

Supplementary table 1. Filtered variants that segregate with the phenotype in families: Br-F3 and Br-F4

Family	AAChange.refGene	esp6500 si	1000g 2012	609Brazil	snp137	avsift	PP2 HDIV	LRT	Mutation Taster	Mutation Assessor	GERP++
Br-F3											
nonsynonymous SNV	CELA3A:NM_005747:exon8:c.803C>A:p.A268E	0	0	0.000000	rs80099629	0.05	B	U	N	neutral	-0.002
nonsynonymous SNV	RASSF5:NM_182663:exon1:c.113C>T:p.P38L	0	0	0.000000	.	0	B	U	N	neutral	0.712
frameshift insertion	ALK:NM_004304:exon19:c.3091_3092insCC:p.L1031fs	0	0	0.000000	.						
nonsynonymous SNV	SDAD1:NM_018115:exon21:c.1919C>T:p.S640L	0	0	0.000000	.	0.01	P	D		low	4.7
nonsynonymous SNV	COL9A1:NM_001851:exon37:c.2552A>G:p.N851S	0	0	0.000000	.	0.13	B	D	N	neutral	5.43
nonsynonymous SNV	NUPL2:NM_007342:exon4:c.457A>G:p.I153V	0.000615	0.0005	0.000821	rs141747889	0.29	B	D	N	low	5.47
nonsynonymous SNV	DDC:NM_000790:exon9:c.908A>C:p.K303T	0	0	0.000000	.	0	D	D		high	5.2
frameshift insertion	PDK4:NM_002612:exon10:c.1001dupT:p.L334fs	0	0	0.000000	.						
nonsynonymous SNV	C10orf68:NM_024688:exon14:c.1096C>T:p.H366Y	0.000077	0	0.000000	rs200088472	0	P		N	low	-1.09
frameshift insertion	ASCC1:NM_001198798:exon3:c.157dupG:p.E53fs	0	0	0.000000	.						
stopgain SNV	LRIT1:NM_015613:exon3:c.609G>A:p.W203X	0	0	0.000000	rs201482815	0.08		D	A		5.91
nonsynonymous SNV	HIPK3:NM_005734:exon2:c.371G>A:p.R124Q	0	0	0.000000	.	0.01	D	D	D	medium	5.65
nonsynonymous SNV	DAGLA:NM_006133:exon20:c.2252G>A:p.R751H	0	0	0.000000	.	0.07	B	D	N	neutral	3.1
nonsynonymous SNV	C2CD2L:NM_014807:exon14:c.2014G>C:p.A672P	0	0	0.000000	.	0.13	P	N	D	neutral	3.31
frameshift deletion	IFT88:NM_006531:exon15:c.1257_1260del:p.419_420del	0	0	0.000000	.						
nonsynonymous SNV	C14orf101:NM_017799:exon1:c.116T>C:p.V39A	0	0	0.000000	.	0.01	B	N	N	low	5.14
nonsynonymous SNV	GOLGA6D:NM_001145224:exon7:c.530G>C:p.C177S	0	0	0.000000	rs201109551	1	B			neutral	0.589
nonsynonymous SNV	PER1:NM_002616:exon22:c.3583C>T:p.R1195W	0.000154	0.0005	0.000000	rs200744636	0.01	D	D	D	low	5.67
nonsynonymous SNV	KRBA2:NM_213597:exon2:c.1223T>C:p.F408S	0	0	0.000000	.	0.34	B		N	low	-1.02
nonsynonymous SNV	MYOCD:NM_153604:exon10:c.1846G>A:p.E616K	0	0	0.001642	.	0.22	D	N	D	low	5.6
nonsynonymous SNV	DHRS7B:NM_015510:exon4:c.475G>C:p.D159H	0	0	0.000000	rs200571409	0.16	B	D	D	neutral	5.63
nonsynonymous SNV	ATP6V0A1:NM_005177:exon19:c.2120T>C:p.F707S	0	0	0.000000	.	0	D	D	D	medium	4.92
nonsynonymous SNV	NLRP2:NM_017852:exon6:c.662C>G;p.T221R	0	0	0.000000	.	0	P	N	P	medium	0.562
nonsynonymous SNV	LZTR1:NM_006767:exon8:c.742G>A:p.G248R	0	0	0.000000	.	0	D	D	D	medium	5.6
Br-F4											
nonsynonymous SNV	LZTR1:NM_006767:exon9:c.850C>T:p.R284C	0	0	0.000000	.	0	D	D	D	medium	4.14