

ID	SNP/INDEL	Gene	Inheritance_DB	Phenotype	Positive_Reason
WP0001	c.1777G>T;p.E593*;	ASXL3	AD	DD+CA	SNP;
WP0010	c.1925_1926del;p.E642Vfs*5;	CTNNB1	AD	DD+CA	INDEL;
WP0016	c.2053G>A;p.V685M;	SMARCA2	AD	DD+CA	SNP;
WP0026	c.2503_2507dup;p.K836Nfs*7;	UBE3A	AD	DD+CA	INDEL;
WP0043	c.2755_2757del;p.V919del;	ATP1A3	AD	CA	INDEL;
WP0049	c.1147T>C;p.F383L;	GABRG2	AD	DD	SNP;
WP0056	c.1983_1984insTCCTA;p.M662Sfs*2;	CTNNB1	AD	DD	INDEL;
WP0057	c.5425C>T;p.R1809C;	NF1	AD	CA	SNP;
WP0059	c.7296C>A;p.C2432*;	ACAN	AD	CA	SNP;
WP0064	c.1579C>T;p.R527X;	FOXP1	AD	DD	SNP;
WP0094	c.896dupA;p.E299fs;	PTEN	AD	late onset	INDEL;
WP0103	c.629C>T;p.T210M;	KCNB1	AD	DD	SNP;
WP0107	c.3070C>T;p.R1024X;	KAT6A	AD	CA	SNP;
WP0111	c.2581C>T;p.R861W;	CHD3	AD	DD	SNP;
WP0116	c.184T>G;p.Y62D;	PTPN11	AD	DD+CA	SNP;
WP0117	c.3762delT;p.N1254fs;	ASXL1	AD	DD+CA	INDEL;
WP0120	c.11797C>T;p.Q3933X;	KMT2D	AD	CA	SNP;
WP0123	c.577C>T;p.R193W;	CACNA1G	AD	DD+CA	SNP;
WP0126	c.8181_8183del;p.E2727del;	PIEZO2	AD	CA	INDEL;
WP0146	c.139C>T;p.R47X;	WAC	AD	DD+CA	SNP;
WP0153	c.392_395dup;p.G133fs;	HIST1H1E	AD	DD	INDEL;
WP0155	c.1100A>C;p.Q367P;	CBL	AD	DD	SNP;
WP0159	c.1123T>C;p.C375R;	ATL1	AD	CA	SNP;
WP0160	c.763G>A;p.V255I;	TUBB4A	AD	DD+CA	SNP;
WP0161	c.3429dupG;p.P1144fs;	ANKRD11	AD	DD	INDEL;
WP0164	c.391C>A;p.R131S;	KCNK9	AD	DD	SNP;
WP0167	c.5640_5647del;p.P1880fs;	KAT6B	AD	DD+CA	INDEL;
WP0172	c.4681C>T;p.Q1561X;	NSD1	AD	DD+CA	SNP;
WP0173	c.8500delT;p.S2834fs;	KMT2D	AD	DD+CA	INDEL;
WP0175	c.934C>T;p.R312C;	KCNB1	AD	DD+CA	SNP;
WP0187	c.260delT;p.I87fs;	MAT1A	AD	CA	INDEL;
WP0196	c.1187delT;p.L396fs;	PAX3	AD	CA	INDEL;
WP0205	c.12209_12210del;p.S4070fs;	KMT2D	AD	DD+CA	INDEL;
WP0207	c.10992_11000del;p.3664_3667del;	SYNE1	AD	CA	INDEL;
WP0228	c.475G>A;p.D159N;	TPM1	AD	CA	SNP;
WP0252	c.4379G>A;p.R1460Q	FGFR3	AD	DD+CA	SNP
WP0256	c.587G>A;p.R196H;	ACTB	AD	DD+CA	SNP;
WP0257	c.10748dupG;p.G3583fs;	PKD1	AD	CA	INDEL;
WP0260	c.2153G>C;p.R718P;	COMP	AD	DD	SNP;
WP0271	c.431G>A;p.R144Q;	KCNQ2	AD	DD	SNP;
WP0273	c.76+1G>T.;	SALL1	AD	CA	SNP;
WP0281	c.776A>G;p.Y259C;	TGFBR2	AD	CA	SNP;
WP0282	c.640C>T;p.R214C;	GABRA1	AD	DD	SNP;
WP0284	c.1522A>G;p.M508V;	PTPN11	AD	DD+CA	SNP;
WP0290	c.293G>A;p.G98E;	TUBB2A	AD	CA	SNP;
WP0291	c.548A>C;p.E183A;	PPP1CB	AD	DD	SNP;
WP0296	c.1118dupC;p.P373fs;	BCL11A	AD	DD	INDEL;
WP0297	c.368G>A;p.R123H;	TUBA1A	AD	DD+CA	SNP;
WP0298	c.2852+1G>A.;	MYT1L	AD	DD+CA	SNP;
WP0303	c.1375delG;p.D459fs;	CTNNB1	AD	DD+CA	INDEL;
WP0315	c.1393delA;p.K465fs;	DYRK1A	AD	DD+CA	INDEL;
WP0316	c.1123_1143delinsGGCGCCAG CGAGCAGCAGCT;p.S375GfsX47;	SOX11	AD	DD	INDEL
WP0336	c.3321delA;p.S1107fs;	FLG	AD	CA	INDEL;
WP0337	c.3584+5G>A.;	CREBBP	AD	DD+CA	SNP;
WP0338	c.1345T>C;p.S449P;	DNMT3A	AD	DD	SNP;
WP0340	c.758T>C;p.L253P;	PIK3R2	AD	DD	SNP;

WP0341	c.2525A>G:p.N842S;	CDK13	AD	DD+CA	SNP;
WP0346	c.2738_2739del:p.Y913fs;	FBXO11	AD	DD	INDEL;
WP0349	c.1138G>A:p.G380R;	FGFR3	AD	DD+CA	SNP;
WP0353	c.5740C>T:p.R1914C;	NSD1	AD	DD+CA	SNP;
WP0356	c.2532G>A:p.W844X;	GRIN2B	AD	DD	SNP;
WP0362	c.937C>T:p.Q313X;	DYRK1A	AD	DD+CA	SNP;
WP0366	c.748G>A:p.E250K;	PSTPIP1	AD	CA	SNP;
WP0367	c.224G>A:p.R75Q;	GJB2	AD	CA	SNP;
WP0369	c.2050G>A:p.E684K;	PDE4D	AD	CA	SNP;
WP0374	c.2056G>A:p.V686M;	GRIN2B	AD	DD+CA	SNP;
WP0380	c.274C>T:p.R92W;	NR5A1	AD	CA	SNP;
WP0386	c.606_610del:p.I202fs;	KIF11	AD	DD+CA	INDEL;
WP0388	c.2880_2883del:p.C960fs;	MED13L	AD	DD+CA	INDEL;
WP0390	c.392G>A:p.R131H;	KCNK9	AD	DD+CA	SNP;
WP0395	c.341G>A:p.R114H;	FGF12	AD	DD	SNP;
WP0399	c.292G>C:p.G98R;	TUBB2B	AD	DD+CA	SNP;
WP0401	c.4234_4235insCT:p.P1412fs;	NF1	AD	DD	INDEL;
WP0403	c.1100_1101insGAT:p.K367delinsKM;	SYT1	AD	DD	INDEL;
WP0407	c.1217+2T>A.;	DEPDC5	AD	CA	SNP;
WP0415	c.239T>C:p.I80T;	GNB1	AD	DD+CA	SNP;
WP0418	c.331A>G:p.K111E;	NR2F1	AD	DD	SNP;
WP0429	c.506G>T:p.C169F;	KCNQ2	AD	DD+CA	SNP;
WP0433	c.2967delT:p.N989fs;	SETBP1	AD	DD	INDEL;
WP0434	c.5788+5G>A.;	FBN1	AD	CA	SNP;
WP0456	c.1400G>A:p.R467H;	SPAST	AD	CA	SNP;
WP0458	c.1034T>C:p.L345P;	PTEN	AD	DD	SNP;
WP0459	c.484A>G:p.N162D;	PURA	AD	DD+CA	SNP;
WP0463	c.792del:p.F264Lfs*23;	STK11	AD	CA	INDEL;
WP0464	c.1185+1G>A.;	NF1	AD	CA	SNP;
WP0465	c.1963G>C:p.G655R;	COL1A2	AD	CA	SNP;
WP0477	c.1166G>A:p.R389H;	SATB2	AD	DD	SNP;
WP0480	c.1516C>T:p.R506X;	ATP6V1B2	AD	DD+CA	SNP;
WP0486	c.654_672del:p.Q219fs;	GATAD2B	AD	DD	INDEL;
WP0493	c.1850_1851insCGGCAT:p.619_620dup	GRIN1	AD	DD+CA	INDEL;
WP0496	c.1715G>A:p.W572X;	SYNGAP1	AD	DD	SNP;
WP0501	c.4663-2A>G.;	TSC2	AD	CA	SNP;
WP0503	c.1690G>T:p.E564*;	SYNGAP1	AD	DD	SNP;
WP0504	c.419_420insCT:p.L140fs;	COL1A1	AD	CA	INDEL;
WP0510	c.2911G>C:p.D971H;	CHD3	AD	DD	SNP;
WP0518	c.2425G>A:p.E809K;	WFS1	AD	DD+CA	SNP;
WP0520	c.7727C>A:p.S2576*;	SRCAP	AD	DD+CA	SNP;
WP0531	c.124C>T:p.L42F;	MAP2K1	AD	DD	SNP;
WP0549	c.2084G>A:p.R695Q;	GRIN2A	AD	DD	SNP;
WP0555	c.2619_2620del:p.V873fs;	SMC3	AD	DD	INDEL;
WP0564	c.5170delC:p.L1724fs;	MED13L	AD	CA	INDEL;
WP0565	c.1466A>G:p.Y489C;	NF1	AD	DD+CA	SNP;
WP0568	c.1516C>T:p.R506*;	WASF1	AD	DD	SNP;
WP0574	c.1732C>T:p.R578C;	MAPK8IP3	AD	DD+CA	SNP;
WP0579	c.3373C>T:p.R1125*;	FBN1	AD	DD+CA	SNP;
WP0586	c.677C>T:p.T226M;	SCN1A	AD	DD	SNP;
WP0594	c.1738C>T:p.R580W;	TCF4	AD	DD	SNP;
WP0600	c.840C>A:p.Y280*;	SYNGAP1	AD	DD	SNP;
WP0601	c.1288G>A:p.E430K;	SCN2A	AD	DD	SNP;
WP0621	c.641G>A:p.G214D;	SCN8A	AD	DD+CA	SNP;
WP0622	c.1391A>G:p.H464R;	USP7	AD	DD+CA	SNP;
WP0623	c.2609dup:p.H871Afs*67;	SYNGAP1	AD	DD	INDEL;
WP0624	c.140G>A:p.R47Q;	CSNK2A1	AD	DD+CA	SNP;
WP0625	c.856C>T:p.R286*;	KAT6A	AD	DD+CA	SNP;

WP0014	c.1064C>T;p.S355L;c.51C>G;p.F17L;	SLC22A5	AR	CA	SNP;SNP;
WP0035	1490G>A;p.S497N;c.1400C>G;p.S467	SLC22A5	AR	CA	SNP;SNP;
WP0039	T>C;p.L179P;c.625_628dup;p.W210S	TREX1	AR	DD	SNP;INDEL;
WP0054	.696G>A;p.W232*;c.605G>A;p.R202H	TH	AR	DD	SNP;SNP;
WP0065	c.514G>A;p.G172S;c.5C>T;p.A2V;	ECHS1	AR	DD+CA	SNP;SNP;
WP0096	55_1156del;p.T385fs;c.1582C>T;p.R5	COASY	AR	DD+CA	INDEL;SNP;
WP0105	1174T>A;p.F392I;c.1162G>A;p.V388N	PAH	AR	CA	SNP;SNP;
WP0109	c.677G>A;p.C226Y;c.318C>A;p.Y106X	GLRA1	AR	DD+CA	SNP;SNP;
WP0118	3478C>T;p.R1160X;c.1003C>T;p.Q335	NPHS1	AR	CA	SNP;SNP;
WP0178	1037T>G;p.L346R;c.1499A>C;p.Q500	IDUA	AR	CA	SNP;SNP;
WP0180	1214G>A;p.G405D;c.1074delT;p.P358	ANTXR2	AR	CA	SNP;INDEL;
WP0184	1286G>T;p.R429L;c.1396delA;p.K466	DLAT	AR	DD+CA	SNP;INDEL;
WP0212	c.2566C>T;p.Q856X;c.2364+1G>A.;	MPDZ	AR	CA	SNP;SNP;
WP0251	2911C>T;p.R971X;c.8872G>T;p.E2958	VPS13B	AR	DD+CA	SNP;SNP;
WP0279	10_522del;p.A170fs;c.1319G>C;p.R44	PANK2	AR	DD	INDEL;SNP;
WP0289	c.2715+1G>A.;;c.1090C>T;p.R364X;	GNPTAB	AR	CA	SNP;SNP;
WP0305	c.527G>C;p.C176S;c.533G>A;p.G178E	GCDH	AR	DD	SNP;SNP;
WP0318	A;p.R133Q;c.286C>T;p.R96C;c.283G>	PARS2	AR	DD	SNP;SNP;SNP;
WP0354	c.518T>A;p.I173N;	CYP21A2	AR	CA	SNP
WP0372	700G;c.7782_7783del;p.Q2594fs;c.17	ASPM	AR	CA	SNP;INDEL;SNP;
WP0375	38T>C;p.L613P;c.1603del;p.V536Cfs*	FSHR	AR	CA	SNP;INDEL;
WP0400	c.962G>A;p.R321H;	RPGRIP1L	AR	CA	SNP;
WP0402	706A>G;p.N236D;c.1157A>G;p.N386	ALDH3A2	AR	DD	SNP;SNP;
WP0411	c.1995+1G>A.;;c.79C>T;p.Q27X;	SNX14	AR	DD	SNP;SNP;
WP0428	c.1264-12T>A.;;c.2168A>G;p.H723R;	SLC26A4	AR	CA	SNP;SNP;
WP0432	c.1A>G;p.M1V;c.567dupT;p.R189fs;	MMACHC	AR	CA	SNP;INDEL;
WP0449	.1238G>C;p.R413P;c.770G>T;p.G257V	PAH	AR	DD	SNP;SNP;
WP0450	254G>T;p.G85V;c.446_449del;p.M149	ACADM	AR	CA	SNP;INDEL;
WP0461	c.419C>T;p.P140L;	PNPT1	AR	DD+CA	SNP;
WP0471	c.6940+1G>T.;;c.11972A>T;p.K3991I;	VPS13B	AR	DD+CA	SNP;SNP;
WP0473	c.823G>A;p.G275R;c.1707+5G>A.;;	SLC26A4	AR	CA	SNP;SNP;
WP0476	0+1G>T.;;c.11843_11844insGTCA;p.L3	VPS13B	AR	DD	SNP;INDEL;
WP0487	.949C>T;p.R317X;c.1198C>T;p.Q400X	TRIP4	AR	DD	SNP;SNP;
WP0507	4C>T;p.R362C;c.985_987delinsAA;p.Y	CYP17A1	AR	CA	SNP;INDEL;
WP0517	171_174del;p.H57fs;c.242G>A;p.W81	METTL23	AR	DD	INDEL;SNP;
WP0525	c.254delA;p.D85fs;	POMC	AR	CA	INDEL
WP0571	6G>A;p.V95I;c.1081_1082del;p.V361P	PARS2	AR	DD	SNP;INDEL;
WP0572	I;p.E1897Nfs*6;c.5540_5541del;p.V18	ASPM	AR	CA	SNP;INDEL;
WP0587	c.2158T>C;p.F720L;c.1797_1798dup	ERCC2	AR	DD+CA	SNP;
WP0588	c.603-1G>A.;;c.390+5G>T.;;	PKHD1	AR	CA	SNP;
WP0589	3467C>T;p.S1156L;c.6370G>T;p.E212	PKHD1	AR	CA	SNP;
WP0597	c.448-1_448del.;;c.710C>T;p.T237M;	PMM2	AR	DD+CA	INDEL;
WP0604	c.640A>G;p.T214A;	IVD	AR	CA	SNP;
WP0607	c.478G>C;p.D160H;	SLC45A2	AR	CA	SNP;
WP0015	c.1042A>G;p.T348A;	EDA	XLR	CA	SNP;
WP0041	c.287G>A;p.W96*;	UBE2A	XLR	DD+CA	SNP;
WP0050	c.789del;p.G264Afs*37;	MECP2	XLD	CA	INDEL;
WP0058	c.1713_1716del;p.S571Rfs*13;	KIAA2022	XLD	DD+CA	INDEL;
WP0068	c.322-1G>T.;;	COL4A5	XLD	CA	SNP;
WP0106	c.2675+1G>C.;;	PCDH19	XL	DD	SNP;
WP0110	c.374A>G;p.N125S;	PCDH19	XL	DD	SNP;
WP0114	c.241+1G>A.;;	UBE2A	XLR	DD+CA	SNP;
WP0115	c.2758C>T;p.Q920X;	KIAA2022	XLD	DD+CA	SNP;
WP0133	c.1045+3_1045+13del.;;	NHS	XLD	CA	INDEL;
WP0145	c.816_819del;p.L272fs;	IL2RG	XLR	CA	INDEL;
WP0152	c.352C>T;p.R118X;	CNKSR2	XL	DD	SNP;
WP0240	c.511C>T;p.R171X;	SLC16A2	XL	DD+CA	SNP;
WP0254	c.447dupA;p.V149fs;	PQBP1	XLR	DD+CA	INDEL;

WP0263	c.1915C>T:p.R639X;	CASK	XLD	DD+CA	SNP;
WP0268	c.577C>T:p.R193W;	SLC35A2	XLD	DD+CA	SNP;
WP0274	c.3241G>A:p.V1081M;	KDM5C	XLR	DD	SNP;
WP0382	c.316A>G:p.K106E;	SRY	YL	CA	SNP;
WP0383	c.99dupC:p.F34fs;	SRY	YL	CA	INDEL;
WP0385	c.979_980del:p.L327fs;	KDM5C	XLR	DD	INDEL;
WP0413	c.1169C>T:p.P390L;	SLC6A8	XLR	DD	SNP;
WP0420	c.538C>T:p.R180X;	MECP2	XLD	DD+CA	SNP;
WP0430	c.838A>G:p.K280E;	PRPS1	XL	CA	SNP;
WP0479	c.587delC:p.P196fs;	RPS6KA3	XLD	DD+CA	INDEL;
WP0512	c.763C>T:p.R255*;	MECP2	XLD	DD	SNP;
WP0526	c.1035C>A:p.C345X;	KIAA2022	XLD	DD	SNP;
WP0533	c.336G>A:p.W112*;	KIAA2022	XLD	DD+CA	SNP;
WP0536	c.1304_1307del:p.V435Afs*15;	RPS6KA3	XLD	DD+CA	INDEL;
WP0543	c.461T>A:p.V154E;	DKC1	XLR	DD+CA	SNP;
WP0613	c.10753C>T:p.Q3585*;	DMD	XLR	DD+CA	SNP;
WP0627	c.1960_1968del:p.V654_E656del;	USP9X	XLD	DD	INDEL;