Supplementary Table 1. Pathogenic interpretation of candidate *SALL1* variants. Y, applicable.

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51174632	CTT	C	c.1499_1500del	p.Lys500ArgfsTer15	0		frameshift_variant	Y	Y		Y						P
16	51174644	G	A	c.1489C>T	p.Gln497Ter	0		stop_gained	Y	Y		Y						P
16	51174785	TGTGTTTG	T	c.1341_1347del	p.Phe447LeufsTer44	0		frameshift_variant	Y	Y		Y						P
16	51175307	G	A	c.826C>T	p.Arg276Ter	0		stop_gained	Y	Y		Y						P
16	51171311	A	AT	c.3686dup	p.Asp1229GlufsTer48	0		frameshift_variant	Y	Y								LP
16	51172925	T	TG	c.3207dup	p.Asn1070GlnfsTer32	0		frameshift_variant	Y	Y								LP
16	51174740	G	A	c.1393C>T	p.Gln465Ter	0		stop_gained	Y	Y								LP
16	51171066	A	T	c.3932T>A	p.Phe1311Tyr	0		missense_variant		Y								VUS-P
16	51171086	C	A	c.3912G>T	p.Glu1304Asp	0		missense_variant		Y								VUS-P
16	51171090	C	A	c.3908G>T	p.Ser1303Ile	0		missense_variant		Y								VUS-P
16	51171227	G	C	c.3771C>G	p.Asn1257Lys	0		missense_variant		Y								VUS-P
16	51171378	C	T	c.3620G>A	p.Gly1207Glu	0		missense_variant		Y	Y							VUS-P
16	51171414	C	T	c.3584G>A	p.Arg1195Gln	0		missense_variant		Y								VUS-P
16	51171478	G	A	c.3535-15C>T		0		splice_polypyrimidine_tract_variant		Y								VUS-P
16	51172624	G	C	c.3509C>G	p.Ala1170Gly	0		missense_variant		Y								VUS-P
16	51172702	GAGA	G	c.3428_3430del	p.Phe1143del	0		inframe_deletion		Y			Y					VUS-P
16	51172798	G	A	c.3335C>T	p.Pro1112Leu	8.79E-06	gnomAD_AF_NFE	missense_variant		Y								VUS-P
16	51172921	T	C	c.3212A>G	p.Gln1071Arg	0		missense_variant		Y								VUS-P
16	51173038	G	T	c.3095C>A	p.Thr1032Lys	0		missense_variant		Y								VUS-P
16	51173045	T	C	c.3088A>G	p.Ile1030Val	0		missense_variant		Y								VUS-P
16	51173150	G	A	c.2983C>T	p.Arg995Trp	8.80E-06	gnomAD_AF_NFE	missense_variant		Y								VUS-P
16	51173399	G	C	c.2734C>G	p.Gln912Glu	0		missense_variant		Y								VUS-P
16	51173556	C	A	c.2577G>T	p.Glu859Asp	0		missense_variant		Y								VUS-P
16	51173563	G	A	c.2570C>T	p.Pro857Leu	0		missense_variant		Y								VUS-P
16	51173569	G	A	c.2564C>T	p.Pro855Leu	0		missense_variant		Y								VUS-P
16	51173873	T	C	c.2260A>G	p.Ser754Gly	0		missense_variant		Y								VUS-P
16	51174769	G	A	c.1364C>T	p.Ala455Val	1.15E-04	gnomAD_AF_AFR	missense_variant			Y							VUS-P

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51174794	A	T	c.1339T>A	p.Phe447Ile	0		missense_variant		Y								VUS-P
16	51174845	G	A	c.1288C>T	p.Pro430Ser	8.79E-06	gnomAD_AF_NFE	missense_variant		Y								VUS-P
16	51174920	G	T	c.1213C>A	p.Pro405Thr	0		missense_variant		Y								VUS-P
16	51174968	T	G	c.1165A>C	p.Asn389His	0		missense_variant		Y								VUS-P
16	51174997	G	A	c.1136C>T	p.Ala379Val	0		missense_variant		Y								VUS-P
16	51175396	A	G	c.737T>C	p.Leu246Ser	0		missense_variant		Y	Y							VUS-P
16	51175489	C	G	c.644G>C	p.Arg215Thr	0		missense_variant		Y								VUS-P
16	51175744	G	C	c.389C>G	p.Pro130Arg	0		missense_variant		Y								VUS-P
16	51175855	T	C	c.278A>G	p.Asn93Ser	0		missense_variant		Y								VUS-P
16	51175964	C	T	c.169G>A	p.Asp57Asn	0		missense_variant		Y								VUS-P
16	51175993	C	T	c.140G>A	p.Arg47Gln	0		missense_variant		Y								VUS-P
16	51176023	G	C	c.110C>G	p.Thr37Ser	0		missense_variant		Y								VUS-P
16	51176042	C	A	c.91G>T	p.Gly31Cys	0		missense_variant		Y								VUS-P
16	51171043	T	C	c.3955A>G	p.Lys1319Glu	1.76E-05	gnomAD_AF_NFE	missense_variant		Y						Y		VUS
16	51171061	G	A	c.3937C>T	p.Arg1313Cys	6.15E-05	gnomAD_AF_AFR	missense_variant										VUS
16	51171099	A	G	c.3899T>C	p.Met1300Thr	0		missense_variant		Y						Y		VUS
16	51171102	T	G	c.3896A>C	p.Lys1299Thr	0		missense_variant		Y				Y		Y		VUS
16	51171125	A	T	c.3873T>A	p.Asn1291Lys	0		missense_variant		Y					Y			VUS
16	51171149	C	T	c.3849G>A	p.Glu1283=	0		synonymous_variant		Y						Y	Y	VUS
16	51171168	C	T	c.3830G>A	p.Gly1277Glu	0		missense_variant		Y				Y		Y		VUS
16	51171201	C	A	c.3797G>T	p.Ser1266Ile	5.44E-05	gnomAD_AF_EAS	missense_variant										VUS
16	51171249	T	C	c.3749A>G	p.Asn1250Ser	0		missense_variant		Y	Y					Y		VUS
16	51171268	C	T	c.3730G>A	p.Gly1244Arg	6.48E-05	gnomAD_AF_NFE	missense_variant										VUS
16	51171314	C	T	c.3684G>A	p.Gly1228=	0		synonymous_variant		Y						Y	Y	VUS
16	51171373	T	G	c.3625A>C	p.Asn1209His	0		missense_variant		Y				Y				VUS
16	51171404	A	G	c.3594T>C	p.Ser1198=	0		synonymous_variant		Y							Y	VUS
16	51171479	G	A	c.3535-16C>T		6.50E-05	gnomAD_AF_NFE	splice_polypyrimidine_tract_variant										VUS
16	51172641	G	A	c.3492C>T	p.Cys1164=	0		synonymous_variant		Y						Y	Y	VUS

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51172781	T	C	c.3352A>G	p.Thr1118Ala	0		missense_variant		Y						Y		VUS
16	51172787	AGGAAGACAGAGGCCCAGACGGGACGTGACT	A	c.3316_3345del	p.His1107_Ser1116del	0		inframe_deletion		Y			Y			Y		VUS
16	51172803	A	T	c.3330T>A	p.Ser1110=	0		synonymous_variant		Y							Y	VUS
16	51172807	G	A	c.3326C>T	p.Pro1109Leu	8.79E-06	gnomAD_AF_NFE	missense_variant		Y						Y		VUS
16	51172809	G	A	c.3324C>T	p.Val1108=	0		synonymous_variant		Y							Y	VUS
16	51172856	C	T	c.3277G>A	p.Val1093Met	5.27E-05	gnomAD_AF_NFE	missense_variant										VUS
16	51172876	T	C	c.3257A>G	p.Lys1086Arg	0		missense_variant		Y						Y		VUS
16	51172896	G	A	c.3237C>T	p.Asn1079=	0		synonymous_variant		Y							Y	VUS
16	51172959	G	A	c.3174C>T	p.Ser1058=	0		synonymous_variant		Y						Y	Y	VUS
16	51173010	C	T	c.3123G>A	p.Lys1041=	0		synonymous_variant		Y							Y	VUS
16	51173060	T	C	c.3073A>G	p.Lys1025Glu	0		missense_variant		Y						Y		VUS
16	51173076	G	A	c.3057C>T	p.His1019=	0		synonymous_variant		Y						Y	Y	VUS
16	51173106	T	C	c.3027A>G	p.Thr1009=	0		synonymous_variant		Y						Y	Y	VUS
16	51173150	G	C	c.2983C>G	p.Arg995Gly	0		missense_variant		Y						Y		VUS
16	51173260	T	C	c.2873A>G	p.Asn958Ser	5.44E-05	gnomAD_AF_EAS	missense_variant										VUS
16	51173378	T	C	c.2755A>G	p.Ile919Val	0		missense_variant		Y				Y	Y	Y		VUS
16	51173448	C	T	c.2685G>A	p.Gly895=	0		synonymous_variant		Y							Y	VUS
16	51173460	C	T	c.2673G>A	p.Gly891=	0		synonymous_variant		Y							Y	VUS
16	51173497	T	C	c.2636A>G	p.Glu879Gly	0		missense_variant		Y						Y		VUS
16	51173520	C	G	c.2613G>C	p.Lys871Asn	0		missense_variant		Y				Y				VUS
16	51173580	T	C	c.2553A>G	p.Leu851=	0		synonymous_variant		Y							Y	VUS
16	51173670	T	C	c.2463A>G	p.Leu821=	0		synonymous_variant		Y							Y	VUS
16	51173724	A	C	c.2409T>G	p.Ser803=	0		synonymous_variant		Y						Y	Y	VUS
16	51173766	C	T	c.2367G>A	p.Met789Ile	8.79E-06	gnomAD_AF_NFE	missense_variant		Y						Y		VUS
16	51173792	G	A	c.2341C>T	p.Leu781=	0		synonymous_variant		Y						Y	Y	VUS
16	51173818	T	C	c.2315A>G	p.Gln772Arg	0		missense_variant		Y	Y					Y		VUS
16	51173832	G	A	c.2301C>T	p.Ser767=	8.79E-06	gnomAD_AF_NFE	synonymous_variant		Y							Y	VUS
16	51173846	T	C	c.2287A>G	p.Arg763Gly	2.89E-05	gnomAD_AF_AMR	missense_variant										VUS

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51173863	C	T	c.2270G>A	p.Arg757His	5.44E-05	gnomAD_AF_EAS	missense_variant			Y					Y		VUS
16	51174069	C	A	c.2064G>T	p.Thr688=	0		synonymous_variant		Y						Y	Y	VUS
16	51174070	G	A	c.2063C>T	p.Thr688Met	8.80E-06	gnomAD_AF_NFE	missense_variant		Y						Y		VUS
16	51174222	C	G	c.1911G>C	p.Ala637=	8.84E-06	gnomAD_AF_NFE	synonymous_variant		Y							Y	VUS
16	51174241	G	A	c.1892C>T	p.Thr631Ile	1.77E-05	gnomAD_AF_NFE	missense_variant		Y					Y			VUS
16	51174315	A	G	c.1818T>C	p.Gly606=	0		synonymous_variant		Y							Y	VUS
16	51174321	G	T	c.1812C>A	p.Asn604Lys	0		missense_variant		Y					Y			VUS
16	51174360	G	C	c.1773C>G	p.Val591=	0		synonymous_variant		Y							Y	VUS
16	51174363	T	C	c.1770A>G	p.Ser590=	0		synonymous_variant		Y						Y	Y	VUS
16	51174375	G	A	c.1758C>T	p.Ser586=	0		synonymous_variant		Y						Y	Y	VUS
16	51174413	C	T	c.1720G>A	p.Glu574Lys	1.09E-04	gnomAD_AF_EAS	missense_variant										VUS
16	51174444	G	A	c.1689C>T	p.Leu563=	0		synonymous_variant		Y							Y	VUS
16	51174467	C	T	c.1666G>A	p.Gly556Ser	2.89E-05	gnomAD_AF_AMR	missense_variant										VUS
16	51174501	G	A	c.1632C>T	p.Thr544=	0		synonymous_variant		Y						Y	Y	VUS
16	51174512	A	C	c.1621T>G	p.Trp541Gly	0		missense_variant		Y				Y				VUS
16	51174531	T	C	c.1602A>G	p.Pro534=	0		synonymous_variant		Y						Y	Y	VUS
16	51174634	T	C	c.1499A>G	p.Lys500Arg	0		missense_variant		Y				Y				VUS
16	51174783	C	T	c.1350G>A	p.Lys450=	0		synonymous_variant		Y						Y	Y	VUS
16	51174820	G	C	c.1313C>G	p.Ala438Gly	0		missense_variant		Y					Y			VUS
16	51174837	A	G	c.1296T>C	p.Asn432=	0		synonymous_variant		Y						Y	Y	VUS
16	51174838	T	C	c.1295A>G	p.Asn432Ser	8.79E-06	gnomAD_AF_NFE	missense_variant		Y				Y				VUS
16	51174843	T	G	c.1290A>C	p.Pro430=	0		synonymous_variant		Y							Y	VUS
16	51174926	G	A	c.1207C>T	p.Pro403Ser	1.63E-04	gnomAD_AF_OTH	missense_variant										VUS
16	51174942	G	C	c.1191C>G	p.Ser397=	0		synonymous_variant		Y							Y	VUS
16	51174972	C	G	c.1161G>C	p.Ala387=	0		synonymous_variant		Y						Y	Y	VUS
16	51175020	T	G	c.1113A>C	p.Ser371=	0		synonymous_variant		Y						Y	Y	VUS
16	51175042	T	C	c.1091A>G	p.His364Arg	0		missense_variant		Y					Y	Y		VUS
16	51175100	C	A	c.1033G>T	p.Ala345Ser	0		missense_variant		Y				Y	Y	Y		VUS

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51175103	A	G	c.1030T>C	p.Leu344=	0		synonymous_variant		Y							Y	VUS
16	51175144	A	G	c.989T>C	p.Ile330Thr	0		missense_variant		Y				Y	Y	Y		VUS
16	51175178	T	C	c.955A>G	p.Ile319Val	0		missense_variant		Y				Y	Y	Y		VUS
16	51175281	C	T	c.852G>A	p.Thr284=	8.79E-06	gnomAD_AF_NFE	synonymous_variant		Y							Y	VUS
16	51175281	C	G	c.852G>C	p.Thr284=	0		synonymous_variant		Y							Y	VUS
16	51175318	T	C	c.815A>G	p.Gln272Arg	0		missense_variant		Y					Y			VUS
16	51175442	C	T	c.691G>A	p.Glu231Lys	0		missense_variant		Y	Y					Y		VUS
16	51175459	G	A	c.674C>T	p.Ala225Val	0		missense_variant		Y						Y		VUS
16	51175557	G	A	c.576C>T	p.Asn192=	0		synonymous_variant		Y							Y	VUS
16	51175595	C	T	c.538G>A	p.Gly180Arg	3.27E-05	gnomAD_AF_SAS	missense_variant										VUS
16	51175620	C	T	c.513G>A	p.Ala171=	0		synonymous_variant		Y						Y	Y	VUS
16	51175666	C	T	c.467G>A	p.Ser156Asn	1.78E-05	gnomAD_AF_NFE	missense_variant		Y					Y			VUS
16	51175745	G	A	c.388C>T	p.Pro130Ser	0		missense_variant		Y					Y			VUS
16	51175752	C	T	c.381G>A	p.Val127=	0		synonymous_variant		Y						Y	Y	VUS
16	51175800	G	A	c.333C>T	p.Ser111=	0		synonymous_variant		Y							Y	VUS
16	51175865	G	C	c.268C>G	p.Pro90Ala	0		missense_variant		Y					Y			VUS
16	51175886	C	T	c.247G>A	p.Glu83Lys	8.81E-06	gnomAD_AF_NFE	missense_variant		Y					Y			VUS
16	51176001	G	A	c.132C>T	p.Val44=	0		synonymous_variant		Y							Y	VUS
16	51185104	C	G	c.49G>C	p.Glu17Gln	0		missense_variant		Y					Y	Y		VUS
16	51185110	C	G	c.43G>C	p.Asp15His	0		missense_variant		Y						Y		VUS
16	51171055	C	T	c.3943G>A	p.Val1315Met	1.23E-04	gnomAD_AF_AFR	missense_variant								Y		VUS-B
16	51171278	C	T	c.3720G>A	p.Ala1240=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51171367	C	T	c.3631G>A	p.Val1211Ile	1.40E-04	gnomAD_AF_NFE	missense_variant								Y		VUS-B
16	51172617	C	T	c.3516G>A	p.Thr1172=	1.41E-04	gnomAD_AF_NFE	synonymous_variant									Y	VUS-B
16	51172806	C	T	c.3327G>A	p.Pro1109=	5.01E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51172902	G	A	c.3231C>T	p.Pro1077=	1.85E-04	gnomAD_AF_AFR	synonymous_variant									Y	VUS-B
16	51172924	T	C	c.3209A>G	p.Asn1070Ser	5.27E-05	gnomAD_AF_NFE	missense_variant							Y			VUS-B
16	51173281	G	A	c.2852C>T	p.Ala951Val	4.01E-05	gnomAD_AF_AFR	missense_variant							Y			VUS-B

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51173346	C	T	c.2787G>A	p.Leu929=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51173564	G	T	c.2569C>A	p.Pro857Thr	5.44E-05	gnomAD_AF_EAS	missense_variant								Y		VUS-B
16	51173898	C	T	c.2235G>A	p.Thr745=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51173901	G	A	c.2232C>T	p.Thr744=	3.27E-05	gnomAD_AF_SAS	synonymous_variant									Y	VUS-B
16	51174093	C	T	c.2040G>A	p.Leu680=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51174225	C	T	c.1908G>A	p.Thr636=	2.82E-05	gnomAD_AF_AMR	synonymous_variant									Y	VUS-B
16	51174260	C	T	c.1873G>A	p.Glu625Lys	5.78E-05	gnomAD_AF_AMR	missense_variant								Y		VUS-B
16	51174537	G	A	c.1596C>T	p.Ile532=	1.63E-04	gnomAD_AF_OTH	synonymous_variant									Y	VUS-B
16	51174905	C	T	c.1228G>A	p.Gly410Arg	3.27E-05	gnomAD_AF_SAS	missense_variant								Y		VUS-B
16	51175485	G	A	c.648C>T	p.Cys216=	9.65E-05	gnomAD_AF_ASJ	synonymous_variant									Y	VUS-B
16	51175551	G	A	c.582C>T	p.Asn194=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51175638	G	C	c.495C>G	p.Ser165=	5.44E-05	gnomAD_AF_EAS	synonymous_variant									Y	VUS-B
16	51175661	T	G	c.472A>C	p.Ser158Arg	1.70E-04	gnomAD_AF_OTH	missense_variant							Y			VUS-B
16	51175721	C	T	c.412G>A	p.Gly138Ser	6.53E-05	gnomAD_AF_SAS	missense_variant							Y			VUS-B
16	51175728	G	A	c.405C>T	p.Ser135=	3.27E-05	gnomAD_AF_SAS	synonymous_variant									Y	VUS-B
16	51175736	T	C	c.397A>G	p.Asn133Asp	3.27E-05	gnomAD_AF_SAS	missense_variant							Y			VUS-B
16	51175749	C	A	c.384G>T	p.Glu128Asp	3.27E-05	gnomAD_AF_SAS	missense_variant							Y			VUS-B
16	51175782	G	A	c.351C>T	p.Asn117=	1.55E-04	gnomAD_AF_OTH	synonymous_variant									Y	VUS-B
16	51172824	G	T	c.3309C>A	p.Thr1103=	3.27E-05	gnomAD_AF_SAS	synonymous_variant								Y	Y	LB
16	51173262	G	A	c.2871C>T	p.Ala957=	5.44E-05	gnomAD_AF_EAS	synonymous_variant								Y	Y	LB
16	51173280	C	T	c.2853G>A	p.Ala951=	5.01E-05	gnomAD_AF_EAS	synonymous_variant								Y	Y	LB
16	51173805	C	T	c.2328G>A	p.Thr776=	5.44E-05	gnomAD_AF_EAS	synonymous_variant								Y	Y	LB
16	51174144	C	T	c.1989G>A	p.Leu663=	7.92E-05	gnomAD_AF_NFE	synonymous_variant								Y	Y	LB
16	51174186	G	A	c.1947C>T	p.Cys649=	1.39E-04	gnomAD_AF_OTH	synonymous_variant								Y	Y	LB
16	51174941	C	T	c.1192G>A	p.Ala398Thr	9.80E-05	gnomAD_AF_SAS	missense_variant							Y	Y		LB
16	51175080	C	T	c.1053G>A	p.Pro351=	6.16E-05	gnomAD_AF_AFR	synonymous_variant								Y	Y	LB
16	51175114	T	C	c.1019A>G	p.Asn340Ser	8.01E-05	gnomAD_AF_AFR	missense_variant							Y	Y		LB
16	51175392	G	A	c.741C>T	p.Ile247=	1.20E-04	gnomAD_AF_AFR	synonymous_variant								Y	Y	LB

CHROM	POS	REF	ALT	HGVSc (NM_002968.3)	HGVSp (NC_002959.2)	gnomAD_AF_POPMAX	MAF population	VEP_Consequence	PVS1	PM2	PP3	PP4	PM4	BS4	BP4	BP5	BP7	Variant classification
16	51175632	T	C	c.501A>G	p.Thr167=	6.16E-05	gnomAD_AF_AFR	synonymous_variant								Y	Y	LB
16	51175649	C	T	c.484G>A	p.Gly162Ser	1.31E-04	gnomAD_AF_SAS	missense_variant							Y	Y		LB
16	51175727	C	T	c.406G>A	p.Gly136Ser	1.21E-04	gnomAD_AF_AFR	missense_variant							Y	Y		LB
16	51175887	G	A	c.246C>T	p.Pro82=	9.92E-05	gnomAD_AF_ASJ	synonymous_variant								Y	Y	LB