CGT Exome v3.2.3

Patient Information		Sample Information		Clinic Information	
Unique pat id.:	0060838 - 15429318	**Sample type:**	Blood (EDTA)	**Clinic:**	IVI BUENOS AIRES
Patient name:		**Date of draw:**	11/05/2022	**Clinician:**	FERNANDO NEUSPILLER
Patient DOB:		**Date of receipt:**	23/05/2022		
Gender:	Female	**Report date/time:**	24/06/2022 09:22		
Ethnic group:	Caucasian				
Indication:	No family history				

TEST RESULTS

POSITIVE

The individual is carrier of:

Thalassemia, alpha-

Gene :	HBA	**Allele:**	Het
DNA Change:	-α3.7	**Inheritance:**	AR
Protein change:	-	**OMIM phenotype:**	604131
ACMG classification:	Pathogenic		

Thyroid dyshormonogenesis, type 6

Gene :	DUOX2	**Allele:**	Het
DNA Change:	NM_014080.4:c.1825C>T	**Inheritance:**	AR
Protein change:	p.Pro609Ser	**OMIM phenotype:**	607200
ACMG 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 a few or all of these variants and this information may be significant for them and for their offspring.
If a patient and partner, or gamete source, 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 is also the case for a negative personal result when a partner or a gamete donor is a carrier for one or more of these analyzed genes. However, due to test limitations present for any genetic test, this low risk is not zero (see limitations section and informed consent)

LOW COVERAGE VARIANTS

There are no low coverage 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) and 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

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 (bioinformatic pipeline v1.0). QC parameters require that more than 99.7% of the tested variants have coverage greater than the minimum read depth (7x). 4. Complementary testing by other techniques for: a) SMN1 gene: exon 7 deletion; b) CYP21A2 gene: frequent mutations; c) HBA1/HBA2 genes: frequent deletions; d) FMR1 gene: CGG repeat sizing (females only); e) DMD gene: frequent deletions/duplications; f) F8 gene: intron 22 inversion (females only); g) FXN gene: GAA repeat sizing.

TEST LIMITATIONS

The CGT test only includes analysis of the specific variants included into the list (list of variants analyzed are available by request), 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. For copy number variation analysis, when a normal result is obtained (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 (silent carrier). 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. In the general population there is a 3-5% risk for bith defects caused by genetic and/or non-genetic factors not detected by this type of test. 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 ARGENTINA S.A . 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.
*IGENOMIX SPAIN holds CLIA Certificate of Compliance: #99D2146167. Part of this test has been outsourced to a referral laboratory whose QMS is based on high Quality Standards, periodically monitored by Igenomix SPAIN and audited by independent external parties.

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 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
	
Ana Cervero PhD 2703-CV Laboratory Director	**Sofia Villanueva** Embryologist

This test or part of this test has been outsourced to a referral Laboratory (IGENOMIX Group) CLIA #99D2146167

Thalassemia, alpha-

What is Thalassemia, alpha-?

Alpha-thalassemia follows an autosomal recessive pattern of inheritance and is caused by pathogenic variants in the HBA1 gene located on chromosomal region 16p13.3. The age of onset is infantile. It is characterized by impaired synthesis of alpha-globin chains leading to a variable clinical picture depending on the number of affected alleles. The disease can be classified into clinical subtypes of increasing severity: silent alpha thalassemia, alpha thalassemia trait (or alpha thalassemia minor), hemoglobin H disease (HbH), and Hb Bart's hydrops fetalis (see these terms). A rare form called alpha-thalassemia-intellectual deficit syndrome has also been identified (see these terms). Alpha thalassemia trait causes microcytosis and hypochromia with absent or mild anemia (often detected on routine blood tests), generally with no other symptoms. HbH patients develop moderate hemolytic anemia with variable amounts of HbH along with occasionally severe splenomegaly, sometimes complicated by hypersplenism. Hb Bart's hydrops fetalis involves a severe deficiency in alpha-globin with serious developmental implications. Alpha-thalassemia-intellectual deficit syndrome is characterized by very mild to severe anemia associated with developmental abnormalities. The prevalence is 1:10,000-5:10,000.

Which is the next step if I am a carrier of Thalassemia, alpha-?

If you are carrier of Thalassemia, alpha- is important that your partner (or gamete donor) test to see if she/he carries the same genetic disease.

What if my partner isn't a carrier?

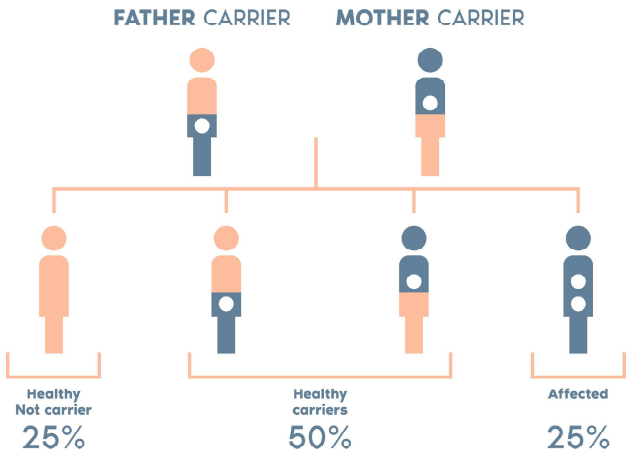
If your partner test negative for Thalassemia, alpha-, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. In consequence, It exists a residual risk of having unknown or undetectable pathogenic variants by current technology.

What if both are carriers de Thalassemia, alpha-?

When both parents are carriers of Thalassemia, alpha-, 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 candidates donors and choose one negative for the same disease.



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

Thyroid dyshormonogenesis, type 6

What is Thyroid dyshormonogenesis, type 6?

Thyroid dyshormonogenesis, type 6 (TDH6) is a disorder due to thyroid dyshormonogenesis, causing hypothyroidism, goiter, and variable mental deficits derived from unrecognized and untreated hypothyroidism.

Which is the next step if I am a carrier of Thyroid dyshormonogenesis, type 6?

If you are carrier of Thyroid dyshormonogenesis, type 6 is important that your partner (or gamete donor) test to see if she/he carries the same genetic disease.

What if my partner isn't a carrier?

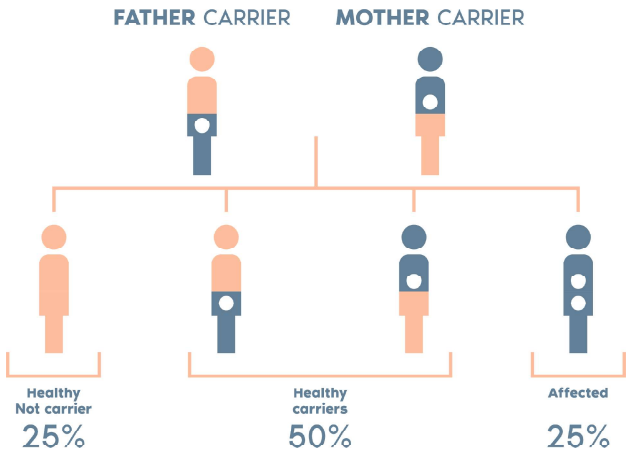
If your partner test negative for Thyroid dyshormonogenesis, type 6, the possibility of having an affected child is very low, significantly lower than the incidence of disease in the general population. However, there is not a test capable of detecting all existing pathogenic variants. In consequence, It exists a residual risk of having unknown or undetectable pathogenic variants by current technology.

What if both are carriers de Thyroid dyshormonogenesis, type 6?

When both parents are carriers of Thyroid dyshormonogenesis, type 6, 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 candidates donors and choose one negative for the same disease.



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

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCC2, ABCC6, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACTA1, ACY1, ADA, ADA2, ADAM9, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS18, ADAMTS2, ADAMTSL2, ADAMTSL4, ADAR, ADAT3, ADGRG1, ADGRG6, ADGRV1, ADK, ADSL, ADSS1, AFF2, AFG3L2, AFP, AGA, AGBL5, AGK, AGL, AGPAT2, AGPS, AGRN, AGT, AGTR1, AGXT, AHCY, AHI1, AICDA, AIMP1, AIMP2, AIPL1, AIRE, AK1, AK2, AKR1C2, AKR1D1, ALAD, ALB, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPK3, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMBN, AMH, AMHR2, AMN, AMPD1, AMPD2, AMT, ANGPTL3, ANKS6, ANO10, ANO5, ANTXR1, ANTXR2, AP1S1, AP1S2, AP3B1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOC2, APOE, APRT, APTX, AQP2, AR, ARFGEF2, ARG1, ARHGDIA, ARHGEF18, ARL13B, ARL2BP, ARL6, ARMC9, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ATAD1, ATF6, ATIC, ATM, ATOH7, ATP13A2, ATP2A1, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1E1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AURKC, AVIL, B2M, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAT2, BCHE, BCKDHA, BCKDHB, BCKDK, BCL10, BCS1L, BEST1, BFSP1, BHLHA9, BIN1, BLM, BLNK, BLOC1S3, BLOC1S6, BLVRA, BMP1, BMPER, BMPR1B, BOLA3, BPGM, BPNT2, BRAT1, BRF1, BRIP1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C12orf65, C19orf12, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C2CD3, C3, C5, C6, C7, C8B, C8orf37, CA12, CA2, CA5A, CA8, CABP2, CABP4, CACNA1D, CACNA2D4, CAD, CALCRL, CANT1, CAPN1, CAPN3, CARD11, CARD9, CARS2, CASQ2, CASR, CAST, CAT, CATSPER1, CAVIN1, CBLIF, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC115, CCDC174, CCDC39, CCDC40, CCDC65, CCDC8, CCDC88C, CCN6, CCNO, CD19, CD247, CD27, CD2AP, CD320, CD36, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDAN1, CDC14A, CDC45, CDCA7, CDH11, CDH23, CDH3, CDHR1, CDIN1, CDK10, CDK5RAP2, CDSN, CDT1, CEBPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP19, CEP290, CEP41, CEP55, CEP57, CEP78, CEP83, CERKL, CERS3, CFAP43, CFAP53, CFD, CFH, CFI, CFL2, CFTR, CHAT, CHKB, CHM, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1, CHUK, CIB2, CIITA, CILK1, CISD2, CIT, CKAP2L, CLCF1, CLCN1, CLCN2, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN10, CLDN14, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM2, CNNM4, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A1, COL11A2, COL13A1, COL17A1, COL18A1, COL1A2, COL25A1, COL27A1, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, CP, CPA6, CPAMD8, CPLANE1, CPLX1, CPS1, CPT1A, CPT2, CR2, CRADD, CRB1, CRB2, CRBN, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYAB, CRYBB1, CRYBB3, CSF2RB, CSF3R, CSPP1, CSTA, CSTB, CTC1, CTH, CTNS, CTPS1, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CUL4B, CUL7, CWC27, CWF19L1, CYB5A, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP24A1, CYP26B1, CYP26C1, CYP27A1, CYP27B1, CYP2R1, CYP2U1, CYP4F22, CYP4V2, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCHS1, DCLRE1C, DCPS, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDRGK1, DDX11, DDX59, DENND5A, DES, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHH, DHODH, DHPS, DHTKD1, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLL3, DMD, DMGDH, DMP1, DMXL2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJB2, DNAJC12, DNAJC19, DNAJC21, DNAJC6, DNAL1, DNASE1L3, DNM1L, DNM2, DNMT3B, DOCK2, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DONSON, DPAGT1, DPH1, DPM1, DPM2, DPM3, DPY19L2, DPYD, DPYS, DRAM2, DRC1, DSG1, DSG4, DSP, DST, DSTYK, DTNBP1, DUOX2, DUOXA2, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, DYSF, DZIP1L, EARS2, ECEL1, ECHS1, ECM1, EDA, EDAR, EDARADD, EDN1, EDN3, EDNRB, EFEMP2, EFL1, EGFR, EGR2, EIF2AK3, EIF2AK4, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4A3, ELAC2, ELMO2, ELOVL4, ELP1, ELP2, EMC1, EMD, EML1, EMP2, ENAM, ENO3, ENPP1, ENTPD1, EOGT, EPB41, EPB42, EPCAM, EPG5, EPM2A, EPRS1, EPS8L2, ERAL1, ERBB3, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERLIN1, ERLIN2, ESCO2, ESPN, ESR1, ESRRB, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXPH5, EXT1, EXTL3, EYS, F10, F11, F13A1, F13B, F2, F5, F7, F8, F9, FA2H, FADD, FAH, FAM126A, FAM161A, FAM20A, FAM20C, FAN1, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBP1, FBXL4, FBXO7, FDXR, FECH, FERMT1, FERMT3, FEZF1, FGA, FGB, FGD1, FGD4, FGF23, FGF3, FGG, FH, FIBP, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLG, FLI1, FLNB, FLVCR1, FLVCR2, FMN2, FMO3, FMR1, FOLR1, FOXE1, FOXE3, FOXN1, FOXRED1, FRAS1, FREM1, FREM2, FRRS1L, FSHB, FSHR, FTCD, FTL, FTO, FTSJ1, FUCA1, FUT8, FXN, FYCO1, FZD6, G6PC, G6PC3, G6PD, GAA, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GAN, GAS8, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GCM2, GCNT2, GCSH, GDAP1, GDF1, GDF5, GDF6, GFER, GFM1, GFPT1, GGCX, GH1, GHR, GHRHR, GHSR, GINS1, GIPC3, GJA1, GJB1, GJB2, GJB6, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLIS2, GLIS3, GLRA1, GLRB, GLRX5, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAT1, GNAT2, GNB5, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNRHR, GNS, GORAB, GOSR2, GOT2, GP1BA, GP1BB, GP6, GP9, GPAA1, GPC6, GPD1, GPHN, GPI, GPIHBP1, GPR143, GPR179, GPR68, GPSM2, GPT2, GPX4, GRHL2, GRHPR, GRID2, GRIK2, GRIN1, GRIP1, GRK1, GRM1, GRM6, GRN, GRXCR1, GSC, GSS, GTF2H5, GTPBP2, GTPBP3, GUCY2C, GUCY2D, GUF1, GUSB, GYG1, GYS1, GYS2, GZF1,

H6PD, HAAO, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HAX1, HBA, HBA1, HBA2, HBB, HCFC1, HELLS, HEPACAM, HERC1, HERC2, HES7, HESX1, HEXA, HEXB, HFM1, HGD, HGF, HGSNAT, HIBCH, HIKESHI, HINT1, HJV, HK1, HLCS, HMGCL, HMGCS2, HMOX1, HMX1, HNMT, HOGA1, HOXA1, HOXB1, HOXC13, HPCA, HPD, HPGD, HPRT1, HPS1, HPS3, HPS4, HPS5, HPS6, HPSE2, HR, HSD11B2, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HTRA1, HTRA2, HYAL1, HYDIN, HYLS1, IARS1, IBA57, ICOS, IDH3B, IDS, IDUA, IER3IP1, IFNGR1, IFNGR2, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGFALS, IGFBP7, IGHMBP2, IGLL1, IHH, IKBKB, IL10RA, IL10RB, IL11RA, IL12B, IL12RB1, IL17RA, IL17RC, IL1RAPL1, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, ILDR1, IMPA1, IMPG2, INPP5E, INPP5K, INPPL1, INS, INSR, INTS1, INVS, IQCB1, IQCE, IRAK4, IRF8, IRX5, ISCA1, ISCA2, ISCU, ISG15, ITCH, ITGA2B, ITGA3, ITGA6, ITGA7, ITGA8, ITGB2, ITGB3, ITGB4, ITGB6, ITK, ITPA, ITPR1, IVD, IYD, JAGN1, JAK3, JAM3, JUP, KANK2, KARS1, KATNB1, 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You can review the list of mutations analysed for the above genes on our website: https://cgt.igenomix.com/diseases-list. If you are unable to access this list digitally, please contact our customer support department for more information.

GLOSARY

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 sex chromosome X. Men with a pathogenic variant have the disease. Women with a pathogenic variant are carriers and generally asymptomatic or with 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:

Mutations present in the two copies of a gene.

- Homozygous mutation (Hom.):

  Each copy of the gene have the same mutation. Generally, it is associated to clinical symptoms.

- Compound heterozygous (Het.):

  Each copy of the gene has a different mutation. Generally, this is associated with clinical symptoms. This situation is referred as having variants "in trans".

Mutation present in one copy of a gene.

- Heterozygous mutation (Het.):
  Only one copy of a gene has a mutation. There is another normal gene copy and the individual is a carrier of the condition. Generally, this is not associated with clinical symptoms.

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

CNV:

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

LARGE GENE CONVERSION:

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