Omica Labs

PANEL DE CARDIOLOGÍA OMICALABS

Aorta

ABCC6[1], ABL1[2], ACTA2[3], ADAMTS10[4], ADAMTS17[5], ADAMTS2[6], ATP7A[7], B3GAT3[8], BGN[9], CBS[10], COL3A1[11], COL4A5[12], COL5A1[13], COL5A2[13], EFEMP2[14], ELN[15], ENPP1[15,16], FBN1[17], FBN2[18], FKBP14[18,19], FLNA[20], FOXE3[21], GATA5[22], HCN4[22,23], LOX[24], MAT2A[25], MED12[26], MFAP5[27], MYH11[28], MYLK[29], NOTCH1[30], PLOD1[31], PLOD3[32], PRKG1[33],ROBO4[34], SKI[33,35], SLC2A10[36], SMAD2, SMAD3[37], SMAD4[38], SMAD6[39], TGFB2[40], TGFB3[41], TGFBR1[42], TGFBR2[43],THSHD5[44], XYLT1[45],XYLT2[45]

Arritmia

ABCC9[46] ,AKAP9[47] ,ANK2[48] ,BAG3[49] ,CACNA1C[50] ,CACNB2[51] ,CALM1[52] ,CALM2 [52],CALM3[52] ,CASQ2[53] ,CAV3[54] ,CDH2 [55],CTNNA3[56] ,DBH [57],DES[58] ,DSC2[59] ,DSG2[60] ,DSP[61] ,FLNC [62],GATA4[63],GATA6[64] ,GNB5[65], HADHA[66] ,HCN4 [67],HERG[68,69],JAG1[70],JUP[71] ,KCNA5[72] ,KCNE1[68] ,KCNE2[68] ,KCNH2[69] ,KCNJ2[73] ,KCNJ5[74] ,KCNQ1[69],KVLQT1[68] ,LDB3[75] ,LEMD2[76] ,LMNA[77] ,MYH6[78] ,MYH7[79] ,MYL4 [80],NKX2-5[70] ,NOS1AP[81] ,NUP155[82] ,PKP2 [83],PLN[84] ,PPA2[85] ,PRKAG2[86] ,RBM20 [87] ,RYR2[88] ,SCN1B[88,89] ,SCN3B[90] ,SCN5A[68] ,TANGO2[91] ,TBX1[70],TBX5[92] ,TECRL[92,93] ,TGFB3[94] ,TMEM43 [95],TNNI3[96] ,TNNI3K[97] ,TNNT2[98] ,TRDN [99],TRPM4 [100],TTN[101], ZFPM2[102]

Fibrilación auricular

ABCC9[103],CACNB2,CASR[103],CAV1[103],CUX2[103],E169K[103], GATA6[64],GJA5[103], HCN4[103], KCNA5[103],KCND3[103], KCNE1[103],KCNE2[103],KCNE3[103],KCNE4[103],KCNE5[103],KCNH2[103], KCNJ2[103], KCNJ5[103], KCNJ8[103],KCNN3[103],KCNQ1[103], LDB3[75], LMNA[104], PRRX1[103],MYL4[105],NPPA[106],NUP155[103], RYR2[107], SCN10A[103], SCN1B[103], SCN2B[103],SCN3B[103], SCN4B[103],SCN5A[103], TBX5[92],ZFHX3[103].

Síndrome de Brugada

CACNA1C[108], CACNB2[109],GPD1L[110], HCN4[111], KCND3[112],KCNE3[113],SCN5A[114],SCN1B[115], SCN3B[116], TRPM4[100]

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Miocardiopatías

Genes seleccionados del 'Atlas of Cardiac Genetic Variation[117]', sólo se han incluido genes con un exceso significativo de mutaciones en pacientes con miocardiopatía.

Miocardiopatía hipertrófica

ACTC1[224], ACTN2[224],ANK2[225], CSRP3[224],CRYAB[225],DES[225], FLNC[225], FHL1[225], GLA,LAMP2[225], MYBPC3[224], MYH7[224],MYOZ2[225], MYL2[224], MYL3[224], MYOZ2[224],PLN[225], PRKAG2[225], PTPN11[225], TCAP[224], TNNC1[224], TNNI3[224], TNNT2[224], TPM1[224], TTN[224],TTR[225].

Miocardiopatía dilatada

ABCC9[188], ACTC1[188], ACTN2[188], ANKRD1[188], BAG3[188], CSRP3[188], CTF1[188], DES[188], DSP[188], DSG2[188], DTNA[188], EYA4[188], FLNC[188], GATAD1[188], ILK[188], JPH2[188], LAMA4[188], LDB3[188], LMNA[189], LRRC10[188], MIB1[188], MYBPC3[188], MYH6[189], MYH7[189], MYL2[188], MYL3[188], MYPN[189], NEBL[188], NEXN[188], NPPA[188], NKX2-5[188], OBSCN[188], PDLIM3[188], PLN[188], PLEKHM2[188], PKP2[188], PRDM16[188], PSEN1[188], PSEN2[188], RBM20[189], SCN5A[189], SGCD[188], TBX20[188], TCAP[188], TNNC1[188], TNNT2[189], TNNI3[188], TPM1[189], TTN[189], VCL[188]

Miocardiopatía arritmogénica del ventrículo derecho

DES, DSC2, DSG2, DSP, JUP, PKP2

Miocardiopatía no compactada del ventrículo izquierdo

ACTC1[226], ALPK3[226], DES[226], DMD[226], DMPK[226], DSP[226], DTNA[226], HCN4[226], LAMP2[226], LDB3[226], LMNA[226], MIB1[226], MYBPC3[226], MYH7[226], NKX2.5[226], NNT[226], NONO[226], OBSCN[226], PKP2[226], PLEKHM2[226], PLN[226], PRDM16[226], RBM20[226], RYR2[226], SCN5A[226], TAZ[226], TBX20[226], TBX5[226], TMEM70[226], TNNT2[226], TPM1[226], TTN[226].

Taquicardia ventricular catecolaminérgica polimórfica

ANK2[48], CALM1[118], CALM2[52], CALM3[119], CASQ2[53], KCNJ2[120], RYR2[107],

TECRL[93], TRDN[121].

PANEL DE CARDIOLOGÍA OMICALABS

Cardiopatía estructural congénita

ABL1[2], ACTA2[122], ACTC1[123], ACTG1[124], ACVR1[125], ACVR2B[125], ADAM17[125],ADAMTS10[4], ADAMTS17[5], AFF4[126],APC[125], ARHGAP31[127], B3GAT3[128], BCL9[125], BCOR[129], BMPR2[125], BRAF[125], CBL[130], CDK13[131], CDKN1C[132], CFAP53[133], CHD4[134], CHD7[135], CREBBP[125], CRELD1[136], CTC1[137],CITED2[125], DCHS1[125],DHCR7[138], DLL4[139], DOCK6[140], DVL1[125],EDN1[125],EFTUD2[141], EHMT1[142], ELN[15], ENG[143], EOGT[144], EP300[125],ETS1[125], EVC[145], EVC2[145], FKTN[146], FLNA[20], FLT4[147], FOXC1[125],FOXC2[125], FOXH1[125],FOXJ1[125],FOXO1[125], FOXP1[125], GATA4[125], GATA5[125], GATA6[125], GDF1[125], GPC3[148], HAND1[125], HAND2[125],HES1[125],HEY2[125], HNRNPK[149], HOXA1[150], HRAS[151], JAG1[125],JARID2[125], KDM6A[152], KMT2D[153], KYNU[154], LZTR1[155], MED12[26], MED13L[26], MEGF8[156], MEIS2[157], MMP21[158], MSX1[125],MYCN[159], MYRF[160], NEXN[161],NFATC1[125], NIPBL[162], NKX2-5[125], NKX2-6[163], NODAL[164], NONO[165], NOTCH1[125], NOTCH2[125],NR1D2[125], NR2F2[125], NSD1[166],PCDHA9[125], PKD1L1[167], PLD1[168], PPP1CB[169], PRDM6[170], PRKD1[171], PTPN11[125], PUF60[172], RAB23[173], RECQL4[174], RERE[175],RBPJ[125],RFX3[125], RIT1[176], SMAD6[125],SMARCB1[177], SMC1A[178], SMC3[178], SOS1[125], SOS2[155], STAG2[179], STRA6[180], TAB2[181], TBX1[125],TBX2[125], TBX20[125],TBX3[125], TBX5[125], TGDS[182],TGFB2[125],TGFB3[125],TGFBR1[125],TGFBR2[125], TLL1[183], TMEM94[184], TWIST1[185], VEGFA[125],ZEB2[186], ZFPM2[125], ZIC3[187].

Síndrome de Ehlers-Danlos

ADAMTS2[190], B3GALT6[190], B4GALT7[190], C1R[190], C1S[190], CHST14[190], COL12A1[190], COL1A1[190], COL1A2[190], COL3A1[190], COL5A1[190], DSE[190], FKBP14[190], PLOD1[190], PRDM5[190], SLC39A13[190], TNXB[190], ZNF469[190].

Telangiectasia hemorrágica hereditaria

ACVRL1[191], ENG[191], EPHB4[192], GDF2[193], RASA1[194], SMAD4[191]

Heterotaxia y Situs Inverus

ACVR2B[195], ANKS6[196],ARMC4[197], C21ORF59[198], CCDC103[199], CCDC114[200], CCDC39[201], CCDC40[202], CFAP53[133], CITED2[203], DNAAF1[204], DNAAF2[205], DNAAF3[206], DNAAF5[207], DNAH11[208], DNAH5[200], DNAI1[209], DNAI2[200], DNAL1[210],

DYX1C1[210,211], GDF1[212], INVS[213], LEFTY2[203], LRRC6[214], MMP21[158], NODAL[164], PIH1D3[215], PKD1L1[167], SPAG1[216], ZIC3[187], ZMYND10[214]

Dislipidemia

ABCA1[217], ABCG5[218], ABCG8[218], ALMS1[217,219], APOA1[220], APOA5[218], APOB[218], APOC2[218], APOC3[221], APOE[222], CREB3L3[223], CYP27A1[218], GPD1, GPIHBP1[218], LDLR[218], LDLRAP1[218], LIPA[218], LMF1[218], LPL[218], PCSK9[218]

Síndrome de Liddle

SCNN1A[227], SCNN1B[227], SCNN1G[227]

Síndrome QT largo

AKAP9[228], ANK2[228], CACNA1C[228], CALM1[228], CALM2[228], CAV3[228], KCNE1[228], KCNE2[228], KCNH2[228], KCNJ2[228], KCNJ5[228], KCNQ1[228], SCN4B[228], SCN5A[228], SNTA1[228].

Hipertensión pulmonar arterial

ACVRL1[229], AQPD1[229], ATP13A3[229], BMPR1B[229], BMPR2[229], CAV1[229], EIF2AK4[229], ENG[229], GDF2[229], KCNA5[229], KCNK3[229], KLF2[229], SMAD1[229], SMAD4[229], SMAD9[229], SOX17[229], TBX4[229].

Síndrome QT corto

CACNA1C[230], CACNB2[230], CACNA2D1[230],KCNH2[230], KCNQ1[230], KCNJ1 [230]

Anexo I- Bibliografía

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