Grupo argenomics

presenta el portfolio de

DIAGNÓSTICO NEUROGENÓMICO

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argenomics

Fundación Investigar

OPTIMIZACIÓN SECUENCIAL POR ETAPAS
de estudios genómicos

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DISCAPACIDAD INTELECTUAL *1

TRASTORNO DEL ESPECTRO AUTISTA CON CI < 70

Cariotipo alta resolución
FRAGIL X (especialmente varones)
Secuenciación MECP2 (especialmente mujeres)

Resultado negativo

aCGH (Microarrays)

Resultado negativo

Panel Intelectual Disabillity

Resultado negativo

Secuenciación Exoma en Tríos
(Búsqueda de mutaciones De Novo)

EPILEPSIAS *2

CONV. FEBRILES ATÍPICAS | INVOLUCIÓN INTELECTUAL | INVOLUCIÓN MOTORA

SCN1A - SECUENCIACIÓN + MLPA

Resultado negativo

MUJER

PCDH19

Resultado negativo

COMIENZO < 1 AÑO

Panel Encefalopatías Epilepticas

Resultado negativo

Exoma Probando | Exoma Trío

ATAXIAS *3

ESPORÁDICO
(Historia familiar negativa)

Friedreich (FxN)

Resultado negativo

EDAD INICIO > 50 AÑOS

SI → SCA6 Premutación FRAGIL X

NO

Resultado negativo

Panel Ataxias ampliado

Resultado negativo

EXOMA

HISTORIA FAMILIAR POSITIVA

RECESIVO
(1 Generación afectada)

Friedreich (FxN)

Resultado negativo

Panel Ataxias recesivas

Resultado negativo

EXOMA

DOMINANTE
(>1 Generación afectada)

NO

DEMENCIA → SCA17

INICIO >50 AÑOS → SCA6

RÁPIDA EVOLUCIÓN → Secuenciación PRNP

RETINOPATÍA → SCA7

Resultado negativo | Resultado negativo

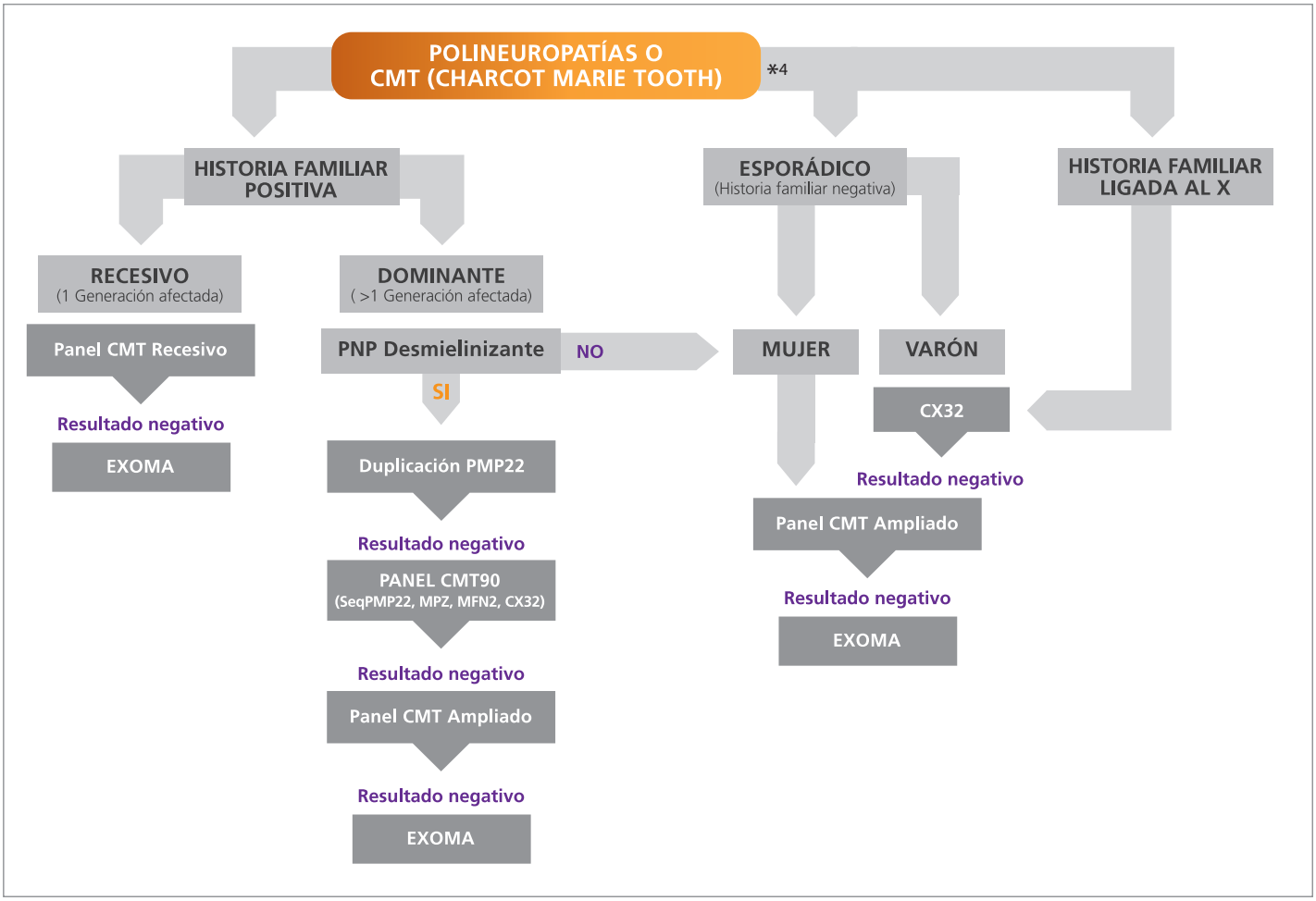
SCA1 SCA2 SCA3

Resultado negativo

Panel Ataxias dominante

Resultado negativo

EXOMA



ESTUDIOS MOLECULARES POR ENFERMEDAD

ATAXIAS *3

- PANEL DE ATAXIAS POR NGS

(BEAN1, ATN1, CACNA1A, NOP56, PPP2R2B, TBP, AFG3L2, DNMT1, FGF14, IFRD1, ITPR1, KCNC3, KCND3, PDYN, PRKCG, SPTBN2, TGM6, TTBK2, APTX, SETX, ATM, TTPA, STUB1, ANO10)

- Panel SCA FULL

(SCA1, SCA2, SCA3, SCA6, SCA7 y SCA17)

- Panel de Ataxias Recesivas

(AOA1 (APTX), AOA2 (SETX), ATM, AVED (TTPA), STUB1, ANO10)

- Marcadores individuales

SCA1 - SCA2 - SCA3 - SCA6 - SCA7 - SCA17 - Friedreich - POLG

EPILEPSIAS DE COMIENZO TEMPRANO *2

- PANEL DE EPILEPSIAS GENÉTICAS POR NGS

(ADSL, ALDH7A1, ALG13, ARHGEF9, ARX, ATP1A2, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DNAJC5, DYNC1H1, EFHC1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GATM, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, KANSL1, KCNH2, KCNJ10, KCNQ2, KCNQ3, KCTD7, LGI1, LIAS, MBD5, MECP2, MEF2C, MFSD8, NHLRC1, NRXN1, PCDH19, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, RBFOX1, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SLC25A22, SLC2A1, SLC6A8, SLC9A6, SPTAN1, STXBP1, SYN1, SYNGAP1, TBC1D24, TCF4, TPP1, TSC1, TSC2, UBE3A, WWOX, ZEB2)

- Marcadores individuales

SCN1A - PCDH19

PARAPARESIAS ESPÁSTICAS HEREDITARIAS

- PANEL DE HSP POR NGS

(ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ATL1, BSCL2, C12orf65, CYP7B1, ERLIN2, FA2H, GJC2, HSPD1, KDM5C, KIAA0196, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SACS, SLC16A2, SPAST, SPG11, SPG20, SPG21, SPG7, ZFYVE26, C19orf12, CCT5, SLC33A1)

LEUCODISTROFIAS / LEUCOENCEFALOPATÍAS

- PANEL DE LEUCODISTROFIAS POR NGS

(AARS2, ABCD1, ACOX1, ADAR, ADCK3, AIMP1, ALDH3A2, ATPAF2, ATP7A, ATP7B, ARSA, ASPA, BCAP31, BCS1L, BEST1, CLCN2, COL4A1, COQ2, COQ9, COX10, COX15, CSF1R, CYP7B1, CYP27A1, C10orf2, DARS2, DGUOK, D2HGDH, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ERCC2, ERCC3, ERCC6, ERCC8, ETFDH, FA2H, FAM126A, FUCA1, GALC, GBE1, GFAP, GFM1, GJA1, GJC2, GLA, GLB1, GTF2H5, HEPACAM, HEXA, HSD17B4, HSPD1, HTRA1, L2HGDH, LMNB1, MCOLN1, MLC1, MPLKIP, MRPS16, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFAF1, NDUFV1, NOTCH3, NPC1, NPC2, OCLN, OCRL, PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHGDH, PHYH, PLP1, POLG, POLG2, POLR3A, POLR3B, PPT1, PRF1, PSAP, PSAT1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RRM2B, SAMHD1, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHB, SLC16A2, SLC17A5, SLC25A4, SLC25A12, SOX10, SPAST, SPG7, SPG11, SPG20, SPG21, STX11, SUCLA2, SUMF1, SURF1, TACO1, TUFM, TREM2, TREX1, TYMP, TYROBP, UNC13D, ZFYVE26, STXBP2)

- Marcadores individuales

CADASIL - CADASIL FULL - Alexander (GFA)

MOVIMIENTOS ANORMALES

- PANEL DE ENFERMEDAD DE PARKINSON FAMILIAR

(SNCA, LRRK2, VPS35, PARK2, PINK1, PARK7, ATP13A2, PLA2G6, FBXO7, DNAJC6)

- PANEL DE NBIA

(ATP13A2, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45)

- PANEL DE DISTONIAS

(TOR1A, THAP1, GCH1, TH, SPR, SLC2A1, PRRT2, PNKD, TUBB4A, SGCE, ATP1A3, PRKRA)

- Sindromes individuales

PARKIN, SCNA, PINK, LRRK2 (mutación G20195), Enfermedad Prionica Familiar, DYT1, PANK2, Huntington

DISCAPACIDAD INTELECTUAL / TRASTORNO DEL ESPECTRO AUTISTA *1

- Estudios Individuales

Cariotipo de Alta Resolución – Frágil X – MECP2 – aCGH

MIOPATÍAS / MIASTENIA / ENF. MOTONEURONA

- PANEL DE MIASTENIAS CONGÉNITAS POR NGS
(AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DPAGT1, DOK7, GFPT1, MUSK, RAPSN, SCN4A)

- PANEL DE DISTROFIAS DE CINTURA POR NGS
(ANO5, CAPN3, CAV3, DES, DNAJB6, DYSF, FKRP, ISPD, LMNA, MYOT, PNPLA2, SGCA, SGCB, SGCD, SGCG, TCAP, TRIM32, TTN)

- PANEL DE MIOPATÍAS METABÓLICAS POR NGS
(ABHD5, ACADVL, AGL, CPT2, ENO3, ETFA, ETFB, ETFDH, GAA, GBE1, GYG1, GYS1, LDHA, LPIN1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PRKAG2, PYGM, SLC22A5, SLC25A20, TAZ)

- PANEL DE MIOPATÍAS CONGÉNITAS POR NGS
(ACTA1, BIN1, CFL2, CNTN1, DNM2, KBTBD13, KLHL40, MTM1, MYF6, MYH7, NEB, RYR1, SEPN1, TNNT1, TPM2)

- PANEL DE DISTROFIAS CONGÉNITAS POR NGS
(B3GNT1, CHKB, COL6A1, COL6A2, COL6A3, COL12A1, DAG1, DPM1, DPM3, FKTN, FKRP, GOSR2, GTDC2, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMT1, POMT2)

- PANEL DE MIOPATÍAS HEREDITARIAS POR NGS
(ABHD5, ACADVL, ACTA1, AGL, AGRN, ANO5, B3GNT1, BIN1, CAPN3, CAV3, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, DAG1, DES, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM3, DYSF, ENO3, ETFA, ETFB, ETFDH, FKRP, FKTN, GAA, GBE1, GFPT1, GOSR2, GTDC2, GYG1, GYS1, ISPD, ITGA7, KBTBD13, KLHL40, LAMA2, LARGE, LDHA, LMNA, LPIN1, MTM1, MUSK, MYF6, MYH7, MYOT, NEB, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, POMGNT1, POMT1, POMT2, PRKAG2, PYGM, RAPSN, RYR1, SCN4A, SEPN1, SGCA, SGCB, SGCD, SGCG, SLC22A5, SLC25A20, TAZ, TCAP, TNNT1, TPM2, TRIM32, TTN)

- Sindromes Individuales
Enfermedad de Kennedy – CPT2 (Exones HotSpot) – DMD – Paralisis Periódicas Hipokalemicas (Mutaciones frecuentes en CACNA15 y SCN4A) – Emerina (EMD) – Distrofia FascioEscapuloHumeral

TRASTORNOS MITOCONDRIALES

- PANEL NUCLEOMITO POR NGS
(C8orf38, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA7, NDUFA8, NDUFA9, NDUFB1, NDUFB3, NDUFB6, NDUFB9, NDUFC2, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, SDHA, SDHB, SDHC, SDHD, UQCRB, UQCRQ, COX4I1, COX4I2, COX6B1, COX7A1, COX7A2, ATP5E, AIFM1, ECSIT, FOXRED1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NUBPL, SDHAF1, SDHAF2, BCS1L, COX10, COX15, SCO1, SCO2, SURF1, ATPAF2, TMEM70, AARS2, C12orf65, DARS2, EARS2, EIF2AK3, FARS2, FOXG1, GARS, GFM1, HARS2, KARS, LARS2, LRPPRC, MARS2, MRPS16, MRPS22, MTO1, MTFMT, MTPAP, NARS2, NKX2-1, PDX1, PUS1, RARS2, SARS2, TACO1, TRMU, TSFM, TUFM, YARS2, ABCD1, DNAJC19, KCNJ11, MFSD8, NPC1, NPC2, SLC12A3, SLC16A2, SLC19A2, SLC22A5, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A38, SLC25A4, SLC2A1, SLC2A10, SLC35A1, SLC35C1, SLC3A1, STAR, TIMM8A, UCP1, UCP2, UCP3, AFG3L2, CHKB, DCX, DNM1L, DNM2, HSPD1, LETM1, MAPT, MFN2, MPV17, MUTYH, NEFL, OPA1, OPA3, REEP1, RNASEH2A, RNASEH2B, RNASEH2C, SPG20, SPG7, TOP1MT, WFS1, ARMS2, ATXN7, AUH, CAPN3, COG1, COG7, COG8, CRYAB, DMPK, GDAP1, HAX1, HFE, MECP2, MYH7, PARL, PHB, PNKD, RYR1, SAMHD1, SLC7A9, TMEM126A, TPM2, AASS, ABAT, ABCB7, ABHD5, ACAD8, ACAD9, ACADL, ACADM, ACADSB, ACADVL, ACAT1, ACSF3, ACSL4, ADCK3, AGXT, AK2, AKR1D1, ALAS2, ALDH18A1, ALDH4A1, ALDH5A1, ALDH6A1, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, APTX, ARG1, ASL, ASS1, ATL1, ATM, ATP7B, ATXN10, B4GALT1, BCKDHA, BCKDHB, BRAF, BTD, C10orf2, CAV3, CDKL5, CISD2, CLN3, CLN5, CLN6, CLN8, COQ2, COQ4, COQ5, COQ6, COQ9, CPOX, CPS1, CPT1A, CPT2, CTNS, CTSD, CYB5A, CYB5R3, CYBA, CYBB, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP27A1, CYP27B1, CYP7B1, D2HGDH, DBT, DECR1, DGUOK, DLAT, DLD, DMGDH, DOLK, DPAGT1, DPM1, DPM3, ELOVL4, ETFA, ETFB, ETFDH, ETHE1, FA2H, FASTKD2, FECH, FGF14, FH, FXN, GAA, GAD1, GAMT, GATM, GCDH, GCK, GCSH, GFER, GK, GLA, GLDC, GLRX5, GLUD1, GNPAT, GPD2, GPHN, HADH, HADHA, HADHB, HCCS, HK1, HLCS, HMGCL, HMGCS2, HSD17B10, HSD3B2, IDH2, ISCU, ITPR1, IVD, KCNC3, KIAA0196, KIAA0226, KIF1B, KIF5A, LMBRD1, MAOA, MCCC1, MCCC2, MGAT2, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MPI, MTHFD1, MTRR, MUT, NAGS, NIPA1, OAT, OTC, OXCT1, PAFAH1B1, PANK2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX13, PLP1, PMM2, PNPLA2, PNPLA3, POLG, POLG2, PPOX, PPT1, PREPL, PRKCG, PRODH, RFT1, RMRP, RRM2B, SCN1A, SGCD, SLC29A3, SLC33A1, SLC6A8, SOD1, SPAST, SPG11, SPTBN2, SUCLA2, SUCLG1, SUOX, TAT, TAZ, TK2, TPP1, TTBK2, TYMP, UBE3A, UNG, UROS, XDH, XPNPEP3, ZFYVE26)

- PANEL DE MANTENIMIENTO GENOMA MITOCONDRIAL POR NGS
(AGK, APTX, C10orf2, DGUOK, MFN2, MPV17, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TK2, TYMP)

- SECUENCIACIÓN COMPLETA DEL GENOMA MITOCONDRIAL POR NEXT GENERATION SEQUENCING

- Sindromes Mitocondriales
MELAS (mt3243A>G) en sangre – MELAS (mt3243A>G) en músculo – MERRF (mt8344A>G) en sangre – MERRF (mt8344A>G) en músculo – NARP/LEIGH (mt8993T>C) en sangre – NARP/LEIGH (mt8993T>C) en músculo – LHON (mt.11778A, 3460A, 14484C)

- Genes Individuales
PEO1 – POLG

POLINEUROPATÍAS HEREDITARIAS *4

- PANEL DE POLINEUROPATÍAS HEREDITARIAS POR NGS
(AARS, AIFM1, BSCL2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, HSPB1, HSPB8, LITAF, LMNA, LRSAM1, MED25, MFN2, MPZ, MTMR2, NDRG1, NEFL, PMP22, PRPS1, PRX, RAB7A, SBF2, SH3TC2, TRPV4, YARS)

- PANEL CMT 90% POR NGS
(PMP22, GJB1, MPZ, MFN2)

- Estudios Individuales
Duplicación PMP22

DEMENCIAS

- PANEL DEMENCIAS FULL
(C9orf72, CDH13, CHMP2B, FUS, GRN, MAPT, OPTN, PFN1, PSEN1, PSEN2, SOD1, SQSTM1, TARDBP, TREM2, UBQLN2, VAPB, VCP)

Demencia Alzheimer:

- PANEL ALZHEIMER POR NGS
(APOE, APP, PSEN1, PSEN2)

- Genes Individuales
APOE – APP – PSEN1 – PSEN2

Demencia Frontotemporal:

- PANEL FRONTOTEMPORAL POR NGS
(TAU, PGRN, VCP, TARDBP)

- Genes Individuales
TAU (MAPT) – PGRN – VCP – TARDBP

TRASTORNOS NEUROMETABÓLICOS

- PANEL ION INHERITED DISORDERS POR NGS
(ABCA4, ABCC9, ABCD1, ACADVL, ACTA2, ACTC1, ACTN2, ADA, AIPL1, AIRE, AKAP9, AKR1B1, ALPL, AMT, ANK2, APC, APP, APTX, ARL6, ARSA, ASL, ASPA, ATL1, ATM, ATP2A2, ATP7A, ATP7B, ATXN1, ATXN2, ATXN7, BAG3, BCKDHA, BCKDHB, BEST1, BMPR1A, BTD, BTK, CA4, CACNA1C, CACNB2, CALR3, CAPN3, CASQ2, CAV3, CCDC39, CCDC40, CDH23, CEP290, CERKL, CFTR, CHAT, CHD7, CHEK2, CHM, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNGB1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A5, COL5A1, COL5A2, COL7A1, COL9A1, CRB1, CRX, CTDP1, CTNS, CYP27A1, DBT, DCX, DES, DHCR7, DKC1, DLD, DMD, DNAH5, DNAH9, DNAH11, DNAI1, DNAI2, DNM2, DOK7, DSC2, DSG2, DSP, DYSF, ELN, EMD, ENG, EXT1, EYA1, EYS, F8, F9, FANCA, FANCC, FANCF, FANCG, FBN1, FBXO7, FGFR1, FGFR3, FMO3, FOXL2, FRG1, FRMD7, FSCN2, FXN, GAA, GALT, GATA4, GBA, GBE1, GCSH, GDF5, GJB2, GJB3, GJB6, GLA, GLDC, GNE, GNPTAB, GPC3, GPD1L, GPR143, GUCY2D, HBA2, HBB, HCN4, HEXA, HFE, HIBCH, HMBS, HR, IDS, IDUA, IKBKAP, IL2RG, IMPDH1, ITGB4, JAG1, JUP, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNQ1, KCNQ4, KIAA0196, KLHL7, KRAS, KRT5, KRT14, L1CAM, LAMB3, LAMP2, LDB3, LNA, LRAT, LRRK2, MAPRE2, MAPT, MC1R, MECP2, MED12, MEN1, MERTK, MFN2, MLH1, MMAA, MMAB, MMACHC, MPZ, MSH2, MTM1, MUT, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYO7A, MYOZ2, NF1, NF2, NIPBL, NKX2-5, NPC1, NPC2, NR2E3, NRAS, NSD1, NUDT19, OCA2, OCRL, OTC, PABPN1, PAFAH1B1, PAH, PAX3, PAX6, PCDH15, PEX1, PEX3, PEX5, PEX10, PEX13, PEX14, PEX19, PEX26, PINK1, PKD1, PKD2, PKHD1, PKP2, PLEC, PLN, PLOD1, PMM2, PMP22, POLG, PPT1, PRCD, PRKAG2, PROM1, PRPF8, PRPF31, PRPH2, PSEN1, PSEN2, PTCH1, PTPN11, RAF1, RAG1, RAG2, RAI1, RAPSN, RB1, RDH12, RET, RHO, ROR2, RP9, RPE65, RPGR, RPGRIP1, RPL11, RL35A, RPS6KA3, RPS7, RPS10, RPS19, RPS24, RPS26, RS1, RSPH4A, RSPH9, RYR1, RYR2, SALL4, SCN1B, SCN3B, SCN4B, SCN5A, SCN9A, SEMA4A, SERPINA1, SERPING1, SGCD, SH3BP2, SIX1, SIX5, SLC25A4, SLC25A13, SLC26A4, SMAD3, SMAD4, SNCA, SNRNP200, SNTA1, SOD1, SOS1, SOX9, SPATA7, SPG7, STARD3, TAF1, TAZ, TBX5, TCOF1, TGFBR1, TGFBR2, TMEM43, TNNC1, TNNI3, TNNT1, TNNT2, TNXB, TOPORS, TP53, TPM1, TSC1, TSC2, TTPA, TTR, TULP1, TWIST1, TXNDC3, TYR, USH1C, USH2A, VCL, VHL, WAS, WRN, WT1)

SECUENCIACIÓN DE EXOMA COMPLETO

Paneles multigénicos realizados a través de
NEXT GENERATION SEQUENCING

Para más información o solicitar un estudio genómico:

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