

Supplementary Table S1. Carrier status of variants qualified as likely pathogenic or pathogenic present on the GSA v1 or v3 chips identified in GSA the Romanian cohorts evaluated. The reported numbers reflect heterozygous individuals/allele counts.

Chr:Pos (GRCh37)	Identifier rs (dbSNPv154) NM (GJB2)	ClinVar	GSAv1 Cluj n=416	GSAv3 Craiova n=472	AF 1000G EUR
13:20763039	rs111033327 NM_004004.6:c.*1C>T	Uncertain significance	NA	0	0.001
13:20763071	rs587783647 NM_004004.6:c.647_650del p.Arg216fs	Pathogenic/Likely pathogenic	0	0	-
13:20763088	rs587783646 NM_004004.6:c.632_633del NM_004004.6:c.632_633delGT p.Cys211fs	Pathogenic/Likely pathogenic	0	0	-
13:20763104	rs111033294 NM_004004.6:c.617A>G p.Asn206Ser	Pathogenic/Likely pathogenic	0	0	-
13:20763113	rs76838169 NM_004004.6:c.608T>C p.Ile203Thr	Benign	0	0	0
13:20763121	rs111033335 NM_004004.6:c.592_600 delinsCAGTGTTCATGACATTC p.Val198_Gly200delinsGlnCysSerTer	Pathogenic/Likely pathogenic	0	0	-
13:20763123	rs786204597 NM_004004.6:c.598G>A p.Gly200Arg NM_004004.6:c.598G>T p.Gly200Ter	Pathogenic/Likely pathogenic Likely pathogenic	NA NA	0 0	- -
13:20763170	rs80338950 NM_004004.6:c.551G>A p.Arg184Gln NM_004004.6:c.551G>C p.Arg184Pro	Pathogenic/Likely pathogenic Pathogenic	0 0	0 0	- -
13:20763186	rs28931595				

	NM_004004.6:c.535G>C p.Asp179His	Uncertain significance	0	0	-
	NM_004004.6:c.535G>A p.Asp179Asn	Pathogenic	0	0	-
13:20763222	rs111033360 NM_004004.6:c.499G>A p.Val167Met	Uncertain significance	0	0	0
13:20763228	rs376898963 NM_004004.6:c.493C>T p.Arg165Trp	Uncertain significance	NA	0	0
13:20763234	rs80338949 NM_004004.6:c.487A>C p.Met163Leu	Uncertain significance	0	0	-
	NM_004004.6:c.487A>G p.Met163Val	Uncertain significance	2/416	0	0
13:20763243	rs34988750 NM_004004.6:c.478G>A p.Gly160Ser	Uncertain significance	1/416	1/472	0
13:20763256	rs772264564 NM_004004.6:c.465T>A p.Tyr155Ter	Pathogenic	0	0	-
13:20763265	rs111033420 NM_004004.6:c.456C>T p.Tyr152=	Likely benign	0	0	-
	NM_004004.6:c.456C>A p.Tyr152Ter	Pathogenic/Likely pathogenic	0	0	-
13:20763293	rs104894401 NM_004004.6:c.428G>A p.Arg143Gln	Pathogenic/Likely pathogenic	NA	0	-
13:20763294	rs80338948 NM_004004.6:c.427C>T p.Arg143Trp	Pathogenic	0	0	0
13:20763295	rs397516877 NM_004004.6:c.426C>A p.Phe142Leu	Pathogenic	0	0	-
13:20763296	rs116769964 NM_004004.6:c.425T>C p.Phe142Ser	Uncertain significance	0	0	0
13:20763305	rs76434661				

	NM_004004.6:c.416G>A p.Ser139Asn	Pathogenic	0	0	0.001
13:20763313	rs786204690 NM_004004.6:c.408C>A p.Tyr136Ter	Pathogenic/Likely pathogenic	NA	0	-
	NM_004004.5:c.[134G>A;408C>A]	Pathogenic	NA	0	
13:20763332	rs779018464 NM_004004.6:c.389G>C p.Gly130Ala	Likely pathogenic	NA	0	-
13:20763351	rs397516874 NM_004004.6:c.370C>T p.Gln124Ter	Pathogenic	0	0	0
13:20763356	rs111033295 NM_004004.6:c.365A>T p.Lys122Ile	Pathogenic/Likely pathogenic	0	0	-
13:20763361	rs80338947 NM_004004.6:c.355GAG p.Glu120del	Pathogenic	0	0	-
13:20763366	rs150529554 NM_004004.6:c.355G>T p.Glu119Ter	Pathogenic/Likely pathogenic	NA	0	-
	NM_004004.6:c.355G>A p.Glu119Lys	Uncertain significance	NA	0	0.001
13:20763371	rs111033441 NM_004004.6:c.350A>G p.Asp117Gly	Uncertain significance	0	0	-
13:20763380	rs2274083 NM_004004.6:c.341A>G p.Glu114Gly	Benign	NA	0	0
	NM_004004.5:c.[79G>A;341A>G]	Benign/Likely benign	NA	0	
13:20763382	rs80338946 NM_004004.6:c.339T>G p.Ser113Arg	Uncertain significance	NA	0	-
13:20763394	rs797045596 NM_004004.6:c.314_329del p.Lys105fs	Pathogenic	0	0	-
13:20763395	rs111033253 NM_004004.6:c.313_326del p.Lys105fs	Pathogenic/Likely pathogenic	0	1/472	-

13:20763421	rs111033204 NM_004004.6:c.299_300del p.His100fs	Pathogenic	0	0	-
13:20763423	rs143343083 NM_004004.6:c.298C>T p.His100Tyr	Pathogenic/Likely pathogenic	0	0	-
13:20763438	rs111033299 NM_004004.6:c.283G>A p.Val95Met	Pathogenic/Likely pathogenic	0	0	0
13:20763452	rs80338945 NM_004004.6:c.269T>C p.Leu90Pro	Pathogenic	NA	1/472	-
13:20763471	rs104894409 NM_004004.6:c.250G>T p.Val84Leu	Pathogenic	NA	0	-
	NM_004004.6:c.250G>A p.Val84Met	Pathogenic	NA	0	-
	NM_004004.6:c.250G>C p.Val84Leu	Pathogenic	NA	0	-
13:20763482	rs727504302 NM_004004.6:c.239A>C p.Gln80Pro	Pathogenic/Likely pathogenic	0	0	-
13:20763486	rs80338943 NM_004004.6:c.235del p.Leu79fs	Pathogenic	0	0	0
13:20763490	rs80338944 NM_004004.6:c.231G>A p.Trp77Ter	Pathogenic	0	0	0
13:20763491	rs104894395 NM_004004.6:c.230G>A p.Trp77Ter	Pathogenic/Likely pathogenic	NA	0	-
13:20763492	rs104894397 NM_004004.6:c.229T>C p.Trp77Arg	Pathogenic	NA	0	-
13:20763494	rs111033361 NM_004004.6:c.227T>C p.Leu76Pro	Likely pathogenic	0	0	-
13:20763497	rs28931593 NM_004004.6:c.224G>A	Pathogenic	0	0	-

	p.Arg75Gln				
13:20763498	rs104894402				
	NM_004004.6:c.223C>G	Likely pathogenic	0	0	-
	p.Arg75Gly				
	NM_004004.6:c.223C>T	Pathogenic	0	0	-
	p.Arg75Trp				
13:20763545	rs104894404				
	NM_004004.6:c.176G>A	Likely pathogenic	0	0	-
	p.Gly59Asp				
	NM_004004.6:c.176G>C	Pathogenic	0	0	-
	p.Gly59Ala				
13:20763552	rs111033297				
	NM_004004.6:c.169C>T	Pathogenic	0	0	-
	p.Gln57Ter				
13:20763554	rs80338942				
	NM_004004.6:c.167del	Pathogenic	NA	0	-
	p.Leu56fs				
13:20763563	rs587783645				
	NM_004004.6:c.158G>T	Likely pathogenic	0	0	-
	p.Cys53Phe				
	NM_004004.6:c.158G>A	Pathogenic	0	0	-
	p.Cys53Tyr				
13:20763573	rs28931594				
	NM_004004.6:c.148G>T	Pathogenic	0	0	-
	p.Asp50Tyr				
	NM_004004.6:c.148G>A	Pathogenic	0	0	-
	p.Asp50Asn				
13:20763582	rs104894398				
	NM_004004.6:c.139G>T	Pathogenic	0	0	-
	p.Glu47Ter				
13:20763587	rs72561723				
	NM_004004.5:c.[134G>A;408C>A]	Pathogenic	0	0	-
	NM_004004.6:c.134G>A	Pathogenic/Likely pathogenic			
	p.Gly45Glu		0	0	
13:20763589	rs104894407				
	NM_004004.6:c.132G>A	Pathogenic	0	0	0
	p.Trp44Ter				
	NM_004004.6:c.132G>C	Pathogenic	0	0	-
	p.Trp44Cys				

13:20763590	rs104894413 NM_004004.6:c.131G>A p.Trp44Ter NM_004004.6:c.131G>C p.Trp44Ser	Pathogenic/Likely pathogenic Pathogenic	0 0	0 0	- -
13:20763601	rs561870637 NM_004004.6:c.120A>C p.Ala40=	Likely benign	NA	0	0.003
13:20763602	rs111033296 NM_004004.6:c.119C>G p.Ala40Gly NM_004004.6:c.119C>A p.Ala40Glu	Likely pathogenic Likely pathogenic	0 0	0 0	- -
13:20763612	rs72474224 NM_004004.6:c.109G>T p.Val37Phe NM_004004.6:c.109G>A p.Val37Ile	Likely pathogenic Pathogenic	0 3/416	0 1/472	- 0
13:20763614	rs587783644 NM_004004.6:c.107T>C p.Leu36Pro	Uncertain significance	0	0	-
13:20763620	rs35887622 NM_004004.6:c.101T>G p.Met34Arg NM_004004.6:c.101T>C p.Met34Thr	Likely pathogenic Pathogenic	0 5/416	0 5/472	- 0.0209
13:20763626	rs111033190 NM_004004.6:c.95G>T p.Arg32Leu NM_004004.6:c.95G>A p.Arg32His	Pathogenic/Likely pathogenic Pathogenic	0 0	0 0	- -
13:20763627	rs371024165 NM_004004.6:c.94C>A p.Arg32Ser NM_004004.6:c.94C>T p.Arg32Cys	Pathogenic/Likely pathogenic Pathogenic	NA NA	0 0	- -
13:20763642	rs2274084 NM_004004.5:c.[79G>A;341A>G] NM_004004.6:c.79G>A	Benign/Likely benign Benign	0 4/416	0 7/472	0

	p.Val27Ile				
13:20763650	rs104894396 NM_004004.6:c.71G>A p.Trp24Ter	Pathogenic	2/416	0	0
13:20763677	rs111033217 NM_004004.6:c.44A>C p.Lys15Thr	Pathogenic/Likely pathogenic	0	0	-
13:20763686	rs1801002 NM_004004.6:c.35G>A p.Gly12Asp	Uncertain significance	0	0	-
	NM_004004.6:c.35G>T p.Gly12Val	Pathogenic/Likely pathogenic	0	0	-
13:20763686	rs80338939 NM_004004.6:c.35del p.Gly12fs	Pathogenic	NA	18/472	0.0089
	NM_004004.6:c.35dup p.Val13fs	Pathogenic	NA	0	-
13:20763687	rs104894408 NM_004004.6:c.34G>T p.Gly12Cys	Likely pathogenic	NA	0	0
	NM_004004.6:c.34G>C p.Gly12Arg	Pathogenic	NA	0	-
13:20763702	rs111033451 NM_004004.6:c.19C>T p.Gln7Ter	Pathogenic/Likely pathogenic	0	0	-
13:20763710	rs111033222 NM_004004.6:c.11G>A p.Gly4Asp	Likely benign	NA	0	0
13:20763712	rs111033401 NM_004004.6:c.9G>T p.Trp3Cys	Uncertain significance	0	0	
	NM_004004.6:c.9G>A p.Trp3Ter	Pathogenic/Likely pathogenic	0	0	-
13:20763720	rs111033293 NM_004004.6:c.1A>T p.Met1Leu	Likely pathogenic	0	0	-
	NM_004004.6:c.1A>G p.Met1Val	Pathogenic	0	0	-
13:20764940	rs9552099				

	NM_004004.6):c.-22-1198G>T	not reported	1/416	27+3/472	-
	NM_004004.6):c.-22-1198G>A		0	0	0.828
13:20766921	rs80338940				
	NM_004004.6:c.-23+1G>A	Pathogenic	0	1/472	-