CGT Exome v3.1.2

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0084286 - 15425988	**Sample type:**	Blood	**Clinic:**	WeFIV
Patient name:		**Date of draw:**	18/10/2021	**Doctor:**	FLORENCIA DATRI
Patient DOB:		**Date of receipt:**	22/10/2021		
Ethnic group:	Not specified	**Report date/time:**	10/03/2026		
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Fructose intolerance, hereditary

Gene :	ALDOB	**Allele:**	Het
DNA Change:	NM_000035.3:c.448G>C	**Inheritance:**	AR
Protein change:	p.Ala150Pro	**OMIM phenotype:**	229600
Variant classification:	Pathogenic		

Glanzmann thrombasthenia

Gene :	ITGB3	**Allele:**	Het
DNA Change:	NM_000212.2:c.197T>G	**Inheritance:**	AR
Protein change:	p.Leu66Arg	**OMIM phenotype:**	273800
Variant classification:	Pathogenic		

Stargardt disease type 1; Cone-rod dystrophy type 3

Gene :	ABCA4	**Allele:**	Het
DNA Change:	NM_000350.2:c.658C>T	**Inheritance:**	AR
Protein change:	p.Arg220Cys	**OMIM phenotype:**	248200; 604116
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)

LOW COVERAGE VARIANTS

DYNC2H1:NM_001080463.1:c.7141delG.These variants have a coverage lower than 7X and it is not possible to determine if they are present or not in the sample (non-informative variants).

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

Report's language has been updated by clinic's request.

TEST METHODOLOGY

1.DNA extraction from the biological sample. 2. Next Generation Sequencing of gene regions where known mutations are located (list available at https://cgt.igenomix.com/diseases-list/). 3. Raw data analysis using bioinformatics. QC parameters require that more than 99.7% of the tested variants have coverage greater than the minimum read depth (10x). 4. Complementary testing by other techniques for: a) SMN1 gene: exon 7 deletion; exon 7-8 deletion; b) CYP21A2 gene: frequent mutations (https://cgt.igenomix.com/diseases-list/); c) HBA1/HBA2 genes: frequent deletions (https://cgt.igenomix.com/diseases-list/); d) FMR1 gene: CGG repeat sizing (females only); e) DMD gene: frequent deletions and duplications (females only); f) F8 gene: intron 22 inversion (females only).

TEST LIMITATIONS

The CGT test only includes analysis of the specific variants included into the list at https://cgt.igenomix.com/diseases-list/, and no others. Therefore, the CGT test does not cover all monogenic diseases nor 100% of disease-causing mutations for each tested gene. The test does not include the analysis of conditions associated with mitochondrial DNA, multifactorial, digenic or dominant inheritance. The test does not detect large rearrangements (inversions, deletions and duplications more than 15 nucleotides), mutations located in regulatory regions or intronic regions outside the +/-3bp cut off or in low sequence coverage areas. DNA changes caused by trinucleotide repeat expansions are not detected, except those indicated in the methodology section. Finally, if our assessment of a variant fails to meet our QC parameters due to low coverage, a result for the variant(s) will not be issued. The analytical detection rate is higher than 99%. The clinical sensitivity varies among conditions (e.g.: for HEXB gene, 30% of affected patients are carriers of a 16 kb deletion that is not included in the test). The sensitivity for SMN1 is approximately 96% because point mutations or small ins/del are not analyzed and, for a normal result (2 copies detected), it is not possible to be certain that the two copies are each in one of the two alleles (non-carrier) or if both are in the same allele (cis) and no copies in the other (carrier).
A negative result for the variants included in CGT does not exclude the possibility of being a carrier. The presence of pseudogenes and/or rare polymorphisms and/or homopolymers may lead to false negative or false positive results. A negative result for the CGT variants does not exclude the possibility of a de novo mutation being present 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 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 ARGENTINA S.A 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.

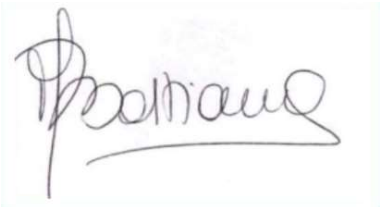
*IGENOMIX SPAIN holds CLIA Certificate of Compliance: #99D2146167.

EXEMPTION CLAUSE OF DIAGNOSTIC LIABILITY

The genetic diagnosis services carried out by IGENOMIX ARGENTINA S.A are exclusively intended to be interpreted by qualified/certified health professionals.

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 ARGENTINA S.A is not responsible for any decisions made or actions undertaken by the contracting party based on the results provided by IGENOMIX ARGENTINA S.A 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
	
Martina Di Bastiano Laboratory Leader	**Arantxa Hervas PhD** 3025-CV Biotechnologist

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

Fructose intolerance, hereditary

What is Fructose intolerance, hereditary?

Hereditary fructose intolerance follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ALDOB gene located on chromosomal region 9q21.3-q22.2. The age of onset is neonatal/infantile. This disease is characterized by severe abdominal pain, vomiting, and hypoglycemia following ingestion of fructose or other sugars metabolised through fructose-1-phosphate. The prevalence is 1:100,000-9:100,000.

What is the next step if I am a carrier of Fructose intolerance, hereditary?

If you are a carrier of Fructose intolerance, hereditary 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?

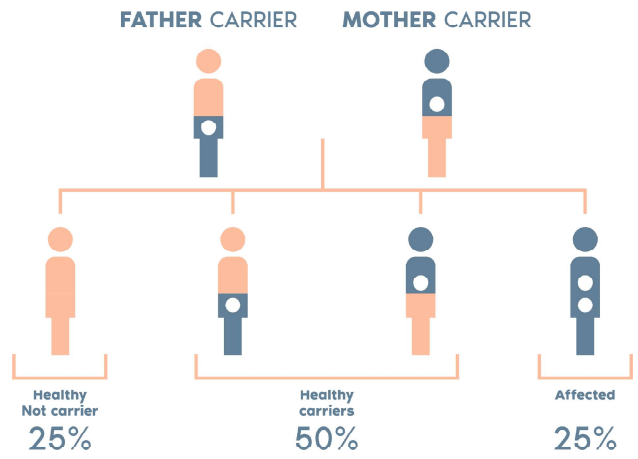
If your partner tests negative for Fructose intolerance, hereditary, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual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

What if both parents are carriers of Fructose intolerance, hereditary?

When both parents are carriers of Fructose intolerance, hereditary, 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.

Glanzmann thrombasthenia

What is Glanzmann thrombasthenia?

Glanzmann thrombasthenia is an autosomal recessive bleeding disorder characterized by failure of platelet aggregation and by absent or diminished clot retraction. The abnormalities are related to quantitative or qualitative abnormalities of the GPIIb/IIIa platelet surface fibrinogen receptor complex resulting from mutations in either the GPIIb or GPIIIa genes (Rosenberg et al., 1997). People with Glanzmann thrombasthenia tend to bruise easily, have frequent nosebleeds (epistaxis), and may bleed from the gums. They may also develop red or purple spots on the skin caused by bleeding underneath the skin (petechiae) or swelling caused by bleeding within tissues (hematoma).

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

If you are a carrier of Glanzmann thrombasthenia 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?

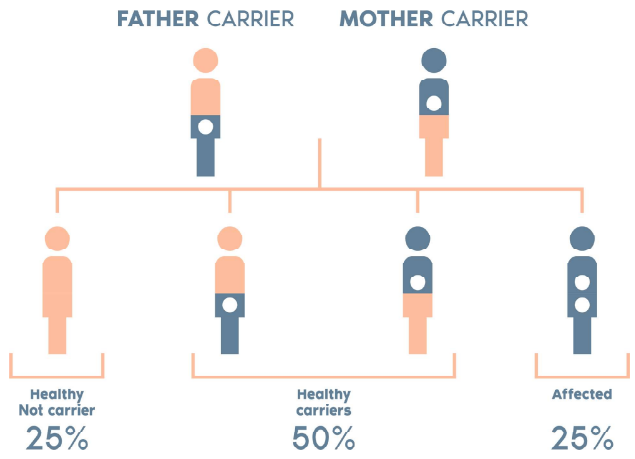
If your partner tests negative for Glanzmann thrombasthenia, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual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

What if both parents are carriers of Glanzmann thrombasthenia?

When both parents are carriers of Glanzmann thrombasthenia, 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.

Stargardt disease type 1; Cone-rod dystrophy type 3

What is Stargardt disease type 1; Cone-rod dystrophy type 3?

Stargardt disease type 1 follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the ABCA4 gene located on chromosomal region 1p22. The age of onset is infantile. This disease is characterized by progressive central vision loss, mild loss of color vision, delayed dark adaptation and macular atrophy with or without paramacular flecks and degeneration of the underlying retinal pigment ephitelium. The estimated prevalence is 1:8,000-10,000. Mutations in the ABCA4 gene account also for 30 to 60 percent of cases of cone-rod dystrophy that are inherited in an autosomal recessive pattern. The problems associated with this condition include a loss of visual sharpness (acuity), an increased sensitivity to light (photophobia), and impaired color vision. These vision problems worsen over time.

What is the next step if I am a carrier of Stargardt disease type 1; Cone-rod dystrophy type 3?

If you are a carrier of Stargardt disease type 1; Cone-rod dystrophy type 3 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?

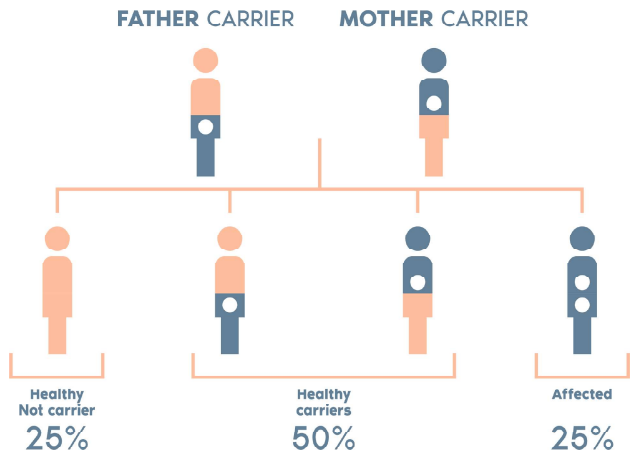
If your partner tests negative for Stargardt disease type 1; Cone-rod dystrophy type 3, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. Therefore, a residual risk remains of having unknown or undetectable pathogenic variants using current technology.

What if both parents are carriers of Stargardt disease type 1; Cone-rod dystrophy type 3?

When both parents are carriers of Stargardt disease type 1; Cone-rod dystrophy type 3, 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 CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, ACO2, APRT, ACTA1, ADK, AMPD1, AMPD2, SLC25A4, FDXR, AGRN, ALB, ALDOA, AFP, NAGA, SLC3A1, ACY1, AGT, AGTR1, ACE, CD19, SERPINC1, ALDH7A1, SERPINA1, IFNGR1, AQP2, RARS1, CYP19A1, DDC, ASNS, ATP2A1, ATP6V1E1, NPR2, SLC4A1, CD40, B2M, BLVRA, CD79A, BMP1, DST, COL17A1, CAD, CDH3, CAST, CACNA1D, CAPN1, CAPN3, CASQ2, CA5A, CAT, TYRP1, CTSD, CP, HSPD1, CLCN1, CYP11A1, COL4A3, COL7A1, COL4A4, COL1A2, COL9A1, COL6A1, COL6A2, COL6A3, COL9A2, COL11A1, COL11A2, COL18A1, COL13A1, MMP2, DCC, C1S, CR2, C3, C5, C8B, GJB2, GJA1, CRYAA, CRYAB, CRYBB3, CNGA1, PPIB, CTPS1, IL10RB, CYC1, POR, CYP11B2, COX6B1, CD55, ALAD, DSP, DES, DSG1, CYP24A1, PCBD1, ERCC2, SLC26A3, EGR2, NT5E, ETFB, TYMP, EDN3, EDNRB, ENO3, ERCC3, ERCC4, ERCC5, SLC1A1, CFH, F13A1, F13B, FTL, FGA, FGB, FGG, FLG, FMO3, FOLR1, FSHR, FSHB, FH, ABAT, GGCX, TACSTD2, MPV17, GCK, SLC2A1, GOT2, SLC2A2, GRIK2, GRIN1, ALDH18A1, GFPT1, EPRS1, GPX4, GLRA1, GLRB, GYS1, GYS2, GNRHR, GRN, CSF3R, GHRHR, GH1, GNAT1, GNAT2, HBA2, HBB, HGF, HSPG2, HK1, HARS1, HOXA1, VSX2, HADHB, IL7R, IGLL1, ADAR, CD79B, ITPR1, IGF1R, IGF1, SOD1, ITGA6, ITGB4, ITGB6, ISG15, INSR, IL1RN, IL2RA, KRT5, KRT14, KRT10, LDHA, LAMB1, LAMC2, LAMB3, LAMA1, LAMB2, LMNA, LARS1, LIFR, PTPRC, LIPE, LHB, LHCGR, LCK, PLOD1, LIM2, MAK, MGP, LAMA2, MARS1, MTR, MITF, MAPT, MTTP, MPZ, MAG, MPL, MYH2, MYO5A, MYBPC1, NDUFV1, IL12B, NEB, NGF, PCSK1, NEFL, TAC3, PNP, LEP, SPINK1, PTH, PTH1R, TAP1, PRF1, PEX2, ABCB4, ALPL, PGM1, GPI, MTHFD1, PHKG2, PSPH, PHKB, SERPINF1, POU1F1, JUP, ENPP1, PLG, SERPINE1, ITGB3, POLE, POLG, KCNE1, INS, PRDX1, PSAP, PROS1, F2, TYK2, ZAP70, PRKCD, PSMB8, EPB42, BCHE, PYCR1, PDHB, RDX, PRPH2, RAG1, RAG2, REN, RD3, RPE65, PDE6A, PDE6B, RLBP1, RARB, RBP4, RBP3, RHO, ROM1, RYR1, AHCY, SAG, STIL, SPR, SLC34A1, SLC5A1, SLC5A2, SPTA1, CSTA, EPCAM, SURF1, CD27, CD3D, CD3E, ITK, TERT, TK2, TG, TSHB, TTN, TFRC, ERBB3, THRB, TGM1, SLC25A1, TNNT1, TH, NTRK1, UQCRC2, UCHL1, DPAGT1, UNG, UGT1A1, 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Gene mean coverage 50x-100x AK1, AK2, APOE, BCAT2, CALCRL, CHAT, MME, C1QA, C1QB, C1QC, DHFR, DHODH, ERCC1, SLC6A3, DAG1, EPB41, EDN1, EGFR, CFD, B4GALT1, GPT2, GLUL, GPD1, GSC, CSF2RB, HMOX1, HPCA, IL10RA, ITPA, IFNGR2, MDH2, MPI, NDUFS1, RMRP, TACR3, CHMP1A, FGF3, TAP2, ACP5, PGM3, CD36, GP9, POMC, SFTPB, PDE6G, GRK1, SPARC, SLC9A3, CD3G, CD247, CD81, TRHR, TPI1, TPM3, DDR2, VLDLR, FLI1, HCFC1, SLC6A8, SLC16A2, OPHN1, DLG3, ABCD1, ARX, CHM, PLP1, NR0B1, FTSJ1, ZDHHC9, NDP, RS1, GJB1, PGK1, CDH11, SLC1A4, SLC18A3, SLC25A3, AKR1C2, ALDH1A3, P2RY12, MASP1, SLC11A2, CPT1A, LHX3, ASPH, NECTIN1, IHH, CEBPE, FOXN1, NDST1, LSS, KCNJ11, AMH, TNXB, LEPR, SLC6A9, CLPP, GDF5, CASR, GAMT, TOP3A, SLC10A2, MUSK, SLCO2A1, MYO1E, IRF8, UBE3A, SERPINB8, HESX1, NDUFS7, B4GALNT1, RAX, WNT10B, ARHGDIA, TAPBP, IMPA1, ESRRB, MYD88, ECM1, SCARB2, LAT, WIPF1, CTSC, FUT8, CDSN, PEX10, ORC4, KIF1C, CAVIN1, TRDN, CRADD, TNFRSF11A, BCL10, DPH1, AP1S1, CTSF, DPM2, AIMP1, COX15, CYP7B1, SGPL1, MED17, NDUFA10, PEX11B, GOSR2, GRM6, TBX15, DNAJB2, SLC6A5, KISS1R, STAT5B, AGXT, BLOC1S6, B4GALT7, LAMC3, ST3GAL5, PLPBP, SP110, GRM1, ZNHIT3, BLNK, ZNF423, WARS2, MALT1, MKKS, DGAT1, BRF1, STK4, SPTBN2, CORO1A, STX11, SPINT2, GAS8, MESP2, CHST6, IL21R, ALX4, ABCG8, B4GAT1, CDT1, ATF6, CIB2, CLDN14, PCDH12, PRX, SOST, DCDC2, NUP62, TMC6, TMC8, SLC35C1, ALG1, STIM1, DPM3, DCLRE1C, MBOAT7, PIP5K1C, SLC19A3, IRX5, SLC45A2, SIX6, ESPN, ANTXR1, STRC, EIF2B2, NHP2, CD320, LZTFL1, SFRP4, OPA3, SP7, UPB1, HEXA, JAM3, COG8, ATP6V1A, LIAS, CLN3, HYAL1, MTHFR, MCPH1, ALG12, FYCO1, PTF1A, TMIE, SRD5A2, AIRE, LIPH, DYM, HPS6, EPM2A, ARSA, ADA2, COLEC10, CCNO, HSD3B7, MCFD2, GNPTG, RCBTB1, SLC52A2, ASPA, NHLRC1, CLN5, PUS1, XYLT2, TTC8, EXT1, CHSY1, ADSL, NEU1, LGI4, CHST14, SMOC1, NCF1, GTPBP3, GLIS2, GRHL2, GLDN, PTRH2, PSMC3IP, SLC39A8, SLC39A13, SLC39A14, TSEN15, SLC5A7, GJC2, ARL6, LRIG2, SLC6A19, ARL13B, CABP4, MRPS16, WDR45B, LPAR6, SLC25A22, TTC7A, REEP6, ERCC8, ERCC6, MAP3K20, NECTIN4, NAGLU, CRB2, MARS2, NEK9, MMACHC, RNF216, COL25A1, CLDN19, DNAL1, PRMT7, MCM9, B3GALNT2, MED25, PIGV, PIGW, ORAI1, DOK7, SLC6A17, LINS1, TRAPPC6B, GLYCTK, UBA5, UFM1, PDSS2, MARVELD2, FLAD1, PRCD, GINS1, PDE10A, PIGY, CYP2U1, HYLS1, CNPY3, HYDIN, SLC25A46, RBCK1, FTO, OBSL1, TRMT5, SLC25A26, RUSC2, FAM20C, FAM20A, SLC30A10, FARS2, ERLIN1, ARV1, PGAP1, TBC1D20, LMF1, MTFMT, NKX2-6, PGAP3, ISCU, B9D2, TRAPPC9, MRPS34, OTUD6B, IYD, ZNF469, PNPLA1, ADAMTSL2, CILK1, NDUFAF5, FECH, RSPH9, TBC1D7, DSTYK, RNF168, GPIHBP1, SDHAF1, COQ4, TRNT1, NDUFAF3, SERPINF2, SMG9, CYB5R3, PPP1R15B, MOCOS, USB1, ZC3H14, FEZF1, KIAA1549, SLC52A3, TPRN, HPSE2, SCARF2, ALG11, SOBP, GZF1, BPGM, SPATA5, ZBTB24, MCIDAS, TRIM2, CCDC8, WDR81, HSD11B2, ACSF3, C19orf12, NUP93, ATAD1, BRAT1, LRMDA, KANK2, UVSSA, MPC1, COA6, SNX10, POMGNT2, DHTKD1, TECPR2, DDHD2, POMK, METTL23, CERS3, ADAT3, ANKS6, ARL2BP, TIMMDC1, COQ8B, CEP19, CDIN1, ERCC6L2, NADK2, VPS53, JAGN1, TRMT10A, ELP2, SPRTN, FAR1, LMOD3, TMEM107, LEMD2, PYCR2, ARHGEF18, PRDM12, TMEM199, TANGO2, TBCK, DNAJC21, PYROXD1, TECRL, PRUNE1, CLDN10, ALPK3, ARMC9, MSTO1, IQCE, TRIT1

Gene mean coverage < 50x LRPAP1, CD59, CA8, PCYT1A, ESR1, KDSR, H6PD, GP1BB, HOXB1, HOXC13, HMX1, NRL, WNT1, WNT3, PAX7, PRRX1, ZBTB16, PDXK, CD8A, TF, UQCRB, MECP2, POU3F4, DCX, PAK3, EDA, AP1S2, IDS, AR, CIITA, MEOX1, SCN1B, SMN1, PDX1, GFER, FOXE3, GPR68, DGKE, WNT7A, SOX18, ENTPD1, NKX3-2, EMP2, FADD, FOXE1, PHOX2A, OCLN, CA12, SCO1, SLC33A1, CLDN1, NDUFA9, GYG1, RNASEH1, GNB5, FRRS1L, CRIPT, NEUROG3, CPLX1, S1PR2, KYNU, CYP26B1, RBM8A, GAN, DHH, SLC35A3, FBXL4, KY, EXTL3, NKX6-2, ALX3, PANK2, STAMBP, BSND, MPIG6B, FKRP, MLYCD, AP4S1, TIMM50, TWIST2, CLCF1, SAR1B, CNNM2, CLN8, HES7, XYLT1, CYP26C1, PEX26, TP53RK, NMNAT1, GTF2H5, BTD, MPLKIP, IER3IP1, L2HGDH, GLRX5, BLOC1S3, SLC24A4, ATOH7, KREMEN1, VPS37A, PIGM, DCPS, RSPO4, SLC35D1, SYT14, ISCA1, PPM1K, NHEJ1, CISD2, TXNL4A, ERLIN2, CLMP, NDUFAF6, NDUFA11, RNASET2, CYB5A, POLR1D, MSRB3, CCDC115, TMCO1, B9D1, FKBP14, DDHD1, MTO1, COX20, TMEM231, B3GALT6, IBA57, ISCA2, BHLHA9, OTULIN, SLC7A14, LYRM7, RSPH3, WDR73, CLPB, CEP78, LIPT2

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).