
CarrierGen

Gen		Enfermedad	Variantes
ABCA4	7	Distrofia de conos y bastones 3	c.1222C>T, c.2616_2617delCT, c.3540_3555delGTCTAAGGGTTTCTCC, c.4793C>A, c.5461-10T>C, c.6179T>G, c.763C>T
ABCA4	52	Enfermedad de Stargardt 1	c.1018T>G, c.1225delA, c.1335C>G, c.161G>A, c.161G>T, c.1622T>C, c.1715G>A, c.1760+2T>G, c.1804C>T, c.1938-1G>A, c.194G>A, c.2160+1G>C, c.2300T>A, c.2565G>A, c.2588G>C, c.2690C>T, c.2798A>T, c.286A>G, c.2888delG, c.2971G>C, c.3106G>A, c.3113C>T, c.3210_3211dupGT, c.3322C>T, c.3364G>A, c.3970delG, c.4139C>T, c.4195G>A, c.4222T>C, c.4457C>T, c.4469G>A, c.4539+1G>T, c.4577C>T, c.4594G>A, c.5285C>A, c.52C>T, c.5338C>G, c.5714+5G>A, c.5819T>C, c.5881G>A, c.5912T>G, c.6079C>T, c.6089G>A, c.6148G>C, c.6320G>A, c.634C>T, c.6445C>T, c.6449G>A, c.661G>A, c.5882G>A, c.4852T>A, c.5908C>T
ABCA4	7	Retinosis pigmentaria 19	c.1630_1633dupGAAA, c.1848delA, c.2815G>T, c.2927delT, c.5391_5392delTG, c.2160+1G>T, c.3098delA
ABCD1	43	Adrenoleucodistrofia ligada al X	c.1165C>G, c.1201C>G, c.1202G>A, c.1252C>T, c.1288C>T, c.1390C>T, c.1396C>T, c.1415_1416delAG, c.1429G>T, c.1451C>G, c.1534G>T, c.1544C>T, c.1547T>C, c.1552C>T, c.1552delC, c.1553G>A, c.1586G>A, c.1661G>A, c.1679C>T, c.1771C>T, c.1781-1G>A, c.1802G>A, c.1817C>T, c.1825G>A, c.1849C>T, c.1850G>A, c.1866-10G>A, c.2006_2007delAC, c.253dupC, c.311G>A, c.346G>A, c.421G>A, c.443A>G, c.520T>G, c.521A>G, c.614C>A, c.686T>C, c.796G>A, c.838C>T, c.871G>A, c.874_876delGAG, c.886T>C, c.887A>G
ACAD9	10	Déficit de acil-CoA deshidrogenasa 9	c.1240C>T, c.1249C>T, c.1253A>G, c.1283C>T, c.130T>A, c.151-2A>G, c.1594C>T, c.359delT, c.797G>A, c.976G>C
ACADM	39	Déficit de acil-CoA deshidrogenasa de cadena media	c.1045C>T, c.1052C>T, c.1102_1105delTTAG, c.1124T>C, c.1189T>A, c.127G>A, c.157C>T, c.199T>C, c.216+2T>C, c.233T>C, c.244dupT, c.250C>T, c.343_348delGGATGT, c.347G>A, c.362C>T, c.387+1delG, c.395C>G, c.447G>A, c.449_452delCTGA, c.464T>C, c.577A>G, c.583G>A, c.600-18G>A, c.609A>C, c.616C>T, c.617G>T, c.683C>A, c.698T>C, c.730T>C, c.734C>T, c.742A>G, c.797A>G, c.799G>A, c.850-2A>G, c.881G>C, c.928G>A, c.985A>G, c.999_1011dupTAGAATGAGTTAC, c.926T>G
ACADS	25	Déficit de acil-CoA deshidrogenasa de cadena corta	c.1031A>G, c.1058C>T, c.1095G>T, c.1108A>G, c.1112G>T, c.1138C>T, c.1147C>T, c.1153G>T, c.136C>T, c.164C>T, c.268G>A, c.274G>T, c.310_312delGAG, c.314T>A, c.319C>T, c.323G>A, c.332C>T, c.409C>T, c.417G>C, c.529T>C, c.575C>T, c.682_683delGA, c.826G>A, c.973C>T, c.988C>T
ACADSB	6	Deficiencia de la 2-metilbutiril-CoA deshidrogenasa	c.1159G>A, c.1228G>A, c.303+3A>G, c.443C>T, c.621G>A, c.763C>T
ACADVL	54	Déficit de acil-CoA deshidrogenasa de cadena muy larga	c.1001T>G, c.1096C>T, c.1097G>A, c.1141_1143delGAG, c.1144A>C, c.1182+1G>A, c.1246G>A, c.1316dupG, c.1316G>A, c.1322G>A, c.1357C>T, c.1360G>A, c.1372T>C, c.1375C>T, c.1375dupC, c.1376G>A, c.1405C>T, c.1406G>A, c.1468G>C, c.1531C>T, c.1532G>A, c.1679-6G>A, c.1700G>A, c.1806_1807delCT, c.1837C>T, c.1843C>T, c.1844G>A, c.265C>T, c.272C>A, c.278-1G>A, c.298_299delCA, c.343delG, c.364A>G, c.37C>T, c.388_390delGAG, c.388_391delGAGA, c.433C>T, c.520G>A, c.535G>T, c.538G>A, c.553G>A, c.577G>C, c.637G>A, c.65C>A, c.664G>A, c.685C>T, c.739A>C, c.739A>G, c.753-2A>C, c.779C>T, c.829_831delGAG, c.856A>G, c.881G>A, c.896_898delAGA
ACOX1	4	Deficiencia Acyl-CoA oxidasa peroxisomal	c.442C>T, c.532G>T, c.832A>G, c.926A>G
ADA	33	Inmunodeficiencia severa combinada	c.1079-15T>A, c.219-2A>G, c.220G>T, c.221G>T, c.226C>T, c.248C>A, c.301C>T, c.302G>T, c.320T>C, c.385G>A, c.419G>A, c.43C>G, c.445C>T, c.446G>A, c.454C>A, c.466C>T, c.467G>A, c.529G>A, c.536C>A, c.58G>A, c.596A>C, c.631C>T, c.632G>A, c.643G>A, c.646G>A, c.698C>T, c.704G>A, c.821C>T, c.872C>T, c.890C>A, c.911T>G, c.956_960delAAGAG, c.986C>T
ADAMTS13	18	Trombocitopenia purpura trombotica congénita	c.1193G>A, c.1345C>T, c.1582A>G, c.1783_1784delTT, c.2074C>T, c.2851T>G, c.286C>G, c.2931_2936delTGCCCG, c.304C>T, c.3070T>G, c.331-2_331-1delAG, c.3638G>A, c.3770dupT, c.414+1G>A, c.4143dupA, c.587C>T, c.749C>T, c.803G>C
ADAMTS2	2	Síndrome de Ehlers-Danlos VII	c.2384G>A, c.673C>T

ADK	3	Hipermetioninemia por déficit de adenosina quinasa	c.38G>A, c.653A>C, c.902C>A
ADGRV1	1	Síndrome de Usher tipo 2C	c.5504_5507delTTCC
AGA	26	Aspartilglucosaminuria	c.101_107delGGCCCTT, c.127_127+1insATGCGG, c.179G>A, c.192T>A, c.200_201delAG, c.214T>C, c.299G>A, c.302C>T, c.336delT, c.346C>T, c.373_376delACAC, c.395-8A>G, c.404T>C, c.439T>C, c.44T>G, c.488G>C, c.503G>A, c.677G>A, c.754G>C, c.755G>A, c.770C>T, c.788delT, c.800dupT, c.904G>A, c.916T>C, c.940+1G>T
AGPS	4	Condrodisplasia puntata rizomélica	c.1256G>A, c.1406T>C, c.1703C>T, c.926C>T
AHI1	40	Síndrome de Joubert 3	c.1051C>T, c.1152-2A>G, c.1267C>T, c.1303C>T, c.1328T>A, c.1484G>A, c.1626+1G>A, c.1626+4_1626+5insTTAC, c.1765C>T, c.1897_1898dupGG, c.1917T>A, c.2012C>T, c.2036+1G>T, c.2156A>G, c.2168G>A, c.2172delA, c.2173T>C, c.2174G>A, c.2212C>T, c.2282C>T, c.2495delT, c.2598_2604delAGTGTAT, c.2687A>G, c.662C>G, c.736A>T, c.910dupA, c.985C>T, c.2187_2196delGAGAGAAGAT, c.1614delA, c.2098_2099dupGT, c.1260G>A, c.1976A>T, c.1115A>G, c.1516C>T, c.1052G>T, c.2297G>A, c.2023G>A, c.2361G>T, c.1997A>T, c.2705T>A
AIPL1	7	Amaurosis congénita de Leber 4	c.1010_1011delAG, c.1053_1064delTGCAGAGCCACC, c.589G>C, c.617T>A, c.715T>C, c.784G>A, c.834G>A
ALDOB	19	Intolerancia a la fructosa hereditaria	c.1005C>G, c.1013C>T, c.1027T>C, c.10C>T, c.113-1_115delGGTA, c.136A>T, c.178C>T, c.2T>C, c.324+1G>A, c.324G>A, c.360_363delCAAA, c.442T>C, c.448G>C, c.524C>A, c.548_553delTGGTAC, c.612T>A, c.625-2A>G, c.720C>A, c.865delC
ALG1	11	Trastorno congénito de la glicosilación Ik	c.1025A>C, c.1079C>T, c.1129A>G, c.1187+1G>A, c.1188-2A>G, c.15C>A, c.262T>G, c.434G>A, c.450C>A, c.773C>T, c.826C>T
ALG12	8	Trastorno congénito de la glicosilación Ig	c.1001delA, c.117delG, c.1242C>G, c.200C>T, c.301G>A, c.424T>G, c.437G>A, c.473T>C
ALG2	2	Trastorno congénito de la glicosilación Ii	c.1040delG, c.393G>T
ALG2	2	Síndrome miasténico congénito	c.203T>G, c.214_224delGGGGACTGGCTinsAGTCCCCG
ALG3	5	Trastorno congénito de la glicosilación Id	c.165C>T, c.211T>C, c.353G>A, c.470T>A, c.512G>A
ALG6	6	Trastorno congénito de la glicosilación Ic	c.1432T>C, c.257+5G>A, c.482A>G, c.53G>A, c.897_899delAAT, c.998C>T
ALG8	3	Trastorno congénito de la glicosilación Ih	c.139A>C, c.413delC, c.824G>A
AMACR	2	Déficit de Alfa-metilacil-CoA racemasa	c.154T>C, c.320T>C
AR	38	Insensibilidad a los andrógenos	c.1732G>A, c.1739G>T, c.1748T>A, c.1769-11T>A, c.1771A>T, c.178C>T, c.179dupA, c.180_181delGC, c.1826G>A, c.1937C>A, c.2033T>C, c.2069A>C, c.2123T>G, c.2137C>T, c.2157G>A, c.2222C>G, c.2281_2287delAGGATGCinsTTCGCCCCTGA, c.2291A>G, c.2323C>T, c.2324G>A, c.2343G>T, c.2362A>G, c.2391G>A, c.2395C>G, c.2423T>C, c.2521C>T, c.2522G>A, c.2567G>A, c.2571C>G, c.2596T>C, c.2599G>A, c.2599G>T, c.2610T>G, c.2633C>T, c.2650A>T, c.2667C>T, c.4G>A, c.521T>G
AR	5	Síndrome de insensibilidad parcial a los andrógenos	c.1174C>T , c.1789G>A, c.1823G>A, c.2231G>T, c.1645C>T
ARG1	11	Argininemia	c.263_266delAGAA, c.32T>C, c.365G>A, c.413G>T, c.466-2A>G, c.57+1G>A, c.61C>T, c.703G>C, c.844delC, c.869C>G, c.871C>T
ARL13B	6	Síndrome de Joubert 8	c.65T>G, c.461A>G, c.236G>A, c.246G>A, c.257A>G, c.598C>T
ARL6	11	Síndrome de Bardet-Biedl	c.266C>T, c.272T>C, c.281T>C, c.362G>A, c.364C>T, c.431C>T, c.4G>T, c.506G>C, c.509T>G, c.92C>G, c.92C>T
ARSA	61	Leucodistrofia metacromática	c.1010A>T, c.1108-2A>G, c.1114C>T, c.1115G>A, c.1136C>T, c.1150G>A, c.1174C>T, c.1175G>A, c.1210+1G>A, c.1223_1231delGTGATACCA, c.1229C>T, c.1232C>T, c.1279C>A, c.1283C>T, c.1408_1418delGCAGCTGTGAC, c.1462C>T, c.1489_1492dupCCCC, c.195delC, c.240dupC, c.257G>A, c.263G>A, c.292_293delTCinsCT, c.293C>T, c.302G>T, c.304delC, c.346C>T, c.34delG, c.370G>A, c.410T>C, c.413C>T, c.465+1G>A, c.467G>A, c.470C>G, c.506C>G, c.511G>A, c.542T>G, c.583delT, c.641C>T, c.677C>T, c.697C>A, c.736C>T, c.737G>A, c.739G>A, c.763G>A, c.769G>C, c.827C>T, c.854+1G>A, c.862A>C, c.868C>T, c.869G>A, c.883G>A,

			c.890C>A, c.899T>C, c.905G>T, c.931G>A, c.937C>T, c.938G>A, c.979+1G>A, c.979G>A, c.986C>T, c.991G>T
ARSB	25	Mucopolisacaridosis tipo VI	c.1143-1G>C, c.1143-8T>G, c.1161dupC, c.1178A>C, c.1214G>A, c.1366C>T, c.1450A>G, c.1562G>A, c.215T>A, c.238delG, c.284G>A, c.349T>C, c.389C>T, c.410G>T, c.427delG, c.571C>T, c.589C>T, c.629A>G, c.707T>C, c.743delC, c.753C>G, c.937C>G, c.944G>A, c.971G>T, c.979C>T
ARSE	10	Condrodisplasia puntata tipo 1	c.119T>G, c.1442C>T, c.1475G>A, c.1732C>T, c.1743G>A, c.332G>C, c.349G>A, c.410G>C, c.410G>T, c.733G>C
ARX	11	Lisencefalia ligada al X con anomalías genitales	c.1001_1009delCGTTCACCA, c.1028T>A, c.1058C>G, c.1105G>T, c.1117C>T, c.1372delG, c.232G>T, c.617delG, c.980_983delAACA, c.995G>A, c.998C>A
ARX	5	Encefalopatía epiléptica infantil temprana	c.1471dupC, c.1604dupT, c.34G>T, c.81C>G, c.1465delG
ARX	2	Deficiencia intelectual no sindrómica ligada al X	c.856G>A, c.98T>C
ASL	38	Acidemia argininosuccinica	c.1045_1057delGTCATCTCTACGC, c.1060C>T, c.1079T>C, c.1122dupC, c.1135C>T, c.1153C>T, c.1193C>A, c.1255_1256delCT, c.1297A>C, c.1360C>T, c.1366C>T, c.175G>A, c.257A>C, c.260A>G, c.280C>T, c.283C>T, c.292delG, c.337C>T, c.346C>T, c.35G>A, c.436C>T, c.446+1G>A, c.461T>C, c.524+2T>G, c.532G>A, c.539T>G, c.544C>T, c.545G>A, c.557G>A, c.575_580dupAGCGGA, c.578G>A, c.602+1G>A, c.649C>T, c.718+5G>A, c.762C>A, c.857A>G, c.889C>T, c.925G>A
ASPA	24	Enfermedad de Canavan	c.212G>A, c.237-2A>T, c.244_245delAT, c.2T>C, c.327T>G, c.32delT, c.433-2A>G, c.454T>C, c.541C>A, c.584T>G, c.654C>A, c.692A>G, c.693C>A, c.71A>G, c.746A>T, c.79G>A, c.820G>A, c.827_828delGT, c.838C>T, c.854A>C, c.859G>A, c.863A>G, c.884T>C, c.914C>A
ATM	126	Ataxia Telangiectasia	c.1027_1030delGAAA, c.103C>T, c.1110C>G, c.1235G>C, c.1240C>T, c.1339C>T, c.1369C>T, c.138_141delTTCA, c.1402_1403delAA, c.1463G>A, c.1524delT, c.1564_1565delGA, c.1607+1G>T, c.170G>A, c.1898+2T>G, c.2113delT, c.2250G>A, c.2251-10T>G, c.2284_2285delCT, c.2376+1G>A, c.2413C>T, c.2502dupA, c.2638+2T>G, c.2662G>T, c.2720_2723delGTGT, c.2921+1G>A, c.3002T>A, c.3085dupA, c.3154-2A>G, c.320G>A, c.3245_3247delATCinsTGAT, c.3372C>G, c.3576G>A, c.3626_3627delTT, c.3663G>A, c.3673C>T, c.3712_3716delTTATT, c.3747-1G>C, c.3802delG, c.3836G>A, c.3894dupT, c.3931C>T, c.3993+1G>A, c.3994-2A>G, c.3G>A, c.4052delT, c.4143dupT, c.4198A>T, c.4396C>T, c.4776+2T>C, c.496+5G>A, c.5188C>T, c.5290delC, c.5320-5_5320-2delTCTA, c.538C>T, c.5515C>T, c.5549delT, c.5623C>T, c.5644C>T, c.5712dupA, c.5763-1050A>G, c.5890A>T, c.5908C>T, c.5932G>T, c.6095G>A, c.6100C>T, c.6154G>A, c.6200C>A, c.6228delT, c.6404_6405insTT, c.640delT, c.6415_6416delGA, c.6572+1G>A, c.6679C>T, c.6916_6917delAG, c.6997dupA, c.7010_7011delGT, c.7096G>T, c.7181C>T, c.7271T>G, c.7279_7284delCTTAGG, c.7327C>T, c.742C>T, c.7449G>A, c.7456C>T, c.748C>T, c.7517_7520delGAGA, c.7630-2A>C, c.7638_7646delTAGAATTTC, c.7705_7706delGA, c.7788G>A, c.7789-3T>G, c.7875_7876delTGinsGC, c.7886_7890delTATTA, c.790delT, c.7913G>A, c.7926A>C, c.7927+5delG, c.7967T>C, c.7985T>A, c.7989_7991delTGT, c.802C>T, c.8030A>G, c.8147T>C, c.824delT, c.8264_8268delATAAG, c.8266A>T, c.8293G>A, c.8307G>A, c.8395_8404delTTTCAGTGCC, c.8418+5_8418+8delGTGA, c.8432delA, c.8480T>G, c.8494C>T, c.8578_8580delTCT, c.8711A>G, c.8876_8879delACTG, c.8977C>T, c.8988-1G>C, c.901+1G>A, c.9019G>T, c.9021dupA, c.9022C>T, c.9079dupA, c.9139C>T, c.8873_8874delTT
ATP7B	101	Enfermedad de Wilson	c.1145_1151delCCCAACT, c.122A>G, c.1285+2T>A, c.1285+5G>T, c.1340_1343delAAAC, c.1512dupT, c.1708-1G>C, c.1745_1746delTA, c.1772G>A, c.1782delT, c.1846C>T, c.1877G>C, c.19_20delCA, c.1922T>C, c.1924G>C, c.1934T>G, c.1969A>C, c.2009_2015delATATGCT, c.2035delC, c.2071G>A, c.2122-8T>G, c.2123T>C, c.2128G>A c.2165dupT, c.2267C>T, c.2293G>A, c.2297C>G, c.2304delC, c.2332C>G, c.2333G>T, c.2336G>A, c.2337G>A, c.2356-2A>G, c.2383C>T, c.2513delA, c.2519C>T, c.2532delA, c.254G>T, c.2575+1G>C, c.2605G>A, c.2621C>T, c.2668G>A, c.2731-2A>G, c.2755C>G, c.2762G>A,

			c.2795C>A, c.2804C>T, c.2807T>A, c.2827G>A, c.2828G>A, c.2865+1G>A, c.2906G>A, c.2972C>T, c.2975C>T, c.3007G>A, c.3008C>T, c.3011A>C, c.3053C>T, c.3101A>C, c.314C>A, c.3191A>C, c.3207C>A, c.3244-2A>G, c.3263T>A, c.3301G>A, c.331C>T, c.3359T>A, c.3402delC, c.3443T>C, c.3451C>T, c.3517G>A, c.3552dupT, c.3556+1G>A, c.3556G>A, c.3598C>T, c.3646G>A, c.3649_3654delGTTCTG, c.3659C>T, c.3683G>C, c.3688A>G, c.3694A>C, c.3700G>T, c.3796G>A, c.3809A>G, c.3817C>T, c.3818C>T, c.3895C>T, c.3955C>T, c.4021G>A, c.4039G>A, c.4051C>T, c.4058G>A, c.4088C>T, c.524_525delAA, c.562C>T, c.778dupC, c.813C>A, c.845delT, c.865C>T, c.915T>A, c.2009_2012dupATAT
AUH	8	Aciduria 3-metilglutacónica tipo 1	c.263-2A>G, c.559G>A, c.589C>T, c.650G>A, c.80delG, c.895-1G>A, c.943-2A>G, c.991A>T
B4GALT1	1	Trastorno congénito de la glicosilación tipo 2d	c.1031dupC
BCKDHA	22	Enfermedad de jarabe de arce clásica 1a	c.1036C>T, c.1037G>A, c.117delC, c.117dupC, c.1198A>T, c.1234G>A, c.1310_1311delAC, c.1312T>A, c.288+1G>A, c.470A>C, c.476G>A, c.632C>T, c.659C>T, c.761C>A, c.844G>C, c.853G>C, c.861_868delAGGCCCC, c.868G>A, c.905A>C, c.929C>G, c.940C>T, c.979G>A
BCKDHB	28	Enfermedad de jarabe de arce tipo 1b	c.1016C>T, c.1022T>A, c.1046G>A, c.1065delT, c.1114G>T, c.197-2A>G, c.293T>G, c.356T>G, c.3G>A, c.401T>A, c.487G>T, c.508C>T, c.509G>A, c.526A>T, c.547C>T, c.548G>C, c.554C>T, c.633+1G>T, c.641T>A, c.752T>C, c.799C>T, c.832G>A, c.853C>T, c.93_103delGGCGCGGGGCT, c.93_103dupGGCGCGGGGCT, c.964A>G, c.970C>T, c.633+1G>A
BCS1L	11	Deficiencia aislada de CoQ-citocromo C reductasa	c.103G>C, c.1057G>A, c.133C>T, c.148A>G, c.166C>T, c.296C>T, c.320+1G>T, c.464G>C, c.547C>T, c.550C>T, c.830G>A
BCS1L	1	Síndrome GRACILE	c.232A>G
BCS1L	2	Sindrome de Bjornstad	c.548G>A, c.871C>T
CAPN3	29	Distrofia muscular de cinturas autosómica recesiva tipo 2A	c.1080G>C, c.1322delG, c.133G>A, c.1466G>A, c.1468C>T, c.1469G>A, c.1525G>T, c.1610A>G, c.1715G>A, c.1743_1744delTG, c.1795dupA, c.1838delA, c.2120A>G, c.2212C>T, c.2243G>A, c.2257G>A, c.2306G>A, c.2362_2363delAGinsTCATCT, c.257C>T, c.328C>T, c.550delA, c.598_612delTTCTGGAGTGCTCTG, c.662G>T, c.676G>A, c.956C>T, c.2329A>G, c.1699G>A, c.310G>T, c.2381-1G>A
CASQ2	8	Taquicardia ventricular polimórfica catecolinérgica 2	c.500T>A, c.546delT, c.567C>G, c.62delA, c.919G>C, c.923C>T, c.97C>T, c.539A>G
CBS	21	Homocistinuria clásica	c.233C>G, c.253G>A, c.302T>C, c.306G>C, c.325T>C, c.341C>T, c.346G>A, c.361C>T, c.362G>A, c.374G>A, c.384G>C, c.393G>C, c.415G>A, c.430G>A, c.434C>T, c.442G>A, c.572C>T, c.676G>A, c.689delT, c.700G>A, c.715G>A.
CD40LG	14	Inmunodeficiencia con hiper-IgM tipo 1	c.107T>G, c.31C>T, c.368C>A, c.384T>A, c.386A>G, c.418T>G, c.419G>A, c.464T>C, c.506A>G, c.632C>A, c.680G>T, c.703G>C, c.719_720delAT, c.761C>T
CDH23	26	Síndrome de Usher tipo 1D	c.172C>T, c.193delC, c.288+1G>C, c.3367C>T, c.3481C>T, c.3617C>G, c.3713_3714delCT, c.3842_3844delTGA, c.3880C>T, c.4069C>T, c.4309C>T, c.4488G>C, c.4504C>T, c.5237G>A, c.5712G>A, c.5985C>A, c.6049G>A, c.6050-9G>A, c.6307G>T, c.6968delC, c.7362+5G>A, c.7549A>G, c.7660G>T, c.8497C>G, c.9127C>T, c.9524G>A
CDH23	8	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB212	c.3178C>T, c.3628C>T, c.4021G>A, c.5663T>C, c.6442G>A, c.7823G>A, c.9565C>T, c.6614C>T
CDHR1	3	Distrofia de conos y bastones 15	c.1485+2T>G, c.524dupA, c.270dupC
CDHR1	6	Retinitis pigmentosa 65	c.1463delG, c.1485+2T>C, c.1536T>A, c.338delG, c.476C>A, c.709delG
CEP290	24	Síndrome de Joubert con defecto óculo-renal 5	c.164_167delCTCA, c.1666delA, c.21G>T, c.3175dupA, c.3176delT, c.4393C>T, c.4656delA, c.4966_4967delGA, c.5182G>T, c.5611_5614delCAAA, c.5668G>T, c.5932C>T, c.6072C>A, c.6277delG, c.7341dupA, c.2112delA, c.4384delG, c.3181_3182delAT, c.654T>G, c.5704G>T, c.4522C>T, c.6939C>A, c.1623+1G>A, c.103-1G>T
CEP290	4	Síndrome de Meckel 4	c.1451delA, c.1984C>T, c.3777_3778delAG, c.613C>T
CEP290	11	Amaurosis congénita de leber 10	c.1219_1220delAT, c.1666dupA, c.1860_1863delAAGA, c.2991+1655A>G, c.3904C>T, c.4452_4455delAGAA, c.4705-1G>T, c.4723A>T, c.4882C>T, c.5344C>T, c.6604delA

CERKL	8	Retinosis pigmentaria 26	c.1090C>T, c.238+1G>A, c.420delT, c.598A>T, c.847C>T, c.858delT, c.890T>C, c.1045_1046delAT
CFH	13	Inmunodeficiencia con anomalía de factor H	c.1222C>T, c.1606T>C, c.2876G>A, c.3234G>T, c.3590T>C, c.3628C>T, c.565G>T, c.1291T>A, c.3503T>A, c.3572C>T, c.380G>T, c.481G>T, c.671_673delAGA
CFTR	393	Fibrosis quística	c.1000C>T, c.1006_1007insG, c.1007T>A, c.1013C>T, c.1021_1022dupTC, c.1021T>C, c.1029delC, c.1040G>A, c.1040G>C, c.1046C>T, c.1055G>A, c.1081delT, c.1083delG, c.1093_1094delCT, c.1116+1G>A, c.1117-1G>A, c.1117G>A, c.1130dupA, c.1155_1156dupTA, c.115C>T, c.1175T>G, c.11C>A, c.1202G>A, c.1203G>A, c.1209+1G>A, c.1240C>T, c.1301_1307delCACTTCT, c.1327_1330dupGATA, c.1340delA, c.1364C>A, c.1365_1366delGG, c.1367T>C, c.1373G>T, c.137C>A, c.1393-1G>A, c.1393-2A>G, c.1397C>A, c.1397C>G, c.1399C>T, c.1400T>C, c.1418delG, c.1438G>T, c.1466C>A, c.1475C>T, c.1477_1478delCA, c.1477C>T, c.14C>T, c.1518C>G, c.1519_1521delATC, c.1521_1523delCTT, c.1545_1546delTA, c.1558G>T, c.1572C>A, c.1573C>T, c.1585-1G>A, c.1585-8G>A, c.1601C>A, c.1624G>T, c.164+12T>C, c.164+1G>A, c.1645A>C, c.1646G>A, c.1646G>T, c.1647T>G, c.1648G>T, c.1650delA, c.1651G>A, c.165-1G>A, c.1652G>A, c.165-3C>T, c.1654C>T, c.1657C>T, c.1658G>A, c.1673T>C, c.1675G>A, c.1679+1G>C, c.1679G>A, c.1679G>C, c.1680-1G>A, c.1680-886A>G, c.1682C>A, c.1684G>C, c.1687T>A, c.1705T>G, c.1706A>G, c.170G>A, c.171G>A, c.1721C>A, c.1753G>T, c.1766+1G>A, c.1766+1G>C, c.1766+3A>G, c.1766+5G>T, c.178G>T, c.1792_1798delAAAACTA, c.1853T>C, c.1865G>A, c.1911delG, c.1923_1931delCTCAAAACTinsA, c.1943A>T, c.1973_1985delGAAATTCAATCCTinsAGAA, c.1973_1985delGAAATTCAATCCTinsAGAAA, c.1986_1989delAACT, c.19G>T, c.1A>G, c.200C>T, c.2012delT, c.2017G>T, c.2036G>A, c.2051_2052delAAinsG, c.2052delA, c.2052dupA, c.2053C>T, c.2053dupC, c.2125C>T, c.2128A>T, c.2146A>T, c.2175dupA, c.2195T>G, c.220C>T, c.2215delG, c.2249C>T, c.2290C>T, c.2291delG, c.2335C>T, c.233dupT, c.2353C>T, c.2374C>T, c.2423_2424dupAT, c.2443G>T, c.2453delT, c.2463_2464delTG, c.2464G>T c.2479G>T, c.2490+1G>A, c.2491G>T, c.2506G>T, c.2537G>A, c.2538G>A, c.2547C>A, c.254G>A, c.2551C>T, c.2583delT, c.2589_2599delAATTTGGTGCT, c.262_263delTT, c.263T>G, c.2657+5G>A, c.2658-1G>C, c.2668C>T, c.2700T>A, c.271G>A, c.273+1G>A, c.273+3A>C, c.273+4A>G, c.2735C>A, c.2737_2738insG, c.2738A>G, c.2739T>A, c.274-1G>A, c.274G>A, c.274G>T, c.2780T>C, c.2810dupT, c.2825delT, c.2834C>T, c.2845C>T, c.2875delG, c.2896delA, c.2908G>C, c.2909G>A, c.292C>T, c.2930C>T, c.2939T>A, c.293A>G, c.2988+1G>A, c.2988G>A, c.2989-1G>A, c.2989-2A>G, c.2T>C, c.3002_3003delTG, c.3022delG, c.3038C>A, c.3039delC, c.3067_3072delATAGTG, c.310delA, c.3139_3139+1delGG, c.313delA, c.3140-26A>G, c.3154T>G, c.3160C>G, c.3161delA, c.3181G>C, c.3184_3188dupCTATG, c.3194T>C, c.3196C>T, c.3197G>A, c.3199G>A, c.3205G>A, c.3209G>A, c.3212A>C, c.3222T>A, c.3230T>C, c.325_327delTATinsG, c.3254A>G, c.3266G>A, c.326A>G, c.3276C>A, c.3276C>G, c.328G>C, c.3294G>A, c.3302T>A, c.3304A>T, c.3310G>A, c.3368-2A>G, c.3380G>A, c.3409A>G, c.3468G>A, c.3469-20T>C, c.3472C>T, c.3484C>T, c.3492dupT, c.349C>T, c.350G>A, c.350G>T, c.3528delC, c.3532_3535dupTCAA, c.3536_3539delCCAA, c.355A>G, c.3587C>G, c.358G>A, c.3605delA, c.3611G>A, c.3612G>A, c.3659delC, c.366T>A, c.3691delT, c.3700A>G, c.3712C>T, c.3717+4A>G, c.3717+5G>A, c.3718-1G>A, c.3718-2477C>T, c.3718-3T>G, c.3731G>A, c.3744delA, c.3746G>A, c.3747delG, c.3752G>A, c.3761T>A, c.3763T>C, c.3764C>A, c.3767dupC, c.3773dupT, c.3846G>A, c.3848G>T, c.3857T>C, c.3873+1G>A, c.3873G>C, c.3883_3886delATTT, c.3883delA, c.3889dupT, c.3907A>C, c.3908delA, c.3908dupA, c.3909C>G, c.3937C>T, c.3947G>A, c.3963+1G>A, c.4003C>T, c.4046G>A, c.4056G>C, c.4077_4080delTGTTinsAA, c.4086dupT, c.409delC, c.4111G>T, c.4144C>T, c.416A>T, c.4197_4198delCT, c.4234C>T, c.4242+1G>A, c.424delA, c.4251delA, c.429delT, c.4300_4301dupAG, c.4364C>G, c.4426C>T, c.442delA,

			c.446G>T, c.455T>G, c.489+1G>T, c.496A>G, c.50delT, c.50dupT, c.53+1G>T, c.531delT, c.532G>A, c.535C>A, c.543_546delTAGT, c.571T>G, c.577G>T, c.579+1G>T, c.579+3A>C, c.579+5G>A, c.580-1G>T, c.595C>T, c.613C>T, c.617T>G, c.650A>G, c.653T>A, c.658C>T, c.680T>G, c.743+1G>C, c.744-14_744-3delTGATTGATTTAC, c.79G>T, c.803delA, c.805_806delAT, c.825C>G, c.828C>A, c.830G>A, c.850dupA, c.860dupA, c.861_865delCTTAA, c.868C>T, c.935_937delTCT, c.948delT, c.987delA, c.988G>T, c.4C>T, c.57G>A, c.88C>T,c.164+1G>T, c.164+2T>C, c.164+4dupT, c.174_177delTAGA, c.223C>T, c.296C>T, c.330C>A, c.489+3A>G, c.494T>C, c.579+3A>G, c.601G>A, c.647G>A, c.1001G>A, c.1487G>A, c.1679+1G>A, c.1680A>C, c.1703delT, c.1766+1G>T, c.2601dupA, c.2645G>A, c.2900T>C, c.3039dupC, c.3124C>T, c.3208C>T, c.3293G>A, c.3435G>A, c.3468+5G>A, c.3476C>T, c.3717G>A, c.3717+40A>G, c.3873+2T>C, c.3988C>T, c.4127_4131delTGGAT, c.4242+1G>T, c.148T>C, c.1075_1079delCAAACinsAAAAA, c.1670delC, c.1687T>G, c.1736A>G, c.1763A>T, c.1826A>G, c.1841A>G, c.1943delA, c.2158C>T, c.3041A>G, c.2936A>T, c.3158C>T, c.3297C>A, c.3872A>G, c.3883_3884insG, c.4004T>C, c.4036_4042delCTAAGCC, c.933C>G, c.175dupA, c.470_483delTTAGTTTGATTTAT, c.717delG, c.720_741del22, c.1680-877G>T, c.1692delA, c.2143C>T, c.2859_2890del32, c.3217dupT, c.3808G>A
CFTR	18	Ausencia congénita bilateral de los vasos deferentes	c.1327G>T, c.166G>A, c.1727G>C, c.2657+2_2657+3insA, c.2763_2764dupAG, c.2813T>G, c.2991G>C, c.3095A>G, c.3454G>C, c.4147dupA, c.4231C>T, 5T, c.349C>G, c.509G>A, c.794T>G, c.1538A>G, c.1001G>T, c.958T>G, c.3458T>A
CHST6	9	Distrofia macular corneal	c.277C>A, c.304T>G, c.329A>G, c.392C>T, c.521A>G, c.599T>G, c.609C>A, c.820G>A, c.827T>C
CLCN1	37	Miotonía congénita autosómica recesiva	c.1013G>A, c.1238T>G, c.1283T>C, c.1412C>T, c.1438C>A, c.1439C>T, c.1453A>G, c.1488G>T, c.1495G>A, c.1592C>T, c.1649C>T, c.1655A>G, c.1667T>A, c.180+3A>T, c.2330delG, c.2680C>T, c.2795C>T, c.2926C>T, c.382A>G, c.394A>T, c.409T>G, c.501C>G, c.566C>T, c.568_569delGGinsTC, c.577G>A, c.592C>G, c.689G>A, c.803C>T, c.847C>T, c.857T>C, c.870C>G, c.871G>A, c.920T>C, c.929C>T, c.937G>A, c.950G>A, c.774+1G>A
CLDN14	6	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB29	c.167G>A, c.242G>A, c.254T>A, c.301G>A, c.398delT, c.694G>A
CLN3	46	Lipofuscinosis ceroide neuronal 3	c.1000C>T, c.1001G>A, c.1054C>T, c.1056+3A>C, c.1056G>C, c.105G>A, c.1195G>T, c.1198-1G>T, c.1247A>G, c.125+5G>A, c.126-1G>A, c.1272delG, c.1A>C, c.222+5G>C, c.233dupG, c.265C>T, c.302T>C, c.370dupT, c.374G>A, c.378_379dupCC, c.400T>C, c.424delG, c.461-13G>C, c.461-1G>C, c.472G>C, c.482C>G, c.485C>G, c.494G>A, c.49G>T, c.509T>C, c.533+1G>A, c.558_559delAG, c.560G>C, c.565G>C, c.575G>A, c.586dupG, c.597C>A, c.622dupT, c.631C>T, c.883G>A, c.883G>T, c.906+5G>A, c.944dupA, c.963-1G>T, c.979C>T, c.988G>T
CLN5	33	Lipofuscinosis ceroide neuronal 5	c.1026C>A, c.1054G>T, c.1071_1072delCT, c.1072_1073delTT, c.1083delT, c.1103_1106delAACA, c.1121A>G, c.1137G>T, c.1175_1176delAT, c.225G>A, c.291dupC, c.335G>A, c.335G>C, c.377G>A, c.433C>T, c.524T>G, c.526dupA, c.565C>T, c.575A>G, c.593T>C, c.595C>T, c.613C>T, c.619T>C, c.620G>C, c.669dupC, c.671G>A, c.672delG, c.772T>G, c.835G>A, c.919delA, c.924_925delAT, c.935G>A, c.955_970delGGAAATGAAACATCTG
CLN6	15	Lipofuscinosis ceroide neuronal 6	c.139C>T, c.17G>C, c.200T>C, c.214G>T, c.268_271dupAACG, c.307C>T, c.308G>A, c.316dupC, c.368G>A, c.395_396delCT, c.461_463delTCA, c.542+5G>T, c.663C>G, c.794_796delCCT, c.7delG
CLN8	24	Lipofuscinosis neuronal 8	c.181_183delAAG, c.209G>A, c.227A>G, c.320T>G, c.415C>T, c.46C>A, c.470A>G, c.473A>G, c.509C>T, c.562_563delCT, c.581A>G, c.610C>T, c.611G>T, c.637_639delTGG, c.661G>A, c.66delG, c.709G>A, c.70C>G, c.766C>G, c.789G>C, c.792C>G, c.88delG, c.88G>C, c.499G>T
CLRN1	11	Síndrome de Usher tipo 3	c.118T>G, c.144T>G, c.149_152delCAGGinsTGTCCAAT, c.189C>A, c.301_305delGTCAT, c.359T>A, c.368C>A, c.449T>C,

			c.459_461delATT, c.502dupA, c.528T>G
CLRN1	1	Retinitis pigmentosa 61	c.92C>T
CNGA1	6	Retinosis pigmentaria 49	c.1271G>A, c.1540C>T, c.191delG, c.1972delA, c.238G>T, c.959C>T.
CNGB1	6	Retinosis pigmentaria 45	c.1589C>G, c.2544dupG, c.2957A>T, c.3150delG, c.3462+1G>A, c.413-1G>A
CNGB3	16	Acromatopsia 3	c.1006G>T, c.1063C>T, c.1119G>A, c.112C>T, c.1148delC, c.1304C>T, c.1578+1G>A, c.1672G>T, c.391C>T, c.446_447insT, c.607C>T, c.644-1G>C, c.646C>T, c.819_826delCAGACTCC, c.886_896delACTTCTACAAAinsT, c.991-3T>G
CNGB3	1	Degeneración macular juvenil	c.1405T>G
COG7	1	Trastorno congénito de la glicosilación tipo IIe	c.323dupT
COG8	1	Trastorno congénito de la glicosilación tipo IIh	c.1611C>G
COL1A2	7	Síndrome de Ehlers-Danlos tipo cardiaco valvular	c.1404+1G>A, c.293dupC, c.3105+2T>C, c.3601G>T, c.577G>C, c.2512G>A, c.838G>T
COL4A3	6	Síndrome de Alport autosómico recesivo	c.345delG, c.4420_4424delCTTTT, c.4421T>C, c.4441C>T, c.4571C>G, c.898G>A
COL4A4	7	Síndrome de Alport autosómico recesivo	c.2420delG, c.3601G>A, c.3713C>A, c.4129C>T, c.4715C>T, c.4923C>A, c.71+1G>A
COL7A1	41	Epidermolisis bullosa distrifca autosómica recesiva	c.1732C>T, c.2471dupG, c.336C>G, c.3840delC, c.4039G>C, c.4119+1G>T, c.425A>G, c.4373C>T, c.4556G>A, c.4783G>C, c.4888C>T, c.4919delG, c.497dupA, c.4980+1G>T, c.4991G>C, c.5096C>T, c.5287C>T, c.5443G>A, c.5532+1G>A, c.5820G>A, c.6091G>A, c.6101G>A, c.6127G>A, c.6187C>T, c.6205C>T, c.6527dupC, c.6573+1G>A, c.6573+1G>C, c.6859G>A, c.6946G>A, c.7344G>A, c.7411C>T, c.7930-1G>C, c.7957G>A, c.8245G>A, c.8371C>T, c.8393T>A, c.8440C>T, c.8441-15_8445delTGCTCTTGGCTCCAGGACCC, c.8479C>T, c.8524_8527+10delGAAGGTGAGGACAG
COX15	2	Cardioencefalomiopatía infantil fatal debido a deficiencia de citocromo c oxidasa 2	c.1030T>C, c.649C>T
CPS1	9	Déficit de carbamil-fosfato sintetasa, hiperamonemia	c.1010A>G, c.130C>T, c.1631C>T, c.1913G>C, c.2359C>T, c.2407C>A, c.2945G>A, c.3037_3039delGTG, c.697C>T
CPT1A	30	Déficit de carnitina palmitoiltransferasa 1A	c.1027T>G, c.1069C>T, c.1079A>G, c.1241C>T, c.1339C>T, c.1361A>G, c.1364A>C, c.1393G>T, c.1425G>A, c.1436C>T, c.1451T>C, c.1493A>G, c.1494T>G, c.1600delC, c.1737C>A, c.1876-1G>A, c.2028+3_2028+6delAAGT, c.2126G>A, c.2129G>A, c.2156G>A, c.298C>T, c.367C>T, c.478C>T, c.912C>G, c.941C>T, nc.946C>G, c.948delG, c.96T>G, c.281+1G>A, c.222C>A
CPT2	35	Déficit de carnitina palmitoiltransferasa II	c.110_111dupGC, c.1145G>A, c.1148T>A, c.1239_1240delGA, c.1342T>C, c.1348A>T, c.1360G>T, c.1369A>T, c.149C>A, c.1507C>T, c.1511C>T, c.1646G>A, c.1649A>G, c.1657G>A, c.1737delC, c.1763C>G, c.1784delC, c.1810C>T, c.1883A>C, c.1891C>T, c.1925_1937delAGGCCTTAGAAGA, c.338C>T, c.359A>G, c.370C>T, c.38delG, c.452G>A, c.481C>T, c.520G>A, c.638A>G, c.641T>C, c.680C>T, c.691C>T,c.886C>T, c.983A>G, c.534_558del25insT
CRB1	12	Retinosis pigmentaria 12, autosómica recesiva	c.2234C>T, c.2290C>T, c.2330_2336delCAAACTC, c.2416G>T, c.2783G>A, c.2843G>A, c.2983G>T, c.3122T>C, c.3299T>C, c.3541T>C, c.484G>A, c.936T>G
CRB1	12	Amaurosis Congénita de leber 8	c.1180T>C, c.1576C>T, c.2401A>T, c.2688T>A, c.3299T>G, c.3307G>A, c.3997G>T, c.4121_4130delCAACTCAGGG, c.613_619delATAGGAA, c.998G>A, c.1612_1613insCTTA, c.807dupA
CRTAP	6	Osteogénesis imperfecta tipo 7	c.118_133delGAGCTGATGCCGCTCGinsTACCC, c.118G>T, c.3G>A, c.471+2C>A, c.561T>G, c.826C>T
CRX	4	Amaurosis congénita de Leber 7	c.124G>A, c.268C>T, c.425A>G, c.529delG
CTNS	23	Cistinosis nefropática	c.1015G>A, c.18_21delGACT, c.225+5_225+6delGTinsCC, c.283G>T, c.292dupA, c.329G>T, c.382C>T, c.397A>T, c.414G>A, c.416C>T, c.473T>C, c.506G>A, c.544T>C, c.561+1delG, c.589G>A, c.613G>A, c.614_616delACG, c.646dupA, c.696dupC, c.809_811delCCT, c.853-3C>G, c.922G>A, c.969C>G
CTSD	7	Lipofuscinosis ceroide neuronal	c.1149G>C, c.1196G>A, c.299C>T, c.446G>T, c.470C>T, c.685T>A, c.764dupA
CTSK	6	Picnodisostosis	c.154A>T, c.236G>A, c.436G>C, c.721C>T, c.926T>C, c.990A>G

CYP11B1	13	Hiperplasia suprarrenal congénita por déficit de 11-beta-hidroxilasa	c.1103C>A, c.1121G>A, c.124C>T, c.125C>T, c.1269T>G, c.1331G>Ac.1343G>A, c.264G>Ac.281C>T, c.347G>A, c.397A>C, c.953C>T, c.956C>T
CYP17A1	24	Hiperplasia suprarrenal congénita por déficit de 17-alfa-hidroxilasa	c.1024C>A, c.1039C>T, c.1040G>A, c.1073G>A, c.1084C>T, c.1216T>C, c.1226C>G, c.1247G>A, c.1283C>T, c.1435_1438dupATCC, c.160_162delTTC, c.278T>G, c.286C>T, c.287G>A, c.316T>C, c.340T>G, c.347A>T, c.374G>A, c.436+5G>T, c.51G>A, c.601T>A, c.715C>T, c.81C>A, c.985T>G
CYP21A2	17	Hiperplasia suprarrenal congénita por déficit de 21-hidroxilasa	c.1069C>T, c.1217G>A, c.1360C>T, c.1451_1452delGGinsC, c.293-13C>G, c.332_339delGAGACTAC, c.518T>A, c.701G>A, c.709_716delATCGTGGA, c.713T>A, c.844G>C, c.844G>T, c.874G>A, c.923dupT, c.92C>T, c.955C>T, c.710_719delTCGTGGAGATinsACGAGGAGAA
CYP27A1	52	Xantomatosis cerebrotendinosa	c.1016C>T, c.1016C>T, c.1061A>G, c.1183C>A, c.1183C>T, c.1184+1G>A, c.1184G>A, c.1185-1G>T, c.1202C>G, c.1209C>G, c.1213C>T, c.1214G>A, c.1222G>T, c.1238T>A, c.1263+1G>A, c.1263+5G>T, c.1264-1G>A, c.1342C>T, c.1415G>C, c.1420C>T, c.1421G>A, c.1435C>G, c.1435C>T, c.305delC, c.355delC, c.373_379delCCAGTAC, c.379C>T, c.380G>A, c.409C>T, c.410G>A, c.433G>A, c.434G>A, c.435G>T, c.446+1G>A, c.475C>T, c.583G>T, c.5dupC, c.646G>C, c.647-1G>T, c.691C>T, c.73delG, c.745C>T, c.752C>A, c.776A>G, c.779G>A, c.808C>T, c.819delT, c.844+1G>A, c.845-1G>A, c.850A>T, c.863delA, c.944_948delTGGCC
D2HGDH	6	Aciduria D-2-hidroxiglutárica	c.1123G>T, c.1315A>G, c.1331T>C, c.1333_1334delAC, c.440T>G, c.566C>T
DBT	24	Enfermedad de jarabe de arce 2	c.1017+1delG, c.1033G>A, c.1169A>G, c.1193T>C, c.1202T>C, c.1209+5G>C, c.1232C>A, c.1281+1G>T, c.1355A>G, c.1448G>T, c.251G>A, c.294C>G, c.363_364delCT, c.434-15_434-4delTTACCTTGTTAC, c.581C>G, c.670G>T, c.75_76delAT, c.788T>C, c.827T>G, c.871C>T, c.901C>T, c.939G>C, c.272_275delCAGT, c.126T>G
DCLRE1C	7	Inmunodeficiencia combinada grave por déficit de DCLRE1C	c.1350_1356delAGATTGT, c.1669dupA, c.194C>T, c.241C>T, c.403G>A, c.597C>A, c.780+1delG
DCLRE1C	2	Síndrome de Omenn	c.103C>G, c.2T>C
DFNB59	7	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB59	c.113dupT, c.122delA, c.161C>T, c.499C>T, c.547C>T, c.726delT, c.988delG
DHCR7	60	Síndrome de Smith-Lemli-Opitz	c.1039G>A, c.1054C>T, c.1055G>A, c.1079T>C, c.111G>A, c.1139G>A, c.1190C>T, c.1210C>T, c.1228G>A, c.1295A>G, c.1327C>T, c.1337G>A, c.1342G>A, c.1351T>C, c.1384T>C, c.1396G>A, c.1406G>C, c.1424T>C, c.1426T>C, c.151C>T, c.1A>G, c.203T>C, c.278C>T, c.292C>T,c.296T>C, c.326T>C, c.356A>T, c.3G>A, c.412+3A>T, c.443T>G, c.452G>A, c.453G>A, c.461C>G, c.461C>T, c.470T>C, c.502T>A, c.506C>T, c.523G>C, c.532A>T, c.536C>T, c.545G>T, c.575C>T, c.670G>A, c.682C>T, c.725G>A, c.728C>G, c.730G>A, c.744G>T, c.818T>G, c.839A>G, c.841G>A, c.852C>A, c.861C>A, c.866C>T, c.906C>G, c.907G>A, c.964-1G>C, c.970T>C, c.976G>T, c.724C>T
DHDDS	1	Retinosis pigmentaria 59	c.124A>G
DLD	11	Enfermedad de la orina de jarabe de arce tipo 3	c.1081A>G, c.1123G>A, c.1178T>C, c.1382G>A, c.140T>C, c.1436A>T, c.1444A>G, c.1463C>T, c.1483A>G, c.214A>G, c.685G>T
DMD	188	Distrofia muscular de Duchenne	c.10019G>A, c.10033C>T, c.10086+1G>A, c.10108C>T, c.10108C>T, c.1012G>T, c.10133delA, c.10141C>T, c.10171C>T, c.10223+1G>A, c.10223+1G>C, c.10453delC, c.10454delT, c.10624_10625delCC , c.1093C>T, c.1150-2A>C, c.1150-2delA, c.1261C>T, c.1286C>A, c.1324C>T, c.1331+1G>A, c.1332-9A>G, c.133C>T, c.1388G>A, c.1465C>T, c.1482+1G>A, c.1489C>T, c.1529_1530delTC, c.1533_1536delTCAC, c.160_162delCTC, c.1615C>T, c.161T>G, c.1663C>T, c.1683G>A, c.1704+1G>C, c.178C>T, c.1886C>G, c.1912C>T, c.1990C>T, c.199G>T, c.2032C>T, c.2125C>T, c.2137C>T, c.2215G>T, c.2281_2285delGAAAA, c.2302C>T, c.2332C>T, c.2380+1G>A, c.2407C>T , c.2419C>T, c.2479G>T, c.2484T>A, c.253C>T, c.2601_2602delAA, c.2611A>T, c.2623-3C>G, c.2650C>T, c.265-2A>G, c.2797C>T, c.2803+1G>A , c.2804-2A>T, c.2808dupT, c.280delA, c.2816T>A, c.2866C>T, c.2929dupC, c.2933_2934delGA, c.2956C>T, c.2991C>G , c.3076G>T, c.3087G>A, c.3121C>T, c.3151C>T,

			c.3246_3247insTTTCTAAAAA, c.3259C>T, c.3295C>T, c.3427C>T, c.3433-5_3434delGGAAGGT, c.3580C>T, c.358-2A>G, c.4117C>T, c.433C>T, c.4375C>T, c.4405C>T , c.4471_4472delAA, c.4518+5G>A, c.4523_4535delTGTATAAAAGTCT, c.4545_4549delGAAGT, c.4606G>T , c.4675-2A>G, c.4729C>T, c.4870C>T, c.4918delA, c.4996C>T , c.503C>A , c.5124_5127delGAAA, c.5131C>T, c.5134C>T, c.530+1delG, c.5314A>T, c.5350G>T, c.5353C>T, c.5461G>T, c.547dupT, c.5530C>T, c.5551C>T, c.5554C>T, c.5602_5605delAGAA, c.5641C>T, c.565C>T, c.5697delA, c.572C>G, c.5773G>T, c.583C>T, c.5899C>T, c.5899C>T, c.5917C>T, c.5922+3G>C, c.5938G>T, c.5985T>G, c.6072T>A, c.6128_6131delATAG, c.615T>A , c.6182delC, c.6237_6238delCC, c.6283C>T, c.6292C>T , c.6373C>T, c.6391_6392delCA, c.649+1G>T, c.6611dupA, c.676_678delAAG, c.6762+1G>T, c.6906G>A, c.691T>A, c.6943G>T, c.6955C>T, c.6964delG, c.6986dupA, c.7229G>A, c.724C>T, c.7309+1G>A, c.748G>T, c.7657C>T, c.7682G>A, c.7755G>A, c.7817G>A, c.8027+2T>A, c.8038C>T, c.8064_8065delTA, c.8086delC, c.8218-2A>G, c.8357G>A, c.8390+2T>C, c.8443C>T, c.8464C>T, c.8608C>T, c.8656C>T, c.8680G>T, c.8713C>T, c.8912_8913delTC, c.8944C>T, c.8970_8971delGA, c.9148C>T, c.9158_9161delTTTC, c.9164-1G>A, c.9164-1G>T, c.9197C>A, c.9204_9207delCAAA, c.9216C>A, c.9337C>T, c.9346C>T, c.9361+1G>A, c.9380C>G, c.9471_9474delTTAT , c.9551dupA, c.9564-1G>A, c.9568C>T, c.9851G>A, c.9854_9863delTGAGACTGGA, c.9862G>T, c.9938_9941dupGTAA, c.9978C>G, c.9975-2delA, c.5363C>G, c.7683G>A, c.8374_8375delAA, c.8069T>G Análisis deleciones/duplicaciones
DMD	26	Distrofia muscular tipo Becker	c.10262C>T, c.10412T>A, c.10509_10510delAG, c.10554-2A>G, c.137A>T, c.14_15delAAinsT, c.1483-1G>C, c.1812+1G>A, c.2169-4_2169-1delTTAGinsCTTT, c.32-2A>T, c.3432+1G>A, c.3432+3A>G, c.355C>T, c.3779_3785delCTTTGGAinsGG, c.3940C>T, c.3940C>T, c.4240C>T, c.4996_5004delCGAGCAGAAinsT, c.5287C>T, c.5287C>T, c.7105G>T, c.7402G>T, c.8668G>A, c.6000T>A, c.6982A>T, c.8652_8653delCT
DMD	14	Miocardiopatía dilatada familiar aislada 3B	c.31+1G>T, c.31+1G>A, c.5640T>A, c.5671A>T, c.5807T>A, c.6014_6017delCTCA, c.3697delC, c.4071G>C, c.6834delT, c.6936delA, c.7764dupT, c.7771G>T, c.7922delA, c.8358G>A
ESPN	2	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB 36	c.1988_1991delAGAG, c.2470_2473delTCAG
ESRRB	2	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB 35	c.1024G>T, c.329C>T
ETFA	5	Acidemia glutárica 2A	c.346G>A, c.470T>G, c.797C>T, c.809_811delTAG, c.963+1delG
ETFB	4	Acidemia glutárica 2B	c.382G>A, c.490C>T, c.491G>A, c.614_616delAGA
ETFDH	23	Acidemia glutárica 2C	c.1001T>C, c.1130T>C, c.1351G>C, c.1367C>T, c.1448C>T, c.1531G>A, c.1773_1774delAT, c.1809G>A, c.1832G>A, c.1852T>C, c.213_215delTGT, c.250G>A, c.2T>C, c.36delA, c.380T>A, c.380T>C, c.405+3A>T, c.413T>G, c.508G>T, c.51dupT, c.524G>A, c.524G>T, c.643G>A
EYS	33	Retinosis pigmentaria 25	c.1211dupA, c.1750G>T, c.179delT, c.2194C>T, c.2710_2726delGACTGTGAAGATATGGT, c.3329C>G, c.3443+1G>T, c.4045C>T, c.4120C>T, c.4350_4356delTATAGCT, c.4829_4832delCATT, c.490C>T, c.4957dupA, c.5202_5203delGT, c.5857G>T, c.6416G>A, c.6557G>A, c.6714delT, c.6794delC, c.7095T>G, c.7793G>A, c.7919G>A, c.8012T>A, c.8376_8379dupTGCA, c.8408dupA, c.8648_8655delCATGCAGA, c.8805C>A, c.8834G>A, c.9186_9187delCA, c.9277_9278dupGG, c.9286_9295delGTAAATATCG, c.9299_9302delCTCA, c.9405T>A
F8	187	Hemofilia A	c.1026T>A, c.1043G>C, c.104A>G, c.1063C>T, c.1077_1080delTGAA, c.1078_1079delGA, c.1171C>T, c.1172G>A, c.1175C>A, c.1175C>T, c.1214T>G, c.121G>T, c.1226A>G, c.1293delG, c.1293G>T, c.1331A>G, c.1336C>T, c.1348T>A, c.1418A>G, c.1475A>G, c.1481T>C, c.1492G>A, c.1596dupG, c.1630G>A, c.1636C>T, c.1648C>G, c.1648C>T,

			c.1649G>A, c.1660A>G, c.1682A>G, c.1726G>T, c.1729T>C, c.1730C>T, c.1750C>A, c.1754T>C, c.1786T>C, c.1804C>T, c.1808G>T, c.1812G>C, c.1814A>C, c.1834C>T, c.1892A>G, c.1957G>A, c.1958T>C, c.1965C>G, c.1988C>T, c.199_200delAA, c.2029T>C, c.210_212delTGT, c.2149C>T, c.2167G>A, c.2215G>A, c.266G>A, c.275G>T, c.2945dupA, c.2962_2963delAG, c.311T>A, c.3169G>A, c.323A>C, c.328A>G, c.350T>G, c.3548_3549delAA, c.3637delA, c.364_365delGT, c.3692delC, c.388G>C, c.396A>C, c.398A>G, c.404A>G, c.410C>T, c.4121_4124delTAGA, c.4296_4300delTTCTC, c.4328_4331delAAGA, c.4379delA, c.43C>T, c.4825dupA, c.4858delC, c.491G>T, c.493C>T, c.5096A>T, c.5113C>T, c.5122C>T, c.5123G>A, c.5143C>G, c.5143C>T, c.5167G>A, c.5183A>G, c.5305G>A, c.5323T>G, c.5325G>C, c.5336G>A, c.5372T>C, c.5398C>G, c.5398C>T, c.5399G>A, c.5408C>A, c.541G>A, c.5422C>T, c.5443C>T, c.5526G>A, c.5530C>T, c.5533A>C, c.554A>C, c.5558C>T, c.5593G>A, c.5593G>T, c.5600A>G, c.5618C>G, c.566C>T, c.5677C>T, c.5710G>A, c.5719dupA, c.5821A>G, c.5822A>G, c.5878C>T, c.5879G>A, c.5879G>T, c.5882G>A, c.5900G>A, c.592T>G, c.5936G>T, c.5938C>T, c.5953C>T, c.5961delA, c.6016G>T, c.6046C>T, c.6049delG, c.6113A>G, c.6193T>C, c.6263C>T, c.6278A>G, c.6360T>G, c.6371A>G, c.6403C>T, c.6404G>C, c.6412_6413delTC, c.6413C>A, c.6464_6465delAA, c.6496C>T, c.6506G>A, c.6515C>A, c.6518C>T, c.6532C>T, c.6533G>A, c.6533G>T, c.6544C>T, c.6545G>A, c.6554T>C, c.6631G>C, c.665A>T, c.6682C>G, c.6682C>T, c.6683G>A, c.6683G>T, c.6699delG, c.670G>T, c.670G>T, .6744G>T, c.6794_6795delAG, c.6794A>G, c.6865C>T, c.688_689delGA, c.6914_6918delATCAA, c.6955C>T, c.6956C>T, c.6967C>T, c.6968G>A, c.6976C>T, c.6977G>A, c.6977G>T, c.77T>G, c.797G>A, c.822G>A, c.832G>A, c.849delT, c.854T>G, c.872A>G, c.881C>T, c.896A>T, c.89A>T, c.902G>A, c.902G>T, c.923C>T, c.935T>C, c.940A>G, c.943delG, c.980T>C
F9	66	Hemofilia B	c.1009G>C, c.1025C>T, c.1058T>C, c.1064G>T, c.1069G>A, c.1070G>A, c.1088G>T, c.1120G>T, c.1135C>T, c.1136G>A, c.1144T>C, c.1150C>T, c.1151G>C, c.1151G>T, c.1180A>G, c.1187G>C, c.1189G>C, c.1217C>T, c.1228G>C, c.1231A>G, c.1232G>T, c.1240C>A, c.1256T>A, c.128G>A, c.1307C>A, c.1307C>T, c.1324G>A, c.133_134insT, c.1357T>C, c.1369A>T, c.169C>T, c.217G>A, c.218A>T, c.223C>T, c.224G>A, c.237A>C, c.277+2T>C, c.278-3A>G, c.278A>G, c.287A>C, c.301C>G, c.316G>A, c.328G>A, c.329A>G, c.479G>C, c.484C>T, c.496A>T, c.540_541delAG, c.571C>T, c.572G>A, c.655C>T, c.676C>T, c.677G>A, c.682G>C, c.682G>T, c.709C>T, c.755G>C, c.804T>G, c.82T>C, c.835G>A, c.872A>T, c.880C>T, c.881G>A, c.892C>T, c.917A>G, c.998C>T
FAH	27	Tirosinemia tipo 1	c.1009G>A, c.1021C>T, c.1027G>T, c.103G>A, c.1062+5G>A, c.1069G>T, c.1090G>T, c.1141A>G, c.1190delA, c.192+1G>T, c.192G>T, c.401C>A, c.456G>A, c.47A>T, c.520C>T, c.554-1G>T, c.607-1G>A, c.607-6T>G, c.698A>T, c.707-1G>A, c.707-1G>C, c.782C>T, c.786G>A, c.836A>G, c.836A>G, c.939delC, c.982C>T
FGD4	8	Enfermedad de Charcot-Marie-Tooth tipo 4H	c.1325G>A, c.1329C>A, c.1756G>T, c.1762-2A>G, c.670C>T, c.823C>T, c.893T>C, c.893T>G
FH	5	Aciduria fumárica	c.1084G>C, c.1127A>C, c.1431_1433dupAAA, c.521C>G, c.697C>T
FIG4	10	Enfermedad de Charcot-Marie-Tooth tipo 4J	c.1141C>T, c.122T>C, c.1666dupA, c.2459+1G>A, c.290-2A>T, c.50T>C, c.547C>T, c.737G>A, c.831_838delTAAATTTG, c.904G>A
FIG4	2	Síndrome de Yunis-Varon	c.311G>A, c.524T>C
FKRP	1	Síndrome de Walker-Warburg	c.953G>A
FKRP	23	Distrofia muscular de cinturas autosómica recesiva tipo C	c.1141delG, c.1154C>A, c.1213G>T, c.1323T>G, c.1343C>T, c.1364C>A, c.1387A>G, c.1486T>A, c.160C>T, c.162_165dupGGAG, c.1A>G, c.387_390dupACCT, c.400C>T, c.545A>G, c.663C>A, c.764G>A, c.826C>A, c.899T>C, c.919T>A, c.926A>G, c.946C>A, c.947C>G, c.970G>C
FMR1	1	Síndrome del X-frágil	c.911T>A, EXP
FLVCR1	3	Enfermedad de astas posteriores, ataxia - Retinosis pigmentaria	c.361A>G, c.574T>C, c.721G>A

FOXN1	1	Inmunodeficiencia severa de linfocitos T - alopecia congénita - distrofia ungueal	c.763C>T
FUCA1	9	Fucosidosis	c.1138G>T, c.1160G>A, c.1229T>G, c.1279C>T, c.244C>T, c.393T>A, c.464C>T, c.648C>A, c.790C>T
FXN	9	Ataxia de Friedreich	c.11_12delTC, c.157delC, c.317T>G, c.371_376delATGTCTinsTACACCTTGAGGACA, c.385-2A>G, c.389G>T, c.3G>T, c.460A>T, c.517T>G
G6PD	31	Déficit de glucosa-6-fosfato-deshidrogenasa	c.1003G>A, c.1048G>C, c.108_110delCAT, c.1082C>T, c.1089C>A, c.1156A>G, c.1159C>T, c.1178G>A, c.1180G>C, c.1229G>A, c.1316G>C, c.1339G>A, c.1360C>T, c.1376G>T, c.1388G>A, c.143T>C , c.202G>A, c.376A>G, c.383T>C, c.487G>A, c.493A>G, c.542A>T, c.563C>T, c.593G>C, c.637G>T, c.680G>T, c.806G>A, c.844G>C, c.871G>A, c.964T>C, c.968T>C
GAA	75	Enfermedad de almacenamiento de glucógeno por déficit de maltasa ácida (enfermedad de Pompe)	c.1004G>A, c.1064T>C, c.1115A>T, c.1124G>T, c.1128_1129delGGinsC, c.1156C>T, c.118C>T, c.1309C>T, c.1316T>A, c.1408_1410delAAC, c.1437G>A, c.1438-1G>C, c.1441T>C, c.1445C>T, c.1465G>A, c.1548G>A, c.1552-3C>G, c.1556T>C, c.1561G>A, c.1585_1586delTCinsGT, c.1634C>T, c.1655T>C, c.172C>T, c.1754+1G>A,c.1799G>A, c.1826dupA, c.1843G>A, c.1912G>T, c.1927G>A, c.1933G>A, c.1933G>C, c.1933G>T, c.1935C>A, c.1942G>A, c.1979G>A, c.1A>G, c.2012T>G, c.2014C>T, c.2015G>A, c.2024_2026delACA, c.2041-1G>A, c.2066_2070dupAGCCG, c.2078dupA, c.2104C>T, c.2140delC, c.2173C>T, c.2237G>A, c.2501_2502delCA, c.2544delC, c.2560C>T, c.258dupC, c.2608C>T, c.2646+2T>A, c.2707_2709delAAG, c.307T>G, c.-32-13T>G, c.343C>T, c.365delT, c.525_526delTG, c.525delT, c.546G>A, c.546G>C, c.569G>A, c.655G>A, c.670C>T, c.710C>T,c.768dupT, c.784G>A, c.853C>T, c.854C>G, c.877G>A, c.896T>C, c.896T>G, c.925G>A, c.953T>C
GALC	37	Enfermedad de Krabbe	c.1004A>G, c.1153G>T, c.1472delA, c.1541T>C, c.1586C>T, c.1591C>T, c.1592G>A, c.1630G>A, c.1657G>A, c.1695delT, c.169G>A, c.1700A>C, c.1723_1724insT, c.1796T>G, c.195G>C, c.2002A>C, c.2056T>C, c.205C>T, c.236G>A, c.246A>G, c.331G>A, c.334A>G, c.349A>G, c.388G>A, c.430delA, c.489G>A, c.599C>A, c.628A>T, c.658C>T, c.683_694delATCTCTGGGAGTinsCTC, c.749T>C, c.857G>A, c.908+1G>A, c.908C>T, c.953C>G, c.955delT, c.956A>G
GALE	11	Déficit de galactosa epimerasa	c.101A>G, c.269G>A, c.280G>A, c.308A>G, c.505C>T, c.548T>C, c.715C>T, c.770A>G, c.905G>A, c.937C>A, c.956G>A
GALK1	7	Déficit de galactoquinasa	c.1031C>T, c.1045G>A, c.1144C>T, c.238G>T, c.766C>T, c.82C>A, c.94G>A
GALNS	33	Mucopolisacaridosis tipo 4A	c.1019G>A, c.106_111delCTGCTC, c.1156C>T, c.1171A>G, c.1175C>T, c.139G>A, c.1417C>T, c.1460A>G, c.1485C>G, c.1559G>A, c.178G>A, c.205T>G, c.280C>G, c.337A>T, c.413T>C, c.415G>A, c.421T>A, c.451C>A, c.463G>A, c.477G>A, c.485C>T, c.542A>G, c.612C>G, c.689G>A, c.704C>A, c.740G>A, c.776G>A, c.860C>T, c.871G>A, c.898+1G>C, c.901G>T, c.935C>G, c.953T>G
GALT	211	Galactosemia clásica	c.1001A>G, c.1006A>T, c.100T>A, c.1014C>G, c.1018G>A, c.1018G>T, c.1024C>A, c.1030C>A, c.1034C>A, c.1047delC, c.1048A>G, c.1052delC, c.1057C>T, c.1059+56C>T, c.107C>T, c.1098C>A, c.1108C>T, c.1132A>G, c.1138T>C, c.113A>C, c.1140A>C, c.130G>A, c.130G>T, c.134C>T, c.136_140delGCTCA, c.152G>A, c.152G>T, c.160C>T, c.163G>T, c.18delC, c.197C>A, c.197C>T, c.199C>T, c.1A>G, c.200G>A, c.203A>C, c.207_214delCCCTCTCA, c.220_221delCT, c.220_221insG, c.221T>C, c.238C>T, c.241G>A, c.247G>A, c.253-2A>G, c.25C>T, c.265T>C, c.265T>G, c.27G>C, c.285T>G, c.289_291delAAC, c.290A>G, c.290A>G, c.292G>A, c.292G>C, c.308A>G, c.328+2T>C, c.329-2A>C, c.334dupA, c.337G>A, c.341A>T, c.350T>C, c.354A>C, c.367C>G, c.367C>T, c.368G>A,c.374T>C, c.377+1G>T, c.379A>G, c.386T>C, c.389G>A, c.392T>G, c.394C>T, c.396C>A, c.400delT, c.404C>G, c.404C>T, c.410dupT, c.413C>T, c.416T>C, c.41delCinsTT, c.425T>A, c.425T>C, c.428C>T, c.442C>G, c.442C>T, c.443G>A, c.448G>C, c.452T>C, c.460T>G, c.462G>A, c.482T>C, c.490C>T, c.496C>G, c.499T>C, c.502_504delGTG, c.505C>A, c.507+2T>C, c.509T>A, c.509T>C, c.512T>C, c.524G>A, c.536G>A, c.539G>T, c.541T>G, c.542C>T, c.547C>A, c.552C>A, c.553C>T, c.554C>A,

			c.554C>T, c.556C>T, c.563A>G, c.564+1G>A, c.565-2A>G, c.574A>G, c.575G>A, c.580T>C, c.584T>C, c.594T>G, c.595G>A, c.598delC, c.601C>T, c.602G>A, c.607G>A, c.610C>T, c.611G>C, c.619C>T, c.626A>C, c.626A>G, c.634C>T, c.635A>C, c.650T>C, c.652C>G, c.652delC, c.658dupG, c.658G>A, c.667C>A, c.676C>G, c.677T>C, c.67A>G, c.680T>C, c.687+2T>C, c.687G>T, c.691C>T, c.692G>A, c.697G>C, c.745T>C, c.747G>A, c.748C>A, c.752A>C, c.752A>G, c.756G>T, c.768_770delGCC, c.770C>T, c.772C>T, c.775C>T, c.776G>A, c.779_790delATGTGCGGCGGC, c.785G>C, c.790_792delCTAinsTAG, c.793C>G, c.812A>G, c.814C>G, c.815G>A, c.820+13A>G, c.821-2A>G, c.824delT, c.82G>A, c.833T>A, c.836T>A, c.844C>G, c.854A>G, c.855G>T, c.857A>G, c.864C>T, c.865C>T, c.866T>G, c.871G>A, c.872A>T, c.881T>A, c.882delT, c.883C>A, c.90G>C, c.91C>A, c.920C>A, c.922G>A, c.947G>A, c.948G>A, c.949delC, c.950A>G, c.951G>T, c.952delC, c.957C>A, c.958G>A, c.95T>A, c.961C>T, c.967T>C, c.967T>G, c.970C>T, c.974C>T, c.976delC, c.979delC, c.980T>C, c.983G>A, c.986C>T, c.989C>T, c.98G>C, c.997C>G, c.997C>T, c.998G>A, c.998G>T
GBA	34	Enfermedad de Gaucher forma fetal	c.1060G>C, c.1085C>T, c.115+1G>A, c.1171G>C, c.1174C>G, c.1192C>T, c.1228C>G, c.1240G>T, c.1246G>A, c.1297G>T, c.1343A>T, c.1397T>G, c.1504C>T, c.1505G>A, c.1505G>A, c.1549G>A, c.1604G>A, c.254G>A, c.259C>T, c.354G>C, c.475C>T, c.481C>T, c.533delC, c.680A>G, c.703T>C, c.721G>A, c.73delC, c.751T>C, c.763T>G, c.764T>A, c.84dupG, c.866G>C, c.887G>A, c.983C>T
GBA	9	Enfermedad de Gaucher tipo 1	c.1043C>T, c.1053G>T, c.1141T>C, c.1208G>C, c.1226A>G, c.1319C>T, c.431T>G, c.476G>A, c.625C>T
GBA	12	Enfermedad de Gaucher tipo 2	c.1049A>G, c.1090G>A, c.1309G>T, c.1342G>C, c.1361C>G, c.1448T>C, c.508C>T, c.509G>T, c.586A>C, c.667T>C, c.870C>A, c.896T>C
GBA	1	Enfermedad de Gaucher tipo 3	c.754T>A
GCDH	36	Déficit de glutaril-CoA deshidrogenasa	c.1015A>G, c.1031C>T, c.1060G>A, c.1093G>A, c.1147C>T, c.1148G>A, c.1156C>G, c.1168G>C, c.1198G>A, c.1204C>T, c.1205G>A, c.1213A>G, c.1239C>A, c.1240G>A, c.1244-2A>C, c.1247C>T, c.1262C>T, c.262C>T, c.271+1G>A, c.383G>A, c.416C>T, c.442G>A, c.482G>A, c.533G>A, c.542A>G, c.572T>C, c.636-1G>A, c.680G>C, c.743C>T, c.764C>T, c.769C>T, c.856C>T, c.877G>A, c.883T>C, c.914C>T, c.947C>A
GDAP1	1	Enfermedad de Charcot-Marie-Tooth autosómica recesiva con ronquera	c.373C>T
GDAP1	6	Enfermedad de Charcot-Marie-Tooth intermedia autosómica recesiva tipo A	c.311-1G>A, c.347T>G, c.501delA, c.579+1G>A, c.692C>T, c.715C>T
GDAP1	8	Enfermedad de Charcot-Marie-Tooth tipo 4A	c.469A>C, c.482G>A, c.487C>T, c.581C>G, c.652C>G, c.769C>T, c.844C>T, c.92G>A
GFM1	6	Hepatoencefalopatía por déficit combinado de la fosforilación oxidativa tipo 1	c.139C>T, c.1487T>G, c.2011C>T, c.521A>G, c.688G>A, c.748C>T
GJB2	78	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB	c.101T>G, c.107T>C, c.109G>A, c.109G>C, c.119C>A, c.119C>G, c.131G>A, c.134G>A, c.139G>T, c.162C>A, c.164C>A, c.167delT, c.169C>T, c.172C>G, c.175G>A, c.176_191delGCTGCAAGAACGTGTG, c.194A>G, c.19C>T, c.1A>G, c.208C>G, c.229T>C, c.-23+1G>A, c.230G>A, c.231G>A, c.235delC, c.238C>T, c.239A>C, c.-23G>T, c.246C>G, c.250G>C, c.250G>T, c.269dupT, c.269T>C, c.280_284dupCACGT, c.283G>A, c.290dupA, c.298C>T, c.299_300delAT, c.299A>T, c.310_323delAGGAAGTTCATCAA, c.313_326delAAGTTCATCAAGGG, c.334_335delAA, c.339T>G, c.355G>A, c.35delG, c.35G>T, c.365A>T, c.370C>T, c.389G>C, c.400T>C, c.402delG, c.408C>A, c.413G>A, c.416G>A, c.427C>T, c.439G>A, c.44A>C, c.456C>A, c.465T>A, c.476A>T, c.506G>A, c.508_511dupAACG, c.51_62delCACCAGCATTGGinsA, c.516G>A, c.535G>A, c.550C>T, c.551G>C, c.557C>T, c.596C>T, c.598G>A, c.598G>T, c.617A>G, c.632_633delGT, c.647_650delGATA, c.71G>A, c.94C>T, c.9G>A, c.-259C>T
GLA	146	Enfermedad de Fabry	c.101A>G, c.1020G>A, c.1023A>C, c.1024C>T, c.1025G>A, c.1033_1034delTC, c.1037delG, c.104G>A, c.1057_1058delAT, c.1066C>T, c.1067G>C, c.1072_1074delGAG, c.1072G>A, c.107T>G, c.1081G>A, c.1081G>C, c.1085C>T, c.1087C>T,

			c.1088G>A, c.1095T>A, c.1117G>A, c.1118G>A, c.1124G>C, c.1157A>C, c.1176G>T, c.1188delC, c.118C>T, c.1192G>T, c.1196G>C, c.1225C>T, c.1228A>G, c.1229C>A, c.1229C>T, c.1235_1236delCT, c.125T>C, c.127G>A, c.131G>A, c.137A>G, c.146G>C, c.166T>G, c.190A>T, c.194G>C, c.195-1G>T, c.19G>T, c.235G>T, c.239G>A, c.242G>A, c.256T>C, c.272T>A, c.272T>C, c.274G>T, c.281G>A, c.285G>T, c.334C>T, c.335G>A, c.337T>A, c.337T>C, c.352C>T, c.369+1G>A, c.369+2T>G, c.370-2A>G, c.386T>C, c.41T>C, c.422C>T, c.424T>C, c.427G>C, c.436C>T, c.466G>A, c.469C>T, c.484T>C, c.485G>A, c.540G>T, c.548-1G>A, c.548-2A>G, c.548G>T, c.561G>A, c.58G>C, c.593T>C, c.59C>A, c.605G>A, c.606T>G, c.610T>C, c.613C>A, c.614C>T, c.620A>G, c.62T>C, c.638A>G, c.640-801G>A, c.641C>T, c.644A>G, c.647A>G, c.657C>G, c.658C>T, c.661C>T, c.666C>A, c.677G>A, c.679C>T, c.680G>A, c.680G>C, c.704C>G, c.707G>A, c.713G>A, c.724A>G, c.724A>T, c.730G>A, c.734G>A, c.748C>T, c.758T>C, c.761T>C, c.784T>C, c.791A>T, c.797A>T, c.801+3A>G, c.801G>A, c.802-2A>G, c.802-3_802-2delCA, c.806T>C, c.806T>G, c.815A>G, c.823C>T, c.830G>A, c.835C>G, c.847C>T, c.861G>A, c.865A>T, c.886A>G, c.888G>A, c.890C>T, c.899T>C, c.901C>T, c.902G>A c.916C>T, c.950T>G, c.961C>G, c.966C>A, c.973G>A, c.974G>A, c.979C>A, c.980A>G, c.980A>T, c.982G>A, c.982G>C, c.983G>C, c.98A>G, c.996_999delACAG, c.999+2T>C
GLB1	4	Gangliosidosis GM1 tipo 1	c.1343A>T, c.1480-2A>G, c.1771T>A, c.1772A>G
GLB1	34	Gangliosidosis GM1 tipo 2	c.1051C>T, c.1068+1G>T, c.1077delA, c.1321G>A, c.1325G>A, c.1369C>T, c.1370G>A, c.145C>T, c.152T>C, c.1549G>T, c.1577dupG, c.1646C>T, c.171C>G, c.1733A>G, c.175C>T, c.1768C>T, c.1769G>A, c.176G>A, c.202C>T, c.203G>A, c.245C>T, c.276G>A, c.367G>A, c.438_440delTCT, c.442C>A, c.442C>T, c.464T>G, c.591dupT, c.601C>T, c.602G>A, c.622C>T, c.75+2dupT, c.922T>C, c.947A>G
GLB1	10	Mucopolisacaridosis tipo 4B	c.1223A>C, c.1313G>A, c.1444C>T, c.1445G>A, c.1498A>G, c.1527G>T, c.247T>C, c.808T>G, c.817_818delTGinsCT, c.817T>C
GM2A	5	Gangliosidosis GM2, variante AB	c.160G>T, c.164C>T, c.333delC, c.412T>C, c.506G>C
GNPTAB	77	Mucolipidosis tipo 2	c.1001G>T, c.1090C>T, c.1120T>C, c.1123C>T, c.118-2A>G, c.1191_1194dupGCTG, c.1206dupT, c.121delG, c.1331dupG, c.136C>T, c.1399delG, c.1402T>A, c.1519C>T, c.1581delC, c.163dupT, c.171delA, c.1759C>T, c.1774G>A, c.1959_1962delTAGT, c.1965delC, c.1999_2000insT, c.1999G>T, c.2089dupC, c.2188delTinsAAA, c.2189delT, c.2220_2221dupGA, c.2249dupA, c.2275_2276delAA, c.2354T>G, c.2422delC, c.2427delC, c.2533C>T, c.2544delA, c.2550_2554delGAAAA, c.2591_2592insG, c.2659dupA, c.2664C>G, c.2681G>A, c.2693dupA, c.3002T>C, c.3053A>G, c.3061C>T, c.3091C>T, c.310C>T, c.3173C>G, c.3231_3234dupCTAC, c.3232delT, c.3249+1G>C, c.3250-2A>G, c.3252delA, c.3310delG, c.3330dupA, c.3336-1G>C, c.3410T>A, c.3428dupA, c.3434+1G>A, c.344_345delCA, c.3443_3446delTTTG, c.3474_3475delTA, c.3487_3490delACAG, c.3503_3504delTC, c.3523_3529delATGTTCC, c.3565C>T, c.3613C>T, c.3741_3744delAGAA, c.441delC, c.571+3A>C, c.616_619delACAG, c.625_629delAGGGG c.637-1G>A c.648_651delAGAA c.749dupA c.755_759delCCTCT c.850delA c.857dupA, c.914dupA, c.940C>T
GNPTAB	28	Mucolipidosis tipo 3	c.1000C>T, c.1001G>A, c.10A>C, c.1196C>T, c.1208T>C, c.1220A>C, c.1285-2A>G, c.1325G>A, c.1381T>G, c.1385dupA, c.1514G>A, c.168T>A, c.2053_2057delTCAAC, c.232_234delGTT, c.242G>T, c.2574_2575delGA, c.2693delA, c.2715+1G>A, c.2715+2T>G, c.2777A>C, c.2866C>T, c.2867A>G, c.3458A>G, c.44C>A, c.517_518insA, c.569A>T, c.771G>A, c.832C>T
GNRHR	20	Hipogonadismo hipogonadotropo normosómico congénito 7	c.268G>A, c.30_31delTCinsAA, c.30T>A, c.317A>G, c.31C>A, c.350T>G, c.386C>A, c.392T>C, c.416G>A, c.504T>A, c.511G>A, c.523-1G>A, c.651C>A, c.785G>A, c.806C>T, c.842C>T, c.851A>G, c.941T>A, c.94A>G, c.959C>T
GNS	6	Síndrome de Sanfilippo tipo D	c.1063C>T, c.1138_1139insGTCCT, c.1168C>T, c.1169delA, c.1226dupG, c.83delT

GPR143	1	Nistagmus 6	c.266C>T
GPR143	5	Albinismo ocular recesivo ligado al X tipo 1	c.104G>A, c.397T>A, c.397T>C, c.455G>A, c.695C>A
GRXCR1	4	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB25	c.229C>T, c.412C>T, c.627+19A>T, c.628-9C>A
GUCY2D	8	Amaurosis congénita de Leber 1	c.1694T>C, c.2302C>T, c.2513G>A, c.2983C>T, c.3233_3236dupACCA, c.416T>C, c.527T>C, c.622delC
GUSB	25	Mucopolisacaridosis tipo 7	c.1050G>C, c.1061C>T, c.1069C>T, c.1084G>A, c.1144C>T, c.1222C>T, c.1244+1G>A, c.1244C>T, c.1337G>A, c.1429C>T, c.1484A>G, c.1521G>A, c.1586A>G, c.1617C>T, c.1730G>T, c.1831C>T, c.1856C>T, c.1881G>T, c.307C>T, c.398G>C, c.442C>T, c.526C>T, c.530C>T, c.646C>T, c.959A>C
HADH	3	Hiperinsulinismo por déficit de 3-hidroxilacil-CoA dehidrogenasa	c.676T>C, c.706C>T, c.773C>T
HADHA	1	Déficit de 3-hidroxiacil-CoA deshidrogenasa de ácidos grasos de cadena larga	c.1528G>C
HADHA	16	Déficit de proteina trifuncional mitocondrial	c.1025T>C, c.1132C>T, c.157C>T, c.1620+2_1620+6delTAAGG, c.1678C>T, c.1793_1794delAT, c.180+1G>A, c.180+3A>G, c.1967delT, c.2027G>A, c.2132dupC, c.274_278delTCATC, c.703C>T, c.845T>A, c.871C>T, c.914T>A
HADHB	8	Déficit de proteina trifuncional mitocondrial	c.1175C>T, c.1331G>A, c.1364T>G, c.182G>A, c.357dupT, c.740G>A, c.776_777insT, c.788A>G
HBA1	5	Alfa-Talasemia	c.179G>A, c.188_190delTGG, c.207C>G, c.223G>C, c.410T>G, Análisis deleciones
HBA2	17	Alfa-Talasemia	c.118_124delACCAAGAinsTACTTC, c.178G>C, c.186G>C, c.207C>A, c.207C>G, c.301-1G>A, c.314G>A, c.339_350delCCTCCCCGCCGA, c.349G>T, c.377T>C, c.410T>C, c.427T>A, c.427T>C, c.429A>T, c.96-2A>G, c.*92A>G, c.*94A>G, Análisis deleciones
HBB	132	Beta talasemia	c.103G>T, c.108C>A, c.110delC, c.112delT, c.113G>A, c.114_120delGACCCAG, c.114G>A, c.117_118delCC, c.118C>T, c.126_129delCTTT, c.126delC, c.127T>G, c.130G>T, c.135delC, c.143dupA, c.169G>C, c.17_18delCT, c.176dupC, c.182T>A, c.184A>T, c.194delG, c.199A>G, c.19G>A, c.1A>G, c.201delA, c.203_204delTG, c.20delA, c.216dupT, c.217_221delAGTGAinsT, c.217dupA, c.230delC, c.247A>G, c.25_26delAA, c.251delG, c.271G>T, c.277C>A, c.27dupG, c.282_283dupTG, c.287dupA, c.295G>A, c.2T>C, c.2T>G, c.315+1G>A, c.315+2T>C, c.315+4_315+5delAG, c.315G>C, c.316-106C>G, c.316-146T>G, c.316-14T>G, c.316-197C>T, c.316-1G>A, c.316-1G>C, c.316-1G>T, c.316-2A>C, c.316-2A>G, c.320T>G, c.323dupG, c.328delG, c.328G>A, c.332T>C, c.341T>A, c.343_344delCTinsG, c.344T>C, c.347C>A, c.364G>A, c.364G>C, c.364G>T, c.36delT, c.380T>A, c.380T>G, c.383_385delAGG, c.383A>C, c.3G>A, c.404T>A, c.415G>C, c.45dupG, c.46delT, c.47G>A, c.48G>A, c.4delG, c.4G>T, c.51delC, c.52A>T, c.59A>G, c.68_74delAAGTTGG, c.71_73delTTG, c.75T>A, c.79_80insT, c.79G>A, c.79G>A, c.82G>T, c.82G>T, c.85dupC, c.86T>A, c.8A>C, c.90C>T, c.92+1G>A, c.92+1G>C, c.92+1G>T, c.92+2T>A, c.92+2T>C, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.92G>A, c.92G>C, c.93-1_94dupGGC, c.93-1G>A, c.93-1G>C, c.93-21G>A, c.93G>T, c.*113A>G, c.*112A>G, c.*111A>G, c.*110T>C, c.-29G>A, c.-50A>C, c.-78A>C, c.-78A>G, c.-79A>G, c.-80T>A, c.-81A>G, c.-82C>A, c.-136C>G, c.-137C>G, c.-137C>T, c.-137C>A, c.-138C>T, c.-140C>T, c.-142C>T, c.-151C>T
HBB	3	Anemia falciforme	c.20A>T, c.256T>C, c.337T>C
HEXA	97	Enfermedad de Tay-Sachs	c.1003A>T, c.1008G>T, c.1043_1046delTCAA, c.1061_1063delTCT, c.1073+1G>A, c.1074-1G>T, c.1121A>G, c.1123delG, c.1141delG, c.1146+1G>A, c.116T>G, c.1177C>T, c.1178G>C, c.1211_1212delTG, c.1214_1215delAAinsG, c.1260G>C, c.1274_1277dupTATC, c.1292G>A, c.1302C>G, c.1307_1308delTA, c.1351C>G, c.1360G>A, c.1373G>A, c.1385A>T, c.1421+1G>C, c.1422-2A>G, c.1422G>C, c.1426A>T, c.1432G>A, c.1444G>A, c.1451T>C, c.1453T>C, c.1490A>G, c.1495C>T, c.1496G>A, c.1510C>T, c.1510delC, c.1511G>A, c.1511G>T, c.1528C>T, c.1537C>T, c.155C>A, c.173G>A, c.1A>C, c.1A>T, c.2T>C, c.340G>A, c.346+1G>C, c.380T>G, c.409C>T,

			c.413-2A>G, c.426delT, c.459+5G>A, c.508C>T, c.509G>A, c.532C>T, c.533G>A, c.533G>T, c.535C>T, c.538T>C, c.540C>G, c.570+1G>A, c.570+3A>G, c.571-1G>T, c.571-2A>G, c.574G>C, c.590A>C, c.598G>A, c.607T>G, c.611A>G, c.613delC, c.621T>G, c.623A>T, c.629C>T, c.632T>C, c.672+1G>A, c.718_719insT, c.736G>A, c.749G>A, c.772G>C, c.778C>T, c.78G>A, c.796T>G, c.805+1G>A, c.805+1G>C, c.805+2T>C, c.805G>A, c.806-7G>A, c.902T>G, c.915_917delCTT, c.947dupA, c.962_964delGAG, c.964G>A, c.964G>T, c.972T>A, c.986+3A>G, c.986G>A
HGF	1	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB39	c.495G>A
HGSNAT	17	Mucopolisacariosis tipo 3, Síndrome de Sanfilippo tipo C	c.1030C>T, c.1209G>T, c.1250+1G>A, c.1345dupG, c.1411G>A, c.1445T>A, c.1464+1G>A, c.1553C>T, c.1843G>A, c.234+1G>A, c.370A>T, c.372-2A>G, c.398G>C, c.493+1G>A, c.525dupT, c.848C>T, c.962T>G
HIBCH	8	Neurodegeneración por déficit en 3-hidroxisobutiril-CoA-hidrolasa	c.1033G>A, c.1128dupT, c.129dupA, c.196C>T, c.220-9T>G, c.365A>G, c.79-3C>G, c.950G>A
HLCS	12	Multiple carboxylase deficiency	c.1513G>C, c.1519+5G>A, c.1522C>T, c.1648G>A, c.1711G>A, c.1741G>A, c.1993C>T, c.647T>G, c.655dupA, c.710T>C, c.782delG, c.722G>C
HMGCL	9	Aciduria 3-hidroxi-metil-glutárica	c.109G>T, c.122G>A, c.206_207delCT, c.208G>C, c.505_506delTC, c.561+1G>A, c.698A>G, c.835G>A, c.914_915delTT
HPD	4	Tirosinemia tipo 3	c.1005C>G, c.479A>G, c.600C>G, c.774T>G
HSD17B3	11	Pseudohermafroditismo	c.166G>A, c.238C>T, c.239G>A, c.277+4A>T, c.389A>G, c.607-1G>A, c.608C>T, c.695C>T, c.703A>G, c.803G>A, c.845C>T
HSD3B2	7	Hiperplasia suprarrenal congénita por déficit de 3-beta-hidroxiesteroide deshidrogenasa 2	c.1022C>T, c.1119A>C, c.29C>A, c.424G>A, c.512G>A, c.664C>A, c.776C>T
HYLS1	1	Síndrome HYDROLETHALUS	c.632A>G
IDH3B	2	Retinosis pigmentaria 46	c.395T>C, c.589delA
IDS	62	Mucopolisacaridosis tipo 2	c.1003C>T, c.1006+1G>T, c.1007-8T>G, c.1016T>C, c.1025A>C, c.1033T>A, c.1122C>T, c.1132_1133delTT, c.1148delC, c.1181-1G>A, c.1264T>G, c.1265G>A, c.1327C>T, c.133G>C, c.1393C>T, c.1402C>T, c.1403G>A, c.1403G>C, c.1403G>T, c.1425G>A, c.1433A>G, c.1463delT, c.1466G>C, c.1505G>C, c.1508T>A, c.1591C>T, c.181T>C, c.208dupC, c.240+1G>A, c.241-5A>T, c.253G>A, c.262C>T, c.283A>G, c.314_317dupTCAA, c.349_351delTCC, c.359C>A, c.401G>A, c.404A>G, c.411delT, c.425C>A, c.463_464delTTinsCCGTATAGCTGG, c.469C>T, c.479C>G, c.508-1G>A, c.508-1G>C, c.509_510delCA, c.514C>T, c.587T>C, c.592G>A, c.596_599delAACA, c.613G>C, c.683C>A, c.683C>T, c.708+1G>A, c.754_767delGATCCCGAGGTCCC, c.820G>T, c.832_833insATGTTTAAGGGAAG, c.880-8A>G, c.884A>T, c.908_909delCT, c.935G>A, c.998C>T
IDUA	36	Síndrome de Hurler	c.1029C>A, c.1037T>G, c.1045G>A, c.1091C>T, c.1096A>C, c.1205G>A, c.1402+1G>T, c.1469T>C, c.1475G>C, c.152G>A, c.1598C>G, c.1614delG, c.1650+5G>C, c.1799delC, c.1855C>G, c.1855C>T, c.1861C>T, c.1874A>G, c.1898C>G, c.192C>A, c.1960T>G, c.208C>T, c.223G>A, c.266G>A, c.299+1G>A, c.386-2A>G, c.46_57delTCGCTCCTGGCC, c.494-1G>A, c.501C>G, c.590-7G>A, c.613_617dupTGCTC, c.653T>C, c.713T>A, c.898G>A, c.928C>T, c.972+1G>A
IKBKAP	3	Disautonomia Familiar	c.2087G>C, c.2204+6T>C, c.2741C>T
IL2RG	22	Inmunodeficiencia combinada grave	c.186T>A, c.270-15A>G, c.2T>C, c.314A>G, c.341G>A, c.343T>C, c.355A>T, c.452T>C, c.454+1G>A, c.458T>A, c.664C>T, c.670C>T, c.676C>T, c.677G>A, c.703_711dupCAGCATTGG, c.758-1G>C, c.854G>A, c.854G>A, c.865C>T, c.878T>A, c.923C>A, c.924+1G>A
IMPDH1	2	Amaurosis congénita de leber 11	c.568C>T, c.849T>G
IMPG2	12	Retinosis pigmentaria 56	c.2274G>A, c.2716C>T, c.2890C>T, c.3230G>T, c.3262C>T, c.370T>C, c.379C>T, c.513T>G, c.635C>G, c.829-1G>T, c.2412_2413delTG, c.68dupA
INPP5E	18	Síndrome de Joubert 1	c.1132C>T, c.1304G>A, c.1543C>T, c.1688G>A, c.1879C>T, c.907G>A, c.1760delT, c.1897_1898delCA, c.1021G>A, c.1684A>G, c.1064C>T, c.944C>T, c.1162G>T, c.1754G>A,

			c.1249T>C, c.1154G>A, c.1577C>T, c.1468G>T
IVD	16	Acidemia isovalérica	c.1141T>C, c.1145_1147+4delTTGGTGA, c.1183C>T, c.1188delT, c.1208A>G, c.134T>C, c.157C>T, c.158G>C, c.214G>A, c.367G>A, c.465+2T>C, c.466-2A>G, c.466-3_466-2delCAinsGG, c.605G>T, c.860G>A, c.941C>T
JAK3	10	Inmunodeficiencia combinada grave T-B+ por déficit de JAK3	c.1333C>T, c.1695C>A, c.171_173delTGC, c.1744C>T, c.1765G>A, c.1767C>T, c.299A>G, c.308G>A, c.678_679delCT, c.913C>T
KCNJ13	3	Amaurosis congénita de Leber 16	c.158G>A, c.496C>T, c.722T>C
KIF7	1	Síndrome hydrolethalus 2	c.2896_2897delGC
KIF7	7	Síndrome acrocalloso	c.217delG, c.3001C>T, c.3331C>T, c.460C>T, c.587dupT, c.687delG, c.811delG
KIF7	1	Displasia epifisaria múltiple tipo Al-Gazali	c.3179A>G
KIF7	2	Síndrome de Joubert 12	c.2917C>T, c.2944G>T
L1CAM	25	Hidrocefalia con estenosis del acueducto de Silvio	c.1108G>A, c.1354G>A, c.1417C>T, c.1453C>T, c.1792G>A, c.1939+5G>A, c.2092G>A, c.2254G>A, c.2278C>T, c.2351A>G, c.2421_2422delTG, c.2432-19A>C, c.2974C>T, c.3201T>G, c.3458-1G>C, c.3489_3490delTG, c.3581C>T, c.523+12C>T, c.536T>G, c.551G>A, c.719C>T, c.772C>T, c.791G>A, c.807-6G>A, c.924C>T
LHFPL5	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB67	c.250delC, c.380A>G, c.494C>T
LHX3	6	Déficit de hormonas hipofisarias combinado no adquirido con anomalías de la columna vertebral	c.111delT, c.148A>T, c.302_303delGCinsTCCT, c.347A>G, c.644C>T, c.687G>A
LIG4	7	Síndrome LIG4	c.1271_1275delAAAGA, c.1298_1300delAAC, c.1406G>A, c.1512_1513delTC, c.1738C>T, c.2440C>T, c.833G>A
LMNA	75	Enfermedad de Charcot-Marie-Tooth tipo axonal 2B1	c.1039G>A, c.1057C>T, c.1072G>A, c.1129C>T, c.1130G>A, c.1139T>C, c.1146C>T, c.1196G>A, c.127G>A, c.1357C>T, c.1366A>C, c.1381-2A>G, c.1412G>A, c.1477C>T, c.1488+1G>A, c.1489-2A>G, c.1540T>C, c.1580G>A, c.1580G>C, c.1583C>A, c.1583C>G, c.1585G>A, c.1586C>T, c.1589T>C, c.1608+1G>A, c.1608+5G>C, c.1609-12T>G, c.1609-3C>G, c.1620G>A, c.1621C>T, c.169G>C, c.16C>T, c.176T>G, c.1771T>A, c.1786G>A, c.1961dupG, c.1968+1G>A, c.1968+2T>C, c.1968+5G>A, c.1968G>A, c.250G>A, c.254T>C, c.29C>T, c.348dupG, c.356+1G>C, c.398G>C, c.419T>C, c.419T>G, c.428C>T, c.448A>C, c.485T>C, c.513+1G>A, c.513+45T>G, c.569G>A, c.588_596delGCTGCAGAC, c.618C>G, c.624_626delGAA, c.626delA, c.664C>T, c.667G>A, c.674G>A, c.695G>A, c.745C>T, c.746G>A, c.777T>A, c.781_783delAAG, c.784G>A, c.799T>C, c.82C>G, c.859delG, c.892C>T, c.898G>A, c.899A>G, c.91_93delGAG, c.976T>A
LOXHD1	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB77	c.2008C>T, c.4480C>T, c.4714C>T
LRAT	3	Amaurosis congénita de Leber 14	c.217_218delAT, c.233_242delTGTTGGCCCT, c.525T>A
LRTOMT	6	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB63	c.242G>A, c.313T>C, c.328G>A, c.358+4A>C, c.614_617dupAGCT, c.333C>G
MAK	5	Retinosis pigmentaria 62	c.37G>A, c.388A>C, c.497G>A, c.553G>A, c.718C>T
MAN2B1	15	Alfa-manosidosis	c.1109G>A, c.1383C>G, c.1830+1G>C, c.1831-2A>G, c.1858dupA, c.1915C>T, c.1929G>A, c.2013delT, c.2119C>T, c.2165+1G>A, c.2248C>T, c.2278C>T, c.2368C>T, c.2402dupG, c.2426T>C
MARVELD2	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB49	c.1183-1G>A, c.1331+1G>A, c.1498C>T
MCCC1	16	Déficit de 3-metilcrotonil-CoA carboxilasa tipo 1	c.1114C>T, c.1155A>C, c.1193_1194delTG, c.1310T>C, c.1331G>A, c.137-2A>G, c.1380T>G, c.1526delG, c.1594G>C, c.1594G>C, c.1604C>T, c.2079delA, c.841C>T, c.974T>G, c.980C>G, c.640-2A>G
MCCC2	18	Déficit aislado de 3-metilcrotonil-CoA carboxilasa 2	c.1015G>A, c.1309A>G, c.1309A>G, c.1574+1G>A, c.295G>C, c.464G>A, c.499T>C, c.517dupT, c.518C>T, c.568C>T, c.569A>G,

			c.577C>T, c.735dupC, c.803G>C, c.803G>C, c.838G>T, c.929C>G, c.994C>T
MCEE	3	Acidemia metilmalónica por déficit de metilmalonil-CoA epimerasa	c.139C>T, c.178A>C, c.427C>T
MCOLN1	21	Mucolipidosis tipo 4	c.1084G>T, c.1207C>T, c.1210dupT, c.1222_1224delTTC, c.1336G>T, c.1340T>C, c.1388G>A, c.1395C>G, c.1406A>G, c.1453_1463dupGGCCGCAGCAG, c.1704A>T, c.235C>T, c.302_303delTC, c.304C>T, c.317T>C, c.406-2A>G, c.473_474delCC, c.497G>T, c.514C>T, c.694A>C, c.964C>T
MED25	1	Enfermedad de Charcot-Marie-Tooth tipo 2B2	c.1004C>T
MED25	1	Síndrome de catarata congénita - microcefalia - nevus flamígero simple - discapacidad intelectual grave	c.116A>G
MEFV	16	Fiebre mediterranea	c.1432C>T, c.1437C>G, c.1958G>A, c.2040G>A, c.2040G>C, c.2060G>A, c.2076_2078delAAT, c.2080A>G, c.2081_2083delTGA, c.2082G>A, c.2084A>G, c.2177T>C, c.2230G>T, c.2282G>A, c.501G>C, c.800C>T
MERTK	11	Retinosis pigmentaria 38	c.1450G>A, c.1604+2T>G, c.1605-2A>G, c.1951C>T, c.2070_2074delAGGAC, c.2189+1G>T, c.2214delT, c.2262C>G, c.2323C>T, c.325A>T, c.933_935delCTCinsTT
MFRP	11	Microftalmia - Retinosis pigmentaria - foveosquisis - drusen de disco óptico	c.1124+1G>T, c.1150dupC, c.1549C>T, c.1622_1625delTCTG, c.201G>A, c.491_492insT, c.498delC, c.498dupC, c.523C>T, c.545T>C, c.951C>A
MFSD8	13	Lipofuscinosis ceroidea neuronal 7	c.1102G>C, c.1102G>C, c.1141G>T, c.1235C>T, c.1286G>A, c.1438G>A, c.1444C>T, c.362A>G, c.754+2T>A, c.863+3_863+4insT, c.881C>A, c.894T>G, c.929G>A
MGAT2	5	Trastorno congénito de la glicosilación IIA	c.1017T>A, c.711G>C, c.785A>G, c.869C>T, c.952A>G
MLYCD	5	Aciduria malónica	c.1064_1065delTT, c.119T>C, c.475delG, c.560C>G, c.8G>A
MMAA	18	Acidemia metilmalónica vitamina B12 sensible tipo cbl A	c.1076G>A, c.161G>A, c.266T>C, c.283C>T, c.358C>T, c.387C>A, c.397C>T, c.433C>T, c.503delC, c.562G>C, c.586C>T, c.593_596delCTGA, c.620A>G, c.64C>T, c.650T>A, c.653G>A, c.733+1G>A, c.988C>T
MMAB	17	Acidemia metilmalónica vitamina B12 sensible tipo cbl B	c.197-1G>T, c.287T>C, c.290G>A, c.291-1G>A, c.349-1G>C, c.548A>T, c.556C>T, c.563_577dupTGTGCCGCCGGGCCG, c.568C>T, c.569G>A, c.570_572dupCCG, c.571C>T, c.572G>A, c.584G>A, c.656A>G, c.700C>T, c.403G>A
MMACHC	26	Acidemia metilmalónica con homocistinuria tipo cblC	c.217C>T, c.271dupA, c.276G>A, c.276G>T, c.328_331delAACC, c.331C>T, c.347T>C, c.388_390delTAC, c.388T>C, c.389A>G, c.394C>T, c.3G>A, c.420G>A, c.440G>A, c.440G>C, c.445_446delTG, c.464G>A, c.481C>T, c.482G>A, c.547_548delGT, c.600G>A, c.608G>A, c.609G>A, c.615C>G, c.616C>T, c.658_660delAAG
MMADHC	12	Acidemia metilmalónica con homocistinuria tipo cblD	c.160C>T, c.419dupA, c.455dupC, c.545C>A, c.57_64delCTCTTTAG, c.696+3_696+6delGAGT, c.746A>G, c.748C>T, c.776T>C, c.60_61insAT, c.133dup, c.228dupG
MOGS	4	Trastorno congénito de la glicosilación IIb; CDG2B	c.1457G>C, c.1954T>C, c.329G>A, c.370C>T
MPDU1	4	Trastorno congénito de la glicosilación tipo If	c.218G>A, c.221T>C, c.2T>C, c.356T>C
MPI	6	Trastorno congénito de la glicosilación tipo Ib	c.1253G>A, c.166dupC, c.305C>T, c.413T>C, c.656G>A, c.884G>A
MRPS16	1	Déficit combinado de la fosforilación oxidativa tipo 2	c.331C>T
MRPS22	2	Defecto de la fosforilación oxidativa combinada tipo 5	c.509G>A, c.644T>C
MTHFR	50	Homocistinuria por déficit de metilentetrahidrofolato reductasa	c.1004G>A, c.1015T>G, c.1042C>T, c.1060C>T, c.1088G>A, c.1114A>G, c.1129C>T, c.1166G>A, c.1167-2delA, c.1262G>C, c.1320G>A, c.136C>T, c.137G>A, c.1408G>T, c.1516T>G, c.1530+2T>C, c.1606G>T, c.1632+2T>G, c.1683G>A, c.1724T>G, c.1743G>A, c.1752+1G>T, c.1753-18G>A, c.1768delC, c.176G>C, c.1793T>C, c.1883T>C, c.1969T>C, c.202C>G, c.244C>T, c.337G>A, c.379C>T, c.388T>C, c.440A>C, c.470G>A, c.474A>T, c.523G>A, c.547C>T, c.548G>A, c.587G>A, c.643_645delAAG, c.673A>C, c.677_679delTCA, c.760C>T, c.764G>T, c.767T>A, c.769T>G, c.780+1G>A, c.968T>C, c.971A>G
MTMR2	3	Enfermedad de Charcot-Marie-	c.1276C>T, c.1444C>T, c.826G>T

		Tooth tipo 4B1	
MUT	97	Acidemia metilmalónica por deficiencia de CoA mutasa	c.1022dupA, c.1038_1040delTCT, c.1084-1G>A, c.1084-2A>G, c.1097A>G, c.1105C>T, c.1106G>A, c.1108A>C, c.1130C>A, c.1164T>A, c.1181dupT, c.1181T>A, c.1207C>T, c.1271C>T, c.1277G>A, c.1280G>A, c.129G>A, c.1399C>T, c.1420C>T, c.1481T>A, c.1489G>T, c.1553T>C, c.1560+1G>T, c.1630_1631delGGinsTA, c.1655C>T, c.1658delT, c.1741C>T, c.1853T>C, c.1867G>A, c.1871A>G, c.1874A>G, c.1885A>G, c.1924G>C, c.1975C>T, c.19C>T, c.2054T>G, c.2078delG, c.2080C>T, c.2099T>A, c.2107G>C, c.2150G>T, c.2179C>T, c.2193_2196dupTGCC, c.2194_2197delGCCGinsTGGAA, c.2200C>T, c.278G>A, c.280G>A, c.281G>T, c.284C>G, c.299A>G, c.2T>C, c.30dupA, c.313T>C, c.322C>G, c.329A>G, c.330T>G, c.349G>T, c.378C>A, c.385+5G>A, c.-39-1G>A, c.397G>A, c.415G>A, c.467A>T, c.521T>C, c.52C>T, c.55dupG, c.560C>G, c.566A>T, c.572C>A, c.607G>A, c.613_615delGAA, c.630delA, c.643G>A, c.643G>T, c.655A>T, c.671_678dupAATTTATG, c.682C>T, c.689C>G, c.691T>A, c.692dupA, c.693C>G, c.753+2T>A, c.826G>T, c.828G>C, c.842T>C, c.850G>A, c.851G>A, c.88C>T, c.914T>C, c.91C>T, c.927G>A, c.935G>T, c.970G>A, c.974G>A, c.977G>A, c.982C>T, c.1663G>A
MVK	18	Aciduria mevalonica	c.1000G>A, c.1129G>A, c.1162C>T, c.185G>A, c.346T>C, c.417dupC, c.421dupG, c.442G>A, c.494C>T, c.500C>T, c.59A>C, c.604G>A, c.608T>C, c.72dupT, c.803T>C, c.829C>T, c.902A>C, c.928G>C
MYO15A	33	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB3	c.10573delA, c.1185dupC, c.3311dupG, c.3313G>T, c.3336delG, c.3385C>T, c.3685C>T, c.3756+1G>T, c.3866+1G>A, c.4351G>A, c.5492G>T, c.5925G>A, c.6046+1G>A, c.6331A>T, c.6337A>T, c.6340G>A, c.6437G>A, c.7207G>T, c.7226delC, c.7801A>T, c.7893+1G>A, c.8148G>T, c.8183G>A, c.8767C>T, c.5335delC, c.8100delC, c.7006dupC, c.3944G>A, c.4108C>T, c.4780G>C, c.6146C>A, c.6589C>T, c.6178-2A>G
MYO3A	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB30	c.1193C>A, c.1777-12G>A, c.732-2A>G
MYO6	4	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB37	c.3496C>T, c.647A>T, c.737A>G, c.826C>T
MYO7A	68	Síndrome de Usher tipo 1	c.1097T>C, c.1190C>A, c.1344-2A>G, c.1370C>T, c.1401_1403dupGCA, c.1556G>A, c.1690+1G>A, c.1884C>A, c.1900C>T, c.19-1G>A, c.1952T>C, c.1996C>T, c.2005C>T, c.2115C>A, c.2172delC, c.2187+1G>A, c.223delG, c.2283-1G>T, c.2311G>T, c.2863G>A, c.2904G>T, c.3134T>C, c.3476G>T, c.3504-1G>C, c.3508G>A, c.3591_3592delCT, c.3696_3706delAAGGACCTTTG, c.3719G>A, c.3764delA, c.3827C>A, c.397dupC, c.4018G>C, c.401T>A, c.4115T>G, c.4293G>A, c.4442-2A>C, c.448C>T, c.4544_4551delAGATCATGinsCA, c.470+1G>A, c.494C>T, c.496delG, c.5101C>T, c.5392C>T, c.5573T>C, c.5581C>T, c.5617C>T, c.5618G>A, c.5648G>A, c.5660C>T, c.5886_5888delCTT, c.5945G>A, c.5968C>T, c.6062A>G, c.6070C>T, c.631A>G, c.634C>T, c.635G>A, c.640G>A, c.6557T>C, c.6560G>A, c.700C>T, c.722G>A, c.73G>A, c.77C>A, c.93C>A, c.977T>A, c.999T>G, c.458G>A
MYO7A	15	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB2	c.1184G>A, c.133-2A>G, c.1797G>A, c.1952_1953insAG, c.2023C>T, c.3564_3570delTGCCCGG, c.3596dupT, c.5464A>C, c.5824G>A, c.5899C>T, c.6025G>A, c.620A>G, c.6439-2A>G, c.731G>C, c.287C>T
NAGLU	16	Síndrome de Sanfilippo tipo B	c.1444C>T, c.1562C>T, c.1693C>T, c.1694G>A, c.1694G>C, c.1876C>T, c.1927C>T, c.1928G>A, c.1946G>T, c.1949G>A, c.2021G>A, c.503G>A, c.507_516delCGGCCAGGAG, c.700C>T, c.889C>T, c.942C>G
NDRG1	2	Enfermedad de Charcot-Marie-Tooth tipo 4D	c.442C>T, c.538-1G>A
NEB	9	Miopatía nemalínica congénita grave 2	c.1152+1G>A, c.19944G>A, c.21076C>T, c.2211+5G>A, c.25279G>T, c.2784delT, c.3987+1_3987+2delGTinsTG, c.8031_8041delAAATAAACGAG, c.24770_24771delTT
NEU1	11	Sialidosis tipo 1	c.239C>T, c.272T>G, c.649G>A, c.674G>C, c.69G>A, c.718T>C, c.727G>A, c.779T>A, c.87G>A, c.893C>T, c.946C>T

NMNAT1	10	Amaurosis congénita de Leber 9	c.25G>A, c.451G>T, c.457C>G, c.507G>A, c.53A>G, c.619C>T, c.710G>T, c.817A>G, c.838T>C, c.769G>A
NPC1	63	Enfermedad de Niemann-Pick tipo C1	c.1030delT, c.1042C>T, c.1133T>C, c.1211G>A, c.1502A>T, c.1553G>A, c.1553G>A, c.1628C>T, c.1800delC, c.1947+2T>G, c.2054T>C, c.2072C>T, c.2128C>T, c.2177G>C, c.2196dupT, c.2230_2231delGT, c.2302dupG, c.2324A>C, c.2366G>A, c.2621A>T, c.2665G>A, c.2761C>T, c.2764C>T, c.2777C>T, c.2783A>C, c.2795dupA, c.2801G>A, c.2819C>T, c.2842G>A, c.2848G>A, c.2861C>T, c.2873G>A, c.2893C>T, c.2912-3C>G, c.2932C>T, c.2972_2973delAG, c.2974G>A, c.2974G>C, c.2974G>T, c.3019C>G, c.3042-2delA, c.3104C>T, c.3160G>A, c.3175C>T, c.3182T>C, c.3263A>G, c.3265G>A, c.337T>C c.3425T>C, c.3467A>T, c.352_353delAG, c.3557G>A, c.3591+1G>A, c.3611_3614delTTAC, c.3614C>A, c.3639G>C, c.3662delT, c.416dupC, c.530G>A, c.743G>T, c.813_815delCAT, c.3229C>T, c.1142G>A
NPC2	14	Enfermedad de Niemann-Pick tipo C2	c.115G>A, c.133C>T, c.141C>A, c.190+5G>A, c.199T>C, c.27delG, c.295T>C, c.332delA, c.352G>T, c.358C>T, c.3G>C, c.436C>T, c.58G>T, c.82+2T>C
NPHP1	4	Nefronoptisis	c.1027G>A, c.1884+1G>T, c.555dupA, c.80T>A
OCA2	21	Albinismo oculocutáneo tipo 2	c.1001C>T, c.1025A>G, c.1044+1G>T, c.1182G>A, c.1211C>T, c.1327G>A, c.1364+1G>A, c.1427A>G, c.1441G>A, c.1465A>G, c.1503+5G>A, c.157delA, c.1610A>T, c.1842+1G>T, c.1960delG, c.2037G>C, c.2207C>T, c.2228C>G, c.2344G>A, c.2359G>A, c.79G>A
OPA3	3	Aciduria 3-metilglutacónica tipo 3	c.143-1G>C, c.322_339delCAGCGCCACAAGGAGGAG, c.415C>T
OTC	318	Déficit de ornitina transcarbamilasa	c.1005+1G>A, c.1005+1G>T, c.1005+2T>C, c.1005G>A, c.1006-1G>A, c.1006-3C>G, c.1006G>T, c.1009G>C, c.1015G>C, c.1018T>C, c.1022T>C, c.1028C>A, c.1033T>C, c.1033T>G, c.1034A>G, c.1039C>A, c.1042C>T, c.1061T>G, c.106C>T, c.115G>T, c.116G>A, c.118C>T, c.119G>A, c.122A>G, c.127C>T, c.131C>T, c.133C>G, c.134T>C, c.140A>C, c.140A>T, c.140delA, c.140dupA, c.143T>C, c.145A>C, c.148G>A, c.148G>T, c.154G>A, c.154G>T, c.155A>G, c.156A>T, c.158T>C, c.158T>G, c.163T>G, c.167T>C, c.170T>A, c.174G>A, c.179C>T, c.188T>C, c.1A>G, c.1A>T, c.200T>G, c.205C>T, c.216+1G>A, c.216+1G>T, c.217-1G>A, c.227T>C, c.231G>T, c.232C>T, c.236G>A, c.238A>G, c.240G>T, c.243_245delCTT, c.245T>G, c.247G>C, c.248G>A, c.254T>G, c.259G>A, c.264A>T, c.268A>G, c.269G>A, c.270T>G, c.271delA, c.274C>G, c.274C>T, c.275G>A, c.275G>C, c.275G>T, c.277A>G, c.281G>C, c.284T>C, c.29_32delACAA, c.292G>A, c.298+1_298+5delGTAAG, c.298+1G>A, c.298+1G>T, c.298+5G>C, c.299G>A, c.2T>C, c.305C>A, c.314G>T, c.316G>A, c.317G>A, c.317G>T, c.330delT, c.350A>G, c.350A>T, c.359_360delTG, c.364_365insTT, c.376delG, c.377A>G, c.386+1G>A, c.386+1G>T, c.386+2T>C, c.386G>A, c.386G>C, c.386G>T, c.387-2A>C, c.387-2A>G, c.387-2A>T, c.390_392dupATT, c.392T>C, c.394T>C, c.395C>T, c.3G>A, c.403delG, c.404C>A, c.407A>T, c.409G>A, c.416T>C, c.418G>C, c.421C>G, c.421C>T, c.422G>A, c.422G>C, c.425T>A, c.42delT, c.430A>T, c.437C>G, c.443T>C, c.443T>G, c.444G>C, c.444G>T, c.451delC, c.452T>G, c.455C>T, c.460G>T, c.463G>C, c.463G>T, c.464C>A, c.476T>C, c.479T>A, c.479T>C, c.479T>G, c.481A>G, c.482A>G, c.484G>A, c.484G>C, c.485G>A, c.490T>C, c.491C>G, c.493G>T, c.501C>A, c.501C>G, c.503A>C, c.503A>G, c.504T>A, c.505C>G, c.506C>T, c.514A>T, c.516C>G, c.520G>C, c.524A>G, c.524A>T, c.526T>C, c.527A>G, c.532_537delACGCTC, c.533C>T, c.536T>C, c.53delA, c.540+1G>C, c.540+2T>A, c.540+2T>C, c.540G>C, c.541-2A>G, c.542A>G, c.545A>T, c.547T>G, c.548A>G, c.562_563delGG, c.562G>C, c.563G>T, c.571C>T, c.576C>G, c.577T>C, c.577T>G, c.578G>A, c.583G>A, c.586delG, c.586G>A, c.586G>T, c.587A>T, c.589G>A, c.589G>T, c.590G>A, c.593A>T, c.594C>A, c.595A>G, c.596A>G, c.602T>C, c.604C>T, c.605A>C, c.608C>G, c.613A>G, c.617T>G, c.618G>C, c.620G>A, c.621C>A, c.622G>A, c.626C>T, c.628A>C, c.640C>T, c.643C>T, c.645dupT, c.646C>G, c.650C>A, c.658C>G, c.659C>T, c.663+1G>A, c.663+1G>T, c.663+2T>C, c.663G>A, c.663G>C, c.664-1G>A, c.665delG, c.673C>A, c.674C>G, c.674C>T, c.67C>T, c.698C>T, c.700G>T, c.716A>G, c.716A>T, c.717+1G>A, c.717+1G>T, c.717+2T>C, c.717+3A>G, c.717G>A, c.717G>C,

			c.718-2A>G, c.725C>T, c.731_739delTGTTGCTGA, c.731T>A, c.740C>A, c.757G>A, c.757G>C, c.759delA, c.764A>C, c.77+1G>A, c.77+1G>T, c.77+3_77+6delAAGT, c.77+4A>C, c.77+5G>A, c.779T>C, c.77G>A, c.77G>C, c.78-1G>C, c.78-3C>G, c.785C>A, c.785C>T, c.787G>A, c.788A>G, c.790A>G, c.791C>A, c.791C>T, c.793T>C,c.794G>T, c.795G>A, c.799A>C, c.803T>C, c.806G>A, c.808C>T, c.809A>C, c.817_819delGAG, c.818delA, c.829C>T, c.830G>A, c.830G>T, c.835C>T, c.867+1G>A, c.867+1G>T, c.867G>T, c.876delA, c.882delT, c.890_893delACTG, c.892_893delTG, c.893G>C, c.903A>T, c.904C>T, c.905A>G, c.905A>T, c.906C>G, c.906delC, c.907T>C, c.907T>G, c.908G>A, c.912G>T, c.914C>A, c.914C>G, c.928G>T, c.929A>G, c.931G>A, c.941_943delAAG, c.943G>T, c.944T>A, c.944T>G, c.947T>C, c.94C>T, c.953C>T, c.958C>T, c.959G>T, c.962C>A, c.976G>A, c.982G>T, c.988A>G, c.991A>T, c.994T>A, c.995G>A, c.996G>A
OTOA	2	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB22	c.1352G>A, c.1879C>T
OTOF	64	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB9	c.1103_1104delGGinsC, c.1172delA, c.1180dupG, c.1194T>A, c.1236delC, c.1273C>T, c.1469C>A, c.1544T>C, c.1601delC, c.1607G>A, c.1609delG, c.1621G>A, c.1651delG, c.1718T>G, c.1740delC, c.1780G>A, c.1841G>A, c.1886dupA, c.1966delC, c.2122C>T, c.2239G>T, c.2316C>A, c.2348delG, c.2381G>A, c.2485C>T, c.2649C>A, c.2684_2685delGG, c.2732_2735dupAGCT, c.2887C>T, c.2891C>A, c.2905_2923delGCCCGCAGCCTCTTTGCCGinsCTCCGAGCGCA, c.2977_2978delAG, c.3032T>C, c.3239G>C, c.3269C>A, c.3400C>T, c.3413T>C, c.4227+1G>T, c.4275G>A, c.4351G>T, c.4483C>T, c.4491T>A, c.4559G>A, c.4718T>C, c.4799+1G>T, c.4809C>G, c.5011dupT, c.5098G>C, c.5197G>A, c.5384T>G, c.5410_5412delGAG, c.5473C>G, c.5567G>A, c.5575_5576delAA, c.5713-2A>G, c.5800dupC, c.5815C>T, c.5816G>A, c.5960C>G, c.709C>T, c.765G>C, c.897+1G>T, c.5332G>T, c.5332G>T
OXCT1	5	Déficit de succinil-CoA: 3 cetoácido CoA transferasa	c.1367G>T, c.656G>A, c.661G>A, c.848C>G, c.971G>A
PAH	182	Fenilcetonuria	c.1030G>A, c.1033G>T, c.1042C>G, c.1045T>C, c.1065+1G>T, c.1065+3A>G, c.1066-11G>A, c.1066-2A>T, c.1066-3C>T, c.1068C>A, c.1068C>G, c.1076C>G, c.1089delG, c.1092_1094delTCT, c.1092_1106delTCTCCCCCTGGAGCT, c.1099delC, c.110T>C, c.1129delT, c.1139C>T, c.1157A>G, c.116_118delTCT, c.1162G>A, c.1163T>C, c.1166delC, c.1169A>G, c.1172G>C, c.117C>G, c.1180G>T, c.1184C>A, c.1184C>G, c.1197A>T, c.1199+17G>A, c.1199+1G>A, c.1199+1G>C, c.1200-1G>A, c.1208C>T, c.1217T>C, c.1220C>T, c.1222C>T, c.1223G>A, c.1238G>C, c.1241A>G, c.1243G>A, c.1301C>A, c.1315+1G>A, c.1315+2T>C, c.1355dupA, c.1357_*2delTAAAG, c.136G>A, c.140C>T, c.143T>C, c.157C>T, c.164T>C, c.165T>G, c.168_168+1delGGinsAA, c.169G>T, c.194T>C, c.196G>T, c.199T>C, c.1A>G, c.204A>T, c.227A>G, c.232G>A, c.241_256delACCCATTTGGATAAAC, c.242C>A, c.250G>T, c.261C>A, c.284_286delTCA, c.293T>C, c.2T>G, c.311C>A, c.320A>G, c.331C>T, c.350delC, c.355C>T, c.385G>T, c.3G>A, c.400C>T, c.441+1G>A, c.441+5G>T, c.441+6T>A, c.442-1G>A, c.442-2A>C, c.442-5C>G, c.47_48delCT, c.472C>T, c.473G>A, c.482T>C, c.490A>G, c.503delA, c.504C>A, c.505C>T, c.506G>A, c.508C>G, c.509+1G>A, c.526C>T, c.527G>T, c.529G>A, c.533A>G, c.547_548delGAinsTT, c.561G>A, c.569T>C, c.569T>G, c.580_581delCT, c.581T>C, c.58C>T, c.591G>C, c.60+5G>A, c.60+5G>T, c.611A>G, c.632C>T, c.638T>C, c.648C>G, c.662A>G, c.664_665delGA, c.665A>G, c.673C>G, c.682G>A, c.682G>T, c.688G>A, c.691T>C, c.721C>T, c.722delG, c.722G>A, c.722G>T, c.727C>T, c.728G>A, c.731C>T, c.733G>C, c.734T>C, c.737C>A, c.740G>T, c.745C>Tc.754C>Tc.755G>A, c.764T>C, c.770G>T, c.776C>T, c.781C>G, c.781C>T, c.782G>A, c.788T>C, c.796A>C, c.800A>G, c.800A>T, c.806delT, c.809G>A, c.812A>T, c.814G>T, c.818C>T, c.823C>T, c.829T>G, c.836C>T, c.837delC, c.838G>A, c.841C>G, c.842+2T>A, c.842+5G>A, c.842C>T, c.847A>T, c.856G>A, c.869A>T,c.887A>G, c.890G>A, c.896T>G, c.898G>T,

			c.912+1G>A, c.912+2T>C, c.913-2A>G, c.913-7A>G, c.913-8A>G, c.916A>G, c.916delA, c.926C>A, c.926C>T, c.929C>A, c.932T>C, c.935G>T, c.965C>G, c.974A>G, c.977G>A, c.997C>T
PANK2	14	Neurodegeneración asociada a pantotenato-quinasa	c.1051T>C, c.1211A>T, c.1412G>A, c.1413-1G>T, c.1442_1444delGAG, c.1561G>A, c.1583C>T, c.533C>A, c.570C>G, c.700A>G, c.790C>T, c.832C>T, c.856C>T, c.930_936delCTTTTGT
PANK2	3	Síndrome de HARP	c.1292T>C, c.1310T>C, c.1441C>T
PCCA	39	Acidemia propiónica (gen PCCA)	c.105+1G>A, c.1118T>A, c.1190_1193delAATG, c.1192T>C, c.1196G>A, c.1268C>T, c.1284+1G>A, c.134_135delTA, c.1426C>T, c.1598_1601delTTGT, c.1643+1G>A, c.1644-6C>G, c.1676G>T, c.1685C>G, c.1746G>A, c.1747-1G>C, c.183delA, c.184-1G>A, c.1891G>C, c.1899+4_1899+7delAGTA, c.2002G>A, c.2062T>C, c.229C>T, c.284A>G, c.412G>A, c.425G>A, c.431G>T, c.467T>A, c.491T>C, c.548T>G, c.590G>A, c.600+1G>T, c.775_779delCTAAT, c.862A>G, c.863G>A, c.878A>G, c.890A>G, c.923dupT, c.937C>T
PCCB	30	Acidemia propiónica (gen PCCB)	c.1173dupT, c.1210G>A, c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA, c.1228C>T, c.1283C>T, c.1304A>G, c.1398+1G>T, c.1495C>T, c.1498+2T>C, c.1534C>T, c.1538_1540dupCCC, c.1540C>T, c.1556T>C, c.1606A>G, c.183+5G>A, c.280G>T, c.331C>T, c.335G>A, c.337C>T, c.372+2T>C, c.418_429dupAAGATCTGCAAA, c.457G>C, c.493C>T, c.502G>A, c.562G>A, c.683C>T, c.737G>T, c.764-2delA, c.942C>A, c.990dupT
PCDH15	12	Síndrome de Usher tipo 1F	c.1006C>T, c.1088delT, c.158-1G>A, c.1737C>G, c.1927C>T, c.1940C>G, c.2971C>T, c.3717+1G>A, c.3718-2A>G, c.400C>T, c.4257delA, c.7C>T
PCDH15	5	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB	c.1583T>A, c.16delT, c.2367_2369delTGT, c.400C>G, c.785G>A
PDE6A	4	Retinosis pigmentaria 43	c.1032C>A, c.1683G>A, c.1749C>G, c.2053G>A
PDE6B	16	Retinosis pigmentaria 40	c.1060-1G>T, c.1467+1G>C, c.1488delC, c.1540delC, c.1580T>C, c.1591C>T, c.1604T>A, c.1669C>T, c.1678C>T, c.1876G>T, c.1920+2T>C, c.2193+1G>A, c.2419T>A, c.772C>A, c.892C>T, c.2193+1G>T
PDSS1	1	Sordera - encefaloneuropatía - obesidad - valvulopatía	c.924T>G
PEX7	18	Condrodisplasia punctata rizomélica tipo 1	c.120C>G, c.13_19dupTGCGGTG, c.188+1G>C, c.340-10A>G, c.345T>G, c.400G>A, c.40A>C, c.45_52dupGGGACGCC, c.618G>A, c.649G>A, c.653C>T, c.694C>T, c.722A>T, c.74C>T, c.854A>G, c.875T>A, c.903+1G>C, c.-45C>T
P3H1	8	Osteogenesis imperfecta, tipo VIII	c.2055+18G>A, c.1656C>A, c.1473+1G>T, c.1365_1366delAGinsC, c.1102C>T, c.1080+1G>T, c.747delC, c.392C>A
PKHD1	74	Poliquistosis renal autosómica recesiva	c.10174C>T, c.10219C>T, c.10364delC, c.10402A>G, c.10412T>G, c.10444C>T, c.10452dupT, c.10505A>T, c.10637delT, c.10658T>C, c.10664T>A, c.107C>T, c.10856delA, c.10865G>A, c.11284C>A, c.11524C>T, c.11611T>C, c.11612G>A, c.1342G>C, c.1409G>A, c.1458C>A, c.1480C>T, c.1486C>T, c.1529delG, c.1880T>A, c.2269A>C, c.2279G>A, c.2341C>T, c.2414C>T, c.2747A>C, c.2810G>A, c.2854G>A, c.3229-2A>C, c.3367G>A, c.3528dupC, c.353delG, c.370C>T, c.3747T>G, c.3761_3762delCCinsG, c.3766delC, c.383delC, c.4165C>A, c.4220T>G, c.4415G>A, c.4870C>T, c.50C>T, c.5221G>A, c.5498C>T, c.5513A>G, c.5750A>G, c.5895dupA, c.5984A>G, c.657C>T, c.664A>G, c.6992T>A, c.711_714delAATG, c.7350+653A>G, c.7916C>A, c.8011C>T, c.8063G>T, c.8317G>T, c.8407T>C, c.8518C>T, c.8824C>T, c.8829dupC, c.8870T>C, c.9053C>T, c.9107T>G, c.9319C>T, c.9370C>T, c.9530T>C, c.9689delA, c.982C>T, c.707+1G>A
PLOD1	17	Síndrome de Ehlers-Danlos tipo 6	c.1336T>G, c.145C>T, c.1533C>G, c.153dupC, c.1594_1596delGAG, c.1651-2delA, c.1836G>C, c.1999G>A, c.2008C>T, c.2032G>A, c.2117A>G, c.426T>A, c.467-2delA, c.579+1G>A, c.955C>T, c.975+2_975+3insTT, c.979C>T
PLP1	7	Paraplegia espástica tipo 2	c.388C>T, c.409C>T, c.418C>T, c.434G>A, c.509C>T, c.560T>C, c.710T>C
PLP1	17	Enfermedad de Pelizaeus-Merzbacher	c.128C>T, c.169G>T, c.1A>G, c.220G>A, c.2T>C, c.3G>A, c.44C>T, c.467C>T, c.487T>C, c.544A>C, c.607G>C, c.646C>T,

			c.655G>T, c.661G>T, c.671T>C, c.683G>A, c.737G>C
PMM2	35	Trastorno congénito de la glicosilación tipo 1a	c.131T>C, c.193G>T, c.1A>G, c.24delC, c.255+2T>C, c.256-1G>C, c.26G>A, c.317A>T, c.323C>T, c.338C>T, c.349G>C, c.357C>A, c.368G>A, c.385G>A, c.395T>C, c.415G>A, c.422G>A, c.442G>A, c.470T>C, c.484C>T, c.53C>G, c.563A>G, c.620T>C, c.623G>C, c.647A>T, c.652C>G, c.653A>T, c.669C>G, c.677C>G, c.691G>A, c.710C>G, c.710C>T, c.722G>C, c.95_96delTAinsGC, c.95T>G
POLR1C	3	Síndrome de Treacher-Collins	c.835C>T, c.836G>A, c.979A>T
POLR1C	6	Leucodistrofia, hipomielinización 11	c.221A>G, c.326G>A, c.436T>C, c.77C>T, c.883_885delAAG, c.95A>T
POMT1	24	Síndrome de Walker-Warburg	c.1153C>T, c.1241C>T, c.1260_1262delCCT, c.1261_1262delCT, c.132A>C, c.1540C>T, c.1544dupA, c.1746G>C, c.1770G>C, c.1864C>T, c.193G>A, c.1958C>T, c.2005G>A, c.2110dupG, c.2163C>A, c.2167dupG, c.2179_2180delT, c.226G>A, c.418_420delATG, c.430A>G, c.598G>C, c.793C>T, c.907C>T, c.1958C>A
POMT2	15	Síndrome de Walker-Warburg (gen POMT2)	c.1006+1G>A, c.1006+5G>A, c.1057G>A, c.1117G>T, c.1261delC, c.1445G>T, c.1912C>T, c.1941G>A, c.1997A>G, c.2177G>A, c.2242T>C, c.2243G>C, c.248+5G>C, c.593T>A, c.737G>A
POU3F4	15	Sordera mixta ligada al X con gusher perilinfático	c.1000A>G, c.1060delA, c.235C>T, c.341G>A, c.499C>T, c.604A>T, c.607_610delCAAA, c.853_854delAT, c.862_865delAGTG, c.896delA, c.935C>T, c.950dupT, c.950T>G, c.967C>G, c.990A>T
PPT1	64	Lipofuscinosis ceroidea, neuronal 1, CLN1	c.114delG, c.114G>A, c.114G>T, c.117T>A, c.124+1G>A, c.125-15T>G, c.125-2A>G, c.125G>A, c.133T>C, c.134G>A, c.135_137dupTTG, c.163A>T, c.169dupA, c.175delG, c.223A>C, c.234+1G>A, c.235-3T>C, c.236A>G, c.255_257delCTT, c.271_287delCAAGTAACAACAGTGTGinsTT, c.272A>C, c.287G>A, c.29T>A, c.310A>T, c.322G>C, c.325T>G, c.363-3T>G, c.364A>T, c.398delT, c.3G>A, c.413C>T, c.451C>T, c.455G>A, c.456C>A, c.490C>T, c.529C>G, c.533A>T, c.536+1G>A, c.536+2T>C, c.538dupC, c.541G>A, c.541G>T, c.544C>T, c.550G>A, c.558G>A, c.560A>G, c.566C>G, c.627+1G>A, c.628-1G>T, c.644delA, c.656T>A, c.665T>C, c.674T>C, c.683T>G, c.707T>A, c.713C>T, c.727-2A>T, c.739T>C, c.749G>T, c.774dupA, c.871C>T, c.886T>C, c.888G>A, c.914T>C
PRCD	4	Retinosis pigmentaria 36	c.2T>C, c.52C>T, c.5G>A, c.64C>T
PROM1	8	Retinosis pigmentaria 41	c.1354dupT, c.1557C>A, c.1726C>T, c.1841delG, c.2077-521A>G, c.604C>G, c.2309delC, c.1697delA
PROP1	20	Panhipopituitarismo	c.109+1G>T, c.112_124delTCGAGTGCTCCAC, c.150_151delAG, c.150delA, c.157delA, c.217C>T, c.218G>A, c.247C>T, c.263T>C, c.295C>T, c.296G>A, c.2T>C, c.301_302delAG, c.310delC, c.334C>T, c.349T>A, c.358C>T, c.373C>T, c.466dupT, c.582G>A
PRX	6	Enfermedad de Charcot-Marie-Tooth tipo 4F	c.1951G>A, c.2098delG, c.2145T>A, c.2553_2556delTCTC, c.3208C>T, c.586C>T
PRX	6	Enfermedad de Dejerine-Sottas	c.1102C>T, c.1174C>T, c.2289delT, c.247delC, c.2787delC, c.2857C>T
PRPS1	5	Enfermedad de Charcot-Marie-Tooth ligada al X tipo 5	c.129A>C, c.344T>C, c.362C>G, c.46T>C, c.830A>C
PRPS1	1	Síndrome Arts	c.398A>C
PRPS1	7	Sordera neurosensorial no sindrómica ligada al X tipo DFN	c.193G>A, c.259G>A, c.337G>T, c.343A>G, c.869T>C, c.916G>A, c.925G>T
PRPS1	7	Hiperactividad de la fosforribosilpirofosfato-sintetasa	c.154G>C, c.341A>G, c.385C>A, c.424G>C, c.547G>C, c.569C>T, c.579C>G
PSAP	2	Enfermedad de Gaucher atípica	c.1145G>T, c.1288C>T
PSAP	6	Leucodistrofia metacromática debida al deficit de saposina B	c.1046T>C, c.1144T>G, c.1A>T, c.643A>C, c.650C>T, c.722G>C
RAG1	12	Inmunodeficiencia combinada con granulomas en la piel	c.1438A>G, c.2005G>A, c.2164G>A, c.2320G>T, c.2326C>T, c.2333G>A, c.2814T>G, c.2923C>T, c.2942A>C, c.322C>T, c.555delG, c.940C>T
RAG1	12	Síndrome de Omenn	c.1186C>T, c.1187G>A, c.1286A>G, c.1566G>T, c.1681C>T, c.1682G>A, c.2210G>A, c.2521C>T, c.256_257delAA, c.2735A>G, c.775delA, c.983G>A
RAG2	12	Inmunodeficiencia combinada con granulomas en la piel	c.104G>C, c.115A>G, c.1352G>C, c.1403_1406delATCT, c.1433G>A, c.217C>T, c.230C>A, c.283G>A, c.374_375delCA, c.547T>C, c.644C>T, c.686G>A
RAG2	5	Síndrome de Omenn	c.123C>G, c.1247G>T, c.1504A>G, c.685C>T, c.854T>G

RAX	3	Anoftalmia - microftalmia aisladas 3	c.439C>T, c.575G>A, c.909C>G
RDH12	18	Amaurosis congénita de Leber 13	c.146C>T, c.152T>A, c.164C>T, c.184C>T, c.295C>A, c.377C>T, c.379G>T, c.451C>A, c.451C>G, c.464C>T, c.523T>C, c.565C>T, c.63_66delCATC, c.658+1G>A, c.677A>G, c.688C>G, c.778delG, c.806_810delCCCTG
RDX	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB24	c.1405dupG, c.1732G>A, c.463C>T
RFT1	7	Trastorno congénito de la glicosilación tipo In	c.1222A>G, c.1325G>A, c.199C>T, c.454A>G, c.887T>A, c.887T>G, c.892G>A
RGR	2	Retinosis pigmentaria 44	c.196A>C, c.454C>A
RHO	47	Retinosis pigmentaria 4	c.1030C>T, c.1033G>A, c.1033G>C, c.1039C>T, c.1040C>A, c.1040C>G, c.1040C>T, c.151G>C, c.152G>C, c.158C>G, c.173C>G, c.173C>T, c.260T>A, c.266G>A,c.269G>A, c.281C>T, c.316G>A, c.316G>T, c.329G>A, c.341G>A, c.403C>T, c.404G>T, c.448G>A, c.44A>G, c.482G>A, c.491C>A, c.50C>T, c.511C>T, c.520G>A, c.533A>G, c.541G>A, c.544G>A, c.562G>A, c.568G>A, c.568G>T, c.569A>G, c.620T>A, c.632A>C, c.659T>G, c.67C>G, c.68C>A, c.745G>T, c.792_794delCTG, c.800C>T, c.875C>A, c.886A>G, c.979_982delCCAC
RP2	9	Retinosis pigmentaria 2	c.16_18delTCC, c.2T>C, c.353G>A, c.353G>T, c.358C>T, c.453C>G, c.453delC, c.76C>T, c.507delT
RPE65	9	Retinosis pigmentaria 20	c.1022T>C, c.1087C>A, c.11+5G>A, c.1102T>C, c.118G>A, c.1355T>G, c.1366delG, c.1543C>T, c.271C>T
RPE65	6	Amaurosis congénita de Leber 2	c.1067delA, c.1292A>G, c.272G>A, c.700C>T, c.907A>T, c.952T>A
RPGR	11	Retinosis pigmentaria 3	c.155-2A>G, c.173_175delTGTinsG, c.179G>T, c.296C>A, c.389T>G, c.469+1G>T, c.517G>C, c.703C>T, c.823G>A, c.1928C>G, c.5203delC
RPGRIP1L	17	Síndrome de Joubert 7	c.1177G>A, c.1721delA, c.1843A>C, c.1975T>C, c.2030C>T, c.2050C>T, c.2083G>C, c.2269delA, c.2305-1G>A, c.2413C>T, c.697A>T, c.757C>T, c.1132delT, c.1709dupA, c.3529C>T, c.1243+1G>A, c.3701+1G>T
RPGRIP1L	5	Síndrome de Meckel 5	c.1033C>T, c.1829A>C, c.2614C>T, c.394A>T, c.723_726delTGAA
SAG	5	Enfermedad de Oguchi	c.523C>T, c.577C>T, c.874C>T, c.916G>T, c.926delA
SBDS	10	Síndrome de Shwachman-Diamond	c.120delG, c.183_184delTAinsCT, c.24C>A, c.258+1G>C, c.258+2T>C, c.297_300delAAGA, c.377G>C, c.505C>T, c.624+1G>C, c.652C>T
SBF2	3	Enfermedad de Charcot-Marie-Tooth tipo 4B2	c.1459C>T, c.2875C>T, c.3586C>T
SEMA4A	2	Retinosis pigmentaria 35	c.1033G>C, c.1049T>G
SERPINA1	19	Déficit de alfa-1 antitripsina	c.1096G>A, c.1145T>G, c.1158delC, c.1177C>T, c.194T>C, c.227_229delTCT, c.230C>T, c.250G>A, c.272G>A, c.347T>A, c.415G>A, c.514G>A, c.538C>T, c.552C>G, c.552delC, c.646+1delG, c.745G>C, c.839A>T, c.863A>T
SGSH	20	Mucopolisacaridosis tipo 3	c.1027dupC, c.1080delC, c.1105G>A, c.1167C>A, c.1298G>A, c.130G>A, c.1339G>A, c.197C>G, c.220C>T, c.235A>C, c.364G>A, c.383C>T, c.416C>T, c.449G>A, c.617G>C, c.675C>G, c.734G>A, c.752G>C, c.877C>T, c.892T>C
SH3TC2	25	Enfermedad de Charcot-Marie-Tooth tipo 4C	c.1178-1G>A, c.1384G>T, c.1747_1748delAG, c.1969G>A, c.1972C>T, c.1982T>C, c.1A>G, c.217_227delGCTGCTCGGAGinsCCAGTAA, c.2491_2492delAG, c.2642A>G, c.2710C>T, c.279G>A, c.2829T>G, c.2860C>T, c.28delG, c.3154C>T, c.3303delG, c.3325C>T, c.3326G>C, c.3341delC, c.3512G>T, c.3601C>T, c.3676-1G>A, c.530-2A>G, c.920G>A
SLC22A5	35	Déficit en la captación de carnitina	c.1051T>C, c.1193C>T, c.1195C>T, c.1196G>A, c.1202dupA, c.1304delG, c.1319C>T, c.1324_1325delGCinsAT, c.1340A>G, c.136C>T, c.1400C>G, c.1403C>G, c.1433C>T, c.1463G>A, c.1556_1559dupACAC, c.248G>T, c.34G>A, c.364G>T, c.396G>A, c.3G>T, c.424G>T, c.43G>T, c.458_459delTG, c.505C>T, c.506G>A, c.51C>G, c.632A>G, c.641C>T, c.695C>T, c.760C>T, c.769C>T, c.844C>T, c.845G>A, c.865C>T, c.95A>G
SLC25A13	12	Citrulinemia tipo 2 forma adulta	c.1177+1G>A, c.1311+1G>A, c.1592G>A, c.15G>A, c.1799dupA, c.1801G>A, c.1801G>T, c.1813C>T, c.495delA, c.615+1G>C, c.674C>A, c.852_855delTATG
SLC25A13	6	Citrulinemia tipo 2 forma neonatal	c.1078C>T, c.1177G>A, c.1763G>A, c.550C>T, c.615+5G>A, c.775C>T

SLC25A15	17	Hiperornitinemia - hiperamonemia - homocitrulinuria	c.110T>G, c.212T>A, c.337G>T, c.446delG, c.535C>T, c.538G>A, c.562_564delTTC, c.564C>G, c.569G>A, c.658G>A, c.79G>A, c.815C>T, c.818T>A, c.823C>T, c.824G>A, c.847C>T, c.95C>G
SLC25A20	7	Déficit de carnitina-acilcarnitina translocasa	c.106-2A>T, c.199-10T>G, c.496C>T, c.576G>A, c.713A>G, c.84delT, c.897dupC
SLC26A2	5	Atelosteogénesis tipo 2	c.1535C>A, c.2144C>T, c.391delC, c.764G>A, c.835C>T
SLC26A2	5	Acondrogénesis tipo 1B	c.1020_1022delTGT, c.1273A>G, c.2033G>T, c.451delT, c.532C>T
SLC26A2	22	Displasia epifisaria múltiple tipo 4	c.1157C>T, c.1242_1245delAAAC, c.1361A>C, c.1394delT, c.1451G>A, c.1650delG, c.1724delA, c.1957T>A, c.1976delT, c.1983delA, c.255delC, c.-26+2T>C, c.331G>T, c.398C>T, c.403C>A, c.469T>A, c.47C>G, c.55G>T, c.700-1G>C, c.705_711delGATGGGC, c.767T>C, c.906_907delCT
SLC26A4	32	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB4	c.1001+1G>T, c.1105A>G, c.1115C>T, c.1174A>T, c.1264-1G>C, c.1336C>T, c.1454C>T, c.1489G>A, c.1520delT, c.1547dupC, c.1586T>G, c.164+1delG, c.165-2A>G, c.1707+5G>A, c.1919G>A, c.1975G>C, c.2027T>A, c.2048T>C, c.2086C>T, c.2162C>T, c.235C>T, c.269C>T, c.281C>T, c.304+2T>C, c.365dupT, c.563T>C, c.58T>C, c.716T>A, c.765+3A>C, c.84C>A, c.916dupG, c.918+2T>C
SLC26A4	27	Síndrome de Prended	c.1001+1G>A, c.1003T>C, c.1079C>T, c.1151A>G, c.1198delT, c.1226G>A, c.1229C>T, c.1246A>C, c.1263+1G>A, c.1284_1286delTGC, c.1334T>G, c.1540C>A, c.1920G>A, c.2000T>G, c.2127delT, c.2168A>G, c.279delT, c.397T>A, c.3G>C, c.412G>T, c.554G>C, c.626G>T, c.68C>A, c.707T>C, c.85G>C, c.890delC, c.919-2A>G
SLC26A5	1	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB61	c.209G>A
SLC35A1	1	Trastorno congénito de la glicosilación IIf; CDG2F	c.277_280delGTGCinsTG
SLC35C1	4	Trastorno congénito de la glicosilación IIc; CDG2C	c.439C>T, c.503_505delTCT, c.91G>T, c.923C>G
SLC45A2	8	Albinismo oculocutáneo tipo 4	c.1082T>C, c.1121delT, c.1273delC, c.1457C>T, c.469G>A, c.563G>T, c.662_664delTCT, c.986delC
SLC6A19	2	Síndrome de Hartnup	c.517G>A, c.718C>T
SMN1	3	Atrofia muscular espinal tipo I	c.835G>T, c.836G>T, c.*3+80T>G deleción Exón 7
SMPD1	53	Enfermedad de Niemann-Pick tipo A y B	c.1076C>A, c.1092-1G>C, c.1111_1112delCT, c.1117C>T, c.1152G>A, c.1154A>G, c.1177T>G, c.1267C>T, c.1314C>A, c.1327C>T, c.1406A>C, c.1420_1421delCT, c.1426C>T, c.1430C>T, c.1451C>A, c.1491_1503delCCGTGTGTACCAA, c.1492C>T, c.1493G>A, c.1493G>T, c.1498T>A, c.1624C>T, c.1630delA, c.1673T>C, c.1734G>C, c.1735G>A, c.1805G>A, c.1829_1831delGCC, c.416T>C, c.475T>C, c.518dupT, c.521_522insT, c.538_539delTT, c.557C>T, c.565_566insC, c.573delT, c.592G>C, c.688C>T, c.689G>A, c.730G>A, c.739G>A, c.740delG, c.742G>A, c.757G>C, c.788T>A, c.795delG, c.842_849dupTCCCCGCA, c.880C>A, c.911T>C, c.940G>A, c.952G>A, c.96G>A, c.995C>G, c.996delC
SPG11	94	Paraplejía espástica autosómica recesiva tipo 11	c.118C>T, c.1203delA, c.1235C>G, c.1282A>T, c.1348dupA, c.1457-2A>G, c.1471_1472delCT, c.1492C>T, c.1549_1550delCT, c.1551_1552delTT, c.1668delT, c.1679C>G, c.1697_1711delATCAGTTTGATCACTinsTACTCCCA, c.1735+3_1735+6delAAGT, c.1837_1838insA, c.1845_1846delGT, c.1951C>T, c.2146C>T, c.2163dupT, c.2198T>G, c.2316+1G>A, c.2358_2359delGA, c.2444+1G>C, c.2444G>T, c.2471dupT, c.2608A>G, c.267G>A, c.268G>T, c.2697G>A, c.2716delC, c.2833A>G, c.2834+1G>T, c.2842dupG, c.2849dupT, c.3075dupA, c.3291+1G>T, c.349G>T, c.359delT, c.3602_3603delAT, c.3664_3665insT, c.3719_3720delTA, c.3741dupA, c.398delG, c.4307_4308delAA, c.442+1G>C, c.4462_4463delGT, c.4668T>A, c.4846C>T, c.5255delT, c.529_533delATATT, c.5399_5402delAGATinsTGGAGGAG, c.5410_5411delTG, c.5456_5457delAG, c.5470C>T, c.5532_5533delCA, c.5623C>T, c.5703delT, c.5769delT, c.5798delC, c.5870C>G, c.5970C>G, c.5974C>T, c.5977C>T, c.5986dupT, c.5989_5992delCTGT, c.5992dupT, c.6091C>T, c.6100C>T, c.6157G>A, c.6451delG, c.6477+4A>G, c.654_655delTTinsG, c.6737_6740delTTGA, c.6739_6742delGAGT, c.6754+2_6754+3dupTG, c.6790dupC,

			c.6832_6833delAG, c.6856C>T, c.6898_6899delCT, c.6899T>C, c.7000-3_7000-2insGGA, c.7023C>A, c.7029dupT, c.704_705delAT, c.7085_7088dupATTA, c.7101dupT, c.7115T>A, c.7151+4_7151+7delAGTA, c.7158dupA, c.733_734delAT, c.869+1G>A, c.6206-1G>C, c.642delT c.7152-1G>C
SPG11	2	Enfermedad de Charcot Marie Tooth autosómica recesiva tipo 2X	c.592C>T, c.6632dupG
SPG7	28	Paraplejía espástica autosómica recesiva tipo 7	c.1045G>A, c.1451_1458delAGAGGCGG, c.1519C>T, c.1529C>T, c.1617delC, c.1636G>A, c.1672A>T, c.1715C>T, c.1729G>A, c.1742_1744delTGG, c.1749G>C, c.1A>G, c.1A>T, c.2026T>C, c.2075G>C, c.2191G>A, c.2216dupA, c.2249C>T, c.233T>A, c.246_248delACA, c.28G>T, c.473_474delTC, c.679C>T, c.698T>C, c.773_774delTG, c.850_851delTTinsC, c.861dupT, c.988-1G>A
SRD5A3	6	Trastorno congénito de la glicosilación tipo Iq	c.286_288delCAAinsTGAGTAAGGC, c.29C>A, c.320G>A, c.424C>T, c.489C>A, c.57G>A
SRD5A3	1	síndrome de Kahrizi	c.204dupC
STAR	13	Hiperplasia suprarrenal congénita lipoide	c.135delT, c.178+2dupT, c.466-11T>A, c.545G>A, c.545G>T, c.551_552insA, c.559G>A, c.577C>T, c.64+1G>T, c.650G>C, c.653C>T, c.749G>A, c.772C>T
STRC	5	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB16	c.2171_2174delTTTG, c.4171C>G, c.4195G>T, c.4351C>T, c.4402C>T
SUCLA2	4	Síndrome de depleción del ADN mitocondrial, forma encefalomiopática con aciduria metilmalónica	c.352G>A, c.534+1G>A, c.751G>A, c.850C>T
SUCLG1	8	Acidosis láctica infantil letal con aciduria metilmalónica	c.152_153delAT, c.254G>C, c.40A>T, c.448C>T, c.509C>G, c.626C>A, c.776G>A, c.97+3G>C
TAT	6	Tirosinemia tipo 2	c.1085G>T, c.1249C>T, c.1297C>T, c.169C>T, c.236-5A>G, c.668C>G
TECTA	7	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB21	c.2428C>T, c.2941+1G>A, c.5509T>C, c.5509T>G, c.5668C>T, c.6062G>A, c.651dupC
TMC1	8	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB11	c.100C>T, c.1165C>T, c.1253T>A, c.1333C>T, c.1543T>C, c.1763+3A>G, c.1939T>C, c.1960A>G
TMEM216	4	Síndrome de Joubert 2	c.217C>T, c.218G>A, c.218G>T, c.398T>G
TMEM216	3	Síndrome de Meckel 2	c.230G>C, c.253C>T, c.341T>G
TMEM67	30	Síndrome de Meckel 3	c.1046T>C, c.1065+1delG, c.1127A>C, c.1319G>A, c.1322G>T, c.1336G>C, c.1351C>T, c.1413-1G>C, c.1413-2A>G, c.1538_1539delAT, c.161A>G, c.1843T>C, c.2002T>C, c.224-2delA, c.2301delT, c.2357G>A, c.2528A>G, c.2542G>T, c.2557A>T, c.2689_2690insTA, c.2754_2756delCTT, c.2897T>C, c.387T>A, c.579delA, c.622A>T, c.653G>C, c.734C>T, c.748G>A, c.755T>C, c.888G>T
TMEM67	10	Síndrome de Joubert 6	c.1538A>G, c.1634G>A, c.2315_2322+4delTGAGTAATGTAAinsGG, c.2322+5delG, c.651+2T>G, c.725A>G, c.730A>G, c.2290C>T, c.2368C>A, c.2801G>A
TMEM67	25	Síndrome COACH	c.1073C>T, c.1081G>T, c.1115C>A, c.1126C>G, c.1321C>T, c.1453C>T, c.1674+3A>G, c.1769T>C, c.1911C>A, c.1961-2A>C, c.244C>T, c.245C>G, c.2498T>C, c.2522A>C, c.2556+1G>T, c.2661+5G>A, c.2825T>G, c.300C>A, c.312+5G>A, c.389C>G, c.515G>A, c.579_580delAG, c.675G>A, c.769A>G, c.978+3A>G
TMEM67	3	Síndrome de nefronoptisis 11	c.2461G>A, c.2461G>C, c.869G>T
TMIE	6	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB6	c.170G>A, c.241C>T, c.250C>T, c.257G>A, c.274C>T, c.94-2_98delAGCCCAGinsC
TMPRSS3	15	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB10	c.1025G>A, c.1159G>A, c.1192C>T, c.1211C>T, c.1276G>A, c.208delC, c.308A>G, c.323-6G>A, c.325C>T, c.413C>A, c.646C>T, c.647G>T, c.727G>A, c.753G>C, c.916G>A
TPP1	24	Lipofuscinosis neuronal ceroide juvenil	c.1015C>T, c.1093T>C, c.1094G>A, c.1266G>C, c.1340G>A, c.1379G>A, c.1551+1G>A, c.196C>T, c.229G>A, c.229G>T, c.311T>A, c.379C>T, c.509-1G>A, c.509-1G>C, c.605C>T, c.616C>T, c.622C>T, c.827A>T, c.833A>G, c.851G>T, c.857A>G, c.887-10A>G, c.89+5G>C, c.972_979delCTATGGAG

TPP1	1	Ataxia espinocerebelosa autosómica recesiva tipo 7	c.1397T>G
TPRN	3	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB79	c.1239G>A, c.1427delC, c.1530delG.
TRDN	3	Taquicardia ventricular polimórfica catecolinérgica 5	c.176C>G, c.53_56delACAG, c.613C>T
TRIOBP	6	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB28	c.1039C>T, c.1741C>T, c.2362C>T, c.3202C>T, c.3349C>T, c.889C>T
TSFM	3	Enfermedad mitocondrial letal debida a una deficiencia de fosforilación oxidativa tipo 3 combinada	c.1007G>A, c.919C>T, c.997C>T
TSHR	28	Hipotiroidismo debido a mutaciones en el receptor TSH	c.100G>A, c.1170T>G, c.1228G>A, c.122G>C, c.1291G>A, c.1358T>C, c.1387A>G, c.1430C>T, c.1514G>A, c.1526T>C, c.1575C>A, c.1637G>A, c.1657G>A, c.1798T>C, c.1887G>T, c.1891T>C, c.1897G>T, c.1915C>T, c.2015G>A, c.202C>T, c.326G>A, c.484C>G, c.500T>A, c.545+3G>C, c.548A>G, c.842G>A, c.928C>T, c.970C>T
TUFM	1	Déficit combinado de la fosforilación oxidativa tipo 4	c.1016G>A
TULP1	7	Amaurosis congénita de Leber 15	c.1102G>T, c.1198C>T, c.1204G>T, c.1256G>A, c.1318C>T, c.180delG, c.725_728delCCAA
TULP1	5	Retinosis pigmentaria 14	c.1145T>C , c.1259G>C, c.1444C>T, c.1471T>C, c.1511_1521delTGCAGTTCGGC
TYR	58	Albinismo oculocutáneo tipo 1A	c.1037-7T>A, c.1064C>T, c.1090A>C, c.1111A>G, c.1112A>C, c.1118C>A, c.1146C>A, c.1147G>A, c.1150C>G, c.115T>C, c.1164delT, c.1177delG, c.1184+1G>A, c.1209G>T, c.1217C>T, c.1234C>G, c.1255G>A, c.125A>G, c.1265G>A, c.1309G>A, c.1336G>A, c.1342G>A, c.140G>A, c.1467dupT, c.1469C>A, c.149C>T, c.1501dupC, c.164G>A, c.1A>G, c.229C>T, c.230G>A, c.242C>T, c.265T>C, c.272G>A, c.286dupA, c.325G>A, c.32G>A, c.446A>G, c.533G>A, c.572delG, c.580delA, c.616G>A, c.61C>T, c.646T>A, c.650G>A, c.661G>A, c.707G>A, c.710delA, c.732_733delTG, c.820-2A>G, c.820-3C>G, c.823G>T, c.832C>T, c.892C>T, c.896G>A, c.902C>T, c.978delA, c.982G>A.
TYRP1	6	Albinismo oculocutáneo tipo 3	c.1057_1060delAACA, c.1067G>A, c.107delT, c.1103delA, c.1120C>T, c.497C>G
UGT1A1	33	Síndrome de Crigler-Najjar tipo 1	c.1006C>T, c.1021C>T, c.1043delA, c.1070A>G, c.1085-2A>G, c.1102G>A, c.1124C>T, c.1143C>G, c.1201G>C, c.1220delA, c.1223dupG, c.1282A>G, c.1309A>T, c.1448G>A, c.1449G>A, c.1456T>G,c.145C>T, c.222C>A, c.392T>C, c.474_475insT, c.513_515delCTT, c.517delC, c.529T>C, c.715C>T, c.801delC, c.835A>T, c.840C>A, c.864+1G>C, c.875C>T, c.877_890delTACATTAATGCTTCinsA, c.923G>A, c.991C>T, c.992A>G
UGT1A1	19	Síndrome de Crigler-Najjar tipo 2	c.101C>A, c.1127A>G, c.1130G>T, c.1186delG, c.118T>C, c.1207C>T, c.1304+1G>T, c.1433C>A, c.396_401delCAACAA, c.44T>G, c.524T>A, c.554A>C, c.576C>G, c.610A>G, c.625C>T, c.722_723delAG, c.864+1G>T, c.881T>C, c.973delG
USH1C	5	Síndrome de Usher tipo 1C	c.1069C>T, c.216G>A, c.238dupC, c.308G>A, c.91C>T
USH1G	8	Síndrome de Usher tipo 1G	c.113G>A, c.1373A>T, c.143T>C, c.186_187delCA, c.394dupG, c.46C>G, c.805C>T, c.832_851delTCGGACGAGGACAGCGTCTC
USH2A	86	Síndrome de Usher tipo 2A	c.1000C>T, c.10190_10191delAA, c.1036A>C, c.10450C>T, c.10544A>G, c.10561T>C, c.10612C>T, c.10636G>A, c.10712C>T, c.10724G>A, c.10759C>T, c.11156G>A, c.11328T>A, c.11411delC, c.11875_11876delCA, c.12067-2A>G, c.1214delA, c.12234_12235delGA, c.1227G>A, c.1256G>T, c.12700A>C, c.12708T>A, c.12868C>T, c.13130C>A, c.13313G>A, c.13316C>T, c.13374delA, c.13576C>T, c.14020A>G, c.14031dupA, c.14180G>A, c.14403C>G, c.14803C>T, c.14911C>T, c.15089C>A, c.15233C>G, c.1558T>C, c.1606T>C, c.1840+1G>A, c.1876C>T, c.1992dupT, c.2135delC, c.2209C>T, c.2299delG, c.2541C>A, c.2898delG, c.2983C>T, c.3129dupT, c.3558delT, c.3788G>A, c.4338_4339delCT, c.4405C>T, c.4429G>T, c.4474G>T, c.4510dupA, c.5329C>T, c.5573-2A>G, c.5581G>A, c.5776+1G>A, c.5857+2T>C, c.5858-1G>A, c.6235A>T, c.6289_6302delATCTATTCAGGCAG, c.6446C>A, c.653T>A, c.6795_6797delATA, c.6862G>T, c.7475C>A, c.7595-3C>G, c.779T>G, c.820C>T, c.8559-2A>G,

			c.8740C>T, c.8890dupT, c.8981G>A, c.9165_9168delCTAT, c.920_923dupGCCA, c.9371+1G>C, c.9469C>T, c.9492_9498delTGATGAG, c.949C>A, c.956G>A, c.9570+1G>A, c.9751T>C, c.9799T>C, c.6470delG
USH2A	10	Retinitis pigmentosa 39	c.10073G>A, c.12574C>T, c.1841-2A>G, c.2276G>T, c.2296T>C, c.2802T>G, c.4321G>T, c.486-1G>A, c.7364G>A, c.1558delT
WHRN	4	Sordera neurosensorial no sindrómica autosómica recesiva, tipo DFNB31	c.2423delG, c.2332C>T, c.1267C>T, c.307C>T
ZFYVE26	6	Paraplejía espástica autosómica recesiva tipo 15	c.1477C>T, c.3935C>A, c.4181G>A, c.4312C>T, c.5422C>T, c.5485-1G>A.