wish to discuss any carrier results with blood relatives, as there is an increased chance that they are also carriers. These results should be interpreted in the context of this individual's clinical findings, biochemical profile, and family history.
- Gene specific notes and limitations may be present. See below.
- Genetic counseling is recommended. Available genetic counselors and additional resources can be found at the National Society of Genetic Counselors (NSGC; <https://www.nsgc.org>)



GENES TESTED:

Custom Beacon Preconception Carrier Screening Panel - Gene

This analysis was run using the Custom Beacon Preconception Carrier Screening Panel gene list. 1 genes were tested with 100.0% of targets sequenced at >20x coverage. For more gene-specific information and assistance with residual risk calculation, see the SUPPLEMENTAL TABLE.

FTCD

METHODS:

Genomic DNA was isolated from the submitted specimen indicated above (if cellular material was submitted). DNA was barcoded, and enriched for the coding exons of targeted genes using hybrid capture technology. Prepared DNA libraries were then sequenced using a Next Generation Sequencing technology. Following alignment to the human genome reference sequence (assembly GRCh37), variants were detected in regions of at least 10x coverage. For this specimen, 100.00% and 100.00% of coding regions and splicing junctions of genes listed had been sequenced with coverage of at least 10x and 20x, respectively, by NGS or by Sanger sequencing. The remaining regions did not have 10x coverage, and were not evaluated. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. To minimize false positive results, any variants that do not meet internal quality standards are confirmed by Sanger sequencing. Variants classified as pathogenic, likely pathogenic, or risk allele which are located in the coding regions and nearby intronic regions (+/- 20bp) of the genes listed above are reported. Variants outside these intervals may be reported but are typically not guaranteed. When a single pathogenic or likely pathogenic variant is identified in a clinically relevant gene with autosomal recessive inheritance, the laboratory will attempt to ensure 100% coverage of coding sequences either through NGS or Sanger sequencing technologies ("fill-in"). All genes listed were evaluated for large deletions and/or duplications. However, single exon deletions or duplications will not be detected in this assay, nor will copy number alterations in regions of genes with significant pseudogenes. Putative deletions or duplications are analyzed using Fulgent Germline proprietary pipeline for this specimen. Bioinformatics: The FPLMv2.0 pipeline was used to analyze this specimen.

LIMITATIONS:

General Limitations

These test results and variant interpretation are based on the proper identification of the submitted specimen, accuracy of any stated familial relationships, and use of the correct human reference sequences at the queried loci. In very rare instances, errors may result due to mix-up or co-mingling of specimens. Positive results do not imply that there are no other contributors, genetic or otherwise, to future pregnancies, and negative results do not rule out the genetic risk to a pregnancy. Official gene names change over time. Fulgent uses the most up to date gene names based on HUGO Gene Nomenclature Committee (<https://www.genenames.org>) recommendations. If the gene name on report does not match that of ordered gene, please contact the laboratory and details can be provided. Result interpretation is based on the available clinical and family history information for this individual, collected published information, and Alamut annotation available at the time of reporting. This assay is not designed or validated for the detection of low-level mosaicism or somatic mutations. This assay will not detect certain types of genomic aberrations such as translocations, inversions, or repeat expansions other than specified genes. DNA alterations in regulatory regions or deep intronic regions (greater than 20bp from an exon) may not be detected by this test. Unless otherwise indicated, no additional assays have been performed to evaluate genetic changes in this specimen. There are technical limitations on the ability of DNA sequencing to detect small insertions and deletions. Our laboratory uses a sensitive detection algorithm, however these types of alterations are not detected as reliably as single nucleotide variants. Rarely, due to systematic chemical, computational, or human error, DNA variants may be missed. Although next generation sequencing technologies and our bioinformatics analysis significantly reduce the confounding contribution of pseudogene sequences or other highly-homologous sequences, sometimes these may still interfere with the technical ability of the assay to identify pathogenic alterations in both sequencing and deletion/duplication analyses. Deletion/duplication analysis can identify alterations of genomic regions which include one whole gene (buccal swab specimens and whole blood specimens) and are two or more contiguous exons in size (whole blood specimens only); single exon deletions or duplications may occasionally be identified, but are not routinely detected by this test. When novel DNA duplications are identified, it is not possible to discern the genomic location or orientation of the duplicated segment, hence the effect of the duplication cannot be predicted. Where deletions are detected, it is not always possible to determine whether the predicted product will remain in-frame or not. Unless otherwise indicated, deletion/duplication analysis has not been performed in regions that have been sequenced by Sanger.



Gene Specific Notes and Limitations

FTCD: The current testing method may not reliably detect copy number variants in exons 8-10 (NM_006657.2) of this gene due to the high GC content.

SIGNATURE:



Dr. Harry Gao, DABMG, FACMG on 4/11/2025
Laboratory Director, Fulgent

DISCLAIMER:

This test was developed and its performance characteristics determined by Fulgent Therapeutics LLC CAP #8042697 CLIA #05D2043189; 4399 Santa Anita Ave., El Monte, CA, 91731. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research. Since genetic variation, as well as systematic and technical factors, can affect the accuracy of testing, the results of testing should always be interpreted in the context of clinical and familial data. For assistance with interpretation of these results, healthcare professionals may contact us directly at **626-350-0537** or by email at info@fulgentgenetics.com. It is recommended that patients receive appropriate genetic counseling to explain the implications of the test result, including its residual risks, uncertainties and reproductive or medical options.

To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes tested on any Beacon panel, please visit the following link:

[Beacon Expanded Carrier Screening Supplemental Table](#)





Patient Information	Specimen Information	Client Information
14644, DONOR DOB: [REDACTED] AGE: [REDACTED] Gender: M Fasting: U Phone: 206.588.1484 Patient ID: 14644 Health ID: 8573034765676522	Specimen: OW647098A Requisition: 0000678 Collected: 06/11/2024 / 12:00 PDT Received: 06/14/2024 / 01:07 PDT Reported: 06/26/2024 / 15:51 PDT	Client #: 98105026 VNLZR00 KUAN, JAMES K SEATTLE SPERM BANK 4915 25TH AVE NE STE 204W SEATTLE, WA 98105-5668

COMMENTS: FASTING:UNKNOWN

Cytogenetic Report

CHROMOSOME ANALYSIS, BLOOD - 14596

Lab:EZ

CHROMOSOME ANALYSIS, BLOOD

Order ID: 24-283254
Specimen Type: Blood
Clinical Indication: GAMETE DONOR

RESULT:
NORMAL MALE KARYOTYPE

INTERPRETATION:
Chromosome analysis revealed normal G-band patterns within the limits of standard cytogenetic analysis.

Please expect the results of any other concurrent study in a separate report.

NOMENCLATURE:
46,XY

ASSAY INFORMATION:

Method: G-Band (Digital Analysis: MetaSyst)
Cells Counted: 20
Band Level: 450
Cells Analyzed: 5
Cells Karyotyped: 5

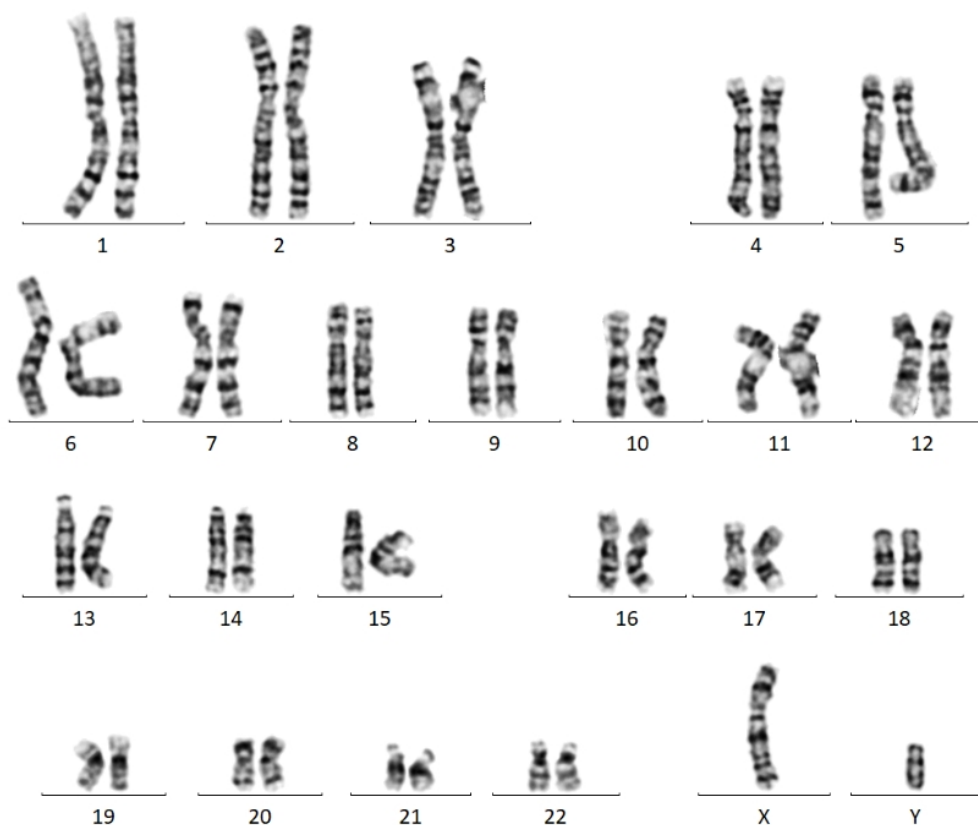
This test does not address genetic disorders that cannot be detected by standard cytogenetic methods or rare events such as low level mosaicism or subtle rearrangements.

Reha M. Toydemir, MD, PhD, FACMG, [site SJC5]

Electronic Signature: 6/26/2024 6:06 PM



Patient Information	Specimen Information	Client Information
14644, DONOR DOB: [REDACTED] AGE: [REDACTED] Gender: M Fasting: U Patient ID: 14644 Health ID: 8573034765676522	Specimen: OW647098A Collected: 06/11/2024 / 12:00 PDT Received: 06/14/2024 / 01:07 PDT Reported: 06/26/2024 / 15:51 PDT	Client #: 98105026 KUAN, JAMES K



PERFORMING SITE:

EZ QUEST DIAGNOSTICS/NICHOLS SJC, 33608 ORTEGA HWY, SAN JUAN CAPISTRANO, CA 92675-2042 Laboratory Director: IRINA MARAMICA,MD,PHD,MBA, CLIA: 05D0643352

Date Collected: 06/11/2024

Date Received: 06/12/2024

Date Reported: 06/14/2024

Fasting: Not Given

Ordered Items: LP+12AC+CBC/D/Plt+UA+Rh+ABO...

General Comments & Additional Information

A courtesy copy of this report has been sent to 2064664696.

Date Collected: 06/11/2024

LP+12AC+CBC/D/Plt+UA+Rh+ABO...

Test	Current Result and Flag	Previous Result and Date	Units	Reference Interval
Glucose ⁰¹	95		mg/dL	70-99
Uric Acid ⁰¹	5.1		mg/dL	3.8-8.4
Therapeutic target for gout patients: <6.0				
BUN ⁰¹	18		mg/dL	6-20
Creatinine ⁰¹	0.90		mg/dL	0.76-1.27
eGFR	119		mL/min/1.73	>59
Calcium ⁰¹	9.5		mg/dL	8.7-10.2
Protein, Total ⁰¹	7.4		g/dL	6.0-8.5
Albumin ⁰¹	5.0		g/dL	4.3-5.2
Bilirubin, Total ⁰¹	0.5		mg/dL	0.0-1.2
Alkaline Phosphatase ⁰¹	79		IU/L	44-121
LDH ⁰¹	148		IU/L	121-224
AST (SGOT) ⁰¹	17		IU/L	0-40
ALT (SGPT) ⁰¹	18		IU/L	0-44
Cholesterol, Total ⁰¹	139		mg/dL	100-199
Triglycerides ⁰¹	70		mg/dL	0-149
HDL Cholesterol ⁰¹	54		mg/dL	>39
LDL Chol Calc (NIH)	71		mg/dL	0-99
LDL/HDL Ratio	1.3		ratio	0.0-3.6
Please Note: ⁰¹				

LDL/HDL Ratio			
		Men	Women
1/2 Avg.Risk		1.0	1.5
Avg.Risk		3.6	3.2
2X Avg.Risk		6.2	5.0
3X Avg.Risk		8.0	6.1

Hgb Fractionation by CE: ⁰²			
Hgb F ⁰²	0.0	%	0.0-2.0
Hgb A ⁰²	97.4	%	96.4-98.8
Hgb A2 ⁰²	2.6	%	1.8-3.2
Hgb S ⁰²	0.0	%	0.0

Interpretation:⁰²

Normal hemoglobin present; no hemoglobin variant or beta thalassemia identified.
Note: Alpha thalassemia may not be detected by the Hgb Fractionation Cascade panel. If alpha thalassemia is suspected, Labcorp offers

Date Collected: 06/11/2024

LP+12AC+CBC/D/Plt+UA+Rh+ABO... (Cont.)

Alpha-Thalassemia DNA Analysis (#511172).

ABO Grouping ⁰¹	O		
Rh Factor ⁰¹	Positive		
	Please note: Prior records for this patient's ABO / Rh type are not available for additional verification.		
. ⁰¹			
CBC, Platelet Ct, and Diff ⁰¹			
WBC ⁰¹	5.3	x10E3/uL	3.4-10.8
▲ RBC ⁰¹	5.98	High	x10E6/uL 4.14-5.80
Hemoglobin ⁰¹	17.2	g/dL	13.0-17.7
▲ Hematocrit ⁰¹	51.2	High	% 37.5-51.0
MCV ⁰¹	86	fL	79-97
MCH ⁰¹	28.8	pg	26.6-33.0
MCHC ⁰¹	33.6	g/dL	31.5-35.7
RDW ⁰¹	11.8	%	11.6-15.4
Platelets ⁰¹	282	x10E3/uL	150-450
Neutrophils ⁰¹	62	%	Not Estab.
Lymphs ⁰¹	27	%	Not Estab.
Monocytes ⁰¹	9	%	Not Estab.
Eos ⁰¹	1	%	Not Estab.
Basos ⁰¹	1	%	Not Estab.
Neutrophils (Absolute) ⁰¹	3.3	x10E3/uL	1.4-7.0
Lymphs (Absolute) ⁰¹	1.4	x10E3/uL	0.7-3.1
Monocytes(Absolute) ⁰¹	0.5	x10E3/uL	0.1-0.9
Eos (Absolute) ⁰¹	0.1	x10E3/uL	0.0-0.4
Baso (Absolute) ⁰¹	0.1	x10E3/uL	0.0-0.2
Immature Granulocytes ⁰¹	0	%	Not Estab.
Immature Grans (Abs) ⁰¹	0.0	x10E3/uL	0.0-0.1
. ⁰¹			
Urinalysis Gross Exam ⁰¹			
Specific Gravity ⁰¹	1.007		1.005-1.030
pH ⁰¹	7.0		5.0-7.5
Urine-Color ⁰¹	Yellow		Yellow
Appearance ⁰¹	Clear		Clear
WBC Esterase ⁰¹	Negative		Negative
Protein ⁰¹	Negative		Negative/Trace
Glucose ⁰¹	Negative		Negative
Ketones ⁰¹	Negative		Negative
Occult Blood ⁰¹	Negative		Negative
Bilirubin ⁰¹	Negative		Negative
Urobilinogen,Semi-Qn ⁰¹	0.2	mg/dL	0.2-1.0
Nitrite, Urine ⁰¹	Negative		Negative
Microscopic Examination ⁰¹	Microscopic not indicated and not performed.		

Disclaimer
The Previous Result is listed for the most recent test performed by Labcorp in the past 5 years where there is sufficient patient demographic data to match the result to the patient. Results from certain tests are excluded from the Previous Result display.

Icon Legend
▲ Out of Reference Range ■ Critical or Alert

Performing Labs
01: SO - Labcorp San Diego, 13112 Evening Creek Dr So Ste 200, San Diego, CA 92128-4108 Dir: Earle Collum, Jr, MD
02: BN - Labcorp Burlington, 1447 York Court, Burlington, NC 27215-3361 Dir: Sanjai Nagendra, MD
For Inquiries, the physician may contact Branch: 800-859-6046 Lab: 858-668-3700

Patient Details	Physician Details	Specimen Details
14644, Donor	J KUAN	Specimen ID: 163-253-1902-0
[REDACTED]	[REDACTED]	Control ID: L2404540256
[REDACTED]	[REDACTED]	Alternate Control Number: L2404540256
Phone: 858-732-8500	Phone: 858-732-8500	Date Collected: 06/11/2024 1244 Local
Date of Birth: [REDACTED]	Account Number: 04087470	Date Received: 06/12/2024 0000 ET
Age: [REDACTED]	Physician ID:	Date Entered: 06/12/2024 0150 ET
Sex: Male	NPI: 1891885745	Date Reported: 06/14/2024 1106 ET
Patient ID:		
Alternate Patient ID:		