Supplementary Table S2. Diagnostic SNVs identified in the cohort.

Sample	Phenotype	Sex	Gene	Variant	Zygosity	Inheritance	Type	Source	Classification
S0002	EP, MA	M	*ATRX*	NM_000489:exon28:c.6254G>A(p.R2085H)	Hemi	XL	missense	NA	P->LP
S0003	EP	M	*TH*	NM_199292:exon7:c.G739A(p.G247S)	Het	AR	missense	Paternal	P
S0003	EP	M	*TH*	NM_199292:exon9:c.G943A(p.G315S)	Het	AR	missense	Maternal	P
S0004	EP	F	*KCNQ2*	NM_172107:exon4:c.533C>T(p.A178V)	Het	AD	missense	De novo	LP
S0004	EP	F	*SCN1A*	NM_001165963:exon25:c.4834G>A(p.V1612I)	Het	AD	missense	Maternal	P
S0006	ID	M	*RAB3GAP2*	NM_012414:exon27:c.3143 A>G(p.H1048R)	Hom	AR	missense	Parental	LP
S0007	EP	F	*MECP2*	NM_001110792:exon3:c.509C>T(p.T170M)	Het	XLD/XLR	missense	De novo	P
S0008	EP	M	*TBC1D24*	NM_001199107:exon2:c.G730A(p.A244T)	Het	AD/AR	missense	Paternal	P
S0008	EP	M	*TBC1D24*	NM_001199107:exon8:c.G1571C(p.R524P)	Het	AD/AR	missense	Maternal	P
S0009	ID	M	*ERCC8*	NM_000082:exon4:c.394_398delTTACA	Hom	AR	frameshift	Maternal	P
S0011	BT, MA	F	*SMARCA4*	NM_001128849:exon10:c.C1615G(p.R539G)	Het	AD	missense	NA	LP
S0012	ID	M	*DDC*	NM_000790:exon6:c.714+4A>T	Hom	AR	splicing	Parental	P
S0025	BT	F	*SCN2A*	NM_021007:exon12:c.1819C>T(p.R607*)	Het	AD	stop gain	De novo	P
S0027	MD	M	*SLC5A5*	NM_000453:exon10:c.1242+1G>A	Hom	AR	splicing	NA	LP
S0029	EP	F	*AP4S1*	NM_001128126:exon6:c.332_334delCAT	Het	AR	inframeshift	paternal	LP
S0029	EP	F	*AP4S1*	NM_001128126:exon2:c.116_118delTC	Het	AR	frameshift	maternal	LP
S0032	EP, MA	F	*PAFAH1B1*	NM_000430:exon6:c.563T>A(p.M188K)	Het	AD	missense	De novo	LP
S0035	MD, EP	M	*ATP7A*	NM_000052:exon8:c.1933C>T(p.R645*)	Hemi	XL	stop gain	Maternal	P
S0037	EP, MA	M	*GRIN1*	NM_007327:exon12:c.1665G>A(p.M555I)	Het	AD	missense	De novo	LP
S0041	MA	M	*ERCC8*	NM_000082.3:exon4:c.394_398delTTACA	Hom	AR	frameshift	De novo	P
S0042	EP	F	*MECP2*	NM_001110792:exon3:c.1078G>T(p.E360*)	Het	XLD/XLR	stop gain	De novo	P

S0045	EP	F	*TSC2*	NM_000548:exon34:c.4375C>T(p.R1459*)	Het	AD	stop gain	NA	P->LP
S0046	MA	M	*PAFAH1B1*	NM_000430:exon4:c.162delA	Het	AD	frameshift	NA	P->LP
S0052	EP, MA	M	*STXBP1*	NM_003165:exon14:c.1216C>T(p.R406C)	Het	AD	missense	NA	P->LP
S0056	BT	F	*MECP2*	NM_004992:exon4:c.401C>A(p.S134Y)	Het	XLD/XLR	missense	De novo	P
S0060	EP	F	*CDKL5*	NM_003159:exon7:c.416A>G(p.E139G)	Het	XLD	missense	NA	P->LP
S0062	EP	M	*STXBP1*	NM_003165:exon18:c.1702G>A(p.G568S)	Het	AD	missense	De novo	LP
S0063	BT, EP	F	*KCNQ2*	NM_172107:exon17:c.2326delC	Het	AD	frameshift	De novo	LP
S0064	MA	F	*CREBBP*	NM_004380:exon5:c.1279_1280delTG	Het	AD	frameshift	De novo	LP
S0065	EP, MA	M	*TSC2*	NM_000548:exon30:c.3412C>T(p.R1138*)	Het	AD	stop gain	Not-paternal	P->LP
S0066	MA	F	*EHMT1*	NM_024757:exon26:c.3553dupT	Het	AD	frameshift	De novo	LP
S0067	EP, MA	M	*SPG11*	NM_025137:exon13:c.2321A>C(p.E774A)	Het	AR	missense	Paternal	LP
S0067	EP, MA	M	*SPG11*	NM_025137:exon16:c.3023A>G(p.Y1008C)	Het	AR	missense	Maternal	LP
S0071	EP	M	*SCN8A*	NM_014191:exon17:c.2932A>G(p.S978G)	Het	AD	missense	NA	P->LP
S0075	EP, MA	F	*TUBA1A*	NM_006009:exon4:c.1175A>G(p.D392G)	Het	AD	missense	De novo	LP
S0083	EP	M	*SLC2A1*	NM_006516:exon2:c.73C>T(p.Q25*)	Het	AD/AR	stop gain	Paternal	LP
S0084	ID	M	*ATP1A3*	NM_152296:exon9:c.1003A>C(p.T335P)	Het	AD	missense	NA	P->LP
S0085	ID	F	*SETD5*	NM_001080517:exon19:c.2882G>A(p.R961K)	Het	AD	missense	NA	LP
S0086	MD	F	*KANK1*	NM_015158:exon3:c.1484C>T(p.A495V)	Het	AD	missense	NA	LP
S0090	MA	M	*FA2H*	NM_024306:exon6:c.956A>G(p.H319R)	Hom	AR	missense	Parental	LP
S0091	ID	F	*MECP2*	NM_004992:exon4:c.674C>G(p.P225R)	Het	XLD/XLR	missense	De novo	P
S0092	EP	M	*SCN1A*	NM_001165963:exon15:c.2825_2826insT	Het	AD	frameshift	NA	LP
S0093	EP, MA	F	*GALC*	NM_000153:exon1:c.184_186insAGC	Het	AR	inframeshift	Maternal	LP
S0093	EP, MA	F	*GALC*	NM_000153:exon16:c.1901T>C(p.L634S)	Het	AR	missense	Paternal	P
S0094	EP	M	*DCC*	NM_005215:exon2:c.143A>C(p.D48A)	Het	AD	missense	NA	LP

S0095	EP	M	*HEXA*	NM_000520:exon5:c.533G>A(p.R178H)	Het	AR	missense	Paternal	P
S0095	EP	M	*HEXA*	NM_000520:exon6:c.571-1G>T	Het	AR	splicing	Maternal	P
S0096	MA	M	*CC2D2A*	NM_001080522:exon27:c.3383T>C(p.L1128P)	Het	AR	missense	NA	LP
S0096	MA	M	*CC2D2A*	NM_001080522:exon21:c.2581G>A(p.D861N)	Het	AR	missense	NA	P->LP
S0102	EP	F	*SCN1A*	NM_001165963:exon15:c.2927T>A(p.M976K)	Het	AD	missense	De novo	P
S0103	EP, MA	M	*GLB1*	NM_000404:exon16:c.2006dupA	Het	AR	frameshift	Maternal	LP
S0103	EP, MA	M	*GLB1*	NM_000404:exon14:c.1369C>T(p.R457*)	Het	AR	stop gain	Paternal	P
S0104	ID	F	*GALC*	NM_000153:exon1:c.136G>T(p.D46Y)	Het	AR	missense	Maternal	P
S0104	ID	F	*GALC*	NM_000153:exon14:c.1586C>T(p.T529M)	Het	AR	missense	Paternal	P
S0105	MA	M	*ARID1B*	NM_020732:exon20:c.5542delC	Het	AD	frameshift	NA	P->LP
S0110	EP	M	*SCN9A*	NM_002977:exon13:c.1985A>G(p.Y662C)	Het	AD/AR	missense	NA	LP
S0114	ID	M	*FOXG1*	NM_005249:exon1:c.723G>C(p.K241N)	Het	AD	missense	De novo	LP
S0122	ID	F	*SLC2A1*	NM_006516:exon4:c.436G>A(p.E146K)	Het	AD/AR	missense	Paternal	P
S0139	MD	F	*ETFDH*	NM_004453:exon13:c.1691-3C>G	Het	AR	splicing	Maternal	P
S0139	MD	F	*ETFDH*	NM_004453:exon8:c.872T>G(p.V291G)	Het	AR	missense	Paternal	P
S0140	BT	M	*BCL11A*	NM_022893:exon4:c.1483G>T(p.E495*)	Het	AD	stop gain	De novo	LP
S0145	ID	M	*SCN8A*	NM_014191:exon11:c.1588C>T(p.R530W)	Het	AD	missense	NA	P->LP
S0146	ID	F	*CTNNB1*	NM_001904:exon5:c.646_652delGGGACCT	Het	AD	frameshift	De novo	LP
S0152	EP	M	*NF1*	NM_000267:exon34:c.4537C>T(p.R1513*)	Het	AD	stop gain	NA	P->LP
S0157	EP	M	*KCNQ2*	NM_172107:exon8:c.1049A>T(p.N350I)	Het	AD	missense	De novo	P
S0158	MD, MA	M	*SBDS*	NM_016038:exon2:c.258+2T>C	Hom	AR	splicing	Paternal/ Maternal in mosaic	P

S0161	EP, MA	M	*SETBP1*	NM_015559:exon4:c.2602G>A(p.D868N)	Het	AD	missense	De novo	P
S0162	EP	M	*SCN2A*	NM_021007:exon15:c.2558G>A(p.R853Q)	Het	AD	missense	De novo	P
S0175	EP	F	*SCN2A*	NM_021007:exon19:c.3524G>A(p.C1175Y)	Het	AD	missense	Not-Paternal	LP
S0176	ID	F	*PLA2G6*	NM_003560:exon2:c.23T>A(p.V8D)	Het	AR	missense	Paternal	LP
S0176	ID	F	*PLA2G6*	NM_003560:exon2:c.68G>A(p.R23Q)	Het	AR	missense	Maternal	P
S0178	EP	F	*CACNA1A*	NM_001127221:exon25:c.4046G>A(p.R1349Q)	Het	AD	missense	De novo	P
S0183	ID	F	*MAN1B1*	NM_016219:exon13:c.1907_1911del	Het	AR	frameshift	Maternal	LP
S0184	MD, MA	M	*PMM2*	NM_000303:exon5:c.430T>C(p.F144L)	Het	AR	missense	Paternal	P
S0184	MD, MA	M	*PMM2*	NM_000303:exon4:c.317A>T(p.Y106F)	Het	AR	missense	Maternal	P
S0186	MA	M	*NIPBL*	NM_133433:exon21:c.4422-1G>C	Het	AD	splicing	De novo	LP
S0193	EP	F	*KCNQ2*	NM_172107:exon5:c.781T>A(p.F261I)	Het	AD	missense	De novo	LP
S0203	EP	M	*NF1*	NM_000267:exon23:c.3109_3110delTTinsA	Het	AD	missense	NA	LP
S0204	MD	M	*GCDH*	NM_000159:exon4:c.191A>C(p.E64A)	Het	AR	missense	NA	LP
S0204	MD	M	*GCDH*	NM_000159:exon12:c.1244-2A>C	Het	AR	splicing	NA	P->LP
S0207	ID	M	*TPP1*	NM_000391:exon6:c.609dupT	Het	AR	frameshift	Maternal	P
S0207	ID	M	*TPP1*	NM_000391:exon12:c.1548_c.1551dupTGAT	Het	AR	splicing	Paternal	P
S0208	EP	F	*SCN2A*	NM_021007:exon19:c.3521-2A>G	Het	AD	splicing	De novo	LP
S0209	ID	M	*FAM126A*	NM_032581:exon4:c.162_163insGA	Hom	AR	frameshift	Parental	LP
S0210	MA	F	*FOXG1*	NM_005249.4:exon1:c.402delG	Het	AD	frameshift	De novo	LP
S0213	EP	F	*MECP2*	NM_004992:exon4c.1157_1197del41	Het	XLD/XLR	frameshift	De novo	P
S0214	ID	F	*THRA*	NM_199334:exon9:c.1176C>A(p.C392*)	Het	AD	stop gain	De novo	P
S0218	ID	F	*ST3GAL5*	NM_003896:exon6:c.1000delC	Het	AR	frameshift	Maternal	LP
S0218	ID	F	*ST3GAL5*	NM_003896:exon4:c.601G>A(p.G201R)	Het	AR	missense	Paternal	P

S0219	ID	M	*PEX1*	NM_000466:exon8:c.1483+1G>A	Het	AR	splicing	Paternal	LP
S0219	ID	M	*PEX1*	NM_000466:exon10:c.1725dupG	Het	AR	frameshift	Maternal	LP
S0220	ID	M	*TUBB4A*	NM_006087:exon4:c.745G>A(p.D249N)	Het	AD	missense	NA	P->LP
S0221	EP	M	*MFSD8*	NM_152778:exon11:c.1102+2T>C	Hom	AR	splicing	NA	LP
S0225	MD, MA	M	*IDS*	NM_000202:exon7:c.908_909delCT	Hemi	XL	frameshift	De novo	P
S0232	EP	F	*TSC2*	NM_000548:exon30:c.3598C>T(p.R1200W)	Het	AD	missense	De novo	P
S0235	EP	M	*SLC2A1*	NM_006516:exon4:c.377G>A(p.R126H)	Het	AD/AR	missense	De novo	P
S0238	MD, EP	F	*MMACHC*	NM_015506:exon4:c.609G>A(p.W203*)	Hom	AR	stop gain	Parental	P
S0241	MD	F	*ARSA*	NM_000487:exon1:c.224delA	Het	AR	frameshift	Maternal	LP
S0241	MD	F	*ARSA*	NM_000487:exon8:c.1484G>C(p.C495S)	Het	AR	missense	Paternal	LP
S0248	EP	M	*SCN2A*	NM_021007:exon27:c.5045T>C(p.F1682S)	Het	AD	missense	De novo	LP
S0252	MA	M	*PAX6*	NM_000280:exon10:c.781C>T(p.R261*)	Het	AD	stop gain	De novo	P
S0253	MA	F	*SMC1A*	NM_006306:exon6:c.896T>A(p.I299N)	Het	XLD	missense	De novo	LP
S0254	ID	F	*IDUA*	NM_000203:exon5:c.536C>G(p.T179R)	Het	AR	missense	Paternal	P
S0254	ID	F	*IDUA*	NM_000203:exon2:c.265C>T(p.R89W)	Het	AR	missense	Maternal	P
S0261	MD	M	*ATP1A2*	NM_000702:exon21:c.2936C>T(p.P979L)	Het	AD	missense	De novo	P
S0262	EP	F	*SCN1A*	NM_001165963:exon8:c.1150T>C(p.W384R)	Het	AD	missense	NA	P->LP
S0266	MD	F	*SMPD1*	NM_000543:exon5:c.1486+5G>C	Het	AR	splicing	NA	P->LP
S0266	MD	F	*SMPD1*	NM_000543:exon3:c.1144C>T(p.L382F)	Het	AR	missense	NA	P->LP
S0271	MA	F	*NFIX*	NM_001271043:exon2:c.177_178insGCGGATGT	Het	AD	frameshift	De novo	LP
S0275	EP	M	*SCN1A*	NM_001165963:exon14:c.2573T>C(p.L858P)	Het	AD	missense	De novo	LP
S0285	EP, MA	M	*SLC6A8*	NM_005629:exon3:c.626_627delCT	Hemi	XL	frameshift	Maternal	LP
S0291	EP, MA	M	*SLC9A6*	NM_006359:exon6:c.803+1G>A	Hemi	XL	splicing	Maternal	P

S0295	ID	M	*IQSEC2*	NM_001111125:exon5:c.1850G>A(p.R617H)	Hemi	XL	missense	Maternal	LP
S0296	MA	F	*NSD1*	NM_022455:exon18:c.5741G>T(p.R1914L)	Het	AD	missense	De novo	P
S0301	MD	F	*SPAST*	NM_014946:exon8:c.1168A>G(p.M390V)	Het	AD	missense	De novo	P
S0303	ID	M	*ATM*	NM_000051:exon35:c.5188C>T(p.R1730*)	Het	AR	stop gain	Paternal	P
S0303	ID	M	*ATM*	NM_000051:exon28:c.4148_4149delCG	Het	AR	frameshift	Maternal	LP
S0314	ID	F	*NPC1*	NM_000271:exon18:c.2632T>C(p.S878P)	Het	AR	missense	Paternal	LP
S0314	ID	F	*NPC1*	NM_000271:exon8:c.1004_1005delT	Het	AR	frameshift	Maternal	LP
S0315	EP	M	*SCN2A*	NM_021007:exon21:c.3961G>A(p.E1321K)	Het	AD	missense	De novo	P
S0321	BT	M	*AFF2*	NM_002025:exon5:c.1126C>A(p.H376N)	Hemi	XL	missense	Maternal	LP
S0331	MD, MA	M	*SLC12A3*	NM_000339:exon3:c.486_490delTACGGinsA	Het	AR	frameshift	Maternal	LP
S0331	MD, MA	M	*SLC12A3*	NM_000339:exon4:c.506-1G>A	Het	AR	splicing	Paternal	P
S0336	MD, MA	F	*SLC37A4*	NM_001164277:exon6:c.C572T(p.P191L)	Het	AR	missense	Maternal	P
S0336	MD, MA	F	*MED13L*	NM_015335:exon15:c.2590A>T(p.M864L)	Het	AD	missense	De novo	P
S0336	MD, MA	F	*SLC37A4*	NM_001164277:exon3:c.T68G(p.L23R)	Het	AR	missense	Paternal	LP
S0337	MA	F	*OFD1*	NM_003611:exon11:c.1089delT	Het	XLD	frameshift	De novo	LP
S0343	ID	M	*GLB1*	NM_000404.2:exon10:c.956-2A>G	Het	AR	splicing	Paternal	P
S0343	ID	M	*GLB1*	NM_000404:exon14:c.1444C>T(p.R482C)	Het	AR	missense	Maternal	P
S0344	MD	M	*NDUFV1*	NM_007103:exon5:c.658C>T(p.Q220*)	Het	AR	stop gain	Maternal	LP
S0344	MD	M	*NDUFV1*	NM_007103:exon8:c.1156C>T(p.R386C)	Het	AR	missense	Paternal	P
S0350	ID	F	*ATP7B*	NM_000053:exon8:c.2333G>T(p.R778L)	Hom	AR	missense	NA	P
S0352	BT, EP	M	*GABRG2*	NM_000816:exon8:c.968G>A(p.R323Q)	Het	AD	missense	De novo	P
S0362	EP	F	*ALDH5A1*	NM_001080:exon3:c.515G>A(p.R172H)	Hom	AR	missense	Parental	LP
S0370	MA	F	*EHMT1*	NM_024757:exon3:c.554_555delAC	Het	AD	frameshift	De novo	LP
S0371	ID	M	*HUWE1*	NM_031407:exon83:c.12928G>C(p.G4310R)	Hemi	XL	missense	Maternal	P

S0374	MD, EP	F	*PCCA*	NM_000282:exon1:c.59G>A(p.W20*)	Het	AR	stop gain	Paternal	LP
S0374	MD, EP	F	*PCCA*	NM_000282:exon3:c.216del	Het	AR	frameshift	Maternal	LP
S0375	MA	M	*DNM2*	NM_001005360:exon17:c.1862T>C(p.L621P)	Het	AD/AR	missense	De novo	P
S0377	MA	M	*FGFR2*	NM_000141:exon7:c.833G>T(p.C278F)	Het	AD	missense	De novo	P
S0383	EP	F	*CDKL5*	NM_003159:exon8:c.532C>T(p.R178W)	Het	XLD	missense	De novo	P
S0385	EP	F	*TSC2*	NM_000548:exon39:c.5012T>G(p.V1671G)	Het	AD	missense	NA	LP
S0386	MA	M	*CREBBP*	NM_004380:exon31:c.5837dupC	Het	AD	frameshift	De novo	P
S0394	EP	F	*POLG*	NM_002693:exon16:c.2591A>G(p.N864S)	Het	AD/AR	missense	Maternal	P
S0394	EP	F	*POLG*	NM_002693:exon18:c.2890C>T(p.R964C)	Het	AD/AR	missense	Paternal	P
S0394	EP	F	*POLG*	NM_002693:exon18:c.2884G>A(p.A962T)	Het	AD/AR	missense	Paternal	P
S0395	EP	F	*SLC2A1*	NM_006516:exon7:c.884C>T(p.T295M)	Het	AD/AR	missense	De novo	P
S0399	EP, MA	M	*TSC1*	NM_000368:exon15:c.1509_1510insAA	Het	AD	frameshift	Maternal	P
S0401	EP	M	*ARX*	NM_139058:exon5:c.1478G>C(p.S493T)	Hemi	XL	missense	Maternal	LP
S0408	MA	M	*MITF*	NM_000248:exon7:c.635-2A>G	Het	AD	splicing	De novo	LP
S0414	MA	F	*PPP2R5D*	NM_006245:exon5:c.592G>A(p.E198K)	Het	AD	missense	De novo	P
S0416	ID	M	*TUBA1A*	NM_006009:exon4:c.790C>T(p.R264C)	Het	AD	missense	De novo	P
S0417	ID	F	*NOTCH3*	NM_000435:exon11:c.1759C>T(p.R587C)	Het	AD	missense	NA	P->LP
S0418	ID	F	*PHEX*	NM_000444:exon9:c.1016T>A(p.V339E)	Het	XL	missense	Paternal	LP
S0419	ID	F	*ATM*	NM_000051:exon13:c.1899-1G>A	Het	AR	splicing	Maternal	P
S0419	ID	F	*ATM*	NM_000051:exon46:c.6592_6593delCT	Het	AR	frameshift	Paternal	LP
S0435	MA	F	*KMT2D*	NM_003482:exon39:c.11119C>T(p.R3707*)	Het	AD	stop gain	NA	P->LP
S0441	ID	F	*NF1*	NM_000267:exon31:c.4267A>G(p.K1423E)	Het	AD	missense	NA	P->LP
S0443	MD, MA	M	*WFS1*	NM_006005:exon8:c.1230_1233delCTCT(p.V412Sfs*29)	Hom	AD/AR	frameshift	Parental	P
S0445	EP	F	*TSC1*	NM_000368:exon15:c.1888_1891delAAAG(	Het	AD	frameshift	De novo	P

				p.K630Qfs*22)					
S0447	ID	F	*NFIX*	NM_001271043:exon2:c.202_203delCT	Het	AD	frameshift	De novo	LP
S0448	EP	M	*SCN1A*	NM_001165963:exon24:c.4529C>T(p.A1510V)	Het	AD	missense	De novo	LP
S0452	ID	M	*STXBP1*	NM_003165:exon18:c.1697T>C(p.L566P)	Het	AD	missense	De novo	LP
S0463	MD	F	*GCDH*	NM_000159:exon12:c.1244-2A>C	Het	AR	splicing	Paternal	P
S0463	MD	F	*GCDH*	NM_000159:exon12:c.1261G>A(p.A421T)	Het	AR	missense	Maternal	P
S0469	EP	M	*TSC1*	NM_000368:exon15:c.1904_1905delCA	Het	AD	frameshift	NA	P->LP
S0475	ID	F	*MECP2*	NM_004992:exon4:c.473C>T(p.T158M)	Het	XLD/XLR	missense	NA	P->LP
S0476	MD	F	*PAH*	NM_000277:exon3:c.331C>T(p.R111*)	Het	AR	stop gain	NA	P->LP
S0476	MD	F	*PAH*	NM_000277:exon6:c.611A>G(p.Y204C)	Het	AR	missense	NA	P->LP
S0478	EP	F	*MECP2*	NM_004992:exon4:c.422A>G(p.Y141C)	Het	XLD/XLR	missense	NA	P->LP
S0483	MA	M	*GAA*	NM_000152:exon14:c.1935C>A(p.D645E)	Hom	AR	missense	Parental	P
S0488	BT, EP	F	*MECP2*	NM_004992:exon4:c.916C>T(p.R306C)	Het	XLD/XLR	missense	De novo	P
S0491	MD	M	*MUT*	NM_000255:exon10:c.1677-1G>A	Het	AR	splicing	NA	P->LP
S0491	MD	M	*MUT*	NM_000255:exon3:c.729_730insTT	Het	AR	frameshift	NA	P->LP
S0493	EP	F	*TSC1*	NM_000368:exon9:c.813T>G(p.Y271*)	Het	AD	stop gain	De novo	LP
S0500	ID	M	*UBA1*	NM_003334:exon15:c.1617G>A(p.M539I)	Hemi	XL	missense	Maternal	P
S0503	ID	M	*PMP22*	NM_000304:exon4:c.215C>T(p.S72L)	Het	AD/AR	missense	De novo	P
S0506	ID	M	*PDHA1*	NM_000284:exon4:c.379C>T(p.R127W)	Hemi	XLD	missense	De novo	P
S0508	ID	F	*ETFDH*	NM_004453:exon3:c.250G>A(p.A84T)	Hom	AR	missense	Parental	P
S0518	EP	M	*DCX*	NM_178153:exon3:c.560G>T(p.S187I)	Hemi	XL	missense	De novo	LP
S0523	MA	M	*SETD5*	NM_001080517:exon19:c.3089_3090insG	Het	AD	frameshift	NA	LP
S0526	ID	F	*PMP22*	NM_000304:exon3:c.117G>A(p.W39*)	Het	AD/AR	stop gain	De novo	P
S0528	MA	F	*KDM5C*	NM_004187:exon23:c.3863G>A(p.W1288*)	Het	XL	stop gain	De novo	LP
S0533	ID	F	*TH*	NM_199292:exon5:c.580+2T>C	Het	AR	splicing	Maternal	P

S0533	ID	F	*TH*	NM_199292:exon6:c.698G>A(p.R233H)	Het	AR	missense	Paternal	P
S0535	MA	F	*PDHA1*	NM_000284:exon10:c.944A>G(p.D315G)	Het	XLD	missense	De novo	LP
S0537	MD, EP, MA	M	*MTHFR*	NM_005957:exon8:c.1316T>C(p.L439P)	Het	AR	missense	Maternal	LP
S0537	MD, EP, MA	M	*MTHFR*	NM_005957:exon3:c.323C>T(p.S108F)	Het	AR	missense	Paternal	LP
S0547	MA	M	*NIPBL*	NM_015384:exon10:c.1660C>T(p.Q554*)	Het	AD	stop gain	De novo	LP
S0548	MD, MA	M	*JAG1*	NM_000214:exon17:c.2186G>T(p.C729F)	Het	AD	missense	NA	LP
S0551	EP	M	*SCN1A*	NM_001165963:exon10:c.1662G>A(p.Q554Q)	Het	AD	splicing	De novo	P
S0554	MD	F	*DHTKD1*	NM_018706:exon7:c.1219C>T(p.R407C)	Het	AD/AR	missense	NA	LP
S0554	MD	F	*DHTKD1*	NM_018706:exon9:c.1739G>A(p.G580D)	Het	AD/AR	missense	NA	LP
S0560	MD	M	*TPK1*	NM_022445:exon7:c.382C>T(p.L128F)	Het	AR	missense	Maternal	LP
S0560	MD	M	*TPK1*	NM_001042482:exon2:c.19C>T(p.P7S)	Het	AR	missense	Paternal	LP
S0565	EP, MA	F	*ADSL*	NM_000026:exon1:c.127T>C(p.W43R)	Het	AR	missense	Maternal	LP
S0565	EP, MA	F	*ADSL*	NM_000026:exon7:c.778G>A(p.A260T)	Het	AR	missense	Paternal	LP
S0566	MA	M	*TUBB3*	NM_006086:exon4:c.1228G>A(p.E410K)	Het	AD	missense	NA	P->LP
S0579	MD	M	*NDUFAF5*	NM_024120:exon6:c.519+4A>G	Het	AR	splicing	Paternal	LP
S0579	MD	M	*NDUFAF5*	NM_024120:exon7:c.562A>G(p.M188V)	Het	AR	missense	Maternal	LP
S0582	MD, MA	M	*NFU1*	NM_001002755:exon5:c.482C>T(p.A161V)	Het	AR	missense	Maternal	LP
S0582	MD, MA	M	*NFU1*	NM_001002755:exon6:c.544C>T(p.R182W)	Het	AR	missense	Paternal	P
S0593	ID	M	*FA2H*	NM_024306:exon3:c.379C>T(p.R127*)	Hom	AR	stop gain	NA	LP
S0594	EP	F	*TSC1*	NM_000368:exon12:c.1030-2A>G	Het	AD	splicing	De novo	LP
S0595	EP	M	*NF1*	NM_000267:exon16:c.1756_1759delACTA	Het	AD	frameshift	De novo	P
S0600	EP	F	*TSC2*	NM_000548:exon38:c.4909_4910del	Het	AD	frameshift	De novo	P
S0601	MA	F	*SPRED1*	NM_152594:exon2:c.190C>T(p.R64*)	Het	AD	stop gain	Paternal	P

S0615	ID	F	*PYCR1*	NM_006907.3:exon4:c.355C>G(p.R119G)	Het	AR	missense	Paternal	P
S0615	ID	F	*PYCR1*	NM_006907.3:exon4:c.345del(p.R116Gfs*6)	Het	AR	frameshift	Maternal	P
S0617	EP	F	*ALG13*	NM_001099922:exon3:c.320A>G(p.N107S)	Het	XLD	missense	De novo	P
S0620	ID	F	*MECP2*	NM_004992:exon4:c.880C>T(p.R294*)	Het	XLD/XLR	stop gain	NA	P->LP
S0624	EP	F	*GABRA1*	NM_000806:exon8:c.580G>T(p.V194F)	Het	AD	missense	De novo	LP
S0627	MD	M	*GLUD1*	NM_005271:exon7:c.943C>T(p.H315Y)	Het	AD	missense	NA	P->LP
S0632	EP	F	*SCN1A*	NM_001165963:exon11:c.2021A>G(p.D674G)	Het	AD	missense	De novo	P
S0633	BT	F	*MECP2*	NM_004992:exon4:c.763C>T(p.R255*)	Het	XLD/XLR	stop gain	De novo	P
S0636	EP	M	*CACNA1A*	NM_001127221:exon17:c.2137G>A(p.A713T)	Het	AD	missense	De novo	P
S0638	MA	F	*BRAF*	NM_004333:exon6:c.739T>G(p.F247V)	Het	AD	missense	De novo	P
S0651	EP	F	*SCN1A*	NM_001165963:exon21:c.4245T>A(p.F1415L)	Het	AD	missense	De novo	P
S0652	MA	M	*DYNC1H1*	NM_001376:exon27:c.5434-8T>C	Het	AD	splicing	NA	LP
S0652	MA	M	*DYNC1H1*	NM_001376:exon4:c.751C>T(p.R251C)	Het	AD	missense	NA	LP
S0659	ID	M	*ALDH3A2*	NM_000382:exon8:c.1157A>G(p.N386S)	Het	AR	missense	Paternal	P
S0659	ID	M	*ALDH3A2*	NM_000382:exon4:c.638C>T(p.P213L)	Het	AR	missense	Maternal	LP
S0661	MA	F	*TUBA1A*	NM_006009:exon4:c.379G>A(p.D127N)	Het	AD	missense	De novo	LP
S0664	MA	M	*LAMA2*	NM_000426:exon29:c.4226_4226insCCCTGGACCAA	Het	AR	frameshift	NA	LP
S0668	BT	F	*IQSEC2*	NM_001111125:exon8:c.2726_2749dupATGACTTCATCAAGAACCTGAGAG	Het	XLD	splicing	De novo	P
S0671	EP	M	*SLC6A1*	NM_003042:exon6:c.581+2T>C	Het	AD	splicing	De novo	LP
S0672	EP	F	*MEF2C*	NM_002397:exon7:c.766C>T(p.R256*)	Het	AD	stop gain	De novo	LP
S0675	EP	M	*SCN1A*	NM_001165963:exon2:c.337_340delCCCT	Het	AD	frameshift	De novo	P

S0676	EP	F	*PCDH19*	NM_001184880:exon1:c.1019A>G(p.N340S)	Het	XL	missense	De novo	P
S0678	EP	M	*NAA10*	NM_003491:exon6:c.346C>T(p.R116W)	Hemi	XLD/XLR	missense	Maternal	P
S0679	ID	M	*PPP2R5D*	NM_006245:exon5:c.598G>A(p.E200K)	Het	AD	missense	De novo	P
S0680	ID	M	*MAP2K1*	NM_002755:exon3:c.371C>A(p.P124Q)	Het	AD	missense	De novo	P
S0682	MA	M	*BRAF*	NM_004333:exon12:c.1455G>C(p.L485F)	Het	AD	missense	De novo	P
S0685	MD	M	*MMACHC*	NM_015506:exon3:c.394C>T(p.R132*)	Het	AR	stop gain	NA	P->LP
S0685	MD	M	*MMACHC*	NM_015506:exon4:c.481C>T(p.R161*)	Het	AR	stop gain	NA	P->LP
S0687	MA	M	*OPA1*	NM_015560:exon8:c.794_797delTTGA	Het	AD/AR	frameshift	Paternal	P
S0687	MA	M	*OPA1*	NM_015560:exon22:c.2231C>T(p.A744V)	Het	AD/AR	missense	Maternal	LP
S0688	ID	F	*ALDH3A2*	NM_000382:exon8:c.1157A>G(p.N386S)	Hom	AR	missense	NA	P
S0690	MA	M	*PAFAH1B1*	NM_000430:exon5:c.265C>T(p.R89*)	Het	AD	stop gain	De novo	P
S0691	MA	F	*GNPTAB*	NM_024312:exon18:c.3428_3429insA	Het	AR	frameshift	Maternal	P
S0691	MA	F	*GNPTAB*	NM_024312:exon10:c.1284+1G>T	Het	AR	splicing	Paternal	LP
S0692	BT	M	*PAK3*	NM_002578:exon6:c.176-2A>G	Hemi	XLR	splicing	Maternal	LP
S0693	MA	M	*IGHMBP2*	NM_002180:exon2:c.2356delG	Het	AR	frameshift	Paternal	P
S0693	MA	M	*IGHMBP2*	NM_002180:exon2:c.2598_2561delGAAA	Het	AR	frameshift	Maternal	P
S0694	MA	M	*MITF*	NM_000248:exon7:c.640_643delCGAAinsC	Het	AD/AR	inframeshift	NA	P->LP
S0695	MD	M	*BCKDHA*	NM_000709:exon5:c.565C>T(p.R189C)	Het	AR	missense	Maternal	LP
S0695	MD	M	*BCKDHA*	NM_000709:exon2:c.117dupC	Het	AR	frameshift	Paternal	P
S0697	ID	M	*OCRL*	NM_000276:exon20:c.2194delC	Hemi	XLR	frameshift	Maternal	LP
S0699	MD	M	*MUT*	NM_000255:exon2:c.323G>A(p.R108H)	Het	AR	missense	Paternal	P
S0699	MD	M	*MUT*	NM_000255:exon3:c.730_730insTT	Het	AR	frameshift	Maternal	P
S0701	EP	F	*KCNQ2*	NM_172107:exon6:c.907_908delTC	Het	AD	frameshift	De novo	LP
S0706	ID	M	*TCF4*	NM_001083962:exon18:c.1733G>A(p.R578H)	Het	AD	missense	De novo	P
S0707	ID	F	*GABRG2*	NM_000816:exon3:c.316G>A(p.A106T)	Het	AD	missense	De novo	P

S0711	ID	M	*TAF1*	NM_004606:exon21:c.3265C>T(p.R1089C)	Hemi	XLR	missense	De novo	LP
S0712	MD	F	*UBR1*	NM_174916:exon16:c.1850-2A>T	Het	AR	splicing	Maternal	LP
S0712	MD	F	*UBR1*	NM_174916:exon39:c.4290T>G(p.S1430R)	Het	AR	missense	Paternal	LP
S0761	MA	F	*BRAF*	NM_004333:exon16:c.1914T>A(p.D638E)	Het	AD	missense	De novo	P
S0764	MA	M	*BRAF*	NM_004333:exon12:c.1501G>A(p.E501K)	Het	AD	missense	De novo	P
S0768	ID	M	*MECP2*	NM_004992.3:exon3:c.317G>A(p.R106Q)	Hemi	XLD/XLR	missense	NA	P->LP
S0779	ID	M	*ATP1A3*	NM_152296:exon17:c.2401G>A(p.D801N)	Het	AD	missense	De novo	P
S0784	ID	M	*ERCC6*	NM_000124:exon9:c.1834C>T(p.R612*)	Het	AD/AR	stop gain	Paternal	P
S0784	ID	M	*ERCC6*	NM_000124:exon10:c.2144delG	Het	AD/AR	frameshift	Maternal	P
S0789	BT, MA	M	*NLGN4X*	NM_020742:exon4:c.808G>T(p.E270*)	Hemi	XL	stop gain	Maternal	LP
S0790	MA	M	*ARID1B*	NM_020732:exon20:c.5833T>C(p.C1945R)	Het	AD	missense	De novo	LP
S0791	ID	M	*GRIA3*	NM_000828:exon12:c.1919T>C(p.F640S)	Hemi	XLR	missense	De novo	LP
S0792	EP, MA	F	*AP4M1*	NM_004722:exon12:c.952C>T(p.R318*)	Het	AR	stop gain	Paternal	P
S0792	EP, MA	F	*AP4M1*	NM_004722:exon14:c.1127G>A(p.G376D)	Het	AR	missense	Maternal	LP
S0795	ID	M	*CACNA1A*	NM_001127221:exon25:c.4046G>A(p.R1349Q)	Het	AD	missense	De novo	P
S0796	MD, EP	M	*MMACHC*	NM_015506:exon4:c.609G>A(p.W203*)	Hom	AR	stop gain	NA	P
S0801	EP	F	*CACNA1A*	NM_001127221:exon25:c.4031_4039delCCCTCCGAG	Het	AD	inframeshift	De novo	P
S0802	BT	F	*MECP2*	NM_004992:exon4:c.397C>T(p.R133C)	Het	XLD/XLR	missense	De novo	P
S0803	EP	M	*SCN1A*	NM_001165963:exon26:c.5674C>T(p.R1892*)	Het	AD	stop gain	De novo	P
S0804	ID	M	*ARHGEF9*	NM_015185:exon5:c.646C>T(p.R216C)	Hemi	XLR	missense	NA	LP
S0807	EP, MA	M	*PAFAH1B1*	NM_000430:exon6:c.426dupT	Het	AD	frameshift	De novo	LP
S0809	ID	M	*MECP2*	NM_004992:exon4:c.808delC	Hemi	XLD/XLR	frameshift	NA	P->LP
S0819	MD, EP	F	*MMACHC*	NM_015506:exon4:c.609G>A(p.W203*)	Het	AR	stop gain	NA	P->LP

S0819	MD, EP	F	*MMACHC*	NM_015506:exon4:c.567dupT	Het	AR	frameshift	NA	P->LP
S0820	ID	F	*MECP2*	NM_004992:exon4:c.502C>T(p.R168*)	Het	XLD/XLR	stop gain	De novo	P
S0823	EP	M	*SCN2A*	NM_021007:exon22:c.4025T>C(p.L1342P)	Het	AD	missense	De novo	P
S0826	MD	M	*JAG1*	NM_000214:exon4:c.551G>A(p.R184H)	Het	AD	missense	NA	P->LP
S0829	ID	M	*KIAA2022*	NM_001008537:exon3:c.3458dupA	Hemi	XL	frameshift	De novo	LP
S0830	EP	F	*STXBP1*	NM_003165:exon3:c.150_152delCAT	Het	AD	splicing	Not-paternal	P->LP
S0832	MA	F	*BRAF*	NM_004333:exon6:c.755G>C(p.R252P)	Het	AD	missense	De novo	P
S0833	ID	M	*HPRT1*	NM_000194:exon2:c.91_97delGATTTGG	Hemi	XLR	frameshift	Maternal	LP
S0834	EP	F	*TSC2*	NM_000548:exon2:c.133_136delCTGA	Het	AD	frameshift	De novo	P
S0835	MA	M	*TMEM67*	NM_153704:exon3:c.329A>G(p.D110G)	Hom	AR	missense	NA	P
S0836	ID	F	*ACTB*	NM_001101:exon2:c.82C>G(p.R28G)	Het	AD	missense	De novo	LP
S0837	EP	F	*TSC2*	NM_000548:exon41:c.5227C>T(p.R1743W)	Het	AD	missense	De novo	P
S0840	MA	M	*DYNC1H1*	NM_001376:exon76:c.13556C>T(p.A4519V)	Het	AD	missense	De novo	LP
S0843	MD	F	*PEX12*	NM_000286:exon3:c.888_889delCT	Het	AR	frameshift	Paternal	P
S0843	MD	F	*PEX12*	NM_000286:exon2:c.272G>A(p.R91K)	Het	AR	missense	Maternal	P
S0844	MA	M	*NSD1*	NM_022455:exon5:c.2192_2193delCT	Het	AD	frameshift	De novo	LP
S0845	BT, EP, MA	F	*MECP2*	NM_001110792:exon1:c.47_57delGCGAGGAGGAG	Het	XLD/XLR	frameshift	NA	P->LP
S0848	MA	M	*EHMT1*	NM_024757:exon17:c.2607+1G>A	Het	AD	splicing	De novo	LP
S0850	BT	M	*SLC6A8*	NM_005629:exon3:c.409T>G(p.S137A)	Hemi	XLR	missense	NA	LP
S0851	MA	M	*ISPD*	NM_001101426:exon3:c.605C>T(p.S202L)	Het	AR	missense	Maternal	LP
S0851	MA	M	*ISPD*	NM_001101426:exon10:c.1354T>C(p.*452R)	Het	AR	stop lost	Paternal	LP
S0853	ID	M	*NF1*	NM_000267:exon36:c.4992G>A(p.W1664*)	Het	AD	stop gain	NA	P->LP
S0855	ID	M	*KMT2A*	NM_001197104:exon31:c.11072-2_11072-	Het	AD	splicing	De novo	LP

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S0860	ID	F	*TH*	NM_199292:exon4:c.457C>T(p.R153*)	Het	AR	stop gain	NA	P->LP
S0860	ID	F	*TH*	NM_199292:exon6:c.698G>A(p.R233H)	Het	AR	missense	NA	P->LP
S0862	MD	F	*PCCA*	NM_000282:exon22:c.2002G>A(p.G668R)	Het	AR	missense	Paternal	P
S0862	MD	F	*PCCA*	NM_000282:exon5:c.304_317delinsGAGGA (p.H102_A106delinsEE)	Het	AR	frameshift	Maternal	LP
S0870	ID	M	*SYNE1*	NM_033071:exon111:c.20323T>C(p.S6775P)	Het	AD/AR	missense	NA	LP
S0870	ID	M	*SYNE1*	NM_033071:exon112:c.20576T>A(p.I6859N)	Het	AD/AR	missense	NA	LP
S0870	ID	M	*GDAP1*	NM_018972:exon6:c.845G>A(p.R282H)	Het	AD/AR	missense	NA	P->LP
S0871	MA	M	*MFSD8*	NM_152778:exon5:c.217dupA	Het	AR	frameshift	Maternal	P
S0871	MA	M	*MFSD8*	NM_152778:exon13:c.1444C>T(p.R482*)	Het	AR	stop gain	Paternal	P
S0873	MD	M	*TRMU*	NM_018006:exon5:c.521C>T(p.T174I)	Het	AR	missense	Paternal	P
S0873	MD	M	*TRMU*	NM_018006:exon3:c.287A>G(p.N96S)	Het	AR	missense	Maternal	P
S0879	BT	F	*SHANK3*	NM_033517:exon22:c.4946A>C(p.K1649T)	Het	AD	missense	NA	LP
S0879	BT	F	*SHANK3*	NM_033517:exon22:c.4988delAinsTCC	Het	AD	frameshift	NA	LP
S0881	MD	M	*SERAC1*	NM_032861:exon15:c.1658C>T(p.S553L)	Het	AR	missense	Maternal	LP
S0881	MD	M	*SERAC1*	NM_032861:exon14:c.1501+5G>A	Het	AR	splicing	Maternal	P
S0883	ID	M	*DNM1L*	NM_012062:exon10:c.1084G>A(p.G362S)	Het	AD/AR	missense	De novo	P
S0884	MA	M	*CREBBP*	NM_004380:exon4:c.1129_1131delinsCAAT G	Het	AD	frameshift	De novo	LP
S0888	ID	M	*UPF3B*	NM_080632:exon6:c.617_620del	Hemi	XLR	frameshift	Maternal	LP
S0889	MA	F	*NIPBL*	NM_133433:exon12:c.3316C>T(p.R1106*)	Mosaic	AD	stop gain	NA	P->LP
S0891	EP	M	*CACNA1D*	NM_000720:exon37:c.4444G>C(p.V1482L)	Het	AD/AR	missense	NA	LP
S0892	BT	F	*MYT1L*	NM_015025:exon10:c.1283A>G(p.K428R)	Het	AD	missense	NA	LP

S0892	BT	F	*ARID1B*	NM_020732:exon12:c.3223C>T(p.R1075*)	Het	AD	stop gain	NA	P->LP
S0898	MA	F	*BRAF*	NM_004333:exon6:c.770A>G(p.Q257R)	Het	AD	missense	De novo	P
S0908	ID	F	*PRX*	NM_181882:exon5:c.28G>T(p.E10*)	Het	AD/AR	stop gain	NA	LP
S0917	EP	F	*CLN5*	NM_006493:exon2:c.433C>T(p.R145*)	Hom	AR	stop gain	Parental	P
S0918	MA	F	*SMARCA2*	NM_001289399:exon2:c.56C>T(p.T19I)	Het	AD	missense	De novo	LP
S0921	ID	M	*ANKRD11*	NM_013275:exon5:c.290_318del29insC	Het	AD	frameshift	De novo	LP
S0923	BT	M	*CHD2*	NM_001271:exon38:c.5128C>T(p.R1710W)	Het	AD	missense	NA	LP
S0926	ID	F	*SETBP1*	NM_015559:exon4:c.2572G>A(p.E858K)	Het	AD	missense	NA	P->LP
S0939	ID	F	*ATP1A3*	NM_152296:exon17:c.2267G>T(p.R756L)	Het	AD	missense	NA	P->LP
S0940	MA	F	*ASPM*	NM_018136:exon3:c.1592_1595delTAAT	Het	AR	frameshift	Maternal	LP
S0940	MA	F	*ASPM*	NM_018136:exon3:c.561_562delAG	Het	AR	frameshift	Paternal	LP
S0948	MD, BT	M	*KMT2D*	NM_003482:exon14:c.4135_4136delAT	Mosaic	AD	frameshift	NA	P->LP
S0949	MD, EP	M	*SCN8A*	NM_014191:exon27:c.5615G>A(p.R1872Q)	Het	AD	missense	De novo	P
S0960	MA	M	*NAA10*	NM_003491:exon5:c.330T>A(p.H110Q)	Hemi	XLD/XLR	missense	NA	LP
S0961	EP	F	*MYT1L*	NM_015025:exon17:c.2636+1G>A	Het	AD	splicing	De novo	P
S0969	ID	M	*ATRX*	NM_000489:exon7:c.579A>C(p.Q193H)	Hemi	XLD/XLR	missense	NA	LP
S0969	ID	M	*ARID1B*	NM_020732:exon1:c.953G>T(p.G318V)	Het	AD	missense	NA	LP
S0969	ID	M	*MECP2*	NM_004992:exon4:c.419C>T(p.A140V)	Hemi	XLD/XLR	missense	NA	P->LP
S0970	BT	M	*DYRK1A*	NM_001396:exon4:c.475dupT	Het	AD	frameshift	NA	LP
S0974	ID	M	*KIF1A*	NM_004321:exon27:c.2761-1G>C	Het	AD/AR	splicing	NA	LP
S0974	ID	M	*KIF1A*	NM_004321:exon22:c.2108A>G(p.E703G)	Het	AD/AR	missense	NA	LP
S0976	MA	F	*CHD7*	NM_017780:exon17:c.3990-2A>G	Het	AD	splicing	NA	P->LP
S0984	EP	F	*STXBP1*	NM_003165:exon10:c.874C>T(p.R292C)	Het	AD	missense	De novo	P
S0986	ID	F	*HAX1*	NM_006118:exon3:c.430dupG	Het	AR	frameshift	Paternal	P
S0986	ID	F	*HAX1*	NM_006118:exon2:c.216_217insC	Het	AR	frameshift	Maternal	LP
S0993	MD	M	*GCDH*	NM_000159:exon11:c.1173dupG	Het	AR	frameshift	Paternal	P

S0993	MD	M	*GCDH*	NM_000159:exon10:c.1064G>A(p.R355H)	Het	AR	missense	Maternal	P
S0994	ID	M	*KIAA2022*	NM_001008537:exon3:c.2299A>C(p.T767P)	Hemi	XLD	missense	Maternal	LP
S0995	MA	M	*KDM5C*	NM_004187:exon16:c.2353_2354insGAGG	Hemi	XLR	frameshift	NA	LP
S0997	MD	F	*GALT*	NM_000155:exon9:c.904+1G>T	Hom	AR	splicing	NA	P
S0999	EP	F	*SCN1A*	NM_001165963:exon4:c.524C>T(p.A175V)	Het	AD	missense	De novo	P
S1002	MA	F	*CREBBP*	NM_004380:exon24:c.4133+1G>A	Het	AD	splicing	De novo	P
S1004	EP	F	*SCN2A*	NM_021007:exon19:c.3579_3580del	Het	AD	frameshift	De novo	LP
S1006	EP	M	*PIGO*	NM_032634:exon7:c.1329G>A(p.W443*)	Het	AR	stop gain	Paternal	LP
S1006	EP	M	*PIGO*	NM_032634:exon7:c.1382C>A(p.S461Y)	Het	AR	missense	Maternal	LP
S1007	MA	M	*SMC1A*	NM_006306:exon21:c.3146G>A(p.R1049Q)	Hemi	XLD	missense	NA	P->LP
S1008	EP, MA	M	*CDKL5*	NM_003159:exon12:c.1730--_1731dupTG	Hemi	XL	frameshift	De novo	LP
S1009	ID	M	*ARID1B*	NM_020732:exon9:c.2641C>T(p.Q881*)	Het	AD	stop gain	De novo	LP
S1014	MA	F	*ZEB2*	NM_014795:exon8:c.1027C>T(p.R343*)	Het	AD	stop gain	De novo	P
S1015	EP	M	*SCN1A*	NM_001165963:exon26:c.5624T>A(p.V1875D)	Het	AD	missense	De novo	LP
S1016	MA	M	*FBN1*	NM_000138:exon43:c.5284G>A(p.G1762S)	Het	AD	missense	NA	P->LP
S1017	MD	M	*PDHA1*	NM_000284:exon11:c.1159_1162dupAAGT	Hemi	XLD	frameshift	De novo	P
S1018	MA	M	*PLA2G6*	NM_003560:exon13:c.1771C>T(p.R591W)	Het	AR	missense	NA	P->LP
S1018	MA	M	*PLA2G6*	NM_003560:exon7:c.1077G>A(p.S359S)	Het	AR	splicing	NA	P->LP
S1020	EP	M	*PNKP*	NM_007254:exon10:c.874G>A(p.G292R)	Het	AR	missense	Not-maternal	P->LP
S1020	EP	M	*PNKP*	NM_007254:exon6:c.635A>C(p.K212T)	Het	AR	missense	Maternal	LP
S1021	BT	M	*SHANK3*	NM_033517:exon21:c.3457C>T(p.R1153*)	Het	AD	stop gain	NA	LP
S1024	MA	F	*SATB2*	NM_015265:exon9:c.1255C>T(p.Q419*)	Het	AD	stop gain	De novo	LP
S1025	ID	M	*SMARCA2*	NM_003070:exon25:c.3475C>G(p.R1159G)	Het	AD	missense	De novo	P
S1026	ID	F	*VPS13B*	NM_017890:exon5:c.436C>T(p.R146*)	Het	AR	stop gain	Paternal	P

S1026	ID	F	*VPS13B*	NM_017890:exon34:c.5296-2A>G	Het	AR	splicing	Maternal	LP
S1027	EP	M	*STXBP1*	NM_003165:exon9:c.751G>C(p.A251P)	Het	AD	missense	De novo	LP
S1028	MA	M	*ARID1B*	NM_020732:exon20:c.6382C>T(p.R2128*)	Het	AD	stop gain	De novo	P
S1030	EP	M	*TSC1*	NM_000368:exon15:c.1997+1G>T	Het	AD	splicing	Maternal	P
S1032	MD, MA	M	*PMM2*	NM_000303:exon5:c.395T>C(p.I132T)	Het	AR	missense	NA	P
S1034	ID	F	*DNM1L*	NM_012062:exon5:c.446G>A(p.G149E)	Het	AD/AR	missense	De novo	LP
S1035	EP	F	*SYNGAP1*	NM_006772:exon11:c.1718_1719insGCTGC	Het	AD	frameshift	De novo	P
S1036	ID	F	*MECP2*	NM_004992:exon4:c.1454_1457del TTAG	Het	XLD/XLR	frameshift	NA	P->LP
S1037	MA	M	*ZEB2*	NM_014795:exon7:c.904C>T(p.R302*)	Het	AD	stop gain	De novo	P
S1038	MA	M	*KCNK9*	NM_001282534:exon2:c.706G>A(p.G236R)	Het	AD/IM	missense	Maternal	P
S1041	ID	M	*IDS*	NM_000202:exon8:c.1165C>T(p.Q389*)	Hemi	XLR	stop gain	NA	P->LP
S1042	ID	M	*AIFM1*	NM_004208:exon8:c.784G>A(p.G262S)	Hemi	XLR	missense	NA	P->LP
S1043	ID	M	*NFIX*	NM_001271043:exon2:c.114G>A(p.W38*)	Het	AD	stop gain	NA	LP
S1044	ID	F	*ASXL3*	NM_030632:exon12:c.4706_4710delTAAAT	Het	AD	frameshift	De novo	LP
S1045	MA	M	*ANKRD11*	NM_013275:exon5:c.316C>T(p.R106*)	Het	AD	stop gain	NA	LP
S1046	ID	M	*PLP1*	NM_000533:exon3:c.453+2T>C	Hemi	XLR	splicing	De novo	P
S1047	MA	M	*CHD7*	NM_017780:exon34:c.7343C>G(p.S2448*)	Het	AD	stop gain	NA	LP
S1048	ID	F	*MECP2*	NM_004992:exon4:c.808C>T(p.R270*)	Het	XLD/XLR	stop gain	De novo	P
S1049	ID	M	*SHOC2*	NM_007373:exon2:c.4A>G(p.S2G)	Het	AD	missense	NA	P->LP
S1050	MD	M	*ATP7B*	NM_000053:exon8:c.2304dupC	Het	AR	frameshift	Maternal	P
S1050	MD	M	*ATP7B*	NM_000053:exon13:c.2975C>T(p.P992L)	Het	AR	missense	Paternal	P
S1051	ID	M	*PLA2G6*	NM_003560:exon14:c.1982C>T(p.T661M)	Het	AR	missense	Maternal	P
S1051	ID	M	*PLA2G6*	NM_003560:exon12:c.1693A>T(p.K565*)	Het	AR	stop gain	Paternal	LP
S1052	MD, EP	M	*PDHA1*	NM_000284:exon11:c.1132C>T(p.R378C)	Hemi	XL	missense	De novo	P
S1090	ID	F	*KCND3*	NM_004980:exon2:c.869G>A(p.R290Q)	Het	AD	missense	De novo	LP
S1067	MA	M	*NSD1*	NM_022455:exon5:c.1810C>T(p.R604*)	Het	AD	stop gain	De novo	P

S1071	BT	M	*SCN2A*	NM_021007:exon9:c.1117_1118GC>AA(p.A373N)	Het	AD	missense	NA	LP

ID: Isolated DD; ***MA***: DD with malformations; ***EP***: DD with epilepsy; ***BT***: DD with behavioral troubles; ***MD***: DD with metabolic disorder; ***M***: Male; ***F***: Female; ***Hom***: homozygous; ***Het***: heterozygous; ***Hemi***: Hemizygous; ***AD***: autosomal dominant; ***AR***: autosomal recessive; ***XL***: X-linked; ***XLD***: X-linked dominant; ***XLR***: X-linked recessive; ***IM***: imprinted; ***P***: Pathogenic; ***LP***: likely pathogenic;