

Supplementary table 2: A list of all DNMs with calculated CADD, GERP, SIFT, Polyphen2 scores, MutationTaster predictions and allele frequencies from the Exome Sequencing Project (ESP) and 1000 genome project. For MutationTaster predictions, A=Disease causing automatic, D=Disease causing, N=Polymorphism, 0=No prediction. The last column shows the Pubmed ID associated to sequencing projects where the exact same mutation has previously been found.

Chr	Start	Stop	Cons	Ref	Gene	Family	Type	CADD	GERP	SIFT	MutationTaster Pred	Polyphen2	ESP6500
chr3	9490126	9490126	G	T	SETD5	Fam5	Stopgain	46	5,76	0,01	A	0	0
chrX	53423489	53423489	G	A	SMC1A	Fam6	Stopgain	43	4,98	0,09	D	0	0
chrX	18598085	18598085	C	T	CDKL5	Fam2	Stopgain	38	-0,78	1	A	0	0
chr5	161576159	161576159	G	A	GABRG2	Fam8	Nonsynonymous	36	5,88	0	D	1	0
chr12	53702981	53702981	C	T	AAAS	Fam8	Nonsynonymous	35	5,34	0,1	D	0,998	0,000077
chr3	56330352	56330352	C	T	ERC2	Fam5	Nonsynonymous	32	5,86	0,02	D	0,974	0
chr13	75869010	75869010	C	G	TBC1D4	Fam6	Nonsynonymous	27,4	5,55	0,02	D	0,988	0
chr6	33400477	33400477	C	T	SYNGAP1	Fam4	Stopgain	25,9	4,06	1	A	0	0
chr2	166166923	166166923	C	T	SCN2A	Fam10	Nonsynonymous	23,3	5,27	0	D	0,999	0
chr17	42931953	42931953	T	G	EFTUD2	Fam7	Nonsynonymous	21,8	5,14	0,32	D	0,457	0
chr20	62044879	62044879	C	A	KCNQ2	Fam3	Nonsynonymous	20,7	4,99	0	D	1	0
chr19	18990105	18990105	A	T	CERS1	Fam14	Nonsynonymous	20,4	3,71	1	D	0,94	0
chr12	5021751	5021751	C	T	KCNA1	Fam12	Nonsynonymous	20,2	4,9	0	D	1	0
chrX	70349234	70349234	G	A	MED12	Fam16	Nonsynonymous	19,76	5,18	0,11	D	0,921	0
chr10	298399	298399	C	T	ZMYND11	Fam7	Nonsynonymous	19,56	4,32	0	D	0,999	0
chrX	109937496	109937496	G	A	CHRD1	Fam15	Nonsynonymous	19,11	5,15	0	D	0,992	0
chr2	197090524	197090524	G	A	HECW2	Fam21	Nonsynonymous	18,9	3,32	0,01	D	0,99	0
chr3	148459465	148459465	T	C	AGTR1	Fam18	Nonsynonymous	17,58	-10,4	0	D	1	0
chr4	2181074	2181074	T	G	POLN	Fam23	Nonsynonymous	15,26	-1,69	0,15	N	0,383	0
chr9	140053150	140053150	A	C	GRIN1	Fam9	Nonsynonymous	12,87	0,298	0,3	D	0,133	0
chr14	35231162	35231162	A	C	BAZ1A	Fam19	Nonsynonymous	12,35	1,16	0,85	D	0,006	0
chr12	56717668	56717668	G	T	PAN2	Fam22	Nonsynonymous	11,86	5,42	0,82	D	0,055	0
chr11	8752629	8752629	G	C	ST5	Fam11	Nonsynonymous	10,6	2,88	0,09	N	0,013	0
chr6	138576840	138576840	C	T	KIAA1244	Fam6	Synonymous	9,97	2,41	0	0	0	0
chr3	133653644	133653644	C	T	SLCO2A1	Fam13	Synonymous	9,162	5,48	0	0	0	0
chr1	207196417	207196417	T	C	C1orf116	Fam20	Nonsynonymous	8,709	0,303	0,78	N	0,037	0
chr1	11562018	11562018	G	A	PTCH2	Fam3	Synonymous	8,536	-2,8	0	0	0	0
chr15	52074974	52074974	C	T	TMOD2	Fam5	Synonymous	8,227	-11,3	0	0	0	0
chr22	43089346	43089346	C	G	A4GALT	Fam17	Synonymous	7,733	-2,26	0	0	0	0

**MutationTaster predictions: A=Disease causing automatic, D=Disease causing, N=Polymorphism, 0=No prediction.**

1000 Genomes	BP substitution	AA substitution	Mutation previously reported
0	G2158T	E720X	PMID:23020937
0	C2611T	Q871X	None
0	C400T	R134X	PMID:21318334
0	G968A	R323Q	PMID:23708187
0	G895A	G299S	None
0	G769A	E257K	None
0	G3296C	G1099A	None
0	C403T	R135X	None
0	C788T	A263V	PMID:20956790
0	A2230C	I744L	None
0	G1687T	D563Y	PMID:23621294
0	T845A	F282Y	None
0	C1207T	P403S	None
0	G3646A	V1216M	None
0	C1705T	R569W	PMID:25356970
0	C670T	R224C	None
0	C3988T	R1330W	None
0	T643C	Y215H	None
0	A1140C	K380N	None
0	A1191C	R397S	None
0	T4044G	F1348L	None
0	C2107A	Q703K	None
0	C208G	R70G	None
0	C1038T	L346L	None
0	G1845A	A615A	None
0	A692G	H231R	None
0	G969A	K323K	None
0	C681T	H227H	None
0	G612C	L204L	None