

Beta, per allele, SD units of eBMD (95% CI)	Beta, per allele, g/cm ² units of eBMD (95% CI)	P-value	Genotype counts, RR RA AA genotypes	AAF
0.13 (0.08, 0.17)	0.015 (0.009, 0.021)	2.1x10 ⁻⁷	418366 1371 0	0.0016

Figure 1

Genetic exposure	Beta, per allele, SD units of eBMD (95% CI)	Beta, per allele, g/cm ² units of eBMD (95% CI)	P-value	Genotype counts, RR RA AA genotypes	AAF
pLoF variants, AAF < 1%	0.18 (0.08, 0.29)	0.022 (0.009, 0.035)	0.00065	419465 272 0	3.0x10 ⁻⁴

Figure 2

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137557:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.847C>T; c.841C>T; c.847C>T	p.Arg283Cys; p.Arg281Cys; p.Arg283Cys	FALSE	8.46E-06
22:29137557:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.847C>T; c.841C>T; c.847C>T	p.Arg283Cys; p.Arg281Cys; p.Arg283Cys	FALSE	6.87E-05
22:29137557:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.847C>T; c.841C>T; c.847C>T	p.Arg283Cys; p.Arg281Cys; p.Arg283Cys	FALSE	4.64E-06
22:29137557:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.847C>T; c.841C>T; c.847C>T	p.Arg283Cys; p.Arg281Cys; p.Arg283Cys	FALSE	1.72E-05
22:29073216:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.90delC; c.90delC; c.90delC	p.Gly31fs; p.Glu31fs; p.Gly31fs	TRUE	6.48E-06
22:29094283:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.123T>G; c.117T>G; c.123T>G	p.Asp41Glu; p.Asp39Glu; p.Asp41Glu	FALSE	1.16E-06
22:29140320:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1162A>G; c.1207A>G; c.1213A>G	p.Thr388Ala; p.Thr403Ala; p.Thr405Ala	FALSE	8.46E-06
22:29125344:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>T; c.553G>T; c.559G>T	p.Glu187*; p.Glu185*; p.Glu187*	TRUE	6.48E-06
22:29125365:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.580G>A; c.574G>A; c.580G>A	p.Gly194Arg; p.Gly192Arg; p.Gly194Arg	FALSE	3.89E-05

Figure 3

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29125365:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.580G>A; c.574G>A; c.580G>A	p.Gly194Arg; p.Gly192Arg; p.Gly194Arg	FALSE	6.03E-05
22:29125365:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.580G>A; c.574G>A; c.580G>A	p.Gly194Arg; p.Gly192Arg; p.Gly194Arg	FALSE	5.92E-05
22:29125365:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.580G>A; c.574G>A; c.580G>A	p.Gly194Arg; p.Gly192Arg; p.Gly194Arg	FALSE	0.000137438
22:29137644:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.934C>A; c.928C>A; c.934C>A	p.Gln312Lys; p.Gln310Lys; p.Gln312Lys	FALSE	1.16E-06
22:29094396:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.236G>T; c.230G>T; c.236G>T	p.Gly79Val; p.Gly77Val; p.Gly79Val	FALSE	3.48E-06
22:29098872:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.271G>A; c.265G>A; c.271G>A	p.Gly91Arg; p.Gly89Arg; p.Gly91Arg	FALSE	1.72E-05
22:29137405:TC:T	ENST00000400335; ENST00000407188; ENST00000327813	c.699delC; c.693delC; c.699delC	p.Asp234fs; p.Asp232fs; p.Asp234fs	TRUE	2.32E-06
22:29073155:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.28delC; c.28delC; c.28delC	p.Leu10fs; p.Leu10fs; p.Leu10fs	TRUE	6.48E-06
22:29137575:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.895C>T; c.859C>T; c.865C>T	p.Arg289Cys; p.Arg287Cys; p.Arg289Cys	FALSE	4.64E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137575:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.856C>T; c.859C>T; c.865C>T	p.Arg289Cys; p.Arg287Cys; p.Arg289Cys	FALSE	1.16E-06
22:29137575:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.865C>T; c.859C>T; c.865C>T	p.Arg289Cys; p.Arg287Cys; p.Arg289Cys	FALSE	6.48E-06
22:29142018:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1283T>G; c.1328T>G; c.1334T>G	p.Ile428Ser; p.Ile443Ser; p.Ile445Ser	FALSE	1.94E-05
22:29142018:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1283T>G; c.1328T>G; c.1334T>G	p.Ile428Ser; p.Ile443Ser; p.Ile445Ser	FALSE	1.27E-05
22:29094298:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.138G>C; c.132G>C; c.138G>C	p.Gln46His; p.Gln44His; p.Gln46His	FALSE	6.48E-06
22:29125319:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.534G>A; c.528G>A; c.534G>A	p.Trp178*; p.Trp176*; p.Trp178*	TRUE	1.16E-06
22:29140320:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1162A>C; c.1207A>C; c.1213A>C	p.Thr388Pro; p.Thr403Pro; p.Thr405Pro	FALSE	1.16E-06
22:29121374:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.370T>C; c.364T>C; c.370T>C	p.Cys124Arg; p.Cys122Arg; p.Cys124Arg	FALSE	1.16E-06
22:29138623:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.965-1G>A; c.959-1G>A; c.965-1G>A		TRUE	3.48E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29138623:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.965-1G>A; c.959-1G>A; c.965-1G>A		TRUE	6.48E-06
22:29121482:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.477+1G>C; c.471+1G>C; c.477+1G>C		TRUE	1.16E-06
22:29137353:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.643G>A; c.637G>A; c.643G>A	p.Ala215Thr; p.Ala213Thr; p.Ala215Thr	FALSE	1.30E-05
22:29137353:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.643G>A; c.637G>A; c.643G>A	p.Ala215Thr; p.Ala213Thr; p.Ala215Thr	FALSE	8.11E-06
22:29140353:CA:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1196delA; c.1241delA; c.1247delA	p.His399fs; p.His414fs; p.His416fs	TRUE	1.16E-06
22:29121438:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.434C>T; c.428C>T; c.434C>T	p.Thr145Ile; p.Thr143Ile; p.Thr145Ile	FALSE	6.48E-06
22:29138834:G:A	ENST00000407188; ENST00000327813	c.1168+1G>A; c.1174+1G>A		TRUE	1.16E-06
22:29125366:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.581G>T; c.575G>T; c.581G>T	p.Gly194Val; p.Gly192Val; p.Gly194Val	FALSE	8.46E-06
22:29137600:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.890C>T; c.884C>T; c.890C>T	p.Ser297Phe; p.Ser295Phe; p.Ser297Phe	FALSE	6.95E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29138738:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1079A>G; c.1073A>G; c.1079A>G	p.Tyr360Cys; p.Tyr358Cys; p.Tyr360Cys	FALSE	2.32E-06
22:29138738:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1079A>G; c.1073A>G; c.1079A>G	p.Tyr360Cys; p.Tyr358Cys; p.Tyr360Cys	FALSE	6.48E-06
22:29098925:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.324G>A; c.318G>A; c.324G>A	p.Trp108*; p.Trp106*; p.Trp108*	TRUE	1.16E-06
22:29137447:T:TCC	ENST00000400335; ENST00000407188; ENST00000327813	c.738_739dupCC; c.732_733dupCC; c.738_739dupCC	p.Arg247fs; p.Arg245fs; p.Arg247fs	TRUE	2.32E-06
22:29142017:A:AT	ENST00000400335; ENST00000407188; ENST00000327813	c.1288dupT; c.1333dupT; c.1339dupT	p.Tyr430fs; p.Tyr445fs; p.Tyr447fs	TRUE	3.48E-06
22:29094255:TA:T	ENST00000400335; ENST00000407188; ENST00000327813	c.98-2delA; c.92-2delA; c.98-2delA		TRUE	6.48E-06
22:29094328:AT:A	ENST00000400335; ENST00000407188; ENST00000327813	c.169delT; c.163delT; c.169delT	p.Cys57fs; p.Cys55fs; p.Cys57fs	TRUE	1.16E-06
22:29121477:TC:T	ENST00000400335; ENST00000407188; ENST00000327813	c.474delC; c.468delC; c.474delC	p.Phe158fs; p.Phe156fs; p.Phe158fs	TRUE	1.16E-06
22:29140297:GTCTGGCA:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1140_1146delTCTGGCA; c.1185_1191delTCTGGCA; c.1191_1197delTCTGGCA	p.Ala382fs; p.Ala397fs; p.Ala399fs	TRUE	4.64E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29094403:TG:T	ENST00000400335; ENST00000407188; ENST00000327813	c.244delG; c.238delG; c.244delG	p.Glu82fs; p.Glu80fs; p.Glu82fs	TRUE	6.48E-06
22:29138783:G:A	ENST00000400335	c.1123+1G>A		TRUE	1.16E-06
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	6.14E-05
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	1.94E-05
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	0.000113071
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	4.64E-05
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	6.87E-05
22:29138753:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1098delC; c.1092delC; c.1098delC	p.Ser367fs; p.Ser365fs; p.Ser367fs	TRUE	4.23E-05
22:29125269:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.484G>A; c.478G>A; c.484G>A	p.Gly162Arg; p.Gly160Arg; p.Gly162Arg	FALSE	1.16E-06
22:29094387:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.227G>C; c.221G>C; c.227G>C	p.Gly76Ala; p.Gly74Ala; p.Gly76Ala	FALSE	0.000113071

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137495:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.785A>C; c.779A>C; c.785A>C	p.Asp262Ala; p.Asp260Ala; p.Asp262Ala	FALSE	2.32E-06
22:29140285:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1127G>A; c.1172G>A; c.1178G>A	p.Trp376*; p.Trp391*; p.Trp393*	TRUE	1.16E-06
22:29142101:G:A	ENST00000327813	c.1416+1G>A		TRUE	1.16E-06
22:29094273:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.113A>G; c.107A>G; c.113A>G	p.Asn38Ser; p.Asn36Ser; p.Asn38Ser	FALSE	2.90E-05
22:29094273:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.113A>G; c.107A>G; c.113A>G	p.Asn38Ser; p.Asn36Ser; p.Asn38Ser	FALSE	1.30E-05
22:29094273:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.113A>G; c.107A>G; c.113A>G	p.Asn38Ser; p.Asn36Ser; p.Asn38Ser	FALSE	8.46E-06
22:29125282:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.497G>A; c.491G>A; c.497G>A	p.Gly166Asp; p.Gly164Asp; p.Gly166Asp	FALSE	4.53E-05
22:29125282:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.497G>A; c.491G>A; c.497G>A	p.Gly166Asp; p.Gly164Asp; p.Gly166Asp	FALSE	3.44E-05
22:29125282:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.497G>A; c.491G>A; c.497G>A	p.Gly166Asp; p.Gly164Asp; p.Gly166Asp	FALSE	1.69E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29094267:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.107C>T; c.101C>T; c.107C>T	p.Thr36Ile; p.Thr34Ile; p.Thr36Ile	FALSE	5.80E-06
22:29094267:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.107C>T; c.101C>T; c.107C>T	p.Thr36Ile; p.Thr34Ile; p.Thr36Ile	FALSE	1.30E-05
22:29094267:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.107C>T; c.101C>T; c.107C>T	p.Thr36Ile; p.Thr34Ile; p.Thr36Ile	FALSE	1.69E-05
22:29137674:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.964G>C; c.958G>C; c.964G>C	p.Ala322Pro; p.Ala320Pro; p.Ala322Pro	FALSE	1.16E-06
22:29125329:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.544G>T; c.538G>T; c.544G>T	p.Glu182*; p.Glu180*; p.Glu182*	TRUE	6.48E-06
22:29137417:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.707A>G; c.701A>G; c.707A>G	p.Tyr236Cys; p.Tyr234Cys; p.Tyr236Cys	FALSE	6.48E-06
22:29137417:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.707A>G; c.701A>G; c.707A>G	p.Tyr236Cys; p.Tyr234Cys; p.Tyr236Cys	FALSE	4.64E-05
22:29137417:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.707A>G; c.701A>G; c.707A>G	p.Tyr236Cys; p.Tyr234Cys; p.Tyr236Cys	FALSE	6.87E-05
22:29137417:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.707A>G; c.701A>G; c.707A>G	p.Tyr236Cys; p.Tyr234Cys; p.Tyr236Cys	FALSE	2.32E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29125392:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.607G>A; c.601G>A; c.607G>A	p.Asp203Asn; p.Asp201Asn; p.Asp203Asn	FALSE	9.27E-05
22:29125392:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.607G>A; c.601G>A; c.607G>A	p.Asp203Asn; p.Asp201Asn; p.Asp203Asn	FALSE	1.51E-05
22:29125392:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.607G>A; c.601G>A; c.607G>A	p.Asp203Asn; p.Asp201Asn; p.Asp203Asn	FALSE	1.72E-05
22:29137657:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.947T>C; c.941T>C; c.947T>C	p.Phe316Ser; p.Phe314Ser; p.Phe316Ser	FALSE	6.48E-06
22:29137657:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.947T>C; c.941T>C; c.947T>C	p.Phe316Ser; p.Phe314Ser; p.Phe316Ser	FALSE	1.74E-05
22:29140312:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1154T>C; c.1199T>C; c.1205T>C	p.Leu385Pro; p.Leu400Pro; p.Leu402Pro	FALSE	8.46E-06
22:29140312:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1154T>C; c.1199T>C; c.1205T>C	p.Leu385Pro; p.Leu400Pro; p.Leu402Pro	FALSE	6.87E-05
22:29098866:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.265C>G; c.259C>G; c.265C>G	p.Pro89Ala; p.Pro87Ala; p.Pro89Ala	FALSE	1.16E-06
22:29125325:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.540C>G; c.534C>G; c.540C>G	p.Tyr180*; p.Tyr178*; p.Tyr180*	TRUE	1.16E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29125325:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.540C>G; c.534C>G; c.540C>G	p.Tyr180*; p.Tyr178*; p.Tyr180*	TRUE	1.16E-06
22:29138723:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1064C>T; c.1058C>T; c.1064C>T	p.Ser355Phe; p.Ser353Phe; p.Ser355Phe	FALSE	5.80E-06
22:29140353:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1195C>T; c.1240C>T; c.1246C>T	p.His399Tyr; p.His414Tyr; p.His416Tyr	FALSE	8.46E-06
22:29137341:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.632-1G>A; c.626- 1G>A; c.632-1G>A		TRUE	6.48E-06
22:29073204:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.78delC; c.78delC; c.78delC	p.Gly27fs; p.Gly27fs; p.Gly27fs	TRUE	6.48E-06
22:29125288:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.503C>T; c.497C>T; c.503C>T	p.Ala168Val; p.Ala166Val; p.Ala168Val	FALSE	1.16E-06
22:29125297:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.512G>T; c.506G>T; c.512G>T	p.Cys171Phe; p.Cys169Phe; p.Cys171Phe	FALSE	1.16E-05
22:29137492:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.782T>G; c.776T>G; c.782T>G	p.Phe261Cys; p.Phe259Cys; p.Phe261Cys	FALSE	1.16E-06
22:29167105:G:C	ENST00000327813	c.1478G>C	p.Ter493Serext*?	TRUE	2.59E-05
22:29167105:G:C	ENST00000327813	c.1478G>C	p.Ter493Serext*?	TRUE	8.46E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29073228:G:T	ENST00000400335; ENST00000327813	c.97+1G>T; c.97+1G>T		TRUE	2.32E-06
22:29094320:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.160G>A; c.154G>A; c.160G>A	p.Gly54Arg; p.Gly52Arg; p.Gly54Arg	FALSE	1.30E-05
22:29094320:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.160G>A; c.154G>A; c.160G>A	p.Gly54Arg; p.Gly52Arg; p.Gly54Arg	FALSE	4.64E-06
22:29094320:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.160G>A; c.154G>A; c.160G>A	p.Gly54Arg; p.Gly52Arg; p.Gly54Arg	FALSE	2.54E-05
22:29094320:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.160G>A; c.154G>A; c.160G>A	p.Gly54Arg; p.Gly52Arg; p.Gly54Arg	FALSE	6.87E-05
22:29137579:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.869C>T; c.863C>T; c.869C>T	p.Pro290Leu; p.Pro288Leu; p.Pro290Leu	FALSE	8.46E-06
22:29094321:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.161G>A; c.155G>A; c.161G>A	p.Gly54Glu; p.Gly52Glu; p.Gly54Glu	FALSE	1.16E-06
22:29137340:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.632-2A>G; c.626- 2A>G; c.632-2A>G		TRUE	1.16E-06
22:29137521:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.811G>A; c.805G>A; c.811G>A	p.Glu271Lys; p.Glu269Lys; p.Glu271Lys	FALSE	2.32E-06
22:29073213:T:TCGGCCC	ENST00000407188	c.87_91+1dupCCCCGG		TRUE	1.16E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137654:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.944G>A; c.938G>A; c.944G>A	p.Gly315Glu; p.Gly313Glu; p.Gly315Glu	FALSE	2.32E-06
22:29138746:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1087A>G; c.1081A>G; c.1087A>G	p.Thr363Ala; p.Thr361Ala; p.Thr363Ala	FALSE	8.46E-06
22:29137340:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.632-2A>C; c.626- 2A>C; c.632-2A>C		TRUE	0.00010305
22:29094324:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.164A>T; c.158A>T; c.164A>T	p.Lys55Met; p.Lys53Met; p.Lys55Met	FALSE	1.16E-06
22:29137647:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.937G>A; c.931G>A; c.937G>A	p.Ala313Thr; p.Ala311Thr; p.Ala313Thr	FALSE	6.48E-06
22:29137647:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.937G>A; c.931G>A; c.937G>A	p.Ala313Thr; p.Ala311Thr; p.Ala313Thr	FALSE	6.87E-05
22:29137360:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.650G>T; c.644G>T; c.650G>T	p.Gly217Val; p.Gly215Val; p.Gly217Val	FALSE	6.87E-05
22:29094390:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.230A>G; c.224A>G; c.230A>G	p.Glu77Gly; p.Glu75Gly; p.Glu77Gly	FALSE	1.16E-06
22:29167083:CAG:C	ENST00000327813	c.1457_1458delAG	p.Gln486fs	TRUE	3.44E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29138812:GC:G	ENST00000407188; ENST00000327813	c.1148delC; c.1154delC	p.Ala383fs; p.Ala385fs	TRUE	3.89E-05
22:29138812:GC:G	ENST00000407188; ENST00000327813	c.1148delC; c.1154delC	p.Ala383fs; p.Ala385fs	TRUE	6.95E-05
22:29138812:GC:G	ENST00000407188; ENST00000327813	c.1148delC; c.1154delC	p.Ala383fs; p.Ala385fs	TRUE	0.000113071
22:29138812:GC:G	ENST00000407188; ENST00000327813	c.1148delC; c.1154delC	p.Ala383fs; p.Ala385fs	TRUE	4.64E-05
22:29094363:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.203A>G; c.197A>G; c.203A>G	p.Tyr68Cys; p.Tyr66Cys; p.Tyr68Cys	FALSE	6.77E-05
22:29094363:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.203A>G; c.197A>G; c.203A>G	p.Tyr68Cys; p.Tyr66Cys; p.Tyr68Cys	FALSE	1.16E-06
22:29094363:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.203A>G; c.197A>G; c.203A>G	p.Tyr68Cys; p.Tyr66Cys; p.Tyr68Cys	FALSE	5.83E-05
22:29140318:T:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1160T>A; c.1205T>A; c.1211T>A	p.Leu387His; p.Leu402His; p.Leu404His	FALSE	1.16E-06
22:29098926:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.325A>G; c.319A>G; c.325A>G	p.Lys109Glu; p.Lys107Glu; p.Lys109Glu	FALSE	8.46E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29098926:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.325A>G; c.319A>G; c.325A>G	p.Lys109Glu; p.Lys107Glu; p.Lys109Glu	FALSE	1.04E-05
22:29125372:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.587A>T; c.581A>T; c.587A>T	p.His196Leu; p.His194Leu; p.His196Leu	FALSE	6.48E-06
22:29125372:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.587A>T; c.581A>T; c.587A>T	p.His196Leu; p.His194Leu; p.His196Leu	FALSE	3.39E-05
22:29125372:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.587A>T; c.581A>T; c.587A>T	p.His196Leu; p.His194Leu; p.His196Leu	FALSE	2.09E-05
22:29125372:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.587A>T; c.581A>T; c.587A>T	p.His196Leu; p.His194Leu; p.His196Leu	FALSE	1.72E-05
22:29140318:TCA:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1163_1164delCA; c.1208_1209delCA; c.1214_1215delCA	p.Thr388fs; p.Thr403fs; p.Thr405fs	TRUE	2.32E-06
22:29094354:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.194A>G; c.188A>G; c.194A>G	p.Gln65Arg; p.Gln63Arg; p.Gln65Arg	FALSE	2.32E-06
22:29141946:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1211C>T; c.1256C>T; c.1262C>T	p.Ser404Phe; p.Ser419Phe; p.Ser421Phe	FALSE	6.48E-06
22:29094256:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.98-2A>G; c.92-2A>G; c.98-2A>G		TRUE	8.46E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29094256:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.98-2A>G; c.92-2A>G; c.98-2A>G		TRUE	3.48E-06
22:29094391:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.231G>C; c.225G>C; c.231G>C	p.Glu77Asp; p.Glu75Asp; p.Glu77Asp	FALSE	1.16E-06
22:29125384:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.599G>C; c.593G>C; c.599G>C	p.Cys200Ser; p.Cys198Ser; p.Cys200Ser	FALSE	6.87E-05
22:29125384:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.599G>C; c.593G>C; c.599G>C	p.Cys200Ser; p.Cys198Ser; p.Cys200Ser	FALSE	8.59E-05
22:29125384:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.599G>C; c.593G>C; c.599G>C	p.Cys200Ser; p.Cys198Ser; p.Cys200Ser	FALSE	1.16E-06
22:29138687:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1028C>T; c.1022C>T; c.1028C>T	p.Thr343Met; p.Thr341Met; p.Thr343Met	FALSE	2.32E-06
22:29138687:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1028C>T; c.1022C>T; c.1028C>T	p.Thr343Met; p.Thr341Met; p.Thr343Met	FALSE	6.48E-06
22:29138687:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1028C>T; c.1022C>T; c.1028C>T	p.Thr343Met; p.Thr341Met; p.Thr343Met	FALSE	8.46E-06
22:29138737:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1078T>G; c.1072T>G; c.1078T>G	p.Tyr360Asp; p.Tyr358Asp; p.Tyr360Asp	FALSE	6.48E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121450:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.446G>A; c.440G>A; c.446G>A	p.Cys149Tyr; p.Cys147Tyr; p.Cys149Tyr	FALSE	2.32E-06
22:29167083:CA:C	ENST00000327813	c.1457delA	p.Gln486fs	TRUE	2.32E-06
22:29073212:CT:C	ENST00000400335; ENST00000407188; ENST00000327813	c.83delT; c.83delT; c.83delT	p.Leu28fs; p.Leu28fs; p.Leu28fs	TRUE	1.16E-06
22:29121416:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.412A>G; c.406A>G; c.412A>G	p.Ser138Gly; p.Ser136Gly; p.Ser138Gly	FALSE	3.24E-05
22:29121416:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.412A>G; c.406A>G; c.412A>G	p.Ser138Gly; p.Ser136Gly; p.Ser138Gly	FALSE	3.48E-06
22:29121416:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.412A>G; c.406A>G; c.412A>G	p.Ser138Gly; p.Ser136Gly; p.Ser138Gly	FALSE	1.69E-05
22:29121416:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.412A>G; c.406A>G; c.412A>G	p.Ser138Gly; p.Ser136Gly; p.Ser138Gly	FALSE	6.87E-05
22:29098873:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.272G>T; c.266G>T; c.272G>T	p.Gly91Val; p.Gly89Val; p.Gly91Val	FALSE	6.48E-06
22:29094257:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.98-1G>A; c.92-1G>A; c.98-1G>A		TRUE	1.16E-06
22:29098911:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.310G>C; c.304G>C; c.310G>C	p.Asp104His; p.Asp102His; p.Asp104His	FALSE	1.16E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29142057:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1322A>T; c.1367A>T; c.1373A>T	p.Lys441Met; p.Lys456Met; p.Lys458Met	FALSE	1.16E-06
22:29094270:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.110C>A; c.104C>A; c.110C>A	p.Ala37Asp; p.Ala35Asp; p.Ala37Asp	FALSE	2.32E-06
22:29094344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.184G>A; c.178G>A; c.184G>A	p.Glu62Lys; p.Glu60Lys; p.Glu62Lys	FALSE	1.16E-06
22:29121372:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.368G>A; c.362G>A; c.368G>A	p.Gly123Asp; p.Gly121Asp; p.Gly123Asp	FALSE	4.64E-06
22:29142110:T:G	ENST00000400335; ENST00000407188	c.1375T>G; c.1420T>G	p.Ter459Gluext*?; p.Ter474Gluext*?	TRUE	8.46E-06
22:29137559:CT:C	ENST00000400335; ENST00000407188; ENST00000327813	c.851delT; c.845delT; c.851delT	p.Phe284fs; p.Phe282fs; p.Phe284fs	TRUE	2.32E-06
22:29098953:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.352A>C; c.346A>C; c.352A>C	p.Met118Leu; p.Met116Leu; p.Met118Leu	FALSE	2.32E-06
22:29137407:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.697C>T; c.691C>T; c.697C>T	p.Pro233Ser; p.Pro231Ser; p.Pro233Ser	FALSE	1.16E-06
22:29094395:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.235G>A; c.229G>A; c.235G>A	p.Gly79Ser; p.Gly77Ser; p.Gly79Ser	FALSE	6.48E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121359:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.355C>T; c.349C>T; c.355C>T	p.Pro119Ser; p.Pro117Ser; p.Pro119Ser	FALSE	2.32E-06
22:29098893:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.292T>C; c.286T>C; c.292T>C	p.Tyr98His; p.Tyr96His; p.Tyr98His	FALSE	2.32E-06
22:29142029:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1294C>A; c.1339C>A; c.1345C>A	p.Pro432Thr; p.Pro447Thr; p.Pro449Thr	FALSE	1.16E-06
22:29121379:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.375C>G; c.369C>G; c.375C>G	p.Tyr125*; p.Tyr123*; p.Tyr125*	TRUE	1.16E-06
22:29121359:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.355C>A; c.349C>A; c.355C>A	p.Pro119Thr; p.Pro117Thr; p.Pro119Thr	FALSE	7.12E-05
22:29121359:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.355C>A; c.349C>A; c.355C>A	p.Pro119Thr; p.Pro117Thr; p.Pro119Thr	FALSE	2.43E-05
22:29121359:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.355C>A; c.349C>A; c.355C>A	p.Pro119Thr; p.Pro117Thr; p.Pro119Thr	FALSE	0.000135428
22:29094342:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.182A>G; c.176A>G; c.182A>G	p.Asn61Ser; p.Asn59Ser; p.Asn61Ser	FALSE	4.64E-06
22:29138806:ATGGGGGCT:A	ENST00000407188; ENST00000327813	c.1142_1149delITGGGGGCT; c.1148_1155delITGGGGGCT	p.Met381fs; p.Met383fs	TRUE	5.80E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137558:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.848G>A; c.842G>A; c.848G>A	p.Arg283His; p.Arg281His; p.Arg283His	FALSE	1.16E-06
22:29137558:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.848G>A; c.842G>A; c.848G>A	p.Arg283His; p.Arg281His; p.Arg283His	FALSE	1.94E-05
22:29137558:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.848G>A; c.842G>A; c.848G>A	p.Arg283His; p.Arg281His; p.Arg283His	FALSE	0.000113071
22:29098878:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.277G>A; c.271G>A; c.277G>A	p.Val93Met; p.Val91Met; p.Val93Met	FALSE	8.46E-06
22:29098878:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.277G>A; c.271G>A; c.277G>A	p.Val93Met; p.Val91Met; p.Val93Met	FALSE	5.80E-06
22:29098878:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.277G>A; c.271G>A; c.277G>A	p.Val93Met; p.Val91Met; p.Val93Met	FALSE	1.30E-05
22:29094276:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.116G>A; c.110G>A; c.116G>A	p.Gly39Asp; p.Gly37Asp; p.Gly39Asp	FALSE	9.27E-06
22:29094276:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.116G>A; c.110G>A; c.116G>A	p.Gly39Asp; p.Gly37Asp; p.Gly39Asp	FALSE	6.48E-06
22:29125415:TAGTG:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631_631+3del/AGTG; c.625_625+3del/AGTG; c.631_631+3del/AGTG	p.Thr211fs; p.Thr209fs; p.Thr211fs	TRUE	6.48E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137654:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.944G>T; c.938G>T; c.944G>T	p.Gly315Val; p.Gly313Val; p.Gly315Val	FALSE	1.16E-06
22:29137650:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.940C>T; c.934C>T; c.940C>T	p.Gln314*; p.Gln312*; p.Gln314*	TRUE	1.16E-06
22:29137650:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.940C>T; c.934C>T; c.940C>T	p.Gln314*; p.Gln312*; p.Gln314*	TRUE	9.27E-05
22:29137539:ACC:A	ENST00000400335; ENST00000407188; ENST00000327813	c.831_832delCC; c.825_826delCC; c.831_832delCC	p.His278fs; p.His276fs; p.His278fs	TRUE	1.69E-05
22:29137539:ACC:A	ENST00000400335; ENST00000407188; ENST00000327813	c.831_832delCC; c.825_826delCC; c.831_832delCC	p.His278fs; p.His276fs; p.His278fs	TRUE	6.48E-06
22:29137539:ACC:A	ENST00000400335; ENST00000407188; ENST00000327813	c.831_832delCC; c.825_826delCC; c.831_832delCC	p.His278fs; p.His276fs; p.His278fs	TRUE	6.95E-06
22:29140311:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1153C>T; c.1198C>T; c.1204C>T	p.Leu385Phe; p.Leu400Phe; p.Leu402Phe	FALSE	1.04E-05
22:29140311:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1153C>T; c.1198C>T; c.1204C>T	p.Leu385Phe; p.Leu400Phe; p.Leu402Phe	FALSE	2.59E-05
22:29140311:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1153C>T; c.1198C>T; c.1204C>T	p.Leu385Phe; p.Leu400Phe; p.Leu402Phe	FALSE	4.23E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29140311:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1153C>T; c.1198C>T; c.1204C>T	p.Leu385Phe; p.Leu400Phe; p.Leu402Phe	FALSE	6.87E-05
22:29121371:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.367G>C; c.361G>C; c.367G>C	p.Gly123Arg; p.Gly121Arg; p.Gly123Arg	FALSE	1.16E-06
22:29125368:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.583G>T; c.577G>T; c.583G>T	p.Asp195Tyr; p.Asp193Tyr; p.Asp195Tyr	FALSE	7.53E-05
22:29125368:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.583G>T; c.577G>T; c.583G>T	p.Asp195Tyr; p.Asp193Tyr; p.Asp195Tyr	FALSE	1.69E-05
22:29138740:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1081G>C; c.1075G>C; c.1081G>C	p.Val361Leu; p.Val359Leu; p.Val361Leu	FALSE	1.62E-05
22:29073228:G:A	ENST00000400335; ENST00000327813	c.97+1G>A; c.97+1G>A		TRUE	1.16E-06
22:29121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	0.000110035
22:29121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	4.64E-05
22:29121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	6.87E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:2912121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	0.00010363
22:2912121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	0.00020979
22:2912121465:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.461G>A; c.455G>A; c.461G>A	p.Arg154Gln; p.Arg152Gln; p.Arg154Gln	FALSE	0.000565355
22:29098885:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.284C>T; c.278C>T; c.284C>T	p.Pro95Leu; p.Pro93Leu; p.Pro95Leu	FALSE	8.46E-06
22:29098885:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.284C>T; c.278C>T; c.284C>T	p.Pro95Leu; p.Pro93Leu; p.Pro95Leu	FALSE	6.48E-06
22:29098885:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.284C>T; c.278C>T; c.284C>T	p.Pro95Leu; p.Pro93Leu; p.Pro95Leu	FALSE	1.16E-05
22:2912121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	7.62E-05
22:2912121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	4.64E-05
22:2912121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	6.87E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	4.75E-05
22:29121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	9.07E-05
22:29121464:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.460C>T; c.454C>T; c.460C>T	p.Arg154Trp; p.Arg152Trp; p.Arg154Trp	FALSE	0.000113071
22:29142029:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1294C>G; c.1339C>G; c.1345C>G	p.Pro432Ala; p.Pro447Ala; p.Pro449Ala	FALSE	1.16E-06
22:29098908:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.307G>A; c.301G>A; c.307G>A	p.Glu103Lys; p.Glu101Lys; p.Glu103Lys	FALSE	4.64E-05
22:29098908:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.307G>A; c.301G>A; c.307G>A	p.Glu103Lys; p.Glu101Lys; p.Glu103Lys	FALSE	6.87E-05
22:29098908:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.307G>A; c.301G>A; c.307G>A	p.Glu103Lys; p.Glu101Lys; p.Glu103Lys	FALSE	8.46E-06
22:29098908:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.307G>A; c.301G>A; c.307G>A	p.Glu103Lys; p.Glu101Lys; p.Glu103Lys	FALSE	3.89E-05
22:29098908:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.307G>A; c.301G>A; c.307G>A	p.Glu103Lys; p.Glu101Lys; p.Glu103Lys	FALSE	2.55E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29142005:GA:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1273delA; c.1318delA; c.1324delA	p.Ile425fs; p.Ile440fs; p.Ile442fs	TRUE	1.16E-06
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	0.000110106
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	8.35E-05
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	0.000226142
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	0.000152356
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	0.000185443
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	6.87E-05
22:29138713:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1054G>A; c.1048G>A; c.1054G>A	p.Ala352Thr; p.Ala350Thr; p.Ala352Thr	FALSE	0.000395026
22:29138818:A:AT	ENST00000407188; ENST00000327813	c.1153_1154insT; c.1159_1160insT	p.Ser385fs; p.Ser387fs	TRUE	5.80E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29138756:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1097C>G; c.1091C>G; c.1097C>G	p.Pro366Arg; p.Pro364Arg; p.Pro366Arg	FALSE	6.48E-06
22:29167104:T:A	ENST00000327813	c.1477T>A	p.Ter493Argext*?	TRUE	1.16E-06
22:29094282:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.122A>T; c.116A>T; c.122A>T	p.Asp41Val; p.Asp39Val; p.Asp41Val	FALSE	1.16E-06
22:29121443:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.439C>T; c.433C>T; c.439C>T	p.Gln147*; p.Gln145*; p.Gln147*	TRUE	1.16E-06
22:29138759:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1100G>T; c.1094G>T; c.1100G>T	p.Ser367Ile; p.Ser365Ile; p.Ser367Ile	FALSE	8.46E-06
22:29138765:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1106C>T; c.1100C>T; c.1106C>T	p.Pro369Leu; p.Pro367Leu; p.Pro369Leu	FALSE	6.48E-06
22:29138765:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1106C>T; c.1100C>T; c.1106C>T	p.Pro369Leu; p.Pro367Leu; p.Pro369Leu	FALSE	1.16E-06
22:29140308:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1150C>G; c.1195C>G; c.1201C>G	p.Leu384Val; p.Leu399Val; p.Leu401Val	FALSE	8.46E-06
22:29140308:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1150C>G; c.1195C>G; c.1201C>G	p.Leu384Val; p.Leu399Val; p.Leu401Val	FALSE	1.16E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29094257:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.98-1G>T; c.92-1G>T; c.98-1G>T		TRUE	3.48E-06
22:29137524:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.814C>T; c.808C>T; c.814C>T	p.Leu272Phe; p.Leu270Phe; p.Leu272Phe	FALSE	4.29E-05
22:29094387:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.227G>T; c.221G>T; c.227G>T	p.Gly76Val; p.Gly74Val; p.Gly76Val	FALSE	6.48E-06
22:29125342:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.557C>T; c.551C>T; c.557C>T	p.Thr186Ile; p.Thr184Ile; p.Thr186Ile	FALSE	1.16E-06
22:29138717:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1058C>T; c.1052C>T; c.1058C>T	p.Ala353Val; p.Ala351Val; p.Ala353Val	FALSE	1.16E-06
22:29098926:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.325A>T; c.319A>T; c.325A>T	p.Lys109*; p.Lys107*; p.Lys109*	TRUE	1.16E-06
22:29121405:T:A	ENST00000400335; ENST00000407188; ENST00000327813	c.401T>A; c.395T>A; c.401T>A	p.Leu134Gln; p.Leu132Gln; p.Leu134Gln	FALSE	1.16E-06
22:29098934:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.333T>G; c.327T>G; c.333T>G	p.Cys111Trp; p.Cys109Trp; p.Cys111Trp	FALSE	9.16E-05
22:29098934:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.333T>G; c.327T>G; c.333T>G	p.Cys111Trp; p.Cys109Trp; p.Cys111Trp	FALSE	9.72E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29098934:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.333T>G; c.327T>G; c.333T>G	p.Cys111Trp; p.Cys109Trp; p.Cys111Trp	FALSE	0.000206157
22:29098934:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.333T>G; c.327T>G; c.333T>G	p.Cys111Trp; p.Cys109Trp; p.Cys111Trp	FALSE	8.59E-05
22:29125267:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.482C>T; c.476C>T; c.482C>T	p.Ala161Val; p.Ala159Val; p.Ala161Val	FALSE	3.89E-05
22:29137351:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.641G>A; c.635G>A; c.641G>A	p.Gly214Asp; p.Gly212Asp; p.Gly214Asp	FALSE	1.16E-06
22:29137353:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.643G>C; c.637G>C; c.643G>C	p.Ala215Pro; p.Ala213Pro; p.Ala215Pro	FALSE	1.16E-06
22:29098924:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.323G>C; c.317G>C; c.323G>C	p.Trp108Ser; p.Trp106Ser; p.Trp108Ser	FALSE	4.64E-06
22:29140329:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1171G>A; c.1216G>A; c.1222G>A	p.Ala391Thr; p.Ala406Thr; p.Ala408Thr	FALSE	1.16E-06
22:29098866:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.265C>A; c.259C>A; c.265C>A	p.Pro89Thr; p.Pro87Thr; p.Pro89Thr	FALSE	1.16E-06
22:29094296:CAG:C	ENST00000400335; ENST00000407188; ENST00000327813	c.138_139delGA; c.132_133delGA; c.138_139delGA	p.Asn47fs; p.Asn45fs; p.Asn47fs	TRUE	1.16E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137359:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.649G>A; c.643G>A; c.649G>A	p.Gly217Ser; p.Gly215Ser; p.Gly217Ser	FALSE	4.64E-06
22:29137359:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.649G>A; c.643G>A; c.649G>A	p.Gly217Ser; p.Gly215Ser; p.Gly217Ser	FALSE	6.48E-06
22:29137359:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.649G>A; c.643G>A; c.649G>A	p.Gly217Ser; p.Gly215Ser; p.Gly217Ser	FALSE	1.72E-05
22:29142019:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1284T>G; c.1329T>G; c.1335T>G	p.Ile428Met; p.Ile443Met; p.Ile445Met	FALSE	3.48E-06
22:29125417:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631+1G>T; c.625+1G>T; c.631+1G>T		TRUE	1.30E-05
22:29125417:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631+1G>T; c.625+1G>T; c.631+1G>T		TRUE	0.000113071
22:29125417:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631+1G>T; c.625+1G>T; c.631+1G>T		TRUE	8.46E-06
22:29125417:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631+1G>T; c.625+1G>T; c.631+1G>T		TRUE	4.64E-05
22:29125417:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.631+1G>T; c.625+1G>T; c.631+1G>T		TRUE	9.74E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121423:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.419C>T; c.413C>T; c.419C>T	p.Thr140Met; p.Thr138Met; p.Thr140Met	FALSE	3.48E-06
22:29098869:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.268G>C; c.262G>C; c.268G>C	p.Asp90His; p.Asp88His; p.Asp90His	FALSE	4.64E-05
22:29098869:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.268G>C; c.262G>C; c.268G>C	p.Asp90His; p.Asp88His; p.Asp90His	FALSE	2.32E-06
22:29121444:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.440A>T; c.434A>T; c.440A>T	p.Gln147Leu; p.Gln145Leu; p.Gln147Leu	FALSE	2.32E-06
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	7.62E-05
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	0.000185443
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	3.44E-05
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	7.77E-05
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	5.22E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137633:A:T	ENST00000400335; ENST00000407188; ENST00000327813	c.923A>T; c.917A>T; c.923A>T	p.Asp308Val; p.Asp306Val; p.Asp308Val	FALSE	0.000113071
22:29094282:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.122A>G; c.116A>G; c.122A>G	p.Asp41Gly; p.Asp39Gly; p.Asp41Gly	FALSE	8.46E-06
22:29094282:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.122A>G; c.116A>G; c.122A>G	p.Asp41Gly; p.Asp39Gly; p.Asp41Gly	FALSE	6.87E-05
22:29094282:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.122A>G; c.116A>G; c.122A>G	p.Asp41Gly; p.Asp39Gly; p.Asp41Gly	FALSE	4.64E-06
22:29094282:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.122A>G; c.116A>G; c.122A>G	p.Asp41Gly; p.Asp39Gly; p.Asp41Gly	FALSE	6.48E-06
22:29138716:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1057G>T; c.1051G>T; c.1057G>T	p.Ala353Ser; p.Ala351Ser; p.Ala353Ser	FALSE	1.30E-05
22:29138716:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1057G>T; c.1051G>T; c.1057G>T	p.Ala353Ser; p.Ala351Ser; p.Ala353Ser	FALSE	4.64E-06
22:29140294:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1136A>G; c.1181A>G; c.1187A>G	p.Tyr379Cys; p.Tyr394Cys; p.Tyr396Cys	FALSE	6.87E-05
22:29140294:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1136A>G; c.1181A>G; c.1187A>G	p.Tyr379Cys; p.Tyr394Cys; p.Tyr396Cys	FALSE	9.27E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29140294:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1136A>G; c.1181A>G; c.1187A>G	p.Tyr379Cys; p.Tyr394Cys; p.Tyr396Cys	FALSE	1.30E-05
22:29094769:GC:G	ENST00000400335; ENST00000407188; ENST00000327813	c.111delC; c.105delC; c.111delC	p.Asn38fs; p.Asn36fs; p.Asn38fs	TRUE	6.48E-06
22:29125389:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.604G>A; c.598G>A; c.604G>A	p.Gly202Ser; p.Gly200Ser; p.Gly202Ser	FALSE	1.16E-06
22:29137540:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.830C>T; c.824C>T; c.830C>T	p.Thr277Ile; p.Thr275Ile; p.Thr277Ile	FALSE	2.20E-05
22:29137540:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.830C>T; c.824C>T; c.830C>T	p.Thr277Ile; p.Thr275Ile; p.Thr277Ile	FALSE	6.48E-06
22:29094389:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.229G>C; c.223G>C; c.229G>C	p.Glu77Gln; p.Glu75Gln; p.Glu77Gln	FALSE	3.44E-05
22:29098940:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.339A>G; c.333A>G; c.339A>G	p.Ile113Met; p.Ile111Met; p.Ile113Met	FALSE	1.27E-05
22:29098939:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.338T>C; c.332T>C; c.338T>C	p.Ile113Thr; p.Ile111Thr; p.Ile113Thr	FALSE	1.16E-06
22:29121428:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.424A>C; c.418A>C; c.424A>C	p.Asn142His; p.Asn140His; p.Asn142His	FALSE	1.16E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121428:A:C	ENST00000400335; ENST00000407188; ENST00000327813	c.424A>C; c.418A>C; c.424A>C	p.Asn142His; p.Asn140His; p.Asn142His	FALSE	8.46E-06
22:29137350:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.640G>T; c.634G>T; c.640G>T	p.Gly214Cys; p.Gly212Cys; p.Gly214Cys	FALSE	6.48E-06
22:29137350:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.640G>T; c.634G>T; c.640G>T	p.Gly214Cys; p.Gly212Cys; p.Gly214Cys	FALSE	8.46E-06
22:29138770:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1111C>T; c.1105C>T; c.1111C>T	p.Gln371*; p.Gln369*; p.Gln371*	TRUE	1.16E-06
22:29073134:G:GCGCCGCC	ENST00000400335; ENST00000407188; ENST00000327813	c.5_11dupCGCCGCC; c.5_11dupCGCCGCC; c.5_11dupCGCCGCC	p.Arg7fs; p.Arg7fs; p.Arg7fs	TRUE	2.32E-06
22:29140284:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1126T>C; c.1171T>C; c.1177T>C	p.Trp376Arg; p.Trp391Arg; p.Trp393Arg	FALSE	1.16E-06
22:29137404:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.694T>C; c.688T>C; c.694T>C	p.Phe232Leu; p.Phe230Leu; p.Phe232Leu	FALSE	5.80E-06
22:29137552:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.842T>C; c.836T>C; c.842T>C	p.Leu281Pro; p.Leu279Pro; p.Leu281Pro	FALSE	0.0001374
22:29094362:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.202T>C; c.196T>C; c.202T>C	p.Tyr68His; p.Tyr66His; p.Tyr68His	FALSE	1.04E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29094362:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.202T>C; c.196T>C; c.202T>C	p.Tyr68His; p.Tyr66His; p.Tyr68His	FALSE	3.89E-05
22:29094362:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.202T>C; c.196T>C; c.202T>C	p.Tyr68His; p.Tyr66His; p.Tyr68His	FALSE	8.46E-06
22:29094362:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.202T>C; c.196T>C; c.202T>C	p.Tyr68His; p.Tyr66His; p.Tyr68His	FALSE	4.64E-05
22:29094362:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.202T>C; c.196T>C; c.202T>C	p.Tyr68His; p.Tyr66His; p.Tyr68His	FALSE	1.72E-05
22:29125385:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.600T>G; c.594T>G; c.600T>G	p.Cys200Trp; p.Cys198Trp; p.Cys200Trp	FALSE	6.48E-06
22:29094333:T:G	ENST00000400335; ENST00000407188; ENST00000327813	c.173T>G; c.167T>G; c.173T>G	p.Leu58Arg; p.Leu56Arg; p.Leu58Arg	FALSE	1.16E-06
22:29121443:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.439C>A; c.433C>A; c.439C>A	p.Gln147Lys; p.Gln145Lys; p.Gln147Lys	FALSE	1.72E-05
22:29137498:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.788T>C; c.782T>C; c.788T>C	p.Ile263Thr; p.Ile261Thr; p.Ile263Thr	FALSE	8.46E-06
22:29137498:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.788T>C; c.782T>C; c.788T>C	p.Ile263Thr; p.Ile261Thr; p.Ile263Thr	FALSE	6.48E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29137498:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.788T>C; c.782T>C; c.788T>C	p.Ile263Thr; p.Ile261Thr; p.Ile263Thr	FALSE	1.16E-06
22:29073133:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.3G>T; c.3G>T; c.3G>T	p.Met1?; p.Met1? p.Met1?	TRUE	1.16E-06
22:29098922:C:A	ENST00000400335; ENST00000407188; ENST00000327813	c.321C>A; c.315C>A; c.321C>A	p.Tyr107*; p.Tyr105*; p.Tyr107*	TRUE	1.16E-06
22:29137530:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.820G>T; c.814G>T; c.820G>T	p.Asp274Tyr; p.Asp272Tyr; p.Asp274Tyr	FALSE	6.48E-06
22:29125270:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.485G>A; c.479G>A; c.485G>A	p.Gly162Glu; p.Gly160Glu; p.Gly162Glu	FALSE	2.43E-05
22:29125270:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.485G>A; c.479G>A; c.485G>A	p.Gly162Glu; p.Gly160Glu; p.Gly162Glu	FALSE	6.48E-06
22:29125282:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.497G>T; c.491G>T; c.497G>T	p.Gly166Val; p.Gly164Val; p.Gly166Val	FALSE	1.16E-06
22:29125282:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.497G>T; c.491G>T; c.497G>T	p.Gly166Val; p.Gly164Val; p.Gly166Val	FALSE	3.44E-05
22:29138676:CG:C	ENST00000400335; ENST00000407188; ENST00000327813	c.1018delG; c.1012delG; c.1018delG	p.Glu340fs; p.Glu338fs; p.Glu340fs	TRUE	2.54E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29121383:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.379G>A; c.373G>A; c.379G>A	p.Asp127Asn; p.Asp125Asn; p.Asp127Asn	FALSE	1.16E-06
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.000211606
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	4.64E-05
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.000206157
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.000480901
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.000226693
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.00011127
22:29125344:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.559G>A; c.553G>A; c.559G>A	p.Glu187Lys; p.Glu185Lys; p.Glu187Lys	FALSE	0.000339213
22:29094281:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.121G>C; c.115G>C; c.121G>C	p.Asp41His; p.Asp39His; p.Asp41His	FALSE	2.32E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29138677:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1018G>A; c.1012G>A; c.1018G>A	p.Glu340Lys; p.Glu338Lys; p.Glu340Lys	FALSE	1.30E-05
22:29138677:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1018G>A; c.1012G>A; c.1018G>A	p.Glu340Lys; p.Glu338Lys; p.Glu340Lys	FALSE	4.64E-05
22:29138677:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1018G>A; c.1012G>A; c.1018G>A	p.Glu340Lys; p.Glu338Lys; p.Glu340Lys	FALSE	6.87E-05
22:29138677:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1018G>A; c.1012G>A; c.1018G>A	p.Glu340Lys; p.Glu338Lys; p.Glu340Lys	FALSE	1.04E-05
22:29138677:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.1018G>A; c.1012G>A; c.1018G>A	p.Glu340Lys; p.Glu338Lys; p.Glu340Lys	FALSE	8.47E-06
22:29073186:CG:C	ENST00000400335; ENST00000407188; ENST00000327813	c.58delG; c.58delG; c.58delG	p.Ala20fs; p.Ala20fs; p.Ala20fs	TRUE	1.30E-05
22:29141987:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1252C>T; c.1297C>T; c.1303C>T	p.Gln418*; p.Gln433*; p.Gln435*	TRUE	2.32E-06
22:29098920:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.319T>C; c.313T>C; c.319T>C	p.Tyr107His; p.Tyr105His; p.Tyr107His	FALSE	1.94E-05
22:29137671:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.961C>T; c.955C>T; c.961C>T	p.Gln321*; p.Gln319*; p.Gln321*	TRUE	4.64E-06

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29167043:G:A	ENST00000327813	c.1417-1G>A		TRUE	9.27E-06
22:29098884:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.283C>T; c.277C>T; c.283C>T	p.Pro95Ser; p.Pro93Ser; p.Pro95Ser	FALSE	1.51E-05
22:29121372:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.368G>T; c.362G>T; c.368G>T	p.Gly123Val; p.Gly121Val; p.Gly123Val	FALSE	1.85E-05
22:29125396:G:A	ENST00000400335; ENST00000407188; ENST00000327813	c.611G>A; c.605G>A; c.611G>A	p.Gly204Asp; p.Gly202Asp; p.Gly204Asp	FALSE	2.32E-06
22:29094291:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.131G>C; c.125G>C; c.131G>C	p.Gly44Ala; p.Gly42Ala; p.Gly44Ala	FALSE	1.69E-05
22:29094291:G:C	ENST00000400335; ENST00000407188; ENST00000327813	c.131G>C; c.125G>C; c.131G>C	p.Gly44Ala; p.Gly42Ala; p.Gly44Ala	FALSE	1.30E-05
22:29098870:A:G	ENST00000400335; ENST00000407188; ENST00000327813	c.269A>G; c.263A>G; c.269A>G	p.Asp90Gly; p.Asp88Gly; p.Asp90Gly	FALSE	1.72E-05
22:29138755:C:G	ENST00000400335; ENST00000407188; ENST00000327813	c.1096C>G; c.1090C>G; c.1096C>G	p.Pro366Ala; p.Pro364Ala; p.Pro366Ala	FALSE	1.16E-06
22:29094348:CTT:C	ENST00000400335; ENST00000407188; ENST00000327813	c.190_191delTT; c.184_185delTT; c.190_191delTT	p.Phe64fs; p.Phe62fs; p.Phe64fs	TRUE	1.16E-06
22:29094390:A:AG	ENST00000400335; ENST00000407188; ENST00000327813	c.236dupG; c.230dupG; c.236dupG	p.Leu80fs; p.Leu78fs; p.Leu80fs	TRUE	4.64E-05

Figure 3 (cont.)

Genomic coordinates and alleles for genetic variant	Transcript(s)	Coding DNA change	Protein change	Variant classified as pLOF	AAF, fraction of 1
22:29125383:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.598T>C; c.592T>C; c.598T>C	p.Cys200Arg; p.Cys198Arg; p.Cys200Arg	FALSE	1.16E-06
22:29138753:G:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1094G>T; c.1088G>T; c.1094G>T	p.Ser365Ile; p.Ser363Ile; p.Ser365Ile	FALSE	1.16E-06
22:29125296:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.511T>C; c.505T>C; c.511T>C	p.Cys171Arg; p.Cys169Arg; p.Cys171Arg	FALSE	2.32E-06
22:29142029:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1294C>T; c.1339C>T; c.1345C>T	p.Pro432Ser; p.Pro447Ser; p.Pro449Ser	FALSE	8.46E-06
22:29142029:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1294C>T; c.1339C>T; c.1345C>T	p.Pro432Ser; p.Pro447Ser; p.Pro449Ser	FALSE	4.64E-05
22:29142077:C:T	ENST00000400335; ENST00000407188; ENST00000327813	c.1342C>T; c.1387C>T; c.1393C>T	p.Gln448*; p.Gln463*; p.Gln465*	TRUE	1.16E-06
22:29137668:T:C	ENST00000400335; ENST00000407188; ENST00000327813	c.958T>C; c.952T>C; c.958T>C	p.Tyr320His; p.Tyr318His; p.Tyr320His	FALSE	1.16E-06

Figure 3 (cont.)