Genomed

Catálogo de pruebas genómicas
por especialidad

Tejido óseo, piel, inmunología y dismorfología

Gen med

Laboratorio de diagnóstico clínico y genómico

Tejido óseo

Panel de mineralización anormal

Nuestro panel de mineralización anormal incluye osteogénesis imperfecta, osteopetrosis, trastornos de alta y baja densidad ósea y genes de diagnóstico diferencial necesarios para discriminar la causa genética real. Las enfermedades procesables, como la hipofosfatasia, también se incluyen en nuestro panel.

Incluye 94 genes: ABCC6, ALPL, AMER1, ANKH, ANO5, AP2S1, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CASR, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTSK, CYP24A1, CYP27B1, CYP2R1, DMP1, DSPP, ENPP1, FAH, FAM20C, FERMT3, FGF23, FGFR1, FGFR3, FKBP10, GALNT3, GJA1, GNA11, GNAS, GORAB, GPAA1, HPGD, HRAS, IFITM5, KRAS, LEMD3, LRP4, LRP5, MBTPS2, MESD, MTAP, NBAS, NOTCH2, NRAS, OCRL, OSTM1, P3H1, P4HB, PHEX, PLEKHM1, PLOD2, PLS3, PPIB, PTDSS1, PTH1R, SEC24D, SERPINF1, SERPINH1, SGMS2, SH3PXD2B, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC9A3R1, SLCO2A1, SNX10, SOST, SOX9, SP7, SPARC, SQSTM1, TAPT1, TBXAS1, TCIRG1, TGFB1, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, TYROBP, VDR, WNT1, XYLT2, ZBTB20, TENT5A

Gen med

Laboratorio de diagnóstico clínico y genómico

Dermatología

CentoSkin

Solución para pacientes que presentan trastornos de la piel. Nuestro panel incluye genes para hipotricosis, epidermólisis ampollosa e ictiosis congénita, entre otros. Para melanoma, consulte nuestra sección de Oncología.

Incluye 152 genes: ABCA12, ABHD5, ACD, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALOX12B, ALOXE3, AP1S1, AP3B1, AP3D1, APCDD1, ARHGAP31, ARSL, ATP6V0A2, ATP7A, BLOC1S3, BLOC1S6, CASP14, CDSN, CERS3, CHST8, CLDN1, COL17A1, COL7A1, CPOX, CSTA, CTC1, CYP4F22, DKC1, DLL4, DOCK6, DSG1, DSG4, DSP, DST, DTNBP1, EBP, EDA, EDAR, EDARADD, EDN3, EDNRB, EFEMP2, ELN, ELOVL4, EOGT, EPG5, ERCC2, ERCC3, EXPH5, FBLN5, FECH, FERMT1, FLG, FLG2, GJB2, GJB3, GJB4, GJB6, GPR143, GTF2H5, HMBS, HPS1, HPS3, HPS4, HPS5, HPS6, HR, ITGA3, ITGA6, ITGB4, JUP, KCTD1, KDSR, KITLG, KRT1, KRT10, KRT14, KRT2, KRT5, KRT74, KRT85, KRT9, LAMA3, LAMB3, LAMC2, LIPH, LIPN, LORICRIN, LPAR6, LRMDA, LYST, MBTPS2, MC1R, MITF, MLPH, MMP1, MPLKIP, MYH9, MYO5A, NECTIN1, NHP2, NIPAL4, NOP10, NOTCH1, NSDHL, OCA2, PARN, PAX3, PEX7, PHGDH, PHYH, PKP1, PLEC, PNPLA1, POMP, PPOX, PSAT1, PYCR1, RAB27A, RBPJ, RPL21, RTEL1, SDR9C7, SERPINB8, SLC24A5, SLC27A4, SLC38A8, SLC45A2, SNAI2, SNAP29, SNRPE, SOX10, SPINK5, ST14, STS, SUMF1, TERT, TGM1, TGM5, TINF2, TYR, TYRP1, UROD, UROS, USB1, VPS33B, WNT10A, WRAP53, ZMPSTE24

Gen med

Laboratorio de diagnóstico clínico y genómico

Inmunología

CentoInmuno

Solución para la inmunodeficiencia y los trastornos de inmunodeficiencia combinada grave (SCID). Nuestro panel incluye genes dirigidos a la inmunodeficiencia combinada grave, la neutropenia congénita, la deficiencia de anticuerpos primarios, la inmunodeficiencia variable común, la enfermedad granulomatosa crónica, la linfoproliferación autoinmune y la agammaglobulinemia.

Incluye 326 genes: ACD, ACP5, ACTB, ADA, ADA2, ADAR, AICDA, AIRE, AK2, AP1S3, AP3B1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CEBPE, CFAP298, CFB, CFD, CFH, CFHR1, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COPA, CORO1A, CR2, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1C, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE1L3, DNMT3B, DOCK2, DOCK8, DRC1, DTNBP1, ELANE, EPG5, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCN3, FERMT3, FGA, FGB, FGG, FOXN1, FOXP3, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GUCY2C, HAX1, HELLS, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYDIN, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17RA, IL17RC, IL1RN, IL21R, IL2RA, IL2RG, IL36RN, IL7R, INO80, IRAK4, IRF3, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, KRAS, LAMTOR2, LAT, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MASP2, MBL2, MCM4, MEFV, MOGS, MS4A1, MSN, MTHFD1, MVK, MYD88, MYSM1, NBN, NCF1, NCF2, NCF4, NCSTN, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE3, OFD1, OSTM1, OTULIN, PARN, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PMM2, PMS2, PNP, POLA1, POLE, PRF1, PRKCD, PSENEN, PSMB4, PSMB8, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RSPH1, RSPH4A, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEMA3E, SERPING1, SH2D1A, SKIV2L, SLC29A3, SLC35C1, SLC7A7, SMARCAL1, SMARCD2, SP110, SPAG1, SPINK5, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STING1, STK4, STX11, STXBP2, TAFAZZIN, TAP1, TAP2, TAPBP, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERT, TFRC, THBD, TICAM1, TINF2, TLR3, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF11, TPP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10

Gen med

Laboratorio de diagnóstico clínico y genómico

Dismorfología

CentoDismorfia

Diseñado para ayudar a los médicos a diagnosticar a los pacientes que padecen un síndrome dismórfico. El panel incluye craneosinostosis, trastornos craneofaciales, paladar hendido/labio, holoprosencefalia, síndrome de Waardenburg, enfermedad de Hirschsprung, lisencefalia y trastornos de malformación cerebral, entre otros.

Incluye 770 genes: A2ML1, ABCA12, ABCB6, ABCC6, ABL1, ACP5, ACTA1, ACTA2, ACTB, ACTG1, ACVR2B, ADAMTS18, ADAMTS2, ADAMTSL2, ADGRG1, ADGRG6, AEBP1, AFF4, AGPS, AGRN, AHDC1, AHI1, AKR1C4, AKT3, ALDH18A1, ALDH1A3, ALG2, ALMS1, ALPL, ALX1, ALX4, AMELX, AMER1, AMH, AMHR2, AMPD2, ANKH, ANKLE2, ANKRD11, ANKS6, ANO5, ANOS1, AP4M1, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARSL, ARX, ASPM, ASXL1, ASXL3, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1E1, ATP7A, ATR, ATRX, B3GALNT2, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCL11A, BCOR, BGN, BIN1, BMP1, BMP2, BMP4, BMPR1B, BNC2, BRAF, C1R, C1S, CANT1, CASK, CASR, CBL, CBS, CC2D2A, CCDC103, CCDC28B, CCDC39, CCDC40, CCDC65, CCM2, CCN6, CCNO, CDC45, CDH1, CDK13, CDK5RAP2, CDKN1C, CDON, CENPF, CENPJ, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CFAP298, CFAP418, CFAP53, CFL2, CHAT, CHD4, CHD7, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CILK1, CIT, CLCN5, CLP1, CNOT1, CNTNAP1, COASY, COG5, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COX7B, CRB2, CREB3L1, CREBBP, CRELD1, CRIPT, CRPPA, CRTAP, CSGALNACT1, CSPP1, CTNNA2, CTNND1, CUL4B, CUL7, CYB5A, CYP19A1, CYP1B1, CYP26B1, DAG1, DCC, DCHS1, DCX, DDR2, DDX59, DEAF1, DHCR24, DHCR7, DHODH, DLL3, DLL4, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAJB13, DNAL1, DNM2, DOCK6, DOK7, DPAGT1, DPF2, DRC1, DSE, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2LI1, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFNB1, EFTUD2, EGR2, EIF2AK3, EIF2S3, ELN, ENPP1, EOGT, EPG5, ERCC1, ERCC2, ERCC5, ERCC6, ERF, ESCO2, EVC, EVC2, EXOSC3, EXOSC9, EXT1, EXT2, EYA1, FAM20C, FAS, FAT4, FBLN1, FBLN5, FBN1, FBN2, FBXL4, FBXW11, FGD1, FGF10, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLCN, FLNA, FLNB, FOXC1, FOXE1, FOXL2, FRAS1, FREM1, FREM2, FZD6, GAS8, GATA4, GBA, GBE1, GDF1, GDF3, GDF5, GDF6, GDNF, GFPT1, GGCX, GJA1, GLE1, GLI2, GLI3, GMPPB, GNAS, GNPAT, GORAB, GPC3, GPC6, GRHL3, GRIP1, HBA1, HCCS, HDAC8, HES7, HESX1, HEXA, HMX1, HOXA13, HOXD13, HRAS, HSPG2, HUWE1, HYDIN, HYLS1, IER3IP1, IFITM5, IFT122, IFT140, IFT172, IFT27, IFT43, IFT80, IFT81, IGF1R, IHH, IL11RA, INPP5E, INPPL1, INVS, IPO8, IRF6, ITGB4, JAG1, KAT6B, KATNB1, KBTBD13, KDM5C, KDM6A, KIAA0586, KIF11, KIF14, KIF22, KIF2A, KIF5C, KIF7, KIFBP, KIT, KLHL40, KLHL41, KMT2A, KMT2D, KNL1, KRAS, KRIT1, L1CAM, LAMA2, LAMA3, LAMB1, LAMB3, LAMC2, LARGE1, LBR, LFNG, LIFR, LMNA, LMOD3, LMX1B, LOX, LRP2, LRP4, LRP5, LTBP3, LZTFL1, LZTR1, MAB21L2, MACF1, MAFB, MAGEL2, MAP2K1, MAP2K2, MASP1, MATN3, MBTPS2, MCIDAS, MCPH1, MED12, MED13L, MED17, MEGF8, MEIS2, MEOX1, MESP2, MFAP5, MFRP, MFSD2A, MGP, MID1, MITF, MKKS, MKS1, MMP13, MMP15, MMP21, MMP9, MN1, MRAS, MSMO1, MSX1, MSX2, 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, MTM1, MUSK, MYBPC1, MYCN, MYH11, MYH2, MYH3, MYH8, MYLK, MYO18B, MYO7A, MYO9A, NAA10, NALCN, NBAS, NCAPD3, NDE1, NDP, NEB, NECTIN1, NEK1, NEK8, NEK9, NEPRO, NF1, NF2, NHEJ1, NIPBL, NKX2-5, NKX3-2, NME8, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPR2, NR2F2, NRAS, NRG1, NSD1, NSDHL, NSUN2, NT5E, OBSL1, OCRL, ODAD4, OFD1, OPHN1, ORC1, OTX2, P3H1, P4HB, PAFAH1B1, PAPSS2, PAX2, PAX3, PAX6, PAX7, PAX9, PCNT, PDCD10, PDE4D, PDE6D, PEX7, PGM1, PHEX, PHF6, PHF8, PHYH, PIBF1, PIEZO2, PIGA, PIGV, PIP5K1C, PITX2, PKD1L1, PKD2, PKHD1, PLEKHA7, PLK4, PLOD1, PLOD2, PLP1, PMM2, PNKP, PNPLA6, POLA1, POLR1C, POLR1D, POMC, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, PORCN, PPARG, PPIB, PPP1CB, PQBP1, PRDM5, PREPL, PRKAR1A, PRKG1, PRPH2, PRSS56, PTCH1, PTH1R, PTHLH, PTPN11, PXDN, PYCR1, PYCR2, QARS1, RAB18, RAB23, RAB3GAP1, RAB3GAP2, RAD21, RAF1, RAPSN, RARB, RARS2, RASA1, RASA2, RAX, RBBP8, RBP4, RBPJ, RELN, RET, RHO, RIN2, RIPK4, RIPPLY2, RIT1, ROBO3, ROR2, RPGRIP1L, RPL10, RPS6KA3, RSPH1, RSPH3, RSPH4A, RSPH9, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SASS6, SATB2, SBDS, SCN4A, SDCCAG8, SEC24D, SELENON, SEMA3E, SEPSECS, SERPINF1, SERPINH1, SF3B4, SH3PXD2B, SHH, SHOC2, SHROOM4, SIX3, SIX6, SKI, SLC18A3, SLC25A19, SLC26A2, SLC2A10, SLC34A3, SLC35D1, SLC38A8, SLC39A13, SLC5A7, SLC9A6, SMAD2, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMCHD1, SMOC1, SMS, SNAI2, SNAP29, SNTG1, SOS1, SOS2, SOX10, SOX11, SOX2, SOX9, SP7, SPAG1, SPARC, SPECC1L, SPRED1, SPRY4, STAMBP, STIL, STRA6, SUFU, SYT2, TAF6, TBC1D20, TBC1D23, TBC1D24, TBX1, TBX15, TBX2, TBX22, TBX3, TBX5, TBX6, TCF12, TCF4, TCOF1,

Gen med

Laboratorio de diagnóstico clínico y genómico

Dismorfología

TCTN1, TCTN2, TCTN3, TENM3, TENT5A, TFAP2A, TGDS, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TK2, TMCO1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMTC3, TNFRSF11B, TNNI2, TNNT1, TNNT3, TNXB, TOE1, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRIM32, TRIP11, TRMT10A, TRPS1, TRPV4, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TTC21B, TTC8, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TXNL4A, TYR, UBA1, VAMP1, VCAN, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, WDR11, WDR19, WDR35, WDR62, WNT1, WNT5A, WNT7A, XYLT1, YWHAE, ZBTB24, ZC4H2, ZEB2, ZFPM2, ZIC1, ZIC2, ZIC3, ZMYND10, ZNF335, ZNF423, ZNF469, ZNF699, ZSWIM6

Panel de tejido conectivo y trastornos relacionados

Nuestro panel de tejido conectivo y trastornos relacionados proporciona una evaluación profunda en un solo paso de varios genes para detectar diferentes trastornos con fenotipos similares, como el síndrome de Marfan, Loeys-Dietz, cutis laxa, Ehlers-Danlos, síndrome de Stickler y aneurisma aórtico torácico familiar y disección.

Incluye 76 genes: ABCC6, ACTA2, ADAMTS2, ADAMTSL2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GAT3, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, CREB3L1, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLCN, FLNA, GORAB, ITGB4, LAMA3, LAMB3, LAMC2, LOX, LRP2, LTBP3, MBTPS2, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, PYCR1, RIN2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, SP7, SPARC, TENT5A, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, VCAN, WNT1, ZNF469