Genomed

Catálogo de pruebas genómicas
por especialidad

Reproducción y prenatales

Gen med

Laboratorio de diagnóstico clínico y genómico

Genética Reproductiva

Panel de Infertilidad

Nuestro panel de infertilidad se recomienda para pacientes que intentan concebir durante un año o más, con problemas de fertilidad conocidos, que han experimentado más de un aborto espontáneo, con menstruación irregular o ausente, con bajo recuento, forma o movimiento de espermatozoides, o con ausencia de desarrollo. de rasgos sexuales secundarios. Nuestro panel incluye los genes más importantes relacionados con la infertilidad en hombres y mujeres. Conocer la causa exacta de la infertilidad permite tomar mejores decisiones de diagnóstico y posibilita un mejor asesoramiento para las parejas.

Incluye 270 genes: AKR1C4, AMH, AMHR2, ANOS1, AR, ARL13B, ARL6, ARX, ATP6V0A4, ATRX, AURKC, B3GLCT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BMP15, BMP4, BNC2, CATSPER1, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CDKN1C, CEP164, CEP290, CEP41, CFAP298, CFAP300, CFAP418, CFTR, CHD4, CHD7, CREBBP, CUL4B, CUL7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DAZ2, DHCR24, DHCR7, DHH, DMRT1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, DUSP6, DYNC2H1, EFNB1, ENPP1, EPG5, ERAL1, ESCO2, EVC, EVC2, FAT4, FBXL4, FEZF1, FGF10, FGF17, FGF8, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FLRT3, FMR1, FOXJ1, FOXL2, FRAS1, FREM2, FSHB, FSHR, GAS8, GATA4, GLI3, GNRH1, GNRHR, GPC3, GRIP1, HBA1, HCCS, HESX1, HEXA, HFE, HNF1B, HOXA13, HS6ST1, HSD17B3, HSD17B4, HSD3B2, HUWE1, IFT172, IFT27, IL17RD, INPP5E, INSL3, IRF6, KHDC3L, KIF7, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LMNA, LRRC56, LZTFL1, MAMLD1, MAP3K1, MCM9, MED12, MID1, MKKS, MKS1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYO7A, MYRF, NEK1, NEK10, NME8, NPHP1, NPHP3, NR0B1, NR0B2, NR3C1, NR5A1, NSMF, ODAD4, OFD1, OPHN1, PANX1, PATL2, PAX6, PCNT, PCSK1, PDE4D, PEX1, PHF6, PITX2, PLCZ1, PNPLA6, POLR3B, POMC, POR, POU1F1, PPARG, PROK2, PROKR2, PROM1, PROP1, PRPH2, PSMC3IP, PTDSS1, PTPN11, RBBP8, RDH5, RHO, RIPK4, RLBP1, RNF216, ROR2, RPGRIP1L, RPL10, RSPO1, SALL1, SAMD9, SDCCAG8, SEMA3A, SETBP1, SGPL1, SOS1, SOX10, SOX2, SOX3, SOX9, SPAG1, SPECC1L, SPRY4, SRY, STAR, TAC3, TACR3, TBX15, TEX11, TEX15, TLE6, TMEM67, TMEM70, TOE1, TP63, TRAF3IP1, TRAIP, TRIM32, TSPYL1, TTC12, TTC21B, TTC8, TUBB8, TWIST2, UBR1, USP9Y, WDPCP, WDR11, WDR35, WEE2, WNT4, ZMYND10, ZP1, ZP2, ZP3

Genes de expansión : AR, FMR1

CentoScreen

Panel de detección integral de 330 trastornos autosómicos, recesivos y ligados al cromosoma X. Análisis adicionales para síndrome X frágil, atrofia muscular espinal e hiperplasia suprarrenal congénita (genes FMR1, SMN1 y CYP21A2 respectivamente). Brinda la oportunidad de tomar decisiones informadas y revisar la gama de opciones disponibles para guiar el embarazo y la planificación familiar. Incluye evaluación de panel completo con análisis de CNV de 34 genes para cada socio.

Existen 3 tipos

-Solamente 1 paciente (Padre o Madre)

-Pareja que quiere concebir (Padres)

-2 parejas (abuelos y padres) [asociación]

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Prueba prenatal (invasiva y no invasiva)

Diseñado para analizar las aneuploidías cromosómicas del feto después de las 10 semanas de gestación. Se reportan sobrerepresentaciones de los cromosomas 21, 18 y 13, así como las aneuploidías de los cromosomas sexuales XO, XXX, XXY y XYY. Esta prueba de detección no detecta aneuploidías en otros cromosomas no mencionados anteriormente y, por lo tanto, no puede excluir anomalías en estos.

Incluye: AAAS, AARS1, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAN, ACAT1, ACE, ACO2, ACOX1, ACP5, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTL6B, ACTN2, ACVR2B, ACVRL1, ACY1, ADA, ADAM17, ADAM22, ADAMTS19, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRHL2, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGR2, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, AKT1, AKT2, AKT3, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALPL, ALS2, ALX3, ALX4, AMACR, AMER1, AMN, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKRD11, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARL6IP1, ARMC4, ARMC9, ARNT2, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD3A, ATCAY, ATIC, ATL1, ATM, ATOH7, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B3, ATP5MD, ATP6AP2, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATR, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT7, B4GAT1, B9D1, B9D2, BAG3, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BGN, BHLHA9, BICD2, BIN1, BLM, BLNK, BMP1, BMP2, BMP4, BMPER, BMPR1B, BOLA3, BPTF, BRAF, BRAT1, BRCA1, BRCA2, BRD4, BRIP1, BRPF1, BRWD3, BSCL2, BSND, BTD, BTK, BUB1B, C12orf57, C12orf65, C15orf41, C19orf12, C1QBP, C2CD3, C8orf37, CA2, CA5A, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CAD, CAMK2A, CAMK2B, CAMTA1, CANT1, CARD11, CARS2, CASK, CASP10, CASR, CAV1, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC114, CCDC115, CCDC151, CCDC22, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCND2, CCNK, CCNO, CCNQ, CCT5, CD19, CD247, CD2AP, CD3D, CD3E, CD3G, CD40, CD40LG, CD79A, CD79B, CD96, CDC45, CDH1, CDH2, CDH23, CDH3, CDK10, CDK13, CDK19, CDK19, CDK5RAP2, CDKL5, CDKN1C, CDON, CDSN, CDT1, CENPF, CENPJ, CEP104, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CEP83, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CIB2, CILK1, CISD2, CIT, CKAP2L, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COROA1, COX10, COX15, COX20, COX6B1, COX7B, CPLANE1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREB3L1, CREBBP, CRELD1, CRIPT, CRKL, CRLF1, CRPPA, CRTAP, CRX, CRYAA, CRYAB, CSF1R, CSF2RB, CSF3R, CSNK1E, CSNK2A1, CSNK2B, CSPP1, CSTA, CSTB, CTC1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTNS, CTPS1, CTSA, CTSD, CTSK, CTU2, CUL3, CUL4B, CUL7, CWC27, CWF19L1, CXCR4, Cxorf56, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCC, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DDX6, DEAF1, DENND5A, DEPDC5, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHTKD1, DHX16, DHX30, DHX37, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DLL3, DLL4, DLX5, DMD, DMP1, DMPK, DMXL2, DNA2, DNAAF3, DNAAF4, DNAAF5, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC3, DNAJC5, DNM1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPF2, DPM1, DPM2, DPYD, DRC1, DSE, DSG1, DSP, DST, DSTYK, DTNA, DUOX2, DUOXA2, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, EDA, EDAR, EDN3, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFNB1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELAC2, ELANE, ELMO2, ELN, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EMX2, ENG, ENPP1, ENTPD1, EOGT, EP300, EPB42, EPCAM, EPG5, EPHB4, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERLIN1,

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ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC8, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, F10, F13A1, F2, F7, F8, F9, FA2H, FADD, FAH, FAM111A, FAM126A, FAM149B1, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FAS, FASLG, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXW11, FBXW4, FCSK, FEZF1, FGA, FGB, FGD1, FGD4, FGF10, FGF12, FGF3, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FGG, FH, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FN1, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXL2, FOXN1, FOXP1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FTCD, FTL, FTSJ1, FUCA1, FUT8, FXN, G6PC, G6PC3, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GAS8, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GDAP1, GDF1, GDF3, GDF5, GDF6, GDI1, GEMIN4, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJA8, GJB2, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMNN, GMPPA, GMPPB, GNA11, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPT2, GPX4, GREB1L, GRHL2, GRHL3, GRIA2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GSS, GTF2H5, GTPBP1, GTPBP3, GUCY2C, GUF1, GUSB, GYS1, GZF1, H1-4, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HAX1, HBA1, HBA2, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HINT1, HIVEP2, HK1, HLCS, HMGCL, HMGCS2, HMX1, HNF1B, HNF4A, HNRNPH2, HNRNPK, HNRNPU, HOXA1, HOXA13, HOXC13, HOXD13, HPD, HPDL, HPGD, HPRT1, HPS1, HPSE2, HRAS, HSD11B2, HSD17B10, HSD17B4, HSD3B2, HSD3B7, HSPA9, HSPD1, HSPG2, HUWE1, HYAL1, HYDIN, HYLS1, IARS1, IARS2, IBA57, ICOS, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFNGR2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IGF1, IGF1R, IGF2, IGFBP7, IGHMBP2, IGSF1, IHH, IKBKB, IKBKG, IL11RA, IL12RB1, IL1RAPL1, IL21R, IL2RA, IL2RG, IL7R, IMPAD1, INPP5E, INPP5K, INPPL1, INS, INSR, INVS, IPO8, IQCB1, IQSEC1, IQSEC2, IRAK4, IRF6, IRF8, IRX5, ISCA2, ITFG2, ITGA3, ITGA6, ITGA7, ITGA8, ITGB4, ITK, ITPA, ITPR1, IVD, JAG1, JAGN1, JAK3, JAM2, JAM3, KANK1, KANSL1, KARS, KARS1, KAT5, KAT6A, KAT6B, KAT8, KATNB1, KBTBD13, KCNA2, KCNB1, KCNC1, KCNC3, KCNE1, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ6, KCNMA1, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD1, KCTD7, KDM5C, KDM6A, KIAA0586, KIAA1109, KIDINS220, KIF11, KIF1A, KIF1C, KIF22, KIF2A, KIF5A, KIF5C, KIF7, KIFBP, KIRREL3, KLF1, KLHL15, KLHL40, KLHL41, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, KRIT1, KRT10, KRT8, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA3, LAMB1, LAMB2, LAMB3, LAMC2, LAMC3, LAMP2, LARGE1, LARP7, LARS2, LAS1L, LBR, LDB3, LEMD3, LETM1, LFNG, LGI4, LHX3, LHX4, LIAS, LIFR, LIG4, LINS1, LIPA, LIPT1, LMBRD1, LMNA, LMOD3, LMX1B, LONP1, LPIN2, LRBA, LRP2, LRP4, LRP5, LRPPRC, LRRC56, LRRC6, LTBP2, LTBP3, LYRM7, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, MAF, MAFB, MAG, MAGEL2, MAGI2, MAGT1, MALT1, MAMLD1, MAN1B1, MAN2B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAP3K1, MAP3K7, MAPK8IP3, MAPRE2, MARS1, MASP1, MAT1A, MATN3, MBD5, MBOAT7, MBTPS2, MC2R, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MDH2, MECOM, MECP2, MECR, MED12, MED13, MED13L, MED17, MED23, MED25, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESP2, METTL5, MFN2, MFRP, MFSD2A, MFSD8, MGAT2, MGME1, MGP, MICU1, MID1, MIPEP, MIR17HG, MITF, MKKS, MKS1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMP13, MMP21, MMUT, MN1, MNX1, MOCS1, MOCS2, MOGS, MORC2, MORC2, MPDU1, MPDZ, MPI, MPLKIP, MPV17, MPZ, MRE11, MRPL3, MRPL44, MRPS2, MRPS22, MRPS34, MSL3, MSMO1, MSX1, MSX2, MTFMT, MTHFR, MTM1, MTO1, MTOR, MTR, MTRR, MTTP, MUSK, MUTYH, MVK, MYBPC1, MYBPC3, MYCN, MYD88, MYH3, MYH6, MYH7, MYH8, MYH9, MYL3, MYO18B, MYO5A, MYO5B, MYO7A, MYOD1, MYPN, MYT1L, NAA10, NAA15, NACC1, NADSYN1, NAGA, NAGLU, NAGS, NALCN, NANS, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NBN, NCAPD3, NCKAP1L, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB8, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEMF, NEU1, NEUROG3, NEXMIF, NF1, NFASC, NFIA, NFIB, NFIX, NFKBIA, NFU1, NGF, NGLY1, NHEJ1, NHLRC1, NHP2, NHS, NIPAL4, NIPBL, NKX2-1, NKX2-5, NKX2-6, NKX3-2, NKX6-2, NLRC4, NLRP3, NMNAT1, NNT, NODAL, NOG, NONO, NOTCH1, NOTCH2, NOTCH3, NOVA2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR2, NR0B1, NR1H4, NR2F1, NR2F2, NR3C2, NR5A1, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, NT5C2, NT5C3A, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2, NUP107, NUP188, NYX, OBSL1, OCLN, OCRL, ODAPH, OFD1, OGDH, OLA1, OPA1, OPA3, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTC, OTOGL, OTUD5, OTUD6B, OTULIN, OTX2, OXCT1, OXR1, P3H1, P4HB, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PALB2, PAM16, PANK2, PAPSS2, PARN, PARS2, PAX2, PAX3, PAX6, PAX8, PC, PCARE, PCBD1, PCCA, PCCB, PCDH15, PCDH19, PCGF2, PCK1, PCNT, PCYT1A, PCYT2, PDCD10, PDE10A, PDE4D, PDE6D, PDE6G, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PEPD, PERCC1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHACTR1, PHEX, PHF21A, PHF6, PHF8, PHGDH, PHIP, PHKG2, PHOX2B, PIEZO1, PIEZO2, PIGA, PIGG, PIGK, PIGL, PIGN, PIGO, PIGQ, PIGS, PIGT, PIGV, PIGY, PIH1D3, PIK3CA, PIK3CD, PIK3R1, PIK3R2, PISD, PITX1,

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Genética Reproductiva

PITX2, PITX3, PKD1L1, PKHD1, PKLR, PLA2G6, PLAA, PLCB1, PLCB4, PLCE1, PLCG2, PLEC, PLEKHG5, PLG, PLK4, PLOD1, PLOD2, PLOD3, PLP1, PLPBP, PLS3, PMM2, PMP22, PMPCB, PMS2, PNKP, PNP, PNPLA1, PNPLA8, PNPO, PNPT1, POC1A, POC1B, POGZ, POLA1, POLD1, POLE, POLG, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMP, POMT1, POMT2, POR, PORCN, POU1F1, PPA2, PPIB, PPM1D, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM12, PRDM16, PREPL, PRF1, PRICKLE1, PRKAG2, PRKAR1A, PRKCD, PRKD1, PRMT7, PROC, PRODH, PROP1, PROS1, PRPS1, PRRT2, PRRX1, PRSS12, PRSS56, PRUNE1, PRX, PSAP, PSAT1, PSMB8, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTH1R, PTHLH, PTPN11, PTPN14, PTPN23, PTPRC, PTRH2, PTS, PUF60, PURA, PUS1, PUS7, PXDN, PYCR1, PYCR2, PYGL, PYGM, PYROXD1, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB18, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAC2, RAD21, RAD50, RAD51C, RAF1, RAG1, RAG2, RAI1, RALGAPA1, RAPSN, RARB, RARS1, RARS2, RASA1, RAX, RB1, RBBP8, RBCK1, RBM10, RBM8A, RBPJ, RDH11, RELN, REN, RERE, RET, RETREG1, RFT1, RFX5, RFX6, RHOA, RHOBTB2, RIC1, RIMS2, RIN2, RIPK4, RIT1, RLIM, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, ROBO3, ROGDI, ROR2, RORA, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL26, RPL5, RPS19, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RTEL1, RTN4IP1, RTTN, RUNX2, RXYLT1, RYR1, SACS, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1, SARS2, SASS6, SATB2, SBDS, SBF1, SC5D, SCAMP5, SCAPER, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCNN1A, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINF1, SERPINH1, SET, SETBP1, SETD1A, SETD2, SETD5, SETX, SF3B4, SFTPB, SFTPC, SFXN4, SGCA, SGCE, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SHANK1, SHANK2, SHH, SHOC2, SHOX, SHROOM4, SIK1, SIL1, SIM1, SIN3A, SIX1, SIX3, SIX5, SKI, SKIV2L, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC26A2, SLC26A3, SLC26A7, SLC27A4, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMCHD1, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNAP29, SNORD118, SNRPB, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX17, SOX2, SOX3, SOX5, SOX6, SOX9, SP110, SP7, SPAG1, SPART, SPAST, SPATA5, SPECC1L, SPEG, SPG11, SPR, SPRED1, SPTAN1, SPTBN2, SPTBN4, SPTLC2, SQSTM1, SRCAP, SRD5A3, SRP54, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STAR, STAT2, STAT5B, STIL, STIM1, STRA6, STRADA, STS, STX11, STX1B, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SUZ12, SVBP, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAB2, TAC3, TACR3, TAF1, TAF13, TAF2, TAF6, TANGO2, TAOK1, TASP1, TAT, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCF12, TCF20, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TECPR2, TEK, TELO2, TERT, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, TH, THAP1, THOC2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF135, RNF168, ROBO3, ROGDI, ROR2, RORA, RPE65, RPGRIP1, RPGRIP1L, RPIA, RPL10, RPL11, RPL13, RPL26, RPL5, RPS19, RPS26, RPS28, RPS29, RPS6KA3, RPS7, RRM2B, RSPH1, RSPH3, RSPO2, RSPO4, RSPRY1, RTEL1, RTN4IP1, RTTN, RUNX2, RXYLT1, RYR1, SACS, SALL1, SALL4, SAMD9, SAMD9L, SAMHD1, SARS2, SASS6, SATB2, SBDS, SBF1, SC5D, SCAMP5, SCAPER, SCARF2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCNN1A, SCO1, SCO2, SCYL1, SCYL2, SDCCAG8, SDHA, SDHAF1, SDHD, SEC23B, SEC24D, SELENOI, SELENON, SEPSECS, SERAC1, SERPINF1, SERPINH1, SET, SETBP1, SETD1A, SETD2, SETD5, SETX, SF3B4, SFTPB, SFTPC, SFXN4, SGCA, SGCE, SGCG, SGPL1, SGSH, SH2D1A, SH3PXD2B, SHANK1, SHANK2, SHH, SHOC2, SHOX, SHROOM4, SIK1, SIL1, SIM1, SIN3A, SIX1, SIX3, SIX5, SKI, SKIV2L, SLC10A7, SLC12A1, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC26A2, SLC26A3, SLC26A7, SLC27A4, SLC2A1, SLC2A10, SLC2A2, SLC30A10, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC39A13, SLC39A8, SLC4A1, SLC4A11, SLC4A4, SLC52A2, SLC52A3, SLC5A1, SLC5A5, SLC5A6, SLC5A7, SLC6A1, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC9A1, SLC9A6, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMCHD1, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNAP29, SNORD118, SNRPB, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX17, SOX2, SOX3, SOX5, SOX6, SOX9, SP110, SP7, SPAG1, SPART, SPAST, SPATA5, SPECC1L, SPEG, SPG11, SPR, SPRED1, SPTAN1, SPTBN2, SPTBN4, SPTLC2, SQSTM1, SRCAP, SRD5A3, SRP54, SRY, SSR4, ST14, ST3GAL3, ST3GAL5, STAG1, STAG2, STAMBP, STAR, STAT2, STAT5B, STIL, STIM1, STRA6, STRADA, STS, STX11, STX1B, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SUZ12, SVBP, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAB2, TAC3, TACR3, TAF1, TAF13, TAF2, TAF6, TANGO2, TAOK1, TASP1, TAT, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX15, TBX18, TBX20, TBX22, TBX3, TBX4, TBX5, TBXAS1, TCF12, TCF20, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TECPR2, TEK, TELO2, TERT, TFAP2A, TFAP2B, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1,

Gen med

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Genética Reproductiva

TH, THAP1, THOC2, THOC6, THRA, TIMM8A, TINF2, TJP2, TK2, TKT, TLK2, TMCO1, TMEM107, TMEM126B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMEM94, TMPRSS6, TMTC3, TMX2, TNFRSF11A, TNFRSF11B, TNFRSF13B, TNFSF11, TNNT1, TOE1, TOP3A, TP53, TP63, TPI1, TPK1, TPM2, TPO, TPP1, TRAPPC11, TRAPPC2, TRAPPC4, TRAPPC9, TREX1, TRIM2, TRIM32, TRIM37, TRIO, TRIP11, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMU, TRNT1, TRPM1, TRPM6, TRPS1, TRPV3, TRPV4, TRPV6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPYL1, TTC19, TTC21B, TTC26, TTC37, TTC7A, TTC8, TTN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TWNK, TXNL4A, TYMP, TYR, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3A, UBE3B, UBR1, UBTF, UCHL1, UFC1, UFM1, UGDH, UGP2, UGT1A1, UMPS, UNC13D, UNC80, UPB1, UPF3B, UQCC2, UQCRC2, UROC1, UROS, USB1, USH1C, USH1G, USH2A, USP18, USP53, USP9X, UVSSA, VAMP1, VAMP2, VANGL1, VARS2, VDR, VIPAS39, VLDLR, VPS13B, VPS33B, VPS37A, VPS41, VPS45, VPS53, VRK1, VSX2, WAC, WAS, WASF1, WASHC5, WDFY3, WDPCP, WDR11, WDR19, WDR26, WDR34, WDR35, WDR37, WDR45, WDR45B, WDR60, WDR62, WDR73, WDR81, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WT1, WWOX, XIAP, XPA, XPC, XRCC2, XRCC4, XYLT1, XYLT2, YAP1, YARS2, YIF1B, YWHAG, YY1, ZAP70, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFP57, ZFPM2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMIZ1, ZMPSTE24, ZMYM6, ZMYND10, ZMYND11, ZNF335, ZNF423, ZNF462, ZNF711, ZNHIT3

Estudios de un solo gen

46,XX inversión sexual tipo 1 SRY
46,XY inversión sexual tipo 8, modificador de AKR1C4
Deficiencia de aromatasa CYP19A1
Azoospermia inducida por microdeleciones del cromosoma Y AZF
Ausencia congénita bilateral de conductos deferentes CFTR
Criptorquidia RXFP2
Sordera e infertilidad masculina STRC
Sordera e infertilidad masculina, relacionadas con CATSPER2
Trastornos del desarrollo sexual con paladar hendido FOXF2
Deficiencia de hormona foliculoestimulante, aislada FSHB
síndrome de gutmacher HOXA13
Síndrome mano-pie-útero HOXA13
Mola hidatidiforme NLRP7
Mola hidatiforme, recurrente, tipo 2 KHDC3L
Hipogonadismo hipogonadotrópico KISS1
Hipogonadismo hipogonadotrópico NSMF
Hipogonadismo hipogonadotrópico tipo 6 FGF8
Hipogonadismo hipogonadtrópico tipo 14 WDR11
Hipospadias tipo 1, ligada al cromosoma X Arkansas
Hipospadias tipo 2, ligada al cromosoma X MAMLD1
Hipoplasia de células de Leydig tipo 1 LHCGR
Oligo-asteno-teratozoospermia NANOS1
Defecto de maduración de ovocitos ZP1
disfunción de la ovogénesis SOHLH1
Disgenesia ovárica tipo 1 FSHR
Disgenesia ovárica tipo 2 BMP15
Síndrome persistente del conducto de Müller tipo 1 AMH
Síndrome persistente del conducto de Müller tipo 2 AMHR2
Preeclampsia/eclampsia tipo 5 CORIN
Pérdida de embarazo, recurrente, relacionada con C4BPA
Pseudohermafroditismo con ginecomastia HSD17B3
SPGF4 SYCP3
SPGF5 AURKC
SPGF6 ESPATA16
SPGF7 CATSPER1
SPGF8 NR5A1
SPGF9 DPY19L2
Anomalías testiculares con o sin cardiopatía congénita GATA4

Se recomienda la prueba de un solo gen en pacientes que tienen:

- Características clínicas distintivas
- Antecedentes familiares de un trastorno específico.
- Trastornos de un solo gen
- Posible trastorno epigenético
- Posibles trastornos de repetición triple
- Confirmación de prueba familiar