

## Human CAZyme genes polymorphism and risk of IBS: a population-based study

A series of papers in *Gut* recently highlighted genetic variation in the sucrase-isomaltase gene (*SI*; coding for a brush-border disaccharidase) as a likely causative factor in a subset of patients with irritable bowel syndrome (IBS).<sup>1-4</sup> Hypomorphic (dysfunctional) *SI* variants may thus underlie gastrointestinal symptoms in rare recessive forms of congenital SI deficiency (CSID)<sup>5</sup> as well as milder complex (IBS) manifestations,<sup>6</sup> across a broad spectrum of *genetic SI deficiencies* (GSID) that vary in severity and onset of presentation.<sup>7</sup> Moreover, *SI* carrier status has been shown to also affect the response to specific carbohydrate-focused diets, thus providing a rationale for personalising (dietary) therapeutic strategies in IBS.<sup>1,8</sup>

Together with *SI*, a number of human Carbohydrate-Active enZymes (hCAZymes, [www.cazy.org/e355.html](http://www.cazy.org/e355.html)) are involved in the breakdown of polysaccharides during the process of carbohydrate digestion, which is initiated by salivary amylases (AMyS) and finalised in the small intestine by pancreatic AMyS and brush-border disaccharidases *SI*, lactase (*LCT*), maltase-glucoamylase (*MGAM*) and trehalase (*TREH*) (figure 1).<sup>5</sup> Of note, similar to *SI* in GSIDs, mutations in other hCAZymes cause rare genetic forms of carbohydrate maldigestion, while regulatory DNA variations (persistent genotype) influence lactose intolerance in adults.<sup>5</sup> This suggests hCAZyme genes other than *SI* may contribute to IBS predisposition via similar mechanisms (reduced hCAZyme activity increasing IBS risk): we sought to test this hypothesis through the analysis of genetic and health-related data in

**Table 1** Significant associations between hCAZymes genes and risk of IBS

	IBS definition	OR (95% CI)	P value	P <sub>FDR</sub>
Single-gene analyses*				
<i>SI</i>	Self-IBS	1.1 (1.0 to 1.3)	0.0101	0.0296
<i>SI</i>	HA-IBS	1.1 (1.0 to 1.3)	0.0203	0.0296
<i>SI</i>	Rome-IBS	1.1 (1.0 to 1.2)	0.0222	0.0296
<i>AMY1B</i>	HA-IBS	1.7 (1.1 to 2.7)	0.0078	0.0311
<i>AMY2A</i>	HA-IBS	1.5 (1.1 to 2.2)	0.0079	0.0317
Multiple gene analyses†				
Any gene	HA-IBS	1.2 (1.1 to 1.3)	0.0015	0.0059
Any gene	Self-IBS	1.2 (1.0 to 1.3)	0.0030	0.0061
Any gene	Rome-IBS	1.1 (1.0 to 1.2)	0.0143	0.0190
Number of genes affected	HA-IBS	1.2 (1.1 to 1.3)	0.0011	0.0045
Number of genes affected	Self-IBS	1.2 (1.1 to 1.3)	0.0040	0.0079
Number of genes affected	Rome-IBS	1.1 (1.0 to 1.2)	0.0151	0.0201

\*Studying carriers of hCAZyme hypomorphic variants in individual genes.

†Studying carriers of hCAZyme hypomorphic variants in any gene (*AMY1B*, *AMY2A* or *SI*) and as a function of the number of genes affected.

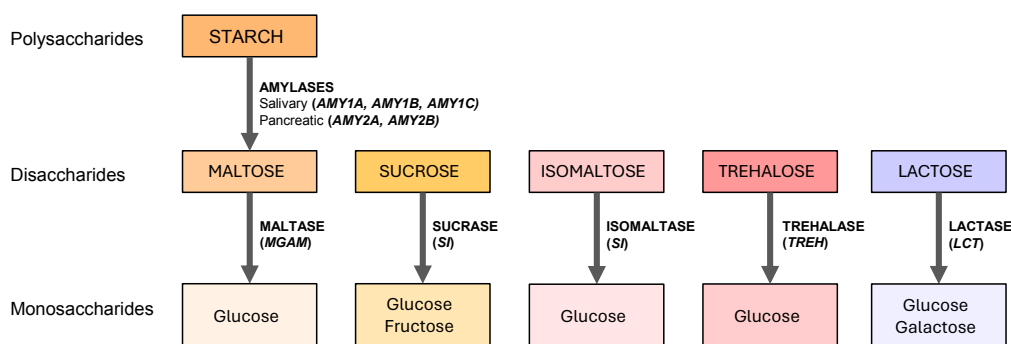
AMY, amylase genes; HA-IBS, Hospital inpatient Admissions IBS; hCAZymes, human Carbohydrate-Active enZymes; IBS, irritable bowel syndrome; Rome-IBS, Rome III Criteria from the Digestive Health Questionnaire IBS; Self-IBS, Self-reported IBS; *SI*, sucrase-isomaltase gene.

366,432 individuals of European ancestry from the large population-based cohort UK Biobank (UKBB) detailed methods' description is provided in online supplemental material).

Patients with IBS were identified across four definitions, based on alternative diagnoses from hospital admissions, general practitioner's notes, Digestive Health Questionnaires including Rome III Criteria and as self-reported condition from health-related questionnaires (online supplemental table S1). Rare (allele frequency <1%) hCAZyme functional (missense, nonsense, read-through and splice-site) DNA variants were extracted from UKBB whole-exome sequencing data for nine genes of interest (*AMY1A*, *AMY1B*, *AMY1C*, *AMY2A*, *AMY2B*, *LCT*, *MGAM*, *SI* and *TREH*), and hypomorphic variants computationally predicted

using stringent criteria from Ensembl Variant Effect Predictor and the pathogenicity classifier AlphaMissense. UKBB participants were stratified into carrier and non-carrier groups for each gene, and hCAZyme-IBS associations were tested via adjusted logistic regression. Additionally, cumulative analyses were carried out for selected genes, in which carrier status was considered collapsing hypomorphic variants from multiple genes into a single group.

In total, 1714 hypomorphic variants were identified across hCAZyme genes (online supplemental table S2). As reported in table 1, when individually tested for association with IBS (based on different definitions), three hCAZyme genes showed significant effects on disease risk after correction for multiple comparisons, namely *SI* (as previously shown),





**Figure 1** Schematic representation of the process of carbohydrate digestion. Reported are the types of carbohydrates, the hCAZymes involved and the corresponding genes studied here.

*AMY1B* (a salivary AMY) and *AMY2A* (a pancreatic AMY). Carriers of a hypomorphic variant in any of these genes were also exposed to increased risk of IBS (table 1), while the strongest association was detected in relation to the number of genes affected by hypomorphic variation (table 1). No other significant associations were detected (not shown).

These results confirm and extend previous findings on the importance of hCAZyme genotype in relation to IBS risk.<sup>6,7</sup> *AMY1B* and *AMY2A* code for AMYs that break down starch into smaller sugars and disaccharides, hence their reduced activity may ultimately lead to excess carbohydrates in the lower bowel where they induce IBS symptoms via osmotic diarrhoea and bacterial fermentation. Of note, *AMY1* and *AMY2* copy numbers (a type of genetic variation not studied here) have been shown to affect AMY activity and starch intake, as well as oral and gut microbiota composition.<sup>9,10</sup>

In summary, we provide additional evidence that hCAZyme genotype is relevant to IBS risk. This warrants replication of current findings, as it holds potential implications for personalising (dietary) therapeutic approaches in IBS.

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**Contributors** MD'A and FB study design, conceptualization and supervision; LT, AZT, CEB and FB statistical and computational analysis; LT, AZT, CEB, FB, IB and MD'A data analysis and interpretation; MD'A obtained funding; MD'A, FB, LT and AZT drafted the

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1       **Human CAZyme genes polymorphism and risk of**  
2       **IBS: a population-based study**

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8       **SUPPLEMENTARY MATERIAL**

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## 21 **Supplementary Methods**

### 22 **UK Biobank data**

23 UK Biobank (UKBB, <https://www.ukbiobank.ac.uk>), is a large-scale biomedical database  
24 that holds biological samples and genetic, lifestyle and health-related information from  
25 500,000 volunteers aged between 40-60 years old.[1] We extracted genetic and  
26 phenotypic data from 366,432 unrelated participants (pairwise kinship coefficient  
27 <0.0442) self-reporting *white-British*, *Irish*, *white ethnic* background (UKBB data field  
28 21000).

29 UKBB participants with irritable bowel syndrome (IBS) were identified based on four  
30 different diagnoses and data types: from electronic medical records (EMRs) of i) hospital  
31 inpatient admissions (HA-IBS; UKBB data field 41202) and ii) general practitioners' notes  
32 (GP-IBS; UKBB data field 42040), according to the Tenth version of the International  
33 Classification of Diseases (ICD10); as self-reported conditions based on touchscreen  
34 questionnaires at enrolment (Self-IBS; UKBB data field 20002); and based on Rome III  
35 Criteria from the Digestive Health Questionnaire (Rome-IBS; UKBB data fields 21025,  
36 21027-8, 21031-4).

37 High quality (UKBB curated) DNA variants from human Carbohydrate-Active enZymes  
38 (hCAZymes; *AMY1A*, *AMY1B*, *AMY1C*, *AMY2A*, *AMY2B*, *LCT*, *MGAM*, *SI*, *TREH*) genes  
39 were extracted from UKBB whole exome sequencing (WES) data (UKBB Category 170).  
40 Top ten principal components from UKBB genotype data (UKBB data field 22020) were  
41 also extracted for all UKBB participants, and included as covariates in association  
42 analyses to control for cryptic relatedness and population substructure (see below).

### 43 **hCAZymes hypomorphic variant classification**

44 GRCh38 genomic coordinates were used to extract rare variants (allele frequency <1%)  
45 from the coding regions of hCAZyme genes of interest. Hypomorphic variants were  
46 identified via a stringent computational analysis based on the Ensembl Variant Effect  
47 Predictor (VEP release 112, [www.ensembl.org/info/docs/tools/vep](http://www.ensembl.org/info/docs/tools/vep)) and annotation with

48 AlphaMissense, the current best-performing pathogenicity classifier  
49 (alphamissense.hegelab.org).[2] Specifically, hypomorphic variants were defined as  
50 having an AlphaMissense score >0.564 (likely pathogenic) or a VEP HIGH impact  
51 classification (stop gained/lost, frameshift and splicing altering variants). Variants  
52 annotated as ambiguous (AlphaMissense score range 0.34-0.564), and corresponding  
53 UKBB carriers were excluded from the analysis to avoid uncertainty, while benign  
54 variants (score <0.34) were ignored.

55 By these means, a total of 1714 hypomorphic variants were identified: 23 in *AMY1A* (0.05  
56 variant/residue), 17 in *AMY1B* (0.03 variant/residue), 32 in *AMY1C* (0.06  
57 variant/residue), 98 in *AMY2A* (0.19 variant/residue), 166 in *AMY2B* (0.32  
58 variant/residue), 265 in *LCT* (0.14 variant/residue), 451 in *MGAM* (0.16 variant/residue),  
59 539 in *SI* (0.3 variant/residue) and 123 in *TREH* (0.21 variant/residue). These were  
60 included in downstream statistical analyses.

### 61 **Association analyses**

62 In order to evaluate the cumulative effect of hypomorphic variants in hCAZyme genes,  
63 collapsing analyses were performed as previously described,[3-7] stratifying UKBB  
64 participants into carriers of hypomorphic variants in individual genes, carriers of  
65 hypomorphic variants in any gene and non-carriers. Hypomorphic variants were  
66 collapsed into single gene groups in order to examine the impact of carrier status on IBS  
67 patients. An additional analysis was carried out to address the effect of carriership of  
68 hypomorphic variants in selected hCAZyme genes (*AMY1B*, *AMY2A* and *SI*) and the  
69 cumulative effect of the number of such affected genes. Collapsing analysis was  
70 modelled based on logistic regression implemented in R (*glm* function) including sex,  
71 age and top 10 genetic principal components as covariates. P-values (one-tailed based  
72 on previous findings) [3-5] were adjusted by false discovery rate (FDR) to control for type  
73 I error in all analyses.

**74 Ethics and data access**

75 UKBB received ethical approval from the competent Research Ethics Committee (REC  
76 reference 11/NW/0382), and the research reported here has been conducted under  
77 application number 17435; PI MD'A). This work is part of a large study on the genetics  
78 of gastrointestinal diseases (GenGIScan project), approved by the local Basque Ethics  
79 Committee (Comité de Ética de la Investigación con medicamentos de Euskadi, code:  
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99 and response to a starch-reduced and sucrose-reduced diet in IBS-D patients.  
100 *Gut* 2024;73: 706-708.

101 **Supplementary Tables****Table S1.** Study subjects: demographics, IBS definitions, and hCAZyme variant carriers.

	<b>Self-IBS</b>	<b>HA-IBS</b>	<b>Rome-IBS</b>	<b>GP-IBS</b>
UK Biobank data field	20002	41202	21025, 21027-8, 21031-4	42040
N	366432	366432	85864	169390
Age (sd)	56.7 (8.0)	56.7 (8.0)	56.3 (7.6)	56.7 (8.0)
% females	53.9	53.9	53.8	54.2
N cases	9702	9332	24470	13562
N controls	356730	357100	61394	155828
AMY1A carriers cases	98	90	256	147
AMY1A carriers controls	3569	3577	627	1525
AMY1B carriers cases	16	20	38	12
AMY1B carriers controls	442	438	80	196
AMY1C carriers cases	4	6	15	11
AMY1C carriers controls	277	275	46	106
AMY2A carriers cases	28	34	62	28
AMY2A carriers controls	848	842	145	370
AMY2B carriers cases	89	95	239	126
AMY2B carriers controls	3699	3693	628	1646
LCT carriers cases	17	24	49	34
LCT carriers controls	675	668	111	302
SI carriers case	134	120	310	193
SI carriers controls	5037	5051	882	2090
MGAM carriers case	348	330	806	454
MGAM carriers controls	11282	11300	1850	4839
TREH carriers cases	74	80	175	105
TREH carriers controls	2675	2669	476	1214

102 **Self-IBS:** self-reported condition from touchscreen questionnaires at enrolment; **HA-IBS:** IBS from hospital  
103 admissions; **Rome-IBS:** Rome III-criteria based diagnosis from Digestive Health Questionnaire; **GP-IBS:**  
104 IBS from general practitioner's notes

105

**Table S2.** hCAZyme hypomorphic variants included in the analyses.

Gene	Genomic position and variation	dbSNP	aa position	aa change	Consequence	VEP/AlphaMissense classification	UKBB AF	Ncarriers
AMY1A	1:103660440:A:G	rs1444701875	320	H/R	missense_variant	pathogenic	0,0000014	1
AMY1A	1:103660461:T:C	-	327	I/T	missense_variant	pathogenic	0,0000014	1
AMY1A	1:103660439:C:T	rs375474128	320	H/Y	missense_variant	pathogenic	0,0000014	1
AMY1A	1:103660434:G:A	rs751946952	318	R/Q	missense_variant	pathogenic	0,0000014	1
AMY1A	1:103660652:G:A	rs770578493	359	W/*	stop_gained	HIGH	0,0000014	1
AMY1A	1:103660466:A:C	-	329	T/P	missense_variant	pathogenic	0,0000027	1
AMY1A	1:103660604:G:A	rs1417084595	343	M/I	missense_variant	pathogenic	0,0000027	2
AMY1A	1:103660474:G:A	-	331	W/*	stop_gained	HIGH	0,0000041	3
AMY1A	1:103660366:G:A	-	295	W/*	stop_gained	HIGH	0,0000055	2
AMY1A	1:103660440:A:AT	rs759859897	320	H/HX	frameshift_variant	HIGH	0,0000068	5
AMY1A	1:103662902:C:T	-	413	R/C	missense_variant	pathogenic	0,0000068	5
AMY1A	1:103660474:G:T	rs769271241	331	W/C	missense_variant	pathogenic	0,0000096	4
AMY1A	1:103660474:G:C	rs769271241	331	W/C	missense_variant	pathogenic	0,0000109	7
AMY1A	1:103660418:A:G	rs771184271	313	N/D	missense_variant	pathogenic	0,0000123	6
AMY1A	1:103660462:A:G	rs771075619	327	I/M	missense_variant	pathogenic	0,0000150	9
AMY1A	1:103660443:G:A	rs1376235977	321	G/D	missense_variant	pathogenic	0,0000164	12
AMY1A	1:103660445:G:A	rs755582791	322	A/T	missense_variant	pathogenic	0,0000246	17
AMY1A	1:103660458:C:G	rs778642937	326	S/C	missense_variant	pathogenic	0,0000355	26
AMY1A	1:103660433:C:T	rs367719363	318	R/*	stop_gained	HIGH	0,0000437	31
AMY1A	1:103660431:A:G	rs138172355	317	Q/R	missense_variant	pathogenic	0,0001187	61
AMY1A	1:103660629:C:T	rs748731907	352	R/*	stop_gained	HIGH	0,0001665	120
AMY1A	1:103660419:A:T	rs150405191	313	N/I	missense_variant	pathogenic	0,0003208	235
AMY1A	1:103660401:C:T	rs201288184	307	A/V	missense_variant	pathogenic	0,0058213	3117

6



AMY1B	1:103691527:C:T	rs1197835323	321	G/D	missense_variant	pathogenic	0,0000027	1
AMY1B	1:103691548:T:C	-	314	H/R	missense_variant	pathogenic	0,0000027	1
AMY1B	1:103691362:C:G	rs1264541618	345	A/P	missense_variant	pathogenic	0,0000027	2
AMY1B	1:103691349:C:A	-	349	G/V	missense_variant	pathogenic	0,0000027	1
AMY1B	1:103691358:T:C	-	346	H/R	missense_variant	pathogenic	0,0000027	1
AMY1B	1:103691604:C:T	-	295	W/*	stop_gained	HIGH	0,0000041	3
AMY1B	1:103691597:C:A	rs768409168	298	G/C	missense_variant	pathogenic	0,0000055	2
AMY1B	1:103691592:C:G	-	299	W/C	missense_variant	pathogenic	0,0000055	4
AMY1B	1:103691487:C:A	rs201703350	-	-	splice_donor_variant	HIGH	0,0000096	7
AMY1B	1:103691488:C:G	rs750169595	334	R/T	missense_variant,splice_region_variant	pathogenic	0,0000123	9
AMY1B	1:103691536:C:T	rs773061354	318	R/Q	missense_variant	pathogenic	0,0000137	10
AMY1B	1:103691537:G:A	rs760326172	318	R/*	stop_gained	HIGH	0,0000150	7
AMY1B	1:103691569:G:A	rs201627077	307	A/V	missense_variant	pathogenic	0,0000191	14
AMY1B	1:103691518:C:T	rs769501303	324	G/E	missense_variant	pathogenic	0,0000218	16
AMY1B	1:103691341:G:A	rs199554865	352	R/*	stop_gained	HIGH	0,0000601	36
AMY1B	1:103691525:C:T	rs140294416	322	A/T	missense_variant	pathogenic	0,0002129	94
AMY1B	1:103691557:A:T	rs549527747	311	V/E	missense_variant	pathogenic	0,0003441	250
AMY1C	1:103754581:G:A	-	321	G/S	missense_variant	pathogenic	0,0000014	1
AMY1C	1:103754561:A:G	rs369196111	314	H/R	missense_variant	pathogenic	0,0000014	1
AMY1C	1:103750511:G:A	-	14	W/*	stop_gained	HIGH	0,0000014	1
AMY1C	1:103750483:G:A	-	5	W/*	stop_gained	HIGH	0,0000014	1
AMY1C	1:103754591:G:A	rs775987541	324	G/E	missense_variant	pathogenic	0,0000027	2
AMY1C	1:103754756:A:G	rs774389336	348	Y/C	missense_variant	pathogenic	0,0000027	1
AMY1C	1:103754798:A:AT	-	362	Y/YX	frameshift_variant	HIGH	0,0000027	1
AMY1C	1:103750552:T:C	-	28	I/T	missense_variant	pathogenic	0,0000027	2
AMY1C	1:103750575:T:C	rs1449635143	36	W/R	missense_variant	pathogenic	0,0000027	2
AMY1C	1:103750546:C:T	-	26	T/I	missense_variant	pathogenic	0,0000027	2
AMY1C	1:103750602:C:T	rs1350376061	45	R/*	stop_gained	HIGH	0,0000027	2

AMY1C	1:103754750:A:G	-	346	H/R	missense_variant	pathogenic	0,0000041	2
AMY1C	1:103754768:G:C	-	352	R/P	missense_variant	pathogenic	0,0000041	2
AMY1C	1:103750588:C:T	-	40	A/V	missense_variant	pathogenic	0,0000041	3
AMY1C	1:103754743:C:T	rs1244427774	344	L/F	missense_variant	pathogenic	0,0000055	4
AMY1C	1:103750597:G:A	-	43	C/Y	missense_variant	pathogenic	0,0000055	4
AMY1C	1:103754816:T:C	rs969510155	-	-	splice_donor_variant	HIGH	0,0000068	5
AMY1C	1:103754517:G:A	-	299	W/*	stop_gained	HIGH	0,0000068	4
AMY1C	1:103750573:G:T	-	35	R/L	missense_variant	pathogenic	0,0000069	5
AMY1C	1:103750571:G:A	rs1246577917	34	W/*	stop_gained	HIGH	0,0000082	6
AMY1C	1:103750542:C:T	rs1238284689	25	R/*	stop_gained	HIGH	0,0000096	7
AMY1C	1:103750519:AT:A	rs1355004560	17	Y/X	frameshift_variant	HIGH	0,0000097	7
AMY1C	1:103754587:G:A	-	323	G/R	missense_variant	pathogenic	0,0000109	8
AMY1C	1:103754759:G:A	rs773043065	349	G/E	missense_variant	pathogenic	0,0000109	8
AMY1C	1:103754622:G:A	rs1181489744	-	-	splice_donor_variant	HIGH	0,0000123	9
AMY1C	1:103750572:C:T	rs938886200	35	R/*	stop_gained	HIGH	0,0000123	9
AMY1C	1:103754584:G:A	rs139911862	322	A/T	missense_variant	pathogenic	0,0000136	10
AMY1C	1:103754780:G:A	-	356	S/N	missense_variant	pathogenic	0,0000191	12
AMY1C	1:103754572:C:T	rs752371105	318	R/*	stop_gained	HIGH	0,0000287	20
AMY1C	1:103754582:G:C	rs779977255	321	G/A	missense_variant	pathogenic	0,0000628	46
AMY1C	1:103754573:G:A	rs757570508	318	R/Q	missense_variant	pathogenic	0,0000628	46
AMY1C	1:103754540:C:T	rs201967863	307	A/V	missense_variant	pathogenic	0,0000683	48
AMY2A	1:103617442:T:G	-	1	M/R	start_lost	HIGH	0,0000014	1
AMY2A	1:103617530:T:A	rs773792622	30	H/Q	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103617546:T:C	-	36	W/R	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103617596:T:A	-	52	F/L	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103617603:G:T	rs776915497	55	V/F	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618004:G:T	-	73	W/C	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618007:G:A	-	74	W/*	stop_gained	HIGH	0,0000014	1

AMY2A	1:103618014:T:A	-	77	Y/N	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618044:A:G	rs1164236404	87	R/G	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618098:G:T	-	105	G/W	missense_variant,splice_region_variant	pathogenic	0,0000014	1
AMY2A	1:103619694:G:A	rs1237825167	218	W/*	stop_gained	HIGH	0,0000014	1
AMY2A	1:103623953:T:C	-	397	W/R	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618074:A:C	-	97	M/L	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618087:G:T	-	101	C/F	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618099:G:T	-	105	G/V	missense_variant,splice_region_variant	pathogenic	0,0000014	1
AMY2A	1:103619612:G:T	rs764700031	191	R/L	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103620602:G:T	-	266	G/C	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618948:G:A	rs1327917758	118	C/Y	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103620651:G:T	rs200554552	282	R/L	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618978:G:A	rs765781153	128	S/N	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618990:G:T	-	132	S/I	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103619047:T:A	-	151	F/Y	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618991:T:G	-	132	S/R	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618994:CT:C	-	134	F/X	frameshift_variant	HIGH	0,0000014	1
AMY2A	1:103619043:G:C	-	150	D/H	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103619043:G:T	-	150	D/Y	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618910:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
AMY2A	1:103618910:G:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
AMY2A	1:103619090:C:A	-	165	N/K	missense_variant	pathogenic	0,0000014	1
AMY2A	1:103618912:T:G	-	106	V/G	missense_variant,splice_region_variant	pathogenic	0,0000014	1
AMY2A	1:103619108:G:C	-	171	Q/H	missense_variant,splice_region_variant	pathogenic	0,0000014	1
AMY2A	1:103617535:T:A	-	32	F/Y	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103617554:T:TA	-	38-39	-/X	frameshift_variant	HIGH	0,0000027	1
AMY2A	1:103619630:A:G	rs755790453	197	Y/C	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103619674:G:C	rs1391062344	212	D/H	missense_variant	pathogenic	0,0000027	2

AMY2A	1:103619691:G:A	rs1229372320	217	M/I	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103617553:A:G	-	38	D/G	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103618014:T:C	-	77	Y/H	missense_variant	pathogenic	0,0000027	1
AMY2A	1:103617601:G:A	rs148925092	54	G/E	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103619705:T:G	rs1256688525	222	I/R	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103623939:CT:C	rs1308679582	392	T/X	frameshift_variant	HIGH	0,0000027	2
AMY2A	1:103620670:G:T	rs772830789	288	K/N	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103618987:G:T	-	131	G/V	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103619025:G:T	-	144	V/F	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103618994:C:A	-	133	Y/*	stop_gained	HIGH	0,0000027	2
AMY2A	1:103618995:T:C	-	134	F/L	missense_variant	pathogenic	0,0000027	2
AMY2A	1:103617953:G:A	rs770998615	-	-	splice_acceptor_variant	HIGH	0,0000041	3
AMY2A	1:103618004:G:A	-	73	W/*	stop_gained	HIGH	0,0000041	3
AMY2A	1:103617558:G:A	-	40	A/T	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103617610:T:C	-	-	-	splice_donor_variant	HIGH	0,0000041	2
AMY2A	1:103617968:T:A	rs143617737	61	N/K	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103618050:G:A	rs747889183	89	G/R	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103617573:C:T	rs182106305	45	R/*	stop_gained	HIGH	0,0000041	3
AMY2A	1:103617582:G:C	-	48	A/P	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103619554:G:T	rs1276826607	172	V/F	missense_variant,splice_region_variant	pathogenic	0,0000041	3
AMY2A	1:103620638:G:A	rs746422923	278	G/S	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103619018:T:G	-	141	F/L	missense_variant	pathogenic	0,0000041	2
AMY2A	1:103619025:G:C	-	144	V/L	missense_variant	pathogenic	0,0000041	2
AMY2A	1:103619046:T:A	rs1179541418	151	F/I	missense_variant	pathogenic	0,0000041	3
AMY2A	1:103619070:G:T	-	159	G/*	stop_gained	HIGH	0,0000041	3
AMY2A	1:103619032:AT:A	-	146	Y/X	frameshift_variant	HIGH	0,0000041	3
AMY2A	1:103617607:A:G	-	56	Q/R	missense_variant,splice_region_variant	pathogenic	0,0000055	3
AMY2A	1:103618047:T:A	-	88	S/T	missense_variant	pathogenic	0,0000055	4

AMY2A	1:103618042:C:G	-	86	T/R	missense_variant	pathogenic	0,0000055	4
AMY2A	1:103619578:C:A	rs372577342	180	L/I	missense_variant	pathogenic	0,0000055	4
AMY2A	1:103623969:G:A	rs758134251	402	R/Q	missense_variant	pathogenic	0,0000068	5
AMY2A	1:103618936:T:C	rs1414076605	114	I/T	missense_variant	pathogenic	0,0000068	5
AMY2A	1:103624223:T:G	rs1272132947	-	-	splice_donor_variant	HIGH	0,0000068	5
AMY2A	1:103617580:T:G	-	47	L/*	stop_gained	HIGH	0,0000082	6
AMY2A	1:103619085:G:T	rs763617994	164	E/*	stop_gained	HIGH	0,0000082	6
AMY2A	1:103617543:C:T	rs566657194	35	R/*	stop_gained	HIGH	0,0000096	5
AMY2A	1:103618006:G:C	rs751752481	74	W/S	missense_variant	pathogenic	0,0000096	6
AMY2A	1:103619041:G:A	rs1376849093	149	W/*	stop_gained	HIGH	0,0000096	7
AMY2A	1:103617583:C:A	-	48	A/D	missense_variant	pathogenic	0,0000109	6
AMY2A	1:103619678:C:T	rs775788005	213	A/V	missense_variant	pathogenic	0,0000109	8
AMY2A	1:103623968:C:T	rs1832269	402	R/*	stop_gained	HIGH	0,0000109	8
AMY2A	1:103617487:AG:A	-	16	Q/X	frameshift_variant	HIGH	0,0000123	8
AMY2A	1:103618941:C:T	rs1172882125	116	H/Y	missense_variant	pathogenic	0,0000123	9
AMY2A	1:103618029:TA:T	rs1381196127	82	Y/X	frameshift_variant	HIGH	0,0000136	10
AMY2A	1:103618031:T:G	rs1002259080	82	Y/*	stop_gained	HIGH	0,0000136	10
AMY2A	1:103620650:C:T	rs768370675	282	R/C	missense_variant	pathogenic	0,0000150	11
AMY2A	1:103617553:A:C	rs1267450005	38	D/A	missense_variant	pathogenic	0,0000164	10
AMY2A	1:103618017:C:A	-	78	Q/K	missense_variant	pathogenic	0,0000164	11
AMY2A	1:103619668:A:G	rs531383627	210	R/G	missense_variant	pathogenic	0,0000177	13
AMY2A	1:103618101:G:A	rs748461150	-	-	splice_donor_variant	HIGH	0,0000177	12
AMY2A	1:103617592:G:C	-	51	G/A	missense_variant	pathogenic	0,0000191	14
AMY2A	1:103623966:A:G	rs1208552687	401	H/R	missense_variant	pathogenic	0,0000191	14
AMY2A	1:103617510:G:T	-	24	G/*	stop_gained	HIGH	0,0000205	12
AMY2A	1:103617529:A:G	rs1453867186	30	H/R	missense_variant	pathogenic	0,0000232	15
AMY2A	1:103617544:G:T	rs1279678965	35	R/L	missense_variant	pathogenic	0,0000287	18
AMY2A	1:103618042:C:T	rs773897479	86	T/I	missense_variant	pathogenic	0,0000287	17

AMY2A	1:103619627:A:AAT	rs779706686	196	E/EX	frameshift_variant	HIGH	0,0000314	22
AMY2A	1:103619608:G:A	rs775489232	190	V/M	missense_variant	pathogenic	0,0000314	23
AMY2A	1:103619653:G:A	rs149883399	205	G/S	missense_variant	pathogenic	0,0000491	36
AMY2A	1:103618989:A:T	rs541443659	132	S/C	missense_variant	pathogenic	0,0000519	37
AMY2A	1:103623968:C:G	rs1832269	402	R/G	missense_variant	pathogenic	0,0001515	111
AMY2A	1:103620550:G:A	rs750258474	-	-	splice_acceptor_variant	HIGH	0,0001569	115
AMY2A	1:103624117:TG:T	rs774596733	415	V/X	frameshift_variant	HIGH	0,0002431	178
AMY2B	1:103571604:T:C	-	1	M/T	start_lost	HIGH	0,0000014	1
AMY2B	1:103571643:G:A	-	14	W/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103571648:C:CA	-	16	Q/QX	frameshift_variant	HIGH	0,0000014	1
AMY2B	1:103571666:C:T	-	22	Q/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103571681:T:C	-	27	S/P	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103571744:G:C	rs1258720765	48	A/P	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103571753:G:A	-	51	G/R	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103571754:G:T	-	51	G/V	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103571765:G:T	rs377503366	55	V/F	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572117:C:T	-	59	P/L	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572163:G:T	-	74	W/C	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572173:C:A	rs765904488	78	Q/K	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572186:A:C	-	82	Y/S	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572192:T:G	-	84	L/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103572207:G:A	-	89	G/E	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103572212:G:T	-	91	E/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103572232:G:A	rs1227216211	97	M/I	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573136:G:A	-	130	C/Y	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573180:C:T	-	145	P/S	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573195:G:T	-	150	D/Y	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573215:T:A	-	156	C/*	stop_gained	HIGH	0,0000014	1

AMY2B	1:103573729:G:T	-	179	G/C	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573738:G:A	rs774051583	182	D/N	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573821:C:G	-	209	F/L	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573823:G:C	-	210	R/T	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573826:T:C	rs369605386	211	L/P	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573841:A:T	rs750913844	216	H/L	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573874:AC:A	-	227	D/X	frameshift_variant	HIGH	0,0000014	1
AMY2B	1:103573903:T:C	-	237	F/L	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574310:T:A	rs1459722379	265	N/K	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574328:C:G	-	271	F/L	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574362:A:G	-	283	K/E	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574367:G:C	-	284	W/C	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575227:T:C	-	295	W/R	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575231:G:A	rs544350905	296	G/E	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575239:T:A	rs774845497	299	W/R	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575255:C:T	rs1478745706	304	S/F	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575260:A:T	-	306	R/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103575284:C:T	-	314	H/Y	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575300:G:T	-	319	G/V	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575302:C:T	rs778916272	320	H/Y	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575308:G:A	-	322	A/T	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575333:T:C	-	330	F/S	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575337:G:T	rs776177641	331	W/C	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103575440:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
AMY2B	1:103575475:C:A	-	346	H/N	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577592:C:G	rs377067814	402	R/G	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577771:G:T	-	424	W/C	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577777:T:G	-	426	D/E	missense_variant	pathogenic	0,0000014	1

AMY2B	1:103577779:A:T	-	427	N/I	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577792:A:C	-	431	Q/H	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577807:A:C	-	436	R/S	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577808:G:A	-	437	G/R	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577813:CAG:C	-	439	R/X	frameshift_variant	HIGH	0,0000014	1
AMY2B	1:103577839:A:G	rs765259995	447	D/G	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103579322:T:A	-	453	L/*	stop_gained	HIGH	0,0000014	1
AMY2B	1:103579361:A:G	-	466	D/G	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103579362:TG:T	-	467	V/X	frameshift_variant	HIGH	0,0000014	1
AMY2B	1:103579394:G:A	-	477	C/Y	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103579500:A:T	-	512	*/Y	stop_lost	HIGH	0,0000014	1
AMY2B	1:103573765:C:A	rs375752225	191	R/S	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574347:G:C	-	278	G/R	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103574360:G:C	-	282	R/P	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103577833:A:C	rs370179572	445	N/T	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103573739:A:G	-	182	D/G	missense_variant	pathogenic	0,0000014	1
AMY2B	1:103571669:C:T	-	23	Q/*	stop_gained	HIGH	0,0000027	2
AMY2B	1:103571762:G:A	-	54	G/R	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103572156:C:CT	-	72	P/PX	frameshift_variant	HIGH	0,0000027	2
AMY2B	1:103572186:A:G	-	82	Y/C	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573062:G:A	rs564253866	-	-	splice_acceptor_variant	HIGH	0,0000027	2
AMY2B	1:103573079:A:T	-	111	D/V	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573214:G:A	-	156	C/Y	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573225:A:C	-	160	S/R	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573799:T:A	rs202103095	202	I/N	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573807:G:C	-	205	G/R	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573819:T:C	rs751750690	209	F/L	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103573835:C:A	-	214	S/Y	missense_variant	pathogenic	0,0000027	2



AMY2B	1:103573849:C:T	-	219	P/S	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103574267:A:C	-	251	D/A	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103574285:T:C	rs370085323	257	I/T	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103574394:G:A	rs746033188	-	-	splice_donor_variant	HIGH	0,0000027	2
AMY2B	1:103575242:G:C	-	300	G/R	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103575243:G:T	rs1437564666	300	G/V	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103575297:G:A	rs374864399	318	R/Q	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103575309:C:A	-	322	A/D	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103577503:G:A	rs761960151	372	W/*	stop_gained	HIGH	0,0000027	2
AMY2B	1:103577737:G:T	-	413	R/L	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103579358:G:A	rs1249415838	465	C/Y	missense_variant	pathogenic	0,0000027	2
AMY2B	1:103579446:TA:T	-	495	S/X	frameshift_variant	HIGH	0,0000027	2
AMY2B	1:103573723:CT:C	-	177	L/X	frameshift_variant	HIGH	0,0000027	2
AMY2B	1:103577592:C:T	rs377067814	402	R/*	stop_gained	HIGH	0,0000027	2
AMY2B	1:103571705:C:T	rs761465185	35	R/*	stop_gained	HIGH	0,0000041	3
AMY2B	1:103572117:C:A	-	59	P/Q	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103573102:G:T	-	119	G/C	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103573170:T:G	rs752704778	141	F/L	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103573227:T:G	-	160	S/R	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103574258:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000041	3
AMY2B	1:103574286:T:G	-	257	I/M	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103575508:T:C	-	357	Y/H	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103577736:C:T	rs767168411	413	R/C	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103577771:G:A	-	424	W/*	stop_gained	HIGH	0,0000041	3
AMY2B	1:103579341:TC:T	-	460	P/X	frameshift_variant	HIGH	0,0000041	3
AMY2B	1:103575229:G:T	-	295	W/C	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103573067:G:C	-	107	R/P	missense_variant	pathogenic	0,0000041	3
AMY2B	1:103571654:T:TC	-	18	S/SX	frameshift_variant	HIGH	0,0000055	4

AMY2B	1:103571704:G:A	-	34	W/*	stop_gained	HIGH	0,0000055	4
AMY2B	1:103573178:T:C	-	144	V/A	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103573934:A:G	rs767610231	247	Q/R	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103574311:G:C	rs1212050010	266	G/R	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103575263:G:A	-	307	A/T	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103577602:A:T	-	405	Q/L	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103577603:A:T	-	405	Q/H	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103577733:T:C	rs774018534	412	F/L	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103573121:G:A	-	125	G/E	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103573730:G:T	-	179	G/V	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103573748:T:G	-	185	L/R	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103573822:A:G	-	210	R/G	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103574382:G:A	rs147458656	289	M/I	missense_variant	pathogenic	0,0000055	4
AMY2B	1:103575461:GAT:G	rs1201479792	341-342	GF/GX	frameshift_variant	HIGH	0,0000055	4
AMY2B	1:103571688:T:C	rs746296699	29	V/A	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103572159:G:A	-	73	W/*	stop_gained	HIGH	0,0000068	5
AMY2B	1:103575241:G:T	rs748614240	299	W/C	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103575335:T:G	rs771258177	331	W/G	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103575506:G:A	-	356	S/N	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103579409:T:A	-	482	I/N	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103573094:A:G	rs775566780	116	H/R	missense_variant	pathogenic	0,0000068	5
AMY2B	1:103571692:T:A	rs763349398	30	H/Q	missense_variant	pathogenic	0,0000082	6
AMY2B	1:103571735:C:T	rs770249517	45	R/*	stop_gained	HIGH	0,0000082	6
AMY2B	1:103574259:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000082	6
AMY2B	1:103571706:G:T	-	35	R/L	missense_variant	pathogenic	0,0000096	7
AMY2B	1:103571718:T:C	rs199581606	39	I/T	missense_variant	pathogenic	0,0000096	7
AMY2B	1:103573237:G:A	rs561651261	164	E/K	missense_variant	pathogenic	0,0000096	7
AMY2B	1:103574334:T:G	-	273	Y/*	stop_gained	HIGH	0,0000109	8

AMY2B	1:103574359:C:T	rs368462934	282	R/C	missense_variant	pathogenic	0,0000109	8
AMY2B	1:103573072:T:C	rs753454981	109	Y/H	missense_variant	pathogenic	0,0000109	8
AMY2B	1:103572120:C:T	-	60	P/L	missense_variant	pathogenic	0,0000123	9
AMY2B	1:103575465:T:G	-	342	F/L	missense_variant	pathogenic	0,0000123	9
AMY2B	1:103577589:C:T	rs760832741	401	H/Y	missense_variant	pathogenic	0,0000123	9
AMY2B	1:103573765:C:T	rs375752225	191	R/C	missense_variant	pathogenic	0,0000123	9
AMY2B	1:103573093:C:G	rs746781812	116	H/D	missense_variant	pathogenic	0,0000136	10
AMY2B	1:103575455:C:A	-	339	A/E	missense_variant	pathogenic	0,0000136	10
AMY2B	1:103571750:A:T	rs534314286	50	K/*	stop_gained	HIGH	0,0000177	13
AMY2B	1:103573241:A:T	rs764923580	165	N/I	missense_variant	pathogenic	0,0000177	13
AMY2B	1:103573262:T:G	rs1553196392	-	-	splice_donor_variant	HIGH	0,0000177	12
AMY2B	1:103574266:G:T	rs759532881	251	D/Y	missense_variant	pathogenic	0,0000191	14
AMY2B	1:103575481:T:C	rs761762278	348	Y/H	missense_variant	pathogenic	0,0000191	14
AMY2B	1:103572243:G:A	rs1209830945	101	C/Y	missense_variant	pathogenic	0,0000205	15
AMY2B	1:103575516:G:A	rs757818865	359	W/*	stop_gained	HIGH	0,0000259	19
AMY2B	1:103575541:G:T	rs201379773	-	-	splice_donor_variant	HIGH	0,0000259	19
AMY2B	1:103577598:C:T	rs558190271	404	R/C	missense_variant	pathogenic	0,0000273	20
AMY2B	1:103574324:A:G	rs771344389	270	E/G	missense_variant	pathogenic	0,0000287	21
AMY2B	1:103575296:C:T	rs757065720	318	R/*	stop_gained	HIGH	0,0000287	21
AMY2B	1:103575273:T:C	rs1279419248	310	F/S	missense_variant	pathogenic	0,0000327	24
AMY2B	1:103575493:C:T	rs760449208	352	R/*	stop_gained	HIGH	0,0000396	29
AMY2B	1:103573850:C:T	rs140826288	219	P/L	missense_variant	pathogenic	0,0000409	30
AMY2B	1:103577579:G:A	rs772402281	397	W/*	stop_gained	HIGH	0,0000437	32
AMY2B	1:103577495:TA:T	-	370	N/X	frameshift_variant	HIGH	0,0000450	33
AMY2B	1:103573766:G:A	rs370144544	191	R/H	missense_variant	pathogenic	0,0000450	33
AMY2B	1:103573204:G:T	rs543472077	153	D/Y	missense_variant	pathogenic	0,0000464	34
AMY2B	1:103577593:G:A	rs768084085	402	R/Q	missense_variant	pathogenic	0,0000505	37
AMY2B	1:103577509:G:A	rs750991144	374	G/E	missense_variant	pathogenic	0,0000887	65

AMY2B	1:103575541:G:A	rs201379773	-	-	splice_donor_variant	HIGH	0,0000942	69
AMY2B	1:103574347:G:A	rs764566458	278	G/S	missense_variant	pathogenic	0,0001010	74
AMY2B	1:103574333:A:G	rs150659982	273	Y/C	missense_variant	pathogenic	0,0002197	161
AMY2B	1:103573922:C:A	rs137860562	243	P/H	missense_variant	pathogenic	0,0004776	350
AMY2B	1:103575230:G:A	rs147215616	296	G/R	missense_variant	pathogenic	0,0031330	2291
LCT	2:135788420:A:T	-	1896	C/*	stop_gained	HIGH	0,0000014	1
LCT	2:135788479:T:TG	rs1241153924	1876-1877	-/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135789661:T:C	-	1825	K/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135789700:A:G	-	1812	F/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135789702:T:C	-	1811	H/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135789745:T:C	-	1797	N/D	missense_variant	pathogenic	0,0000014	1
LCT	2:135790817:G:T	-	1726	P/T	missense_variant	pathogenic	0,0000014	1
LCT	2:135794773:A:T	-	1660	L/Q	missense_variant,splice_region_variant	pathogenic	0,0000014	1
LCT	2:135800730:C:CAGAT	-	1581	L/LSX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135800753:C:G	-	1574	A/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135800768:G:T	-	1569	H/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135803945:C:T	rs756645100	1550	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135803982:A:T	-	1537	N/K	missense_variant	pathogenic	0,0000014	1
LCT	2:135803986:A:G	-	1536	L/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135804103:G:A	-	1497	P/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135804792:A:G	-	1480	L/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135804816:T:C	-	1472	Y/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135804817:A:G	-	1472	Y/H	missense_variant	pathogenic	0,0000014	1
LCT	2:135804872:C:G	-	1453	W/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135804950:G:C	rs1212279051	1427	D/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135805017:A:G	-	1405	I/T	missense_variant	pathogenic	0,0000014	1
LCT	2:135805051:C:G	-	1394	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135805054:C:T	-	1393	E/K	missense_variant	pathogenic	0,0000014	1

LCT	2:135807154:A:G	-	1383	W/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135807179:G:C	-	1374	Y/*	stop_gained	HIGH	0,0000014	1
LCT	2:135807303:C:T	-	1333	G/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135807305:A:C	-	1332	F/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135807385:C:T	rs1489231732	1306	D/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135808524:C:T	rs540136979	1275	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135808558:C:A	-	1263	E/D	missense_variant	pathogenic	0,0000014	1
LCT	2:135808595:C:T	-	1251	W/*	stop_gained	HIGH	0,0000014	1
LCT	2:135808602:CA:C	-	1248	A/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135808662:C:T	rs370971797	1229	D/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135808794:G:A	rs780302947	1185	R/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135808822:C:A	-	1175	R/S	missense_variant	pathogenic	0,0000014	1
LCT	2:135808843:C:T	-	1168	M/I	missense_variant	pathogenic	0,0000014	1
LCT	2:135808863:C:T	rs1156834807	1162	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135808864:G:C	-	1161	N/K	missense_variant	pathogenic	0,0000014	1
LCT	2:135808872:A:G	rs768963883	1159	F/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135808916:T:C	-	1144	D/G	missense_variant	pathogenic	0,0000014	1
LCT	2:135808999:C:G	-	1116	Q/H	missense_variant	pathogenic	0,0000014	1
LCT	2:135809007:G:A	-	1114	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135809013:A:G	rs1490497561	1112	Y/H	missense_variant	pathogenic	0,0000014	1
LCT	2:135809075:T:C	-	1091	Y/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135809131:TA:T	rs762583302	1072	L/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135809155:A:T	-	1064	N/K	missense_variant	pathogenic	0,0000014	1
LCT	2:135809249:C:T	-	1033	W/*	stop_gained	HIGH	0,0000014	1
LCT	2:135809254:T:TC	-	1031	G/GX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135809285:C:A	-	1021	W/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135809301:C:G	-	1016	V/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135809349:A:G	-	1000	Y/H	missense_variant	pathogenic	0,0000014	1

LCT	2:135809399:A:C	-	983	I/S	missense_variant	pathogenic	0,0000014	1
LCT	2:135809408:C:T	-	980	W/*	stop_gained	HIGH	0,0000014	1
LCT	2:135809441:A:G	-	969	L/S	missense_variant	pathogenic	0,0000014	1
LCT	2:135809462:T:C	-	962	D/G	missense_variant	pathogenic	0,0000014	1
LCT	2:135809549:T:C	-	933	D/G	missense_variant	pathogenic	0,0000014	1
LCT	2:135809558:C:T	-	930	S/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135809567:T:A	-	927	K/I	missense_variant	pathogenic	0,0000014	1
LCT	2:135809571:C:A	-	926	G/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135809596:C:G	-	917	Q/H	missense_variant	pathogenic	0,0000014	1
LCT	2:135809923:AG:A	rs1310762569	808	P/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135812474:C:G	-	730	W/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135812545:C:T	rs1037426502	707	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135812568:CA:C	-	699	C/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135812605:T:G	rs778010175	687	T/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135812619:C:T	-	682	G/D	missense_variant	pathogenic	0,0000014	1
LCT	2:135812751:T:A	-	638	D/V	missense_variant	pathogenic	0,0000014	1
LCT	2:135812802:T:A	-	621	E/V	missense_variant	pathogenic	0,0000014	1
LCT	2:135812847:G:T	rs1410039851	606	A/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135812868:A:G	-	599	I/T	missense_variant	pathogenic	0,0000014	1
LCT	2:135812874:A:T	-	597	V/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135812913:T:G	-	584	H/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135817340:C:T	rs1295289020	-	-	splice_donor_variant	HIGH	0,0000014	1
LCT	2:135817426:TG:T	-	541	H/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135817437:C:A	-	537	W/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135817677:C:T	-	457	W/*	stop_gained	HIGH	0,0000014	1
LCT	2:135817817:A:T	rs1367677759	411	W/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135817871:C:T	-	393	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135817885:C:A	-	388	W/L	missense_variant	pathogenic	0,0000014	1

LCT	2:135817963:A:AC	-	362	I/SX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135836636:C:CACTT	-	178	V/VSX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135836659:T:G	-	171	T/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135836689:A:G	-	161	S/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135836803:CA:C	-	122	T/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135836811:A:T	-	120	L/H	missense_variant	pathogenic	0,0000014	1
LCT	2:135836844:GT:G	-	109	T/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135788399:G:T	-	1903	Y/*	stop_gained	HIGH	0,0000014	1
LCT	2:135790733:G:A	-	1754	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135790774:T:G	-	1740	Y/S	missense_variant	pathogenic	0,0000014	1
LCT	2:135794725:T:C	-	1676	D/G	missense_variant	pathogenic	0,0000014	1
LCT	2:135804054:CT:C	-	1513	Q/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135804813:T:G	-	1473	Y/S	missense_variant	pathogenic	0,0000014	1
LCT	2:135804840:T:TA	-	1464	Y/LX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135804867:C:A	-	1455	R/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135807343:C:T	-	1320	D/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135808704:G:A	-	1215	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135808800:TG:T	-	1182	A/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135808819:A:C	-	1176	S/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135808939:G:GA	rs754495130	1136	V/VX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135808961:GC:G	-	1129	A/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135809076:A:T	-	1091	Y/N	missense_variant	pathogenic	0,0000014	1
LCT	2:135809199:A:G	-	1050	F/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135809279:A:T	-	1023	L/Q	missense_variant	pathogenic	0,0000014	1
LCT	2:135812749:C:A	-	639	G/*	stop_gained	HIGH	0,0000014	1
LCT	2:135817933:G:GCCCT	-	372	A/EGX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135818012:G:A	-	346	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135836707:CAT:C	-	154	Y/X	frameshift_variant	HIGH	0,0000014	1

LCT	2:135836887:G:A	-	95	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135837155:C:T	-	5	W/*	stop_gained	HIGH	0,0000014	1
LCT	2:135789717:T:C	-	1806	E/G	missense_variant	pathogenic	0,0000014	1
LCT	2:135794711:T:A	-	1681	N/Y	missense_variant	pathogenic	0,0000014	1
LCT	2:135794777:T:C	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
LCT	2:135803930:C:T	-	1555	G/R	missense_variant,splice_region_variant	pathogenic	0,0000014	1
LCT	2:135809015:TTCTCA:T	rs1198586467	1109-1111	DEK/EX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135809951:C:G	-	799	R/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135812632:C:G	-	678	A/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135812956:C:A	-	570	V/L	missense_variant,splice_region_variant	pathogenic	0,0000014	1
LCT	2:135788356:G:A	rs775774032	1918	Q/*	stop_gained	HIGH	0,0000014	1
LCT	2:135788427:C:T	-	1894	G/E	missense_variant	pathogenic	0,0000014	1
LCT	2:135798099:C:T	-	1636	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135804114:G:T	-	1493	H/Q	missense_variant	pathogenic	0,0000014	1
LCT	2:135804850:T:G	-	1461	T/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135805049:AC:A	-	1394	G/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135836977:GACAA:G	-	63-64	VC/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135788365:G:GT	-	1914-1915	-/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135833167:G:A	rs147426095	222	R/*	stop_gained	HIGH	0,0000014	1
LCT	2:135836605:CT:C	-	188	R/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135805047:G:A	-	1395	A/V	missense_variant	pathogenic	0,0000014	1
LCT	2:135807341:GT:G	-	1320	D/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135812323:C:T	-	781	E/K	missense_variant	pathogenic	0,0000014	1
LCT	2:135790692:G:T	-	1767	Y/*	stop_gained	HIGH	0,0000014	1
LCT	2:135794750:TCTCA:T	rs386833836	1666-1667	SE/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135808460:A:G	-	1296	I/T	missense_variant	pathogenic	0,0000014	1
LCT	2:135798092:T:C	-	1638	Y/C	missense_variant	pathogenic	0,0000014	1
LCT	2:135812313:T:A	-	784	K/M	missense_variant,splice_region_variant	pathogenic	0,0000014	1



LCT	2:135823961:GT:G	-	282	K/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135798106:G:C	-	1633	F/L	missense_variant	pathogenic	0,0000014	1
LCT	2:135833192:T:C	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
LCT	2:135836586:AGG:A	-	194-195	TL/TX	frameshift_variant	HIGH	0,0000014	1
LCT	2:135794717:C:G	-	1679	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135800606:C:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
LCT	2:135822069:C:G	-	313	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135822069:C:T	-	313	G/R	missense_variant	pathogenic	0,0000014	1
LCT	2:135822069:CA:C	-	312	I/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135829652:CAA:C	-	248	L/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135804999:T:G	-	1411	H/P	missense_variant	pathogenic	0,0000014	1
LCT	2:135804069:AT:A	-	1508	N/X	frameshift_variant	HIGH	0,0000014	1
LCT	2:135789747:TC:T	-	1796	D/X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135805008:GT:G	-	1408	T/X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135808661:T:G	rs1211733539	1229	D/A	missense_variant	pathogenic	0,0000027	2
LCT	2:135809436:C:A	-	971	V/L	missense_variant	pathogenic	0,0000027	2
LCT	2:135809551:C:G	-	932	W/C	missense_variant	pathogenic	0,0000027	2
LCT	2:135809551:C:T	-	932	W/*	stop_gained	HIGH	0,0000027	2
LCT	2:135809864:G:C	-	828	S/*	stop_gained	HIGH	0,0000027	2
LCT	2:135812667:A:G	-	666	F/S	missense_variant	pathogenic	0,0000027	2
LCT	2:135812788:A:G	-	626	F/L	missense_variant	pathogenic	0,0000027	2
LCT	2:135817482:A:C	rs1200034961	522	Y/*	stop_gained	HIGH	0,0000027	2
LCT	2:135817484:A:G	-	522	Y/H	missense_variant	pathogenic	0,0000027	2
LCT	2:135836674:C:A	-	166	V/F	missense_variant	pathogenic	0,0000027	2
LCT	2:135836686:A:G	rs755623198	162	F/L	missense_variant	pathogenic	0,0000027	2
LCT	2:135836908:T:C	-	88	K/E	missense_variant	pathogenic	0,0000027	2
LCT	2:135837142:T:TA	rs1408682771	9-10	-/X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135800750:G:A	rs749948532	1575	H/Y	missense_variant	pathogenic	0,0000027	2

LCT	2:135803957:C:A	-	1546	G/C	missense_variant	pathogenic	0,0000027	2
LCT	2:135807239:G:T	-	1354	Y/*	stop_gained	HIGH	0,0000027	2
LCT	2:135808713:T:A	-	1212	R/*	stop_gained	HIGH	0,0000027	2
LCT	2:135808965:A:G	-	1128	W/R	missense_variant	pathogenic	0,0000027	2
LCT	2:135809084:C:T	-	1088	W/*	stop_gained	HIGH	0,0000027	2
LCT	2:135809588:C:A	rs758731636	920	G/V	missense_variant	pathogenic	0,0000027	2
LCT	2:135809603:G:A	-	915	A/V	missense_variant	pathogenic	0,0000027	2
LCT	2:135812513:C:T	rs770253133	717	W/*	stop_gained	HIGH	0,0000027	2
LCT	2:135812842:G:T	-	608	P/T	missense_variant	pathogenic	0,0000027	2
LCT	2:135817535:G:A	rs1558741337	505	Q/*	stop_gained	HIGH	0,0000027	2
LCT	2:135837167:C:A	-	1	M/I	start_lost	HIGH	0,0000027	2
LCT	2:135789720:GA:G	-	1805	S/X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135807396:G:T	-	1302	A/D	missense_variant,splice_region_variant	pathogenic	0,0000027	2
LCT	2:135817620:G:C	-	476	Y/*	stop_gained	HIGH	0,0000027	2
LCT	2:135804041:A:G	-	1518	Y/H	missense_variant	pathogenic	0,0000027	2
LCT	2:135794754:ACT:A	rs1398217038	1666	S/X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135829635:G:T	-	254	C/*	stop_gained	HIGH	0,0000027	2
LCT	2:135788331:G:GA	-	1926	S/FX	frameshift_variant	HIGH	0,0000027	2
LCT	2:135836565:GCTTTTCTGTGGGC:G	-	198-202	AHRKA /X	frameshift_variant	HIGH	0,0000027	2
LCT	2:135808582:C:G	-	1255	R/S	missense_variant	pathogenic	0,0000041	3
LCT	2:135809679:G:A	rs1337972637	890	Q/*	stop_gained	HIGH	0,0000041	3
LCT	2:135812597:GC:G	-	689	R/X	frameshift_variant	HIGH	0,0000041	3
LCT	2:135812601:GA:G	-	688	S/X	frameshift_variant	HIGH	0,0000041	3
LCT	2:135812616:A:T	-	683	L/Q	missense_variant	pathogenic	0,0000041	3
LCT	2:135817393:C:T	-	552	G/D	missense_variant	pathogenic	0,0000041	3
LCT	2:135836780:G:GATGA	rs1417230303	130	I/IHX	frameshift_variant	HIGH	0,0000041	3
LCT	2:135836811:AG:A	-	120	L/X	frameshift_variant	HIGH	0,0000041	3
LCT	2:135788391:C:CA	-	1906	C/LX	frameshift_variant	HIGH	0,0000041	3

LCT	2:135800752:G:A	-	1574	A/V	missense_variant	pathogenic	0,0000041	3
LCT	2:135808825:G:T	-	1174	N/K	missense_variant	pathogenic	0,0000041	3
LCT	2:135808972:A:T	-	1125	S/R	missense_variant	pathogenic	0,0000041	3
LCT	2:135809368:G:T	-	993	N/K	missense_variant	pathogenic	0,0000041	3
LCT	2:135812783:C:T	-	627	M/I	missense_variant	pathogenic	0,0000041	3
LCT	2:135836787:A:C	-	128	M/R	missense_variant	pathogenic	0,0000041	3
LCT	2:135836827:G:A	rs980175497	115	R/*	stop_gained	HIGH	0,0000041	3
LCT	2:135788362:G:A	rs369141275	1916	R/*	stop_gained	HIGH	0,0000041	3
LCT	2:135805026:C:T	-	1402	G/E	missense_variant	pathogenic	0,0000041	3
LCT	2:135809430:C:G	rs749021292	973	A/P	missense_variant	pathogenic	0,0000041	3
LCT	2:135812765:G:C	rs1227458387	633	H/Q	missense_variant	pathogenic	0,0000041	3
LCT	2:135790755:A:ATTT	-	1746	Y/*N	stop_gained	HIGH	0,0000041	3
LCT	2:135808453:C:G	-	1298	E/D	missense_variant	pathogenic	0,0000041	3
LCT	2:135798058:C:A	rs759920127	1649	R/S	missense_variant	pathogenic	0,0000041	3
LCT	2:135829592:C:T	-	-	-	splice_donor_variant	HIGH	0,0000041	3
LCT	2:135805027:C:T	-	1402	G/R	missense_variant	pathogenic	0,0000055	4
LCT	2:135808494:C:A	-	1285	D/Y	missense_variant	pathogenic	0,0000055	4
LCT	2:135809402:C:T	-	982	R/Q	missense_variant	pathogenic	0,0000055	4
LCT	2:135809581:C:T	-	922	W/*	stop_gained	HIGH	0,0000055	4
LCT	2:135809903:A:C	rs1207742583	815	F/C	missense_variant	pathogenic	0,0000055	4
LCT	2:135812475:C:T	-	730	W/*	stop_gained	HIGH	0,0000055	4
LCT	2:135812851:A:C	-	605	W/G	missense_variant	pathogenic	0,0000055	4
LCT	2:135817435:A:T	rs1335546413	538	V/E	missense_variant	pathogenic	0,0000055	4
LCT	2:135817473:G:T	rs373372396	525	F/L	missense_variant	pathogenic	0,0000055	4
LCT	2:135817845:C:G	rs1335321424	401	W/C	missense_variant	pathogenic	0,0000055	4
LCT	2:135817855:T:TC	-	398	E/GX	frameshift_variant	HIGH	0,0000055	4
LCT	2:135836716:C:G	rs114525655	152	A/P	missense_variant	pathogenic	0,0000055	4
LCT	2:135789741:A:G	rs201143582	1798	F/S	missense_variant	pathogenic	0,0000055	4

LCT	2:135804867:C:T	rs867715897	1455	R/H	missense_variant	pathogenic	0,0000055	4
LCT	2:135812404:C:T	rs1558739417	754	G/R	missense_variant	pathogenic	0,0000055	4
LCT	2:135809279:A:G	rs775728891	1023	L/P	missense_variant	pathogenic	0,0000055	4
LCT	2:135794748:C:G	-	1668	K/N	missense_variant	pathogenic	0,0000055	4
LCT	2:135790657:C:G	-	-	-	splice_donor_variant	HIGH	0,0000055	4
LCT	2:135800692:A:G	-	1594	I/T	missense_variant	pathogenic	0,0000068	5
LCT	2:135807129:T:G	rs771753468	1391	Q/P	missense_variant,splice_region_variant	pathogenic	0,0000068	5
LCT	2:135807369:C:G	rs185632404	1311	R/P	missense_variant	pathogenic	0,0000068	5
LCT	2:135809448:G:A	rs930628297	967	R/*	stop_gained	HIGH	0,0000068	5
LCT	2:135807131:A:T	rs121908936	1390	Y/*	stop_gained	HIGH	0,0000068	5
LCT	2:135809688:A:G	rs972582653	887	F/L	missense_variant	pathogenic	0,0000068	5
LCT	2:135803989:G:A	rs747969940	1535	T/M	missense_variant	pathogenic	0,0000068	5
LCT	2:135804889:G:A	rs781290297	1448	R/C	missense_variant	pathogenic	0,0000068	5
LCT	2:135809205:AG:A	-	1047	D/X	frameshift_variant	HIGH	0,0000068	5
LCT	2:135809988:T:A	rs962540601	787	K/*	stop_gained	HIGH	0,0000068	5
LCT	2:135809250:A:G	rs1201370072	1033	W/R	missense_variant	pathogenic	0,0000082	6
LCT	2:135809268:G:A	rs772255416	1027	L/F	missense_variant	pathogenic	0,0000082	6
LCT	2:135809492:G:A	rs866304022	952	A/V	missense_variant	pathogenic	0,0000082	6
LCT	2:135812748:C:A	-	639	G/V	missense_variant	pathogenic	0,0000082	6
LCT	2:135809373:TA:T	-	991	S/X	frameshift_variant	HIGH	0,0000082	6
LCT	2:135808570:C:A	rs867783072	1259	W/C	missense_variant	pathogenic	0,0000082	6
LCT	2:135809921:GA:G	-	809	S/X	frameshift_variant	HIGH	0,0000082	6
LCT	2:135836954:CAGGAA:C	-	71-72	FL/X	frameshift_variant	HIGH	0,0000082	6
LCT	2:135836957:G:T	-	71	F/L	missense_variant	pathogenic	0,0000082	6
LCT	2:135804025:A:C	-	1523	F/C	missense_variant	pathogenic	0,0000082	6
LCT	2:135829667:C:A	-	244	D/Y	missense_variant	pathogenic	0,0000082	6
LCT	2:135790691:A:G	rs201748922	1768	Y/H	missense_variant	pathogenic	0,0000082	6
LCT	2:135808914:G:A	rs765765945	1145	R/*	stop_gained	HIGH	0,0000096	7

LCT	2:135833191:C:T	-	-	-	splice_acceptor_variant	HIGH	0,0000096	7
LCT	2:135808587:G:A	rs761309644	1254	R/*	stop_gained	HIGH	0,0000109	8
LCT	2:135809635:CCGAAA:C	rs756488468	903-904	FR/X	frameshift_variant	HIGH	0,0000109	8
LCT	2:135812372:A:AT	-	764	N/KX	frameshift_variant	HIGH	0,0000109	8
LCT	2:135836976:TG:T	-	65	H/X	frameshift_variant	HIGH	0,0000109	8
LCT	2:135788460:GCTGT:G	rs1397567983	1882-1883	TA/X	frameshift_variant	HIGH	0,0000109	8
LCT	2:135812613:G:A	rs1457912116	684	S/L	missense_variant	pathogenic	0,0000123	9
LCT	2:135817480:G:A	rs769594848	523	A/V	missense_variant	pathogenic	0,0000136	10
LCT	2:135817460:A:G	-	530	F/L	missense_variant	pathogenic	0,0000136	10
LCT	2:135798028:CCGAGACTTGT:C	-	1656-1659	NKSR/X	frameshift_variant,splice_region_variant	HIGH	0,0000151	11
LCT	2:135817420:G:A	rs1412074020	543	P/L	missense_variant	pathogenic	0,0000164	12
LCT	2:135789775:G:A	rs548104619	1787	R/*	stop_gained	HIGH	0,0000164	12
LCT	2:135808985:G:A	rs1268587670	1121	S/L	missense_variant	pathogenic	0,0000164	12
LCT	2:135836934:A:G	rs1483709990	79	L/P	missense_variant	pathogenic	0,0000177	13
LCT	2:135809646:C:T	rs776158841	901	G/R	missense_variant	pathogenic	0,0000177	13
LCT	2:135789763:CTG:C	-	1790	T/X	frameshift_variant	HIGH	0,0000218	16
LCT	2:135804888:C:T	-	1448	R/H	missense_variant	pathogenic	0,0000232	17
LCT	2:135808553:C:A	rs1469063105	1265	G/V	missense_variant	pathogenic	0,0000396	29
MGAM	7:142008679:T:C	-	101	C/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142021716:GA:G	rs1396602292	230	R/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142022270:T:C	-	238	F/S	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142022284:G:A	-	243	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142022294:T:C	rs374712168	246	L/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142022356:G:A	-	267	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142022359:G:A	rs1223926013	268	E/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142022359:G:C	-	268	E/Q	missense_variant	pathogenic	0,0000014	1
MGAM	7:142022413:T:C	-	286	F/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142025075:A:G	-	303	Q/R	missense_variant	pathogenic	0,0000014	1

MGAM	7:142027171:G:A	rs1554461237	347	D/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142027171:G:T	-	347	D/Y	missense_variant	pathogenic	0,0000014	1
MGAM	7:142027172:A:G	-	347	D/G	missense_variant	pathogenic	0,0000014	1
MGAM	7:142027692:T:C	rs782811856	393	M/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142030390:T:C	-	417	M/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142030399:G:T	-	420	R/I	missense_variant	pathogenic	0,0000014	1
MGAM	7:142030446:T:C	-	436	F/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034298:G:A	rs782729423	569	C/Y	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034304:A:G	rs1554464957	571	D/G	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034723:C:T	-	614	T/I	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034756:G:A	rs1554465201	625	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142034797:T:C	-	639	S/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036228:G:A	-	673	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142036233:A:T	-	675	Q/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036239:G:T	-	677	G/V	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036832:C:T	rs1554466078	696	P/S	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036926:T:C	-	727	F/S	missense_variant	pathogenic	0,0000014	1
MGAM	7:142040720:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142040728:C:CA	-	794	Q/QX	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142040807:T:C	-	820	I/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142050721:C:T	-	888	Q/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142050821:C:CA	-	921	P/PX	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142052350:G:C	-	954	W/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142052390:C:T	-	968	P/S	missense_variant	pathogenic	0,0000014	1
MGAM	7:142052391:C:G	-	968	P/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142052950:T:C	-	1042	V/A	missense_variant	pathogenic	0,0000014	1
MGAM	7:142054761:A:G	-	1056	D/G	missense_variant	pathogenic	0,0000014	1
MGAM	7:142054776:G:T	-	1061	R/L	missense_variant	pathogenic	0,0000014	1

MGAM	7:142054779:A:G	-	1062	Y/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142054797:T:C	-	1068	L/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142055565:T:C	-	1108	S/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142055602:TC:T	-	1120	I/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142055646:G:A	rs1299819756	1135	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142055727:G:A	rs866692167	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142056024:CA:C	-	1170	H/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142056039:G:A	rs1487334316	1175	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056070:T:C	-	1185	V/A	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056842:A:C	-	1198	Q/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056859:A:G	-	1204	T/A	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056875:G:A	-	1209	G/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056881:T:A	-	1211	V/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142056936:C:A	-	1229	Y/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142059473:A:T	-	1274	D/V	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142059478:C:CAG	-	1276	Q/QX	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142059488:A:T	-	1279	D/V	missense_variant	pathogenic	0,0000014	1
MGAM	7:142059499:A:G	rs759340776	1283	M/V	missense_variant	pathogenic	0,0000014	1
MGAM	7:142059514:G:T	-	1288	D/Y	missense_variant	pathogenic	0,0000014	1
MGAM	7:142059857:A:G	rs1247460301	1317	D/G	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142059898:T:C	-	1331	F/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142059962:G:A	rs1340523455	1352	G/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142059966:G:T	rs368053105	1353	K/N	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142060313:CT:C	-	1355	W/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142060314:T:C	rs374999893	1355	W/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142060324:T:G	-	1358	F/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142062595:G:A	-	1384	D/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142062623:G:A	-	1393	W/*	stop_gained	HIGH	0,0000014	1

MGAM	7:142062627:G:A	rs747366964	1394	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142062627:G:T	-	1394	W/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142062699:G:A	-	1418	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142062699:G:T	rs368030090	1418	W/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142063508:G:A	-	1423	E/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142063521:T:G	-	1427	F/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142063584:C:T	-	1448	P/L	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142064408:T:C	-	1457	L/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064420:C:A	-	1461	T/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064454:C:G	rs751737247	1472	D/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064456:GC:G	rs756866285	1473	G/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142064458:T:C	rs751562541	1474	S/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064472:C:G	rs371707186	1478	H/Q	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064478:C:G	-	1480	N/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064491:T:A	-	1485	Y/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142064492:A:G	rs367916209	1485	Y/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065421:T:C	rs1192414161	1524	L/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065431:C:G	-	1527	N/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065443:G:T	-	1531	W/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065448:A:T	-	1533	Q/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065458:G:T	rs1248970291	1536	K/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065468:G:T	-	1540	G/C	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142065470:T:C	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142065611:G:T	rs754493483	1548	G/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065612:G:A	-	1548	G/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065615:T:C	-	1549	I/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065621:A:G	rs141258770	1551	Y/C	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142066670:T:G	-	1623	M/R	missense_variant	pathogenic	0,0000014	1



MGAM	7:142066699:G:T	-	1633	V/F	missense_variant	pathogenic	0,0000014	1
MGAM	7:142093495:C:T	-	2373	Q/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142094761:CTG:C	-	2453	C/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142095592:GC:G	-	2496	A/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142096416:G:A	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142099611:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142099707:G:A	-	2615	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142103386:TC:T	-	2711	S/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142027674:A:G	-	387	Y/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142031752:G:T	-	515	E/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142032871:G:GT	-	544	C/CX	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142034301:T:C	-	570	M/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034327:C:A	rs368127800	579	Q/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034352:G:C	-	587	G/A	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034671:G:C	-	597	A/P	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142036827:A:T	-	694	Q/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036912:C:A	-	722	Y/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142036916:T:C	-	724	Y/H	missense_variant	pathogenic	0,0000014	1
MGAM	7:142038577:T:C	rs765827662	760	W/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142040805:C:A	-	819	Y/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142050233:A:G	rs746819971	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142058234:G:T	-	1242	W/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142058261:A:C	-	1251	Y/S	missense_variant	pathogenic	0,0000014	1
MGAM	7:142058264:G:A	rs1160691795	1252	G/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142058274:T:G	rs2961078	1255	N/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142060315:G:T	-	1355	W/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142060374:G:T	rs184909179	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142063499:G:A	rs201271635	1420	D/N	missense_variant,splice_region_variant	pathogenic	0,0000014	1

MGAM	7:142064494:G:A	-	1486	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142066660:T:C	-	1620	Y/H	missense_variant	pathogenic	0,0000014	1
MGAM	7:142066691:G:A	rs765508348	1630	G/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142071069:G:T	-	1713	G/W	missense_variant	pathogenic	0,0000014	1
MGAM	7:142071079:T:C	-	1716	I/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142083335:C:CCGTA	-	2101-2102	-/RX	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142095714:G:A	rs771164603	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142102628:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142102629:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142102646:ACT:A	rs1563231709	2661	L/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142036266:A:G	rs782009801	686	H/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142036892:C:G	rs782806699	716	R/G	missense_variant	pathogenic	0,0000014	1
MGAM	7:142050266:TG:T	-	874	E/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142052297:G:C	-	937	A/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142052986:T:C	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142056944:T:C	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142056944:T:G	rs776108491	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142067409:G:A	-	1663	S/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034355:A:G	-	588	Y/C	missense_variant	pathogenic	0,0000014	1
MGAM	7:142047814:C:A	-	843	A/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142066722:G:A	rs774200745	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142078807:G:C	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142080864:CT:C	rs1439523668	1974	P/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142097591:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142021687:CA:C	-	221	R/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142031704:C:G	rs782494225	499	P/A	missense_variant	pathogenic	0,0000014	1
MGAM	7:142047834:G:A	rs767376034	850	A/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142067426:G:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1

MGAM	7:142086322:G:A	-	2247	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142030671:AG:A	-	462	S/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142047855:G:A	rs753403339	857	D/N	missense_variant	pathogenic	0,0000014	1
MGAM	7:142052984:G:C	-	1053	K/N	missense_variant,splice_region_variant	pathogenic	0,0000014	1
MGAM	7:142025069:G:A	-	301	G/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142040155:G:A	-	786	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142047793:AC:A	-	836	N/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142094655:G:A	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142076827:G:A	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142083415:T:C	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142074149:T:C	-	1751	F/L	missense_variant	pathogenic	0,0000014	1
MGAM	7:142082516:GC:G	-	2072	L/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142036206:CT:C	-	666	P/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142086329:G:A	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142036270:T:A	-	687	N/K	missense_variant	pathogenic	0,0000014	1
MGAM	7:142030644:C:A	rs971296318	453	P/T	missense_variant	pathogenic	0,0000014	1
MGAM	7:142034364:C:A	-	591	A/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065811:C:T	-	1584	H/Y	missense_variant	pathogenic	0,0000014	1
MGAM	7:142021069:C:G	-	182	R/G	missense_variant	pathogenic	0,0000014	1
MGAM	7:142030640:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142074157:T:A	-	1753	D/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142047783:A:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142094496:TG:T	-	2436	G/X	frameshift_variant,splice_region_variant	HIGH	0,0000014	1
MGAM	7:142040142:C:T	-	782	P/S	missense_variant	pathogenic	0,0000014	1
MGAM	7:142025140:A:G	rs782718423	325	N/D	missense_variant	pathogenic	0,0000014	1
MGAM	7:142074161:G:A	rs758303816	1755	G/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142065779:A:C	-	1573	Q/P	missense_variant	pathogenic	0,0000014	1
MGAM	7:142008573:TG:T	-	66	G/X	frameshift_variant	HIGH	0,0000014	1

MGAM	7:142020980:CA:C	-	152	T/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142005531:A:G	rs1554452041	1	M/V	start_lost,splice_region_variant	HIGH	0,0000014	1
MGAM	7:142082500:AC:A	-	2066	H/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142030758:G:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142105886:CTG:C	rs751944457	2753	L/X	frameshift_variant	HIGH	0,0000014	1
MGAM	7:142074084:G:A	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
MGAM	7:142047821:T:A	-	845	D/E	missense_variant	pathogenic	0,0000014	1
MGAM	7:142084550:G:A	-	2138	W/*	stop_gained	HIGH	0,0000014	1
MGAM	7:142034380:G:A	rs782741238	-	-	splice_donor_variant	HIGH	0,0000014	1
MGAM	7:142034789:T:G	rs1563135360	636	L/R	missense_variant	pathogenic	0,0000014	1
MGAM	7:142008646:T:A	rs1358768865	90	C/S	missense_variant	pathogenic	0,0000027	2
MGAM	7:142022326:C:T	rs781972778	257	R/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142027160:G:C	-	343	G/A	missense_variant	pathogenic	0,0000027	2
MGAM	7:142027169:T:C	-	346	L/P	missense_variant	pathogenic	0,0000027	2
MGAM	7:142027640:T:A	rs1554461435	376	W/R	missense_variant	pathogenic	0,0000027	2
MGAM	7:142027641:G:A	-	376	W/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142034331:A:G	-	580	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142034780:G:A	-	633	W/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142036248:A:G	rs367841743	680	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142052384:T:A	rs1186847866	966	C/S	missense_variant	pathogenic	0,0000027	2
MGAM	7:142052419:CT:C	rs1001916370	978	C/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142052420:T:C	rs1394181284	978	C/R	missense_variant	pathogenic	0,0000027	2
MGAM	7:142054869:T:A	-	1092	F/Y	missense_variant	pathogenic	0,0000027	2
MGAM	7:142055590:G:T	rs3024261	1116	S/I	missense_variant	pathogenic	0,0000027	2
MGAM	7:142055620:T:C	-	1126	L/P	missense_variant	pathogenic	0,0000027	2
MGAM	7:142055667:T:C	-	1142	Y/H	missense_variant	pathogenic	0,0000027	2
MGAM	7:142055677:AC:A	-	1145	D/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142055701:T:C	-	1153	M/T	missense_variant	pathogenic	0,0000027	2

MGAM	7:142055702:G:A	rs575188237	1153	M/I	missense_variant	pathogenic	0,0000027	2
MGAM	7:142055728:T:C	-	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142056864:C:A	-	1205	Y/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142059472:G:A	-	1274	D/N	missense_variant,splice_region_variant	pathogenic	0,0000027	2
MGAM	7:142059475:G:A	rs1255039773	1275	V/M	missense_variant	pathogenic	0,0000027	2
MGAM	7:142059907:G:A	rs1212270440	1334	G/S	missense_variant	pathogenic	0,0000027	2
MGAM	7:142064471:A:G	-	1478	H/R	missense_variant	pathogenic	0,0000027	2
MGAM	7:142065442:G:A	-	1531	W/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142096416:G:C	-	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142099664:GA:G	-	2601	D/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142031735:G:C	-	509	W/S	missense_variant	pathogenic	0,0000027	2
MGAM	7:142034688:C:G	rs1554465123	602	F/L	missense_variant	pathogenic	0,0000027	2
MGAM	7:142056016:A:G	rs745962488	1167	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142056866:G:T	rs780595542	1206	R/L	missense_variant	pathogenic	0,0000027	2
MGAM	7:142059922:G:T	-	1339	V/F	missense_variant	pathogenic	0,0000027	2
MGAM	7:142062614:C:T	rs747625145	1390	T/I	missense_variant	pathogenic	0,0000027	2
MGAM	7:142063500:A:C	-	1420	D/A	missense_variant,splice_region_variant	pathogenic	0,0000027	2
MGAM	7:142066715:T:C	-	1638	L/P	missense_variant	pathogenic	0,0000027	1
MGAM	7:142071109:C:T	-	1726	T/I	missense_variant	pathogenic	0,0000027	2
MGAM	7:142082193:C:T	rs529863811	2052	R/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142034765:A:G	rs539459227	628	D/G	missense_variant	pathogenic	0,0000027	2
MGAM	7:142058257:C:T	rs377213841	1250	R/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142058305:A:T	rs559779417	1266	M/L	missense_variant	pathogenic	0,0000027	2
MGAM	7:142059506:G:A	rs759425386	1285	R/Q	missense_variant	pathogenic	0,0000027	2
MGAM	7:142034349:A:G	rs1554464981	586	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142082141:GCA:G	rs1364694211	2035	H/X	frameshift_variant	HIGH	0,0000027	1
MGAM	7:142080790:G:T	-	-	-	splice_acceptor_variant	HIGH	0,0000027	1
MGAM	7:142095563:A:G	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2

MGAM	7:142031714:C:T	-	502	T/I	missense_variant	pathogenic	0,0000027	2
MGAM	7:142036896:A:G	rs782519815	717	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142052985:G:A	-	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142093548:C:A	-	2390	Y/*	stop_gained,splice_region_variant	HIGH	0,0000027	1
MGAM	7:142030653:T:TC	-	456	S/SX	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142036274:C:T	rs1554465805	689	Q/*	stop_gained	HIGH	0,0000027	2
MGAM	7:142030694:CA:C	-	470	R/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142094386:C:T	-	2399	Q/*	stop_gained	HIGH	0,0000027	1
MGAM	7:142076827:G:T	-	-	-	splice_donor_variant	HIGH	0,0000027	1
MGAM	7:142025048:A:T	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2
MGAM	7:142031681:T:C	-	491	V/A	missense_variant,splice_region_variant	pathogenic	0,0000027	2
MGAM	7:142065832:G:A	rs1563188084	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142065726:T:A	-	1555	D/E	missense_variant	pathogenic	0,0000027	2
MGAM	7:142032907:C:A	rs1554464350	556	P/H	missense_variant,splice_region_variant	pathogenic	0,0000027	2
MGAM	7:142005596:GTTGT:G	rs781922105	23-24	LF/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142020982:GC:G	-	153	A/X	frameshift_variant	HIGH	0,0000027	2
MGAM	7:142025150:G:A	rs1340856858	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142021084:G:A	rs138090433	-	-	splice_donor_variant	HIGH	0,0000027	2
MGAM	7:142027220:A:G	rs781905880	363	Y/C	missense_variant	pathogenic	0,0000027	2
MGAM	7:142074083:A:C	-	-	-	splice_acceptor_variant	HIGH	0,0000027	1
MGAM	7:142034789:T:C	-	636	L/P	missense_variant	pathogenic	0,0000027	2
MGAM	7:142008636:TTC:T	-	87	S/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142022291:T:C	-	245	L/P	missense_variant	pathogenic	0,0000041	3
MGAM	7:142022432:C:G	-	292	P/R	missense_variant	pathogenic	0,0000041	3
MGAM	7:142052944:TG:T	-	1040	L/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142055641:G:A	-	1133	G/D	missense_variant	pathogenic	0,0000041	3
MGAM	7:142055645:T:A	-	1134	F/L	missense_variant	pathogenic	0,0000041	3
MGAM	7:142055686:G:T	rs1261220607	1148	W/L	missense_variant	pathogenic	0,0000041	3

MGAM	7:142055721:C:G	-	1160	P/A	missense_variant	pathogenic	0,0000041	3
MGAM	7:142056860:C:T	-	1204	T/I	missense_variant	pathogenic	0,0000041	3
MGAM	7:142056875:G:T	-	1209	G/V	missense_variant	pathogenic	0,0000041	3
MGAM	7:142059590:T:C	-	1313	I/T	missense_variant	pathogenic	0,0000041	3
MGAM	7:142062626:G:A	rs778209488	1394	W/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142063519:C:A	-	1426	S/R	missense_variant	pathogenic	0,0000041	3
MGAM	7:142063571:C:T	rs779261743	1444	P/S	missense_variant	pathogenic	0,0000041	3
MGAM	7:142065340:T:A	-	1497	V/E	missense_variant	pathogenic	0,0000041	3
MGAM	7:142065588:G:A	-	1540	G/D	missense_variant,splice_region_variant	pathogenic	0,0000041	3
MGAM	7:142099613:GGT:G	-	2584	G/X	frameshift_variant,splice_region_variant	HIGH	0,0000041	3
MGAM	7:142027649:G:T	-	379	G/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142036962:G:A	-	739	R/K	missense_variant	pathogenic	0,0000041	3
MGAM	7:142082179:G:A	rs745514815	2047	W/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142052442:GGGAGGTAACCA:G	rs763211623	985-?	-	splice_donor_variant,splice_donor_5th_base_variant,co ding_sequence_variant,intron_variant	HIGH	0,0000041	3
MGAM	7:142056027:C:A	rs1386283834	1171	P/T	missense_variant	pathogenic	0,0000041	3
MGAM	7:142093428:CA:C	-	2351	R/X	frameshift_variant	HIGH	0,0000041	2
MGAM	7:142036202:AC:A	rs1297696796	665	T/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142058247:C:A	-	1246	F/L	missense_variant	pathogenic	0,0000041	3
MGAM	7:142093486:TC:T	rs771156854	2370	S/X	frameshift_variant	HIGH	0,0000041	2
MGAM	7:142031711:A:G	rs782615480	501	Y/C	missense_variant	pathogenic	0,0000041	3
MGAM	7:142052982:A:G	-	1053	K/E	missense_variant,splice_region_variant	pathogenic	0,0000041	3
MGAM	7:142094363:G:A	rs1370453005	-	-	splice_acceptor_variant	HIGH	0,0000041	3
MGAM	7:142036892:C:T	rs782806699	716	R/C	missense_variant	pathogenic	0,0000041	3
MGAM	7:142019248:G:GT	rs781829209	126	S/SX	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142030698:G:A	rs377157038	471	G/S	missense_variant	pathogenic	0,0000041	3
MGAM	7:142062575:G:C	-	1377	R/P	missense_variant	pathogenic	0,0000041	3
MGAM	7:142083387:GA:G	-	2119	E/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142031747:A:G	rs1249964085	513	E/G	missense_variant	pathogenic	0,0000041	3

MGAM	7:142105869:T:A	-	2747	L/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142036188:G:T	-	660	G/V	missense_variant	pathogenic	0,0000041	3
MGAM	7:142079000:C:T	-	1947	Q/*	stop_gained	HIGH	0,0000041	2
MGAM	7:142005627:CT:C	-	33	L/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142021079:T:C	-	185	F/S	missense_variant	pathogenic	0,0000041	3
MGAM	7:142056002:C:G	-	1162	Y/*	stop_gained,splice_region_variant	HIGH	0,0000041	3
MGAM	7:142038616:G:T	-	-	-	splice_donor_variant	HIGH	0,0000041	3
MGAM	7:142065832:G:GT	rs943323068	-	-	splice_donor_variant	HIGH	0,0000041	2
MGAM	7:142094448:G:A	-	2419	W/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142074154:G:A	-	1752	W/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142091919:C:T	rs758163241	2273	R/*	stop_gained	HIGH	0,0000041	3
MGAM	7:142084548:CTG:C	-	2138	W/X	frameshift_variant	HIGH	0,0000041	3
MGAM	7:142022314:C:T	-	253	Q/*	stop_gained	HIGH	0,0000055	4
MGAM	7:142036953:C:A	-	736	T/K	missense_variant	pathogenic	0,0000055	4
MGAM	7:142052441:T:C	-	985	W/R	missense_variant	pathogenic	0,0000055	4
MGAM	7:142054869:T:C	rs769882031	1092	F/S	missense_variant	pathogenic	0,0000055	4
MGAM	7:142059493:G:A	rs748225017	1281	D/N	missense_variant	pathogenic	0,0000055	4
MGAM	7:142063514:T:C	rs748280923	1425	S/P	missense_variant	pathogenic	0,0000055	4
MGAM	7:142065376:C:A	rs750509389	1509	T/N	missense_variant	pathogenic	0,0000055	4
MGAM	7:142065587:G:A	rs755754661	-	-	splice_acceptor_variant	HIGH	0,0000055	4
MGAM	7:142099642:G:A	-	2593	W/*	stop_gained	HIGH	0,0000055	4
MGAM	7:142022351:G:A	-	265	G/D	missense_variant	pathogenic	0,0000055	4
MGAM	7:142040794:C:G	rs368880817	816	R/G	missense_variant	pathogenic	0,0000055	4
MGAM	7:142065607:CTT:C	rs1231325342	1547	F/X	frameshift_variant	HIGH	0,0000055	4
MGAM	7:142025063:T:G	rs1172867548	299	L/W	missense_variant	pathogenic	0,0000055	4
MGAM	7:142093465:C:T	-	2363	Q/*	stop_gained	HIGH	0,0000055	4
MGAM	7:142036843:T:G	rs74975727	699	F/L	missense_variant	pathogenic	0,0000055	4
MGAM	7:142058257:C:G	rs377213841	1250	R/G	missense_variant	pathogenic	0,0000055	4



MGAM	7:142058261:A:G	rs779857945	1251	Y/C	missense_variant	pathogenic	0,0000055	4
MGAM	7:142058282:A:T	rs747497594	1258	E/V	missense_variant	pathogenic	0,0000055	4
MGAM	7:142093468:C:T	rs753394274	2364	Q/*	stop_gained	HIGH	0,0000055	3
MGAM	7:142005656:AG:A	-	43	A/X	frameshift_variant,splice_region_variant	HIGH	0,0000055	4
MGAM	7:142093551:G:A	rs764741565	-	-	splice_donor_variant	HIGH	0,0000055	3
MGAM	7:142034280:T:G	-	563	L/R	missense_variant	pathogenic	0,0000055	4
MGAM	7:142078471:G:A	rs774189961	-	-	splice_donor_variant	HIGH	0,0000055	3
MGAM	7:142038616:G:A	-	-	-	splice_donor_variant	HIGH	0,0000055	4
MGAM	7:142092047:TGTAA:T	-	-	-	splice_donor_variant,splice_donor_region_variant,intro n_variant	HIGH	0,0000055	4
MGAM	7:142040139:GTGCCTGA:G	-	781-783	VPD/X	frameshift_variant	HIGH	0,0000055	4
MGAM	7:142008670:C:T	rs782363163	98	R/*	stop_gained	HIGH	0,0000068	5
MGAM	7:142034811:GC:G	rs782348870	644	L/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142034812:CT:C	-	644	L/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142052436:G:GT	-	983	C/CX	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142056843:G:T	-	1198	Q/H	missense_variant	pathogenic	0,0000068	5
MGAM	7:142064392:T:C	-	1452	S/P	missense_variant	pathogenic	0,0000068	5
MGAM	7:142034750:C:CA	-	623	A/AX	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142058237:CT:C	-	1243	S/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142058268:C:G	-	1253	Y/*	stop_gained	HIGH	0,0000068	5
MGAM	7:142096344:C:T	rs771880163	2541	Q/*	stop_gained	HIGH	0,0000068	5
MGAM	7:142071023:G:A	-	1697	W/*	stop_gained	HIGH	0,0000068	3
MGAM	7:142050258:CT:C	rs747730268	871	L/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142062597:CTT:C	-	1385	F/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142027163:GC:G	rs782300594	344	G/X	frameshift_variant	HIGH	0,0000068	5
MGAM	7:142040154:T:A	-	786	W/R	missense_variant	pathogenic	0,0000068	5
MGAM	7:142074114:C:A	-	1739	A/D	missense_variant	pathogenic	0,0000068	5
MGAM	7:142059596:T:C	-	1315	I/T	missense_variant	pathogenic	0,0000082	6
MGAM	7:142059959:G:A	rs1231688882	1351	W/*	stop_gained	HIGH	0,0000082	6

MGAM	7:142034840:T:G	-	653	M/R	missense_variant,splice_region_variant	pathogenic	0,0000082	6
MGAM	7:142059555:GATCA:G	rs763450912	1302-1303	IN/X	frameshift_variant	HIGH	0,0000082	6
MGAM	7:142050251:AG:A	-	869	V/X	frameshift_variant	HIGH	0,0000082	6
MGAM	7:142065372:A:T	rs767687937	1508	I/F	missense_variant	pathogenic	0,0000082	6
MGAM	7:142052379:TAG:T	-	964-965	ID/IX	frameshift_variant	HIGH	0,0000082	6
MGAM	7:142027126:C:T	rs782785096	332	Q/*	stop_gained	HIGH	0,0000082	6
MGAM	7:142067410:C:G	rs571732744	1663	S/R	missense_variant	pathogenic	0,0000082	6
MGAM	7:142086297:AC:A	rs755470019	2239	Y/X	frameshift_variant	HIGH	0,0000082	6
MGAM	7:142094389:C:T	rs770700844	2400	R/*	stop_gained	HIGH	0,0000082	6
MGAM	7:142065756:C:G	rs113709751	1565	Y/*	stop_gained	HIGH	0,0000083	4
MGAM	7:142019319:G:C	rs782771834	150	G/R	missense_variant,splice_region_variant	pathogenic	0,0000096	7
MGAM	7:142021715:A:G	-	230	R/G	missense_variant	pathogenic	0,0000096	7
MGAM	7:142031739:G:C	rs1190098845	510	W/C	missense_variant	pathogenic	0,0000096	7
MGAM	7:142050804:GA:G	-	916	K/X	frameshift_variant	HIGH	0,0000096	7
MGAM	7:142063578:A:G	rs772187068	1446	Y/C	missense_variant	pathogenic	0,0000096	7
MGAM	7:142063509:A:C	rs780492645	1423	E/A	missense_variant	pathogenic	0,0000096	7
MGAM	7:142056015:T:C	rs537830668	1167	Y/H	missense_variant	pathogenic	0,0000096	7
MGAM	7:142103302:T:TA	rs777914137	2683	Y/*	stop_gained,frameshift_variant	HIGH	0,0000096	7
MGAM	7:142022307:G:C	rs782014529	250	Q/H	missense_variant	pathogenic	0,0000109	8
MGAM	7:142052938:T:A	rs763035134	1038	L/H	missense_variant	pathogenic	0,0000109	8
MGAM	7:142055638:A:G	rs1283465913	1132	Y/C	missense_variant	pathogenic	0,0000109	8
MGAM	7:142058246:T:C	rs777631001	1246	F/S	missense_variant	pathogenic	0,0000109	8
MGAM	7:142065770:G:A	rs1412935226	1570	R/H	missense_variant	pathogenic	0,0000109	6
MGAM	7:142097650:G:T	-	-	-	splice_donor_variant	HIGH	0,0000109	8
MGAM	7:142063509:A:ATGCC	rs752461974	1423	E/DAX	frameshift_variant	HIGH	0,0000123	9
MGAM	7:142093447:A:AC	-	2357	T/TX	frameshift_variant	HIGH	0,0000123	8
MGAM	7:142062583:G:A	rs755905439	1380	V/M	missense_variant	pathogenic	0,0000123	9
MGAM	7:142065728:T:C	-	1556	I/T	missense_variant	pathogenic	0,0000123	9

MGAM	7:142065769:C:T	rs1199033944	1570	R/C	missense_variant	pathogenic	0,0000123	8
MGAM	7:142071084:C:T	rs774222321	1718	P/S	missense_variant	pathogenic	0,0000136	10
MGAM	7:142025114:C:T	-	316	S/F	missense_variant	pathogenic	0,0000136	10
MGAM	7:142047841:G:A	-	852	G/E	missense_variant	pathogenic	0,0000136	10
MGAM	7:142082572:G:C	rs747832072	-	-	splice_donor_variant	HIGH	0,0000136	9
MGAM	7:142082473:A:C	-	-	-	splice_acceptor_variant	HIGH	0,0000137	7
MGAM	7:142040794:C:T	rs368880817	816	R/*	stop_gained	HIGH	0,0000150	11
MGAM	7:142063510:A:C	rs749428982	1423	E/D	missense_variant	pathogenic	0,0000150	11
MGAM	7:142031767:C:T	-	520	Q/*	stop_gained	HIGH	0,0000150	11
MGAM	7:142065379:G:A	rs760340491	1510	R/H	missense_variant	pathogenic	0,0000164	12
MGAM	7:142059505:C:T	rs773299141	1285	R/W	missense_variant	pathogenic	0,0000164	12
MGAM	7:142103268:G:A	rs754695913	-	-	splice_acceptor_variant	HIGH	0,0000164	12
MGAM	7:142065722:C:A	rs369348289	1554	A/E	missense_variant	pathogenic	0,0000164	12
MGAM	7:142055709:C:T	rs779349942	1156	R/*	stop_gained	HIGH	0,0000191	14
MGAM	7:142059479:A:G	rs770457039	1276	Q/R	missense_variant	pathogenic	0,0000191	14
MGAM	7:142008706:G:A	-	-	-	splice_donor_variant	HIGH	0,0000191	14
MGAM	7:142022389:A:ATGAAT	rs782284375	278	M/MN X	frameshift_variant	HIGH	0,0000205	15
MGAM	7:142103398:A:AT	-	2715	M/IX	frameshift_variant	HIGH	0,0000205	15
MGAM	7:142032834:G:T	rs781979872	532	E/*	stop_gained	HIGH	0,0000205	15
MGAM	7:142027204:C:T	rs372045141	358	Q/*	stop_gained	HIGH	0,0000205	15
MGAM	7:142034717:G:A	rs369813631	612	R/H	missense_variant	pathogenic	0,0000218	16
MGAM	7:142040846:G:A	rs750654956	833	S/N	missense_variant,splice_region_variant	pathogenic	0,0000218	16
MGAM	7:142052304:T:C	-	939	I/T	missense_variant	pathogenic	0,0000218	16
MGAM	7:142021043:T:C	rs782028908	173	L/P	missense_variant	pathogenic	0,0000218	16
MGAM	7:142032834:G:A	rs781979872	532	E/K	missense_variant	pathogenic	0,0000218	16
MGAM	7:142062703:G:T	rs371590004	-	-	splice_donor_variant	HIGH	0,0000232	17
MGAM	7:142063578:A:C	-	1446	Y/S	missense_variant	pathogenic	0,0000232	17
MGAM	7:142065736:T:C	rs1381221752	1559	F/L	missense_variant	pathogenic	0,0000232	15

MGAM	7:142036943:C:T	rs782352514	733	R/*	stop_gained	HIGH	0,0000273	20
MGAM	7:142091971:G:A	-	2290	W/*	stop_gained	HIGH	0,0000287	18
MGAM	7:142036257:C:T	rs782294701	683	S/F	missense_variant	pathogenic	0,0000300	22
MGAM	7:142062574:C:T	rs761756111	1377	R/*	stop_gained	HIGH	0,0000300	22
MGAM	7:142067426:G:A	rs189725035	-	-	splice_donor_variant	HIGH	0,0000300	17
MGAM	7:142021085:T:C	rs372438770	-	-	splice_donor_variant	HIGH	0,0000300	22
MGAM	7:142034295:T:C	rs782503061	568	L/P	missense_variant	pathogenic	0,0000314	23
MGAM	7:142040837:C:T	rs1432514373	830	T/I	missense_variant	pathogenic	0,0000327	24
MGAM	7:142038569:A:C	rs763035955	757	Q/P	missense_variant	pathogenic	0,0000341	25
MGAM	7:142034822:AC:A	rs749861352	647	N/X	frameshift_variant	HIGH	0,0000355	26
MGAM	7:142030702:C:G	rs781922241	472	S/*	stop_gained	HIGH	0,0000368	27
MGAM	7:142065793:TA:T	-	1578	Y/X	frameshift_variant	HIGH	0,0000369	25
MGAM	7:142056850:C:T	rs200314253	1201	P/S	missense_variant	pathogenic	0,0000382	28
MGAM	7:142056836:CG:C	rs1337419275	1196	T/X	frameshift_variant	HIGH	0,0000382	28
MGAM	7:142062569:T:C	rs147552510	1375	L/P	missense_variant,splice_region_variant	pathogenic	0,0000382	28
MGAM	7:142027649:G:A	rs558293494	379	G/R	missense_variant	pathogenic	0,0000396	29
MGAM	7:142034321:G:T	rs1263650229	577	G/C	missense_variant	pathogenic	0,0000450	33
MGAM	7:142064400:CAG:C	rs777281085	1455	R/X	frameshift_variant	HIGH	0,0000450	33
MGAM	7:142019226:T:C	rs376282334	119	W/R	missense_variant	pathogenic	0,0000450	33
MGAM	7:142030489:T:C	rs782372862	450	I/T	missense_variant	pathogenic	0,0000491	36
MGAM	7:142063587:G:A	rs373622603	-	-	splice_donor_variant	HIGH	0,0000491	36
MGAM	7:142065378:C:T	rs760873437	1510	R/C	missense_variant	pathogenic	0,0000519	38
MGAM	7:142036893:G:A	rs202196208	716	R/H	missense_variant	pathogenic	0,0000559	41
MGAM	7:142034716:C:T	rs782122165	612	R/C	missense_variant	pathogenic	0,0000573	42
MGAM	7:142063548:G:A	rs761452877	1436	C/Y	missense_variant	pathogenic	0,0000655	48
MGAM	7:142050826:C:T	rs200498907	923	Q/*	stop_gained	HIGH	0,0000669	49
MGAM	7:142065360:C:T	rs200390887	1504	R/*	stop_gained	HIGH	0,0000669	49
MGAM	7:142005635:CAA:C	rs782214553	36	K/X	frameshift_variant	HIGH	0,0000669	49

MGAM	7:142027650:G:A	rs191269779	379	G/E	missense_variant	pathogenic	0,0000696	51
MGAM	7:142036223:C:T	rs374646574	672	R/W	missense_variant	pathogenic	0,0000710	52
MGAM	7:142064498:G:T	rs201310218	1487	W/L	missense_variant	pathogenic	0,0000737	54
MGAM	7:142059583:C:T	rs573145045	1311	R/W	missense_variant	pathogenic	0,0000778	57
MGAM	7:142054778:T:C	rs187047261	1062	Y/H	missense_variant	pathogenic	0,0000805	59
MGAM	7:142031680:G:T	rs781896542	491	V/F	missense_variant,splice_region_variant	pathogenic	0,0000820	60
MGAM	7:142084518:G:T	rs769241743	-	-	splice_acceptor_variant	HIGH	0,0000832	51
MGAM	7:142056869:C:A	rs375460909	1207	T/N	missense_variant	pathogenic	0,0000914	67
MGAM	7:142065751:G:T	rs771962308	1564	E/*	stop_gained	HIGH	0,0001010	60
MGAM	7:142062687:T:G	rs372526101	1414	F/L	missense_variant	pathogenic	0,0001037	75
MGAM	7:142032827:T:G	rs200532183	529	D/E	missense_variant,splice_region_variant	pathogenic	0,0001201	88
MGAM	7:142038570:G:T	rs201786618	757	Q/H	missense_variant	pathogenic	0,0001515	111
MGAM	7:142064430:GGA:G	rs777111709	1465	E/X	frameshift_variant	HIGH	0,0001678	123
MGAM	7:142074086:C:T	rs773318610	1730	R/*	stop_gained,splice_region_variant	HIGH	0,0001720	104
MGAM	7:142056905:G:A	rs202179894	1219	G/E	missense_variant	pathogenic	0,0002115	155
MGAM	7:142065384:A:C	rs199508271	1512	T/P	missense_variant	pathogenic	0,0002879	210
MGAM	7:142052957:C:G	rs752471950	1044	Y/*	stop_gained	HIGH	0,0003329	243
MGAM	7:142022337:C:A	rs376595784	260	S/R	missense_variant	pathogenic	0,0003534	259
MGAM	7:142054886:CG:C	rs777208381	1098	R/X	frameshift_variant	HIGH	0,0006686	490
MGAM	7:142022321:C:T	rs201266519	255	S/F	missense_variant	pathogenic	0,0007136	523
MGAM	7:142052429:C:T	rs146202976	981	R/C	missense_variant	pathogenic	0,0008173	599
SI	3:164987191:T:A	-	1715	D/V	missense_variant	pathogenic	0,0000014	1
SI	3:164991372:C:G	-	1697	A/P	missense_variant	pathogenic	0,0000014	1
SI	3:164991478:C:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:164994340:T:A	-	1586	R/S	missense_variant	pathogenic	0,0000014	1
SI	3:164998548:G:T	-	1511	S/*	stop_gained	HIGH	0,0000014	1
SI	3:164998557:A:G	-	1508	M/T	missense_variant	pathogenic	0,0000014	1
SI	3:164998560:T:G	-	1507	N/T	missense_variant	pathogenic	0,0000014	1

SI	3:164998561:T:A	-	1507	N/Y	missense_variant	pathogenic	0,0000014	1
SI	3:164998584:C:T	rs1376892049	1499	G/E	missense_variant	pathogenic	0,0000014	1
SI	3:165009336:C:A	-	1374	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165009336:C:T	rs1036821027	1374	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165017556:T:A	-	1251	I/F	missense_variant	pathogenic	0,0000014	1
SI	3:165017619:G:A	rs560337003	1230	R/C	missense_variant	pathogenic	0,0000014	1
SI	3:165017627:T:C	rs1216531324	1227	Q/R	missense_variant	pathogenic	0,0000014	1
SI	3:165017819:A:G	-	1192	L/S	missense_variant	pathogenic	0,0000014	1
SI	3:165019604:C:G	rs368953032	1141	G/R	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165019615:T:C	-	1137	D/G	missense_variant	pathogenic	0,0000014	1
SI	3:165019641:C:A	-	1128	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165019641:C:G	-	1128	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165019679:C:T	-	1116	E/K	missense_variant	pathogenic	0,0000014	1
SI	3:165019748:A:G	-	1093	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165030776:C:G	rs375113759	943	C/S	missense_variant	pathogenic	0,0000014	1
SI	3:165030789:C:A	-	939	E/*	stop_gained	HIGH	0,0000014	1
SI	3:165030812:C:T	-	931	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165046893:A:T	-	612	M/K	missense_variant	pathogenic	0,0000014	1
SI	3:165059180:A:C	-	422	Y/*	stop_gained	HIGH	0,0000014	1
SI	3:164982274:GA:G	-	1795	S/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164991384:A:G	rs761065677	1693	C/R	missense_variant	pathogenic	0,0000014	1
SI	3:164992339:A:G	-	1634	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:164992347:C:A	-	1631	G/V	missense_variant	pathogenic	0,0000014	1
SI	3:164994294:C:G	-	1602	A/P	missense_variant	pathogenic	0,0000014	1
SI	3:165009337:C:A	-	1374	W/L	missense_variant	pathogenic	0,0000014	1
SI	3:165009337:C:T	-	1374	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165009339:C:T	-	1373	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165015163:A:G	-	1320	F/S	missense_variant	pathogenic	0,0000014	1

SI	3:165015181:C:T	rs1356567879	1314	G/E	missense_variant	pathogenic	0,0000014	1
SI	3:165016080:C:T	-	1254	D/N	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165017807:A:T	rs1045525232	1196	M/K	missense_variant	pathogenic	0,0000014	1
SI	3:165017838:G:GGTAA	-	1185-1186	-/LX	frameshift_variant	HIGH	0,0000014	1
SI	3:165017843:G:A	-	1184	T/I	missense_variant	pathogenic	0,0000014	1
SI	3:165019609:G:C	-	1139	P/R	missense_variant	pathogenic	0,0000014	1
SI	3:165019616:C:G	-	1137	D/H	missense_variant	pathogenic	0,0000014	1
SI	3:165019616:C:T	rs757669730	1137	D/N	missense_variant	pathogenic	0,0000014	1
SI	3:165019751:C:T	-	1092	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165021266:CA:C	-	1072	F/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165023722:AAG:A	rs751263307	982	S/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165023765:T:TA	-	968	L/LX	frameshift_variant	HIGH	0,0000014	1
SI	3:165030732:G:A	rs745923820	958	R/C	missense_variant	pathogenic	0,0000014	1
SI	3:165030772:A:T	-	944	Y/*	stop_gained	HIGH	0,0000014	1
SI	3:165030780:TA:T	-	941	F/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165030783:A:G	-	941	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165030785:CT:C	-	940	R/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165030786:T:A	-	940	R/*	stop_gained	HIGH	0,0000014	1
SI	3:165030811:C:G	-	931	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165033409:A:G	rs759374070	851	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165033445:C:T	rs1576898573	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165039947:C:G	rs866045548	728	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165041085:G:A	-	672	P/S	missense_variant	pathogenic	0,0000014	1
SI	3:165041085:G:T	-	672	P/T	missense_variant	pathogenic	0,0000014	1
SI	3:165043111:T:C	-	651	Q/R	missense_variant	pathogenic	0,0000014	1
SI	3:165046886:C:A	rs1337199717	614	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:165046896:T:C	-	611	Q/R	missense_variant	pathogenic	0,0000014	1
SI	3:165046922:T:G	rs1462662903	602	L/F	missense_variant	pathogenic	0,0000014	1

SI	3:165046951:C:T	-	593	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165046983:C:T	rs1422477424	582	R/K	missense_variant	pathogenic	0,0000014	1
SI	3:165059272:T:C	rs750073419	392	M/V	missense_variant	pathogenic	0,0000014	1
SI	3:165059900:A:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165059940:CT:C	-	369	K/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164987174:G:A	-	1721	Q/*	stop_gained	HIGH	0,0000014	1
SI	3:164992192:C:A	-	1656	W/C	missense_variant	pathogenic	0,0000014	1
SI	3:164992351:A:G	rs1290014420	1630	W/R	missense_variant	pathogenic	0,0000014	1
SI	3:164992357:A:G	-	1628	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:164992364:GA:G	-	1625	F/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164994317:TA:T	-	1594	Y/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164998542:A:T	rs1219075046	1513	I/N	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165006844:C:T	-	1460	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165015211:T:G	-	1304	E/A	missense_variant	pathogenic	0,0000014	1
SI	3:165016079:T:C	-	1254	D/G	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165017632:TC:T	-	1225	G/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165036461:C:G	-	815	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165039960:T:A	-	724	D/V	missense_variant	pathogenic	0,0000014	1
SI	3:165046959:G:T	-	590	T/K	missense_variant	pathogenic	0,0000014	1
SI	3:165059298:T:C	-	383	D/G	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:164998572:G:A	rs868014810	1503	A/V	missense_variant	pathogenic	0,0000014	1
SI	3:165006851:A:C	-	1457	N/K	missense_variant	pathogenic	0,0000014	1
SI	3:165016057:G:C	-	1261	D/E	missense_variant	pathogenic	0,0000014	1
SI	3:165016081:C:T	rs1279115346	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165021265:C:A	-	1073	G/V	missense_variant	pathogenic	0,0000014	1
SI	3:165021358:T:C	rs770444068	1042	Y/C	missense_variant	pathogenic	0,0000014	1
SI	3:165036429:T:TAG	-	825	T/TX	frameshift_variant	HIGH	0,0000014	1
SI	3:165036473:T:TA	-	810-811	-/X	frameshift_variant	HIGH	0,0000014	1



SI	3:165039931:A:T	-	734	F/I	missense_variant	pathogenic	0,0000014	1
SI	3:165041000:T:C	-	700	Y/C	missense_variant	pathogenic	0,0000014	1
SI	3:165043110:T:A	-	651	Q/H	missense_variant	pathogenic	0,0000014	1
SI	3:165055241:C:A	-	489	E/*	stop_gained	HIGH	0,0000014	1
SI	3:165059040:A:G	-	441	Y/H	missense_variant	pathogenic	0,0000014	1
SI	3:165059901:C:A	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:164982261:AT:A	-	1799	N/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164982335:A:G	rs1444747428	1775	W/R	missense_variant	pathogenic	0,0000014	1
SI	3:164991405:G:T	-	1686	R/S	missense_variant	pathogenic	0,0000014	1
SI	3:164998626:G:A	rs1311477041	1485	S/F	missense_variant	pathogenic	0,0000014	1
SI	3:165006831:A:C	-	1464	M/R	missense_variant	pathogenic	0,0000014	1
SI	3:165015134:A:T	-	1330	C/S	missense_variant	pathogenic	0,0000014	1
SI	3:165015959:A:G	rs752481222	1294	I/T	missense_variant	pathogenic	0,0000014	1
SI	3:165016065:C:T	-	1259	D/N	missense_variant	pathogenic	0,0000014	1
SI	3:165019772:T:A	rs200479964	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165023583:A:AT	-	1029	M/NX	frameshift_variant	HIGH	0,0000014	1
SI	3:165023597:A:C	-	1024	Y/*	stop_gained	HIGH	0,0000014	1
SI	3:165032578:C:A	-	894	E/*	stop_gained	HIGH	0,0000014	1
SI	3:165037963:C:A	-	788	G/V	missense_variant	pathogenic	0,0000014	1
SI	3:165040999:G:GT	-	700	Y/*	stop_gained,frameshift_variant	HIGH	0,0000014	1
SI	3:165043130:G:A	-	645	L/F	missense_variant	pathogenic	0,0000014	1
SI	3:165055252:C:A	-	485	W/L	missense_variant	pathogenic	0,0000014	1
SI	3:165059172:A:T	-	425	I/N	missense_variant	pathogenic	0,0000014	1
SI	3:165059907:G:T	-	381	P/T	missense_variant	pathogenic	0,0000014	1
SI	3:164987216:GT:G	-	1706	K/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164998632:G:A	-	1483	S/F	missense_variant	pathogenic	0,0000014	1
SI	3:164998632:G:T	-	1483	S/Y	missense_variant	pathogenic	0,0000014	1
SI	3:165019717:G:T	-	1103	S/*	stop_gained	HIGH	0,0000014	1

SI	3:165023588:A:AT	rs763342449	1027	N/KX	frameshift_variant	HIGH	0,0000014	1
SI	3:165037964:C:A	-	788	G/*	stop_gained	HIGH	0,0000014	1
SI	3:165037964:C:T	rs1042903450	788	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165041015:A:G	-	695	L/S	missense_variant	pathogenic	0,0000014	1
SI	3:165046926:C:A	-	601	W/L	missense_variant	pathogenic	0,0000014	1
SI	3:165067371:G:A	-	202	Q/*	stop_gained	HIGH	0,0000014	1
SI	3:164982334:C:T	rs1271339714	1775	W/*	stop_gained	HIGH	0,0000014	1
SI	3:164991409:A:T	-	1684	H/Q	missense_variant	pathogenic	0,0000014	1
SI	3:164998603:A:T	-	1493	W/R	missense_variant	pathogenic	0,0000014	1
SI	3:164998646:T:G	-	1478	R/S	missense_variant	pathogenic	0,0000014	1
SI	3:165015974:C:T	-	1289	G/E	missense_variant	pathogenic	0,0000014	1
SI	3:165018020:T:A	-	1157	E/V	missense_variant	pathogenic	0,0000014	1
SI	3:165039909:A:G	rs1167931116	741	L/P	missense_variant	pathogenic	0,0000014	1
SI	3:165046908:G:A	-	607	A/V	missense_variant	pathogenic	0,0000014	1
SI	3:165062424:A:C	rs1283221179	323	F/V	missense_variant	pathogenic	0,0000014	1
SI	3:165062426:T:A	rs751813346	322	D/V	missense_variant	pathogenic	0,0000014	1
SI	3:165065289:A:T	-	260	I/N	missense_variant	pathogenic	0,0000014	1
SI	3:165017610:A:G	-	1233	Y/H	missense_variant	pathogenic	0,0000014	1
SI	3:165018017:TC:T	-	1158	E/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165041041:C:A	-	686	R/S	missense_variant	pathogenic	0,0000014	1
SI	3:165041057:A:G	rs1291880028	681	L/S	missense_variant	pathogenic	0,0000014	1
SI	3:165049166:T:A	-	559	H/L	missense_variant	pathogenic	0,0000014	1
SI	3:164982344:G:GATCC	rs1162243631	1771-1772	-/GX	frameshift_variant	HIGH	0,0000014	1
SI	3:165009395:C:G	-	1355	A/P	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165039900:G:A	-	744	P/L	missense_variant	pathogenic	0,0000014	1
SI	3:165043117:C:T	-	649	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165046918:C:A	rs1471498582	604	D/Y	missense_variant	pathogenic	0,0000014	1
SI	3:165075987:A:T	-	9	L/*	stop_gained	HIGH	0,0000014	1

SI	3:164982329:TC:T	-	1776	G/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165006815:C:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165006847:A:G	-	1459	Y/H	missense_variant	pathogenic	0,0000014	1
SI	3:165015154:C:T	-	1323	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165021227:A:G	rs1223791903	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165032613:C:A	-	882	G/V	missense_variant	pathogenic	0,0000014	1
SI	3:165049211:A:C	-	544	I/S	missense_variant	pathogenic	0,0000014	1
SI	3:164994327:GC:G	-	1590	L/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165015983:C:G	-	1286	R/T	missense_variant	pathogenic	0,0000014	1
SI	3:165058968:C:CA	-	464-465	-/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165065305:A:G	-	255	W/R	missense_variant	pathogenic	0,0000014	1
SI	3:165067469:C:G	rs1351473418	169	R/T	missense_variant	pathogenic	0,0000014	1
SI	3:165075944:TA:T	-	23	I/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164996597:G:T	rs767701775	1544	R/S	missense_variant	pathogenic	0,0000014	1
SI	3:165006918:G:T	rs1444651507	1435	T/K	missense_variant	pathogenic	0,0000014	1
SI	3:165006918:GT:G	-	1435	T/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165006922:T:C	-	1434	R/G	missense_variant	pathogenic	0,0000014	1
SI	3:165049844:C:T	-	515	G/D	missense_variant	pathogenic	0,0000014	1
SI	3:164987154:A:T	-	1727	D/E	missense_variant	pathogenic	0,0000014	1
SI	3:165040940:T:A	-	720	E/V	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165046916:G:T	-	604	D/E	missense_variant	pathogenic	0,0000014	1
SI	3:164982257:CT:C	-	1800	E/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164996626:A:C	-	1534	F/C	missense_variant	pathogenic	0,0000014	1
SI	3:164996626:A:G	rs1236832078	1534	F/S	missense_variant	pathogenic	0,0000014	1
SI	3:165019688:CAT:C	-	1112	Y/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165032619:A:G	-	880	I/T	missense_variant	pathogenic	0,0000014	1
SI	3:165040963:TG:T	-	712	T/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164982379:CCT:C	-	1759-1760	RG/RX	frameshift_variant	HIGH	0,0000014	1

SI	3:164992394:G:T	-	1615	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165069167:C:T	-	95	W/*	stop_gained	HIGH	0,0000014	1
SI	3:165074599:A:G	-	63	C/R	missense_variant	pathogenic	0,0000014	1
SI	3:165018050:T:C	rs760937243	1147	Y/C	missense_variant	pathogenic	0,0000014	1
SI	3:164996625:G:T	-	1534	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165012984:A:G	rs1427782400	1353	V/A	missense_variant	pathogenic	0,0000014	1
SI	3:165012984:A:T	-	1353	V/D	missense_variant	pathogenic	0,0000014	1
SI	3:165032566:GTTGA:G	rs774850525	896-897	NQ/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165068739:TG:T	-	155	P/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165009295:A:C	-	1388	F/C	missense_variant	pathogenic	0,0000014	1
SI	3:165062409:C:T	rs765800369	328	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165055285:T:C	-	474	Y/C	missense_variant	pathogenic	0,0000014	1
SI	3:165016017:A:G	-	1275	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:165016017:A:T	rs760534537	1275	F/I	missense_variant	pathogenic	0,0000014	1
SI	3:165009292:T:G	-	1389	D/A	missense_variant	pathogenic	0,0000014	1
SI	3:165023571:T:A	-	1033	K/M	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165065283:G:A	rs146187373	262	T/I	missense_variant	pathogenic	0,0000014	1
SI	3:165065344:C:A	-	242	G/*	stop_gained	HIGH	0,0000014	1
SI	3:165023569:C:T	rs775933206	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165006942:CT:C	-	1427	R/X	frameshift_variant	HIGH	0,0000014	1
SI	3:164996588:G:T	rs763589746	1547	Q/K	missense_variant	pathogenic	0,0000014	1
SI	3:165018049:A:T	-	1147	Y/*	stop_gained	HIGH	0,0000014	1
SI	3:165007975:A:C	rs574552408	1401	F/L	missense_variant	pathogenic	0,0000014	1
SI	3:164987137:C:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:164994254:TACTC:T	rs1240035419	1614-?	-	splice_donor_variant,coding_sequence_variant	HIGH	0,0000014	1
SI	3:164994255:A:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165049831:TC:T	-	519	G/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165049806:G:T	-	528	P/T	missense_variant	pathogenic	0,0000014	1

SI	3:164987227:C:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165043169:C:T	-	632	A/T	missense_variant	pathogenic	0,0000014	1
SI	3:165055215:C:CA	-	497	V/VX	frameshift_variant	HIGH	0,0000014	1
SI	3:165068823:CT:C	-	127	E/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165009289:C:T	-	1390	G/D	missense_variant	pathogenic	0,0000014	1
SI	3:165018002:T:C	-	1163	H/R	missense_variant	pathogenic	0,0000014	1
SI	3:165040942:A:T	-	719	H/Q	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165062411:A:G	rs1287181714	327	L/P	missense_variant	pathogenic	0,0000014	1
SI	3:165049872:T:C	rs1302522597	506	M/V	missense_variant	pathogenic	0,0000014	1
SI	3:165063492:T:A	-	286	D/V	missense_variant	pathogenic	0,0000014	1
SI	3:165062397:C:A	-	332	E/*	stop_gained	HIGH	0,0000014	1
SI	3:165065415:C:T	rs775308282	218	G/D	missense_variant	pathogenic	0,0000014	1
SI	3:164994324:AG:A	-	1591	P/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165038024:C:A	-	768	G/C	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165037898:A:C	rs778794280	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165043175:C:A	-	630	V/F	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165049805:G:A	rs746292703	528	P/L	missense_variant	pathogenic	0,0000014	1
SI	3:165009278:C:A	rs1395936979	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165013044:T:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165063484:C:A	-	289	G/*	stop_gained	HIGH	0,0000014	1
SI	3:165067338:A:G	rs1185991890	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:164996632:C:T	-	1532	G/D	missense_variant	pathogenic	0,0000014	1
SI	3:165017980:G:T	-	1170	S/R	missense_variant	pathogenic	0,0000014	1
SI	3:165069076:A:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165049793:G:A	-	532	P/L	missense_variant,splice_region_variant	pathogenic	0,0000014	1
SI	3:165069196:C:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165049148:C:T	rs149898910	565	S/N	missense_variant	pathogenic	0,0000014	1
SI	3:165007925:G:A	-	1418	P/L	missense_variant	pathogenic	0,0000014	1

SI	3:164996644:G:A	-	1528	A/V	missense_variant	pathogenic	0,0000014	1
SI	3:165063477:G:A	-	291	S/L	missense_variant	pathogenic	0,0000014	1
SI	3:165065392:A:T	-	226	Y/N	missense_variant	pathogenic	0,0000014	1
SI	3:164996653:T:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165047013:C:G	rs746602450	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165062449:ATATGT:A	-	313-314	TY/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165062451:A:G	rs1324410079	314	Y/H	missense_variant	pathogenic	0,0000014	1
SI	3:165049241:A:C	rs1198725425	534	I/S	missense_variant	pathogenic	0,0000014	1
SI	3:165055205:C:T	-	501	G/R	missense_variant	pathogenic	0,0000014	1
SI	3:165065353:A:G	rs756714861	239	Y/H	missense_variant	pathogenic	0,0000014	1
SI	3:164996774:T:C	rs749013790	-	-	splice_acceptor_variant	HIGH	0,0000014	1
SI	3:165007910:C:T	rs199897248	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165015121:ACCTTTG:A	-	1332-?	-	splice_donor_variant,coding_sequence_variant	HIGH	0,0000014	1
SI	3:164996533:A:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:165075970:C:CA	-	14-15	-/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165039881:GCCTA:G	rs756860453	-	-	splice_donor_variant,splice_donor_5th_base_variant,intron_variant	HIGH	0,0000014	1
SI	3:165039886:C:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1
SI	3:164996748:CCAAA:C	rs1200263910	1521-1522	FG/X	frameshift_variant	HIGH	0,0000014	1
SI	3:165030868:C:G	rs1294620045	-	-	splice_acceptor_variant	HIGH	0,0000018	1
SI	3:165030741:A:G	-	955	C/R	missense_variant	pathogenic	0,0000027	2
SI	3:164998574:A:T	-	1502	Y/*	stop_gained	HIGH	0,0000027	2
SI	3:165017616:A:G	rs1324457122	1231	Y/H	missense_variant	pathogenic	0,0000027	2
SI	3:165017825:C:T	rs769201557	1190	G/E	missense_variant	pathogenic	0,0000027	2
SI	3:165017829:C:T	-	1189	G/R	missense_variant	pathogenic	0,0000027	2
SI	3:165019610:G:T	-	1139	P/T	missense_variant	pathogenic	0,0000027	2
SI	3:165021262:A:T	-	1074	I/N	missense_variant	pathogenic	0,0000027	2
SI	3:165059945:A:T	-	368	V/E	missense_variant	pathogenic	0,0000027	2
SI	3:165069128:A:C	-	108	F/C	missense_variant	pathogenic	0,0000027	2

SI	3:165046944:C:T	-	595	G/E	missense_variant	pathogenic	0,0000027	2
SI	3:165046976:G:C	-	584	F/L	missense_variant	pathogenic	0,0000027	2
SI	3:164991398:C:T	-	1688	G/D	missense_variant	pathogenic	0,0000027	2
SI	3:165068789:A:G	-	139	F/S	missense_variant	pathogenic	0,0000027	2
SI	3:164994287:C:T	-	1604	G/D	missense_variant	pathogenic	0,0000027	2
SI	3:165017653:C:A	-	1218	M/I	missense_variant	pathogenic	0,0000027	2
SI	3:165017667:C:G	-	1214	G/R	missense_variant	pathogenic	0,0000027	2
SI	3:165043114:A:C	-	650	M/R	missense_variant	pathogenic	0,0000027	2
SI	3:165046897:G:T	rs1025991342	611	Q/K	missense_variant	pathogenic	0,0000027	2
SI	3:165017553:G:T	-	1252	P/T	missense_variant	pathogenic	0,0000027	2
SI	3:165043097:A:G	rs1432675903	656	Y/H	missense_variant	pathogenic	0,0000027	2
SI	3:165046996:A:G	-	578	F/L	missense_variant	pathogenic	0,0000027	2
SI	3:165068749:A:AT	-	152	N/KX	frameshift_variant	HIGH	0,0000027	2
SI	3:164982333:C:T	-	1775	W/*	stop_gained	HIGH	0,0000027	2
SI	3:164991427:A:T	-	1678	Y/*	stop_gained	HIGH	0,0000027	2
SI	3:165067454:T:C	rs1163225821	174	H/R	missense_variant	pathogenic	0,0000027	2
SI	3:165068811:T:A	-	132	R/W	missense_variant	pathogenic	0,0000027	2
SI	3:165041024:C:T	-	692	R/H	missense_variant	pathogenic	0,0000027	2
SI	3:165059904:ATG:A	-	381	P/X	frameshift_variant	HIGH	0,0000027	2
SI	3:164998592:G:C	rs1324167161	1496	H/Q	missense_variant	pathogenic	0,0000027	2
SI	3:165023673:A:T	-	999	L/H	missense_variant	pathogenic	0,0000027	2
SI	3:165049179:G:T	-	555	Q/K	missense_variant	pathogenic	0,0000027	2
SI	3:165055238:A:G	-	490	C/R	missense_variant	pathogenic	0,0000027	2
SI	3:164982412:T:G	rs1298467427	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:165015133:C:A	rs138283071	1330	C/F	missense_variant	pathogenic	0,0000027	2
SI	3:165036427:G:T	rs769928395	826	A/D	missense_variant	pathogenic	0,0000027	2
SI	3:165039921:C:T	-	737	G/D	missense_variant	pathogenic	0,0000027	2
SI	3:165049205:A:T	-	546	M/K	missense_variant	pathogenic	0,0000027	2

SI	3:165019691:A:G	-	1112	Y/H	missense_variant	pathogenic	0,0000027	2
SI	3:165021229:A:G	-	1085	I/T	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165036407:C:T	-	833	D/N	missense_variant	pathogenic	0,0000027	2
SI	3:164987224:C:T	rs771806803	1704	R/Q	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165046936:C:G	rs769057782	598	A/P	missense_variant	pathogenic	0,0000027	2
SI	3:164991415:G:C	rs777479828	1682	N/K	missense_variant	pathogenic	0,0000027	2
SI	3:165017999:C:T	-	1164	G/D	missense_variant	pathogenic	0,0000027	2
SI	3:164994328:C:A	-	1590	L/F	missense_variant	pathogenic	0,0000027	2
SI	3:165037921:G:T	-	802	P/Q	missense_variant	pathogenic	0,0000027	2
SI	3:164998674:C:T	rs200481844	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:165009286:A:C	rs1305050775	1391	L/W	missense_variant	pathogenic	0,0000027	2
SI	3:165049188:A:G	rs1034161542	552	W/R	missense_variant	pathogenic	0,0000027	2
SI	3:165037987:A:AT	-	780	M/NX	frameshift_variant	HIGH	0,0000027	2
SI	3:165009385:G:A	-	1358	A/V	missense_variant	pathogenic	0,0000027	2
SI	3:165012996:T:C	-	1349	E/G	missense_variant	pathogenic	0,0000027	2
SI	3:165067493:T:C	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:164996625:G:GA	rs779227756	1534	F/FX	frameshift_variant	HIGH	0,0000027	2
SI	3:165043166:C:T	rs753216085	633	D/N	missense_variant	pathogenic	0,0000027	2
SI	3:165046938:TG:T	-	597	H/X	frameshift_variant	HIGH	0,0000027	2
SI	3:165036476:G:A	rs370537003	810	R/C	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165036476:G:T	-	810	R/S	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165013008:TTA:T	rs1171377862	1344-1345	DK/EX	frameshift_variant	HIGH	0,0000027	2
SI	3:165017760:C:T	-	-	-	splice_donor_variant	HIGH	0,0000027	2
SI	3:165018053:G:A	rs1193249063	1146	S/F	missense_variant	pathogenic	0,0000027	2
SI	3:165018061:T:A	-	1143	K/N	missense_variant	pathogenic	0,0000027	2
SI	3:165036388:C:G	-	-	-	splice_donor_variant	HIGH	0,0000027	2
SI	3:165039076:A:G	-	-	-	splice_donor_variant	HIGH	0,0000027	2
SI	3:165049157:T:C	rs772385303	562	Y/C	missense_variant	pathogenic	0,0000027	2



SI	3:165043079:G:T	-	662	H/N	missense_variant	pathogenic	0,0000027	2
SI	3:165049146:T:A	-	566	M/L	missense_variant	pathogenic	0,0000027	2
SI	3:165063501:C:T	-	283	C/Y	missense_variant	pathogenic	0,0000027	2
SI	3:165065406:A:C	rs1264604545	221	V/G	missense_variant	pathogenic	0,0000027	2
SI	3:164996563:G:A	rs769560102	1555	S/L	missense_variant	pathogenic	0,0000027	2
SI	3:165075997:ATTTC:A	rs772061269	4-5	KK/X	frameshift_variant	HIGH	0,0000027	2
SI	3:165062472:G:A	-	307	Q/*	stop_gained	HIGH	0,0000027	2
SI	3:165032600:ACTGT:A	rs1221816681	885-886	DS/X	frameshift_variant	HIGH	0,0000027	2
SI	3:165055303:C:T	-	468	W/*	stop_gained	HIGH	0,0000027	2
SI	3:164996653:T:A	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:165018067:C:A	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:165043176:C:A	-	-	-	splice_acceptor_variant	HIGH	0,0000027	2
SI	3:165055301:G:T	-	469	P/T	missense_variant	pathogenic	0,0000027	2
SI	3:165007997:T:C	-	1394	D/G	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:164979369:CA:C	-	1826	W/X	frameshift_variant	HIGH	0,0000027	2
SI	3:165049142:GC:G	-	567	A/X	frameshift_variant	HIGH	0,0000027	2
SI	3:165065431:A:G	-	213	F/L	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165049241:A:G	-	534	I/T	missense_variant	pathogenic	0,0000027	2
SI	3:165049126:C:A	-	-	-	splice_donor_variant	HIGH	0,0000027	2
SI	3:165063511:A:C	-	280	F/V	missense_variant	pathogenic	0,0000027	2
SI	3:164996765:C:T	rs1040060732	1516	M/I	missense_variant	pathogenic	0,0000027	2
SI	3:164996533:A:T	-	-	-	splice_donor_variant	HIGH	0,0000027	2
SI	3:165063442:C:T	-	303	E/K	missense_variant,splice_region_variant	pathogenic	0,0000027	2
SI	3:165039878:AGAGCCTACCTGTTTT:A	rs750338611	746-?	-	splice_donor_variant,splice_donor_5th_base_variant,co ding_sequence_variant,intron_variant	HIGH	0,0000028	2
SI	3:165030868:C:A	rs1294620045	-	-	splice_acceptor_variant	HIGH	0,0000036	2
SI	3:164994372:C:G	rs759447493	1576	A/P	missense_variant	pathogenic	0,0000041	3
SI	3:165017817:C:T	rs746636433	1193	D/N	missense_variant	pathogenic	0,0000041	3
SI	3:165059928:T:C	-	374	R/G	missense_variant	pathogenic	0,0000041	3

SI	3:164991401:C:T	-	1687	G/D	missense_variant	pathogenic	0,0000041	3
SI	3:165015169:T:A	rs1193644915	1318	D/V	missense_variant	pathogenic	0,0000041	3
SI	3:165059945:A:G	-	368	V/A	missense_variant	pathogenic	0,0000041	3
SI	3:164982287:C:A	-	1791	G/*	stop_gained	HIGH	0,0000041	3
SI	3:165009363:G:GA	-	1365	F/FX	frameshift_variant	HIGH	0,0000041	3
SI	3:165017634:C:A	-	1225	G/*	stop_gained	HIGH	0,0000041	3
SI	3:165059250:G:A	rs75188774	399	T/I	missense_variant	pathogenic	0,0000041	3
SI	3:165060021:C:A	rs898650750	343	G/*	stop_gained	HIGH	0,0000041	3
SI	3:165068781:C:G	-	142	D/H	missense_variant	pathogenic	0,0000041	3
SI	3:165016064:T:C	-	1259	D/G	missense_variant	pathogenic	0,0000041	3
SI	3:164998570:G:A	rs1164456060	1504	R/*	stop_gained	HIGH	0,0000041	3
SI	3:165041021:T:C	-	693	Y/C	missense_variant	pathogenic	0,0000041	3
SI	3:165055241:C:T	-	489	E/K	missense_variant	pathogenic	0,0000041	3
SI	3:165017777:G:C	-	1206	T/R	missense_variant	pathogenic	0,0000041	3
SI	3:165059907:G:A	-	381	P/S	missense_variant	pathogenic	0,0000041	3
SI	3:165065295:C:T	rs1414419456	258	W/*	stop_gained	HIGH	0,0000041	3
SI	3:165067465:A:T	rs751748355	170	Y/*	stop_gained	HIGH	0,0000041	3
SI	3:165019718:A:G	-	1103	S/P	missense_variant	pathogenic	0,0000041	3
SI	3:165041073:C:CA	-	675-676	-/X	frameshift_variant	HIGH	0,0000041	3
SI	3:165049184:C:T	-	553	G/D	missense_variant	pathogenic	0,0000041	3
SI	3:165007950:A:G	-	1410	C/R	missense_variant	pathogenic	0,0000041	3
SI	3:164991415:G:T	-	1682	N/K	missense_variant	pathogenic	0,0000041	3
SI	3:164992310:TAC:T	-	-	-	splice_donor_variant	HIGH	0,0000041	3
SI	3:164996597:G:C	rs767701775	1544	R/G	missense_variant	pathogenic	0,0000041	3
SI	3:165017990:A:G	-	1167	L/S	missense_variant	pathogenic	0,0000041	3
SI	3:164983053:T:C	-	-	-	splice_acceptor_variant	HIGH	0,0000041	3
SI	3:165062439:C:T	rs749095854	318	G/S	missense_variant	pathogenic	0,0000041	3
SI	3:165017874:C:A	rs757893762	-	-	splice_acceptor_variant	HIGH	0,0000041	3

SI	3:165013027:G:A	rs1203735967	1339	P/S	missense_variant	pathogenic	0,0000041	3
SI	3:165021363:CTTCTT:C	rs765274319	1039-1040	KK/X	frameshift_variant	HIGH	0,0000041	3
SI	3:165040942:A:C	rs1193627786	719	H/Q	missense_variant,splice_region_variant	pathogenic	0,0000041	3
SI	3:165007982:G:C	-	1399	S/*	stop_gained	HIGH	0,0000041	3
SI	3:165063484:C:T	-	289	G/R	missense_variant	pathogenic	0,0000041	3
SI	3:165007914:G:A	-	1422	P/S	missense_variant	pathogenic	0,0000041	3
SI	3:165049813:CAATTT:C	-	524-525	KL/X	frameshift_variant	HIGH	0,0000041	3
SI	3:165055193:C:T	rs1320118665	-	-	splice_donor_variant	HIGH	0,0000041	3
SI	3:165068815:T:A	rs1471839282	130	L/F	missense_variant	pathogenic	0,0000055	4
SI	3:165015215:T:G	-	1303	N/H	missense_variant	pathogenic	0,0000055	4
SI	3:164991428:TA:T	-	1678	Y/X	frameshift_variant	HIGH	0,0000055	4
SI	3:165049175:T:C	rs1368010717	556	Y/C	missense_variant	pathogenic	0,0000055	4
SI	3:165062426:T:C	-	322	D/G	missense_variant	pathogenic	0,0000055	4
SI	3:165041004:G:A	rs1299380975	699	L/F	missense_variant	pathogenic	0,0000055	4
SI	3:165067470:T:C	-	169	R/G	missense_variant	pathogenic	0,0000055	4
SI	3:164998666:G:A	rs1247249531	1472	Q/*	stop_gained	HIGH	0,0000055	4
SI	3:165062432:A:T	-	320	I/N	missense_variant	pathogenic	0,0000055	4
SI	3:164996605:A:G	rs1198502961	1541	L/P	missense_variant	pathogenic	0,0000055	4
SI	3:165041022:A:G	-	693	Y/H	missense_variant	pathogenic	0,0000055	4
SI	3:165037921:G:C	rs781161253	802	P/R	missense_variant	pathogenic	0,0000055	4
SI	3:165065280:C:T	rs143135955	263	R/Q	missense_variant	pathogenic	0,0000055	4
SI	3:165062408:C:T	-	328	G/E	missense_variant	pathogenic	0,0000055	4
SI	3:164994255:A:C	rs762853812	-	-	splice_donor_variant	HIGH	0,0000055	4
SI	3:165013037:C:A	-	1335	W/C	missense_variant	pathogenic	0,0000055	4
SI	3:165055206:A:C	rs776358754	500	D/E	missense_variant	pathogenic	0,0000055	4
SI	3:164992398:C:A	-	-	-	splice_acceptor_variant	HIGH	0,0000055	4
SI	3:165018055:A:T	rs1248872255	1145	N/K	missense_variant	pathogenic	0,0000055	4
SI	3:164979368:C:T	rs1363481742	1826	W/*	stop_gained	HIGH	0,0000055	4

SI	3:165065382:A:T	rs1178804798	229	I/N	missense_variant	pathogenic	0,0000055	4
SI	3:164996642:C:T	-	1529	D/N	missense_variant	pathogenic	0,0000055	4
SI	3:165030866:AC:A	rs777884395	913	V/X	frameshift_variant,splice_region_variant	HIGH	0,0000055	4
SI	3:165032521:C:T	-	-	-	splice_donor_variant	HIGH	0,0000055	4
SI	3:164996553:G:T	-	1558	H/Q	missense_variant	pathogenic	0,0000055	4
SI	3:165059201:C:A	rs1350844385	415	L/F	missense_variant	pathogenic	0,0000068	5
SI	3:165046965:C:T	rs773389871	588	R/H	missense_variant	pathogenic	0,0000068	5
SI	3:164998668:A:C	rs1489696757	1471	L/W	missense_variant	pathogenic	0,0000068	5
SI	3:164987222:G:A	-	1705	Q/*	stop_gained	HIGH	0,0000068	5
SI	3:165017862:G:A	rs898055093	1178	Q/*	stop_gained	HIGH	0,0000068	5
SI	3:165017873:T:A	rs750344760	1174	D/V	missense_variant,splice_region_variant	pathogenic	0,0000068	5
SI	3:165047002:T:A	-	576	K/*	stop_gained	HIGH	0,0000068	5
SI	3:164996627:A:G	rs1187821046	1534	F/L	missense_variant	pathogenic	0,0000068	5
SI	3:164998606:G:A	rs747584061	1492	R/*	stop_gained	HIGH	0,0000068	5
SI	3:165076010:C:T	rs1427661221	1	M/I	start_lost,splice_region_variant	HIGH	0,0000068	5
SI	3:165055284:G:T	-	474	Y/*	stop_gained	HIGH	0,0000068	5
SI	3:164996555:G:C	-	1558	H/D	missense_variant	pathogenic	0,0000068	5
SI	3:165007995:T:C	rs757836000	1395	M/V	missense_variant	pathogenic	0,0000068	5
SI	3:165063543:T:C	rs772079087	-	-	splice_acceptor_variant	HIGH	0,0000068	5
SI	3:164994308:A:G	rs1191812834	1597	M/T	missense_variant	pathogenic	0,0000082	6
SI	3:165046925:C:A	rs1168175370	601	W/C	missense_variant	pathogenic	0,0000082	6
SI	3:165067370:TG:T	rs1476548939	202	Q/X	frameshift_variant	HIGH	0,0000082	6
SI	3:164987225:G:A	rs779803851	1704	R/*	stop_gained,splice_region_variant	HIGH	0,0000082	6
SI	3:165067407:A:G	rs755870756	190	Y/H	missense_variant	pathogenic	0,0000082	6
SI	3:164996596:C:T	rs1340078396	1544	R/H	missense_variant	pathogenic	0,0000082	6
SI	3:165055210:T:G	-	499	Y/S	missense_variant	pathogenic	0,0000082	6
SI	3:165007919:TA:T	-	1420	Y/X	frameshift_variant	HIGH	0,0000082	6
SI	3:165007989:C:T	-	1397	E/K	missense_variant	pathogenic	0,0000082	6

SI	3:164996736:A:G	rs754961413	-	-	splice_donor_variant	HIGH	0,0000082	6
SI	3:165063443:C:T	-	302	M/I	missense_variant,splice_region_variant	pathogenic	0,0000083	6
SI	3:164998583:T:TCCAAGCCAGTGTC CTCC	-	1499	G/GED TGLX	frameshift_variant	HIGH	0,0000096	7
SI	3:165021295:T:TAA	-	1063	Y/FX	frameshift_variant	HIGH	0,0000096	7
SI	3:165017970:C:A	-	1174	D/Y	missense_variant,splice_region_variant	pathogenic	0,0000096	7
SI	3:164979400:CAT:C	rs745476019	1815	N/X	frameshift_variant	HIGH	0,0000096	7
SI	3:165015161:C:A	-	1321	V/F	missense_variant	pathogenic	0,0000109	8
SI	3:164983018:T:G	rs1360094442	1744	Q/P	missense_variant	pathogenic	0,0000109	8
SI	3:165065415:C:A	rs775308282	218	G/V	missense_variant	pathogenic	0,0000109	7
SI	3:165062387:A:C	rs540333172	335	V/G	missense_variant	pathogenic	0,0000109	8
SI	3:164996551:TTG:T	-	1558-1559	HN/QX	frameshift_variant	HIGH	0,0000109	8
SI	3:165062370:C:T	rs759187679	-	-	splice_donor_variant	HIGH	0,0000110	8
SI	3:164987159:A:T	-	1726	W/R	missense_variant	pathogenic	0,0000123	9
SI	3:165019606:G:C	-	1140	P/R	missense_variant	pathogenic	0,0000123	9
SI	3:165021295:TAA:T	rs776569472	1062-1063	LY/LX	frameshift_variant	HIGH	0,0000123	9
SI	3:165063468:A:C	rs767579358	294	V/G	missense_variant	pathogenic	0,0000123	9
SI	3:165055304:A:G	-	468	W/R	missense_variant	pathogenic	0,0000123	9
SI	3:165065355:A:G	-	238	I/T	missense_variant	pathogenic	0,0000123	9
SI	3:165030760:AT:A	rs752264180	948	D/X	frameshift_variant	HIGH	0,0000136	10
SI	3:165059080:G:T	-	427	D/E	missense_variant,splice_region_variant	pathogenic	0,0000136	10
SI	3:164994401:T:C	rs765870223	1566	Q/R	missense_variant	pathogenic	0,0000136	10
SI	3:165009339:C:A	-	1373	W/C	missense_variant	pathogenic	0,0000136	10
SI	3:165040990:A:C	rs1341192568	703	F/L	missense_variant	pathogenic	0,0000136	10
SI	3:165046855:A:G	rs761862749	625	F/L	missense_variant	pathogenic	0,0000136	10
SI	3:165055253:A:T	rs895537564	485	W/R	missense_variant	pathogenic	0,0000136	10
SI	3:165062400:G:A	rs201980143	331	P/S	missense_variant	pathogenic	0,0000137	10
SI	3:165023743:A:G	rs755351227	976	F/L	missense_variant	pathogenic	0,0000150	11
SI	3:165059277:T:C	rs1481268075	390	D/G	missense_variant	pathogenic	0,0000150	11

SI	3:165021254:G:A	rs1000661675	1077	R/*	stop_gained	HIGH	0,0000150	11
SI	3:165060014:G:A	rs1353943703	345	P/L	missense_variant	pathogenic	0,0000150	11
SI	3:164991409:A:C	rs748153693	1684	H/Q	missense_variant	pathogenic	0,0000150	11
SI	3:165017870:A:C	rs1431988029	1175	V/G	missense_variant	pathogenic	0,0000150	11
SI	3:165009305:T:A	rs764620208	1385	K/*	stop_gained	HIGH	0,0000150	11
SI	3:165063471:C:T	rs1255647930	293	G/D	missense_variant	pathogenic	0,0000150	11
SI	3:165063523:C:T	rs371801531	276	G/S	missense_variant	pathogenic	0,0000150	11
SI	3:165046903:A:G	rs1458541922	609	W/R	missense_variant	pathogenic	0,0000164	12
SI	3:165019759:C:T	rs774109373	1089	W/*	stop_gained	HIGH	0,0000164	12
SI	3:164998608:C:T	rs769141131	1491	G/E	missense_variant	pathogenic	0,0000164	12
SI	3:165069173:C:CA	-	93	C/LX	frameshift_variant	HIGH	0,0000164	12
SI	3:165043078:T:C	rs1266738544	662	H/R	missense_variant	pathogenic	0,0000164	12
SI	3:164992178:GTA:G	rs756088593	1660-1661	HT/HX	frameshift_variant,splice_region_variant	HIGH	0,0000164	12
SI	3:165069077:C:T	rs772671498	-	-	splice_donor_variant	HIGH	0,0000164	12
SI	3:165063453:C:T	rs367672931	299	S/N	missense_variant	pathogenic	0,0000164	12
SI	3:164996774:T:A	rs749013790	-	-	splice_acceptor_variant	HIGH	0,0000164	12
SI	3:165059280:A:T	rs150428844	389	I/N	missense_variant	pathogenic	0,0000177	13
SI	3:164987177:C:T	-	1720	A/T	missense_variant	pathogenic	0,0000177	13
SI	3:165046974:A:G	-	585	I/T	missense_variant	pathogenic	0,0000177	13
SI	3:165036457:A:G	-	816	L/P	missense_variant	pathogenic	0,0000177	13
SI	3:164994326:G:T	rs1717913697	1591	P/H	missense_variant	pathogenic	0,0000177	13
SI	3:165063511:ATGTT:A	rs769229932	278-279	QT/X	frameshift_variant	HIGH	0,0000191	14
SI	3:165039940:C:G	rs1011926780	731	D/H	missense_variant	pathogenic	0,0000205	15
SI	3:165017619:G:T	rs560337003	1230	R/S	missense_variant	pathogenic	0,0000218	16
SI	3:165059061:G:A	rs982849547	434	R/*	stop_gained	HIGH	0,0000218	16
SI	3:164992347:C:T	rs371948467	1631	G/D	missense_variant	pathogenic	0,0000218	16
SI	3:165023595:T:G	rs1311777282	1025	H/P	missense_variant	pathogenic	0,0000232	17
SI	3:165046948:A:G	rs765433197	594	S/P	missense_variant	pathogenic	0,0000246	18

SI	3:164992234:C:T	rs779508795	-	-	splice_acceptor_variant	HIGH	0,0000246	18
SI	3:165063472:C:T	rs756366998	293	G/S	missense_variant	pathogenic	0,0000246	18
SI	3:165059297:A:C	rs1212421924	383	D/E	missense_variant,splice_region_variant	pathogenic	0,0000259	19
SI	3:165069136:C:A	rs138564183	105	W/C	missense_variant	pathogenic	0,0000259	19
SI	3:165075991:C:A	rs750431387	8	G/*	stop_gained	HIGH	0,0000259	19
SI	3:164994307:C:T	rs758507059	1597	M/I	missense_variant	pathogenic	0,0000273	20
SI	3:165013041:A:C	rs778216480	1334	V/G	missense_variant,splice_region_variant	pathogenic	0,0000273	20
SI	3:165049844:C:A	rs144972103	515	G/V	missense_variant	pathogenic	0,0000287	21
SI	3:164998575:T:C	rs1382587818	1502	Y/C	missense_variant	pathogenic	0,0000300	22
SI	3:165046929:T:C	rs576510887	600	H/R	missense_variant	pathogenic	0,0000300	22
SI	3:165019621:G:A	rs1412604560	1135	T/I	missense_variant	pathogenic	0,0000314	23
SI	3:164996597:G:A	rs767701775	1544	R/C	missense_variant	pathogenic	0,0000314	23
SI	3:165046891:C:A	rs201055347	613	E/*	stop_gained	HIGH	0,0000327	24
SI	3:164983001:C:T	rs1034557983	-	-	splice_donor_variant	HIGH	0,0000328	24
SI	3:165021251:G:A	rs769259229	1078	R/W	missense_variant	pathogenic	0,0000341	25
SI	3:164996641:T:C	rs539127421	1529	D/G	missense_variant	pathogenic	0,0000341	25
SI	3:165063517:G:A	rs754468827	278	Q/*	stop_gained	HIGH	0,0000341	25
SI	3:165019732:T:G	rs121912611	1098	Q/P	missense_variant	pathogenic	0,0000355	26
SI	3:165037925:C:A	rs200972419	801	E/*	stop_gained	HIGH	0,0000355	26
SI	3:165036475:C:T	rs746487509	810	R/H	missense_variant,splice_region_variant	pathogenic	0,0000355	26
SI	3:165043152:A:AG	rs866887929	637	F/FX	frameshift_variant	HIGH	0,0000382	28
SI	3:165043153:A:AAGAC	rs867598736	637	F/CLX	frameshift_variant	HIGH	0,0000382	28
SI	3:165030869:T:C	rs774405472	-	-	splice_acceptor_variant	HIGH	0,0000409	30
SI	3:165017621:C:T	rs121912614	1229	C/Y	missense_variant	pathogenic	0,0000423	31
SI	3:165063461:TA:T	rs769103567	296	L/X	frameshift_variant	HIGH	0,0000424	31
SI	3:164998630:G:A	rs201282506	1484	R/C	missense_variant	pathogenic	0,0000464	34
SI	3:165046966:G:A	rs201583834	588	R/C	missense_variant	pathogenic	0,0000464	34
SI	3:165036421:C:T	rs748638688	828	G/E	missense_variant	pathogenic	0,0000464	34

SI	3:165041025:G:A	rs371618948	692	R/C	missense_variant	pathogenic	0,0000546	40
SI	3:165043120:C:T	rs375458342	648	R/K	missense_variant	pathogenic	0,0000573	42
SI	3:165069145:AAG:A	rs1319765477	102	L/X	frameshift_variant	HIGH	0,0000587	43
SI	3:165059906:G:A	rs528535943	381	P/L	missense_variant	pathogenic	0,0000655	48
SI	3:165063461:T:TA	rs769103567	296	L/FX	frameshift_variant	HIGH	0,0000712	52
SI	3:164991381:G:T	rs138524473	1694	Q/K	missense_variant	pathogenic	0,0000750	55
SI	3:165075954:A:T	rs771580575	20	V/D	missense_variant	pathogenic	0,0000764	56
SI	3:164994273:G:A	rs200328403	1609	R/*	stop_gained	HIGH	0,0000805	59
SI	3:165007988:T:C	rs754436879	1397	E/G	missense_variant	pathogenic	0,0000819	60
SI	3:165015208:G:A	rs371426683	1305	T/I	missense_variant	pathogenic	0,0000846	62
SI	3:165017675:T:C	rs761276852	-	-	splice_acceptor_variant	HIGH	0,0000969	71
SI	3:165041070:G:GC	rs751534972	676-677	-/X	frameshift_variant	HIGH	0,0001037	76
SI	3:165067440:C:A	rs1231349140	179	E/*	stop_gained	HIGH	0,0001051	77
SI	3:165017806:CAT:C	rs780535026	1196	M/X	frameshift_variant	HIGH	0,0001201	88
SI	3:165009325:A:C	rs148831941	1378	I/S	missense_variant	pathogenic	0,0001501	110
SI	3:165019617:T:G	rs138927818	1136	R/S	missense_variant	pathogenic	0,0001556	114
SI	3:165068788:A:C	rs758407768	139	F/L	missense_variant	pathogenic	0,0001556	114
SI	3:165037957:T:C	rs149382090	790	H/R	missense_variant	pathogenic	0,0001719	126
SI	3:165007927:A:C	rs142090504	1417	Y/*	stop_gained	HIGH	0,0001911	139
SI	3:164994281:G:A	rs376062850	1606	T/I	missense_variant	pathogenic	0,0002088	153
SI	3:165040949:A:T	rs188320908	717	V/D	missense_variant	pathogenic	0,0002129	156
SI	3:165019655:G:A	rs200451408	1124	R/*	stop_gained	HIGH	0,0003234	237
SI	3:165015986:A:G	rs375443860	1285	I/T	missense_variant	pathogenic	0,0003684	270
SI	3:165049235:T:A	rs376816463	536	D/V	missense_variant	pathogenic	0,0004040	296
SI	3:164998629:C:T	rs145246112	1484	R/H	missense_variant	pathogenic	0,0006782	496
SI	3:164983015:A:C	rs79717168	1745	F/C	missense_variant	pathogenic	0,0017646	1291
SI	3:165059937:C:T	rs138434001	371	V/M	missense_variant	pathogenic	0,0036433	2667
SI	3:165021265:C:T	rs121912616	1073	G/D	missense_variant	pathogenic	0,0039408	2885



TREH	11:118658679:C:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
TREH	11:118658689:A:C	-	530	Y/*	stop_gained	HIGH	0,0000014	1
TREH	11:118658706:C:G	-	525	G/R	missense_variant	pathogenic	0,0000014	1
TREH	11:118658734:C:A	rs782090708	-	-	splice_acceptor_variant	HIGH	0,0000014	1
TREH	11:118659396:GC:G	rs1358778941	469	A/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118659399:C:T	-	468	W/*	stop_gained	HIGH	0,0000014	1
TREH	11:118659409:G:A	-	465	P/S	missense_variant	pathogenic	0,0000014	1
TREH	11:118659414:T:C	-	463	D/G	missense_variant	pathogenic	0,0000014	1
TREH	11:118660667:C:A	-	325	R/L	missense_variant	pathogenic	0,0000014	1
TREH	11:118660674:A:G	-	323	S/P	missense_variant	pathogenic	0,0000014	1
TREH	11:118660700:G:A	-	314	A/V	missense_variant	pathogenic	0,0000014	1
TREH	11:118660734:C:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
TREH	11:118660901:C:CT	-	291	S/KX	frameshift_variant	HIGH	0,0000014	1
TREH	11:118661163:G:A	rs782679595	285	P/L	missense_variant	pathogenic	0,0000014	1
TREH	11:118661243:C:G	-	258	W/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118661284:T:G	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
TREH	11:118661392:C:T	-	-	-	splice_donor_variant	HIGH	0,0000014	1
TREH	11:118661474:T:C	-	218	Y/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118661486:C:T	-	214	G/E	missense_variant	pathogenic	0,0000014	1
TREH	11:118661497:GA:G	-	210	V/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118661694:G:A	rs1446204296	187	S/L	missense_variant	pathogenic	0,0000014	1
TREH	11:118661914:C:T	rs529472872	167	G/D	missense_variant	pathogenic	0,0000014	1
TREH	11:118661920:G:A	-	165	P/L	missense_variant	pathogenic	0,0000014	1
TREH	11:118662901:A:G	-	135	W/R	missense_variant	pathogenic	0,0000014	1
TREH	11:118662923:C:T	-	127	W/*	stop_gained	HIGH	0,0000014	1
TREH	11:118662957:AG:A	-	116	L/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118663060:C:G	-	109	W/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118663061:C:T	rs782735682	109	W/*	stop_gained	HIGH	0,0000014	1

TREH	11:118663077:A:G	-	104	W/R	missense_variant	pathogenic	0,0000014	1
TREH	11:118663404:A:C	-	42	V/G	missense_variant	pathogenic	0,0000014	1
TREH	11:118679545:C:T	-	28	C/Y	missense_variant	pathogenic	0,0000014	1
TREH	11:118679550:TG:T	-	26	P/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118658292:C:T	-	583	W/*	stop_gained	HIGH	0,0000014	1
TREH	11:118658398:TC:T	-	548	D/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118658432:C:G	-	537	G/R	missense_variant	pathogenic	0,0000014	1
TREH	11:118658435:A:G	-	536	F/L	missense_variant	pathogenic	0,0000014	1
TREH	11:118658437:C:T	-	535	G/E	missense_variant	pathogenic	0,0000014	1
TREH	11:118658442:C:T	-	-	-	splice_acceptor_variant	HIGH	0,0000014	1
TREH	11:118661957:A:AC	-	152-153	-/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118661968:TG:T	-	149	H/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118662900:C:T	-	135	W/*	stop_gained	HIGH	0,0000014	1
TREH	11:118663359:A:G	-	57	M/T	missense_variant	pathogenic	0,0000014	1
TREH	11:118659019:T:C	rs782373744	-	-	splice_acceptor_variant	HIGH	0,0000014	1
TREH	11:118658915:ATGGCTGAC:A	-	509-512	KSAM/ NX	frameshift_variant	HIGH	0,0000014	1
TREH	11:118659754:T:C	-	438	Y/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118659793:C:T	-	425	C/Y	missense_variant	pathogenic	0,0000014	1
TREH	11:118659866:C:G	rs199814175	401	D/H	missense_variant	pathogenic	0,0000014	1
TREH	11:118659870:C:G	-	399	W/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118659892:C:T	-	392	W/*	stop_gained	HIGH	0,0000014	1
TREH	11:118659899:CTG:C	rs1555144516	389	T/X	frameshift_variant	HIGH	0,0000014	1
TREH	11:118659920:G:A	rs1403388683	383	R/C	missense_variant	pathogenic	0,0000014	1
TREH	11:118659745:A:G	-	-	-	splice_donor_variant	HIGH	0,0000014	1
TREH	11:118659753:G:T	-	438	Y/*	stop_gained	HIGH	0,0000014	1
TREH	11:118659390:A:C	-	471	L/R	missense_variant	pathogenic	0,0000014	1
TREH	11:118659390:A:G	-	471	L/P	missense_variant	pathogenic	0,0000014	1
TREH	11:118659390:AG:A	-	471	L/X	frameshift_variant	HIGH	0,0000014	1

TREH	11:118661409:C:G	-	240	D/H	missense_variant	pathogenic	0,0000027	2
TREH	11:118661475:A:C	-	218	Y/D	missense_variant	pathogenic	0,0000027	2
TREH	11:118663106:A:G	-	94	F/S	missense_variant	pathogenic	0,0000027	2
TREH	11:118663380:T:C	rs1555145376	50	D/G	missense_variant	pathogenic	0,0000027	2
TREH	11:118658427:C:A	rs782543423	538	W/C	missense_variant	pathogenic	0,0000027	2
TREH	11:118658435:A:T	-	536	F/I	missense_variant	pathogenic	0,0000027	2
TREH	11:118660698:C:T	rs374895583	315	G/R	missense_variant	pathogenic	0,0000027	2
TREH	11:118659925:G:T	rs1555144540	381	S/*	stop_gained	HIGH	0,0000027	2
TREH	11:118659799:G:T	-	423	A/D	missense_variant	pathogenic	0,0000027	2
TREH	11:118659903:GT:G	rs781960542	388	N/X	frameshift_variant	HIGH	0,0000027	2
TREH	11:118663423:C:T	rs782398259	36	G/R	missense_variant	pathogenic	0,0000028	2
TREH	11:118661408:T:C	rs1555144937	240	D/G	missense_variant	pathogenic	0,0000041	3
TREH	11:118661487:C:T	rs1555144958	214	G/R	missense_variant	pathogenic	0,0000041	3
TREH	11:118661694:G:C	rs1446204296	187	S/*	stop_gained	HIGH	0,0000041	3
TREH	11:118660558:G:T	-	361	S/R	missense_variant	pathogenic	0,0000041	3
TREH	11:118660662:G:GC	-	326-327	-/X	frameshift_variant	HIGH	0,0000041	3
TREH	11:118658434:A:G	rs782590281	536	F/S	missense_variant	pathogenic	0,0000041	3
TREH	11:118659398:C:T	rs782178104	468	W/*	stop_gained	HIGH	0,0000055	4
TREH	11:118663353:A:G	-	59	L/P	missense_variant	pathogenic	0,0000055	4
TREH	11:118662880:C:A	-	-	-	splice_donor_variant	HIGH	0,0000055	4
TREH	11:118659764:C:G	rs1245870739	435	A/P	missense_variant	pathogenic	0,0000055	4
TREH	11:118659893:A:C	-	392	W/G	missense_variant	pathogenic	0,0000055	4
TREH	11:118659384:T:TCCTGCAGGGGGG CCCAGGC	-	473	D/GLG PPAGX	frameshift_variant	HIGH	0,0000068	5
TREH	11:118661727:T:G	-	176	D/A	missense_variant,splice_region_variant	pathogenic	0,0000068	5
TREH	11:118660667:C:T	rs782048237	325	R/H	missense_variant	pathogenic	0,0000068	5
TREH	11:118661909:A:G	-	169	F/L	missense_variant	pathogenic	0,0000069	5
TREH	11:118663422:CCGTGGCAGTAAAT CTCACTGCAGAGA:C	rs527655595	?-36	-	splice_acceptor_variant,coding_sequence_variant,intro n_variant	HIGH	0,0000069	5

TREH	11:118658724:T:TGACGTCATACTG GG	rs1565522858	518-519	- /PSMT X	frameshift_variant	HIGH	0,0000082	6
TREH	11:118661465:C:T	rs376837165	221	R/Q	missense_variant	pathogenic	0,0000082	6
TREH	11:118662909:T:C	rs1162438659	132	H/R	missense_variant	pathogenic	0,0000082	6
TREH	11:118660623:G:A	rs147240748	340	R/*	stop_gained	HIGH	0,0000082	6
TREH	11:118658425:G:A	rs782243802	539	T/M	missense_variant	pathogenic	0,0000082	6
TREH	11:118661889:C:T	rs1555145077	-	-	splice_donor_variant	HIGH	0,0000082	6
TREH	11:118659825:G:C	rs377041843	414	Y/*	stop_gained	HIGH	0,0000082	6
TREH	11:118658705:C:T	rs191186222	525	G/D	missense_variant	pathogenic	0,0000096	7
TREH	11:118661466:G:A	rs782204994	221	R/W	missense_variant	pathogenic	0,0000096	7
TREH	11:118663339:C:A	-	64	E/*	stop_gained,splice_region_variant	HIGH	0,0000096	7
TREH	11:118658393:A:C	rs1378486820	550	Y/D	missense_variant	pathogenic	0,0000096	7
TREH	11:118659444:G:A	rs781841527	453	T/I	missense_variant	pathogenic	0,0000109	8
TREH	11:118658952:G:A	rs782110462	500	R/*	stop_gained	HIGH	0,0000109	8
TREH	11:118658951:C:G	rs782721212	500	R/P	missense_variant	pathogenic	0,0000123	9
TREH	11:118658974:G:GCTTT	rs782212660	492	F/LKX	frameshift_variant	HIGH	0,0000123	9
TREH	11:118658954:A:T	rs782805459	499	I/N	missense_variant	pathogenic	0,0000136	10
TREH	11:118661483:C:T	rs541953573	215	R/H	missense_variant	pathogenic	0,0000136	10
TREH	11:118661484:G:A	rs535722007	215	R/C	missense_variant	pathogenic	0,0000136	10
TREH	11:118659368:A:G	rs528258475	-	-	splice_donor_variant	HIGH	0,0000136	10
TREH	11:118661646:A:G	rs1555145005	203	L/P	missense_variant	pathogenic	0,0000150	11
TREH	11:118663051:C:A	rs782403674	-	-	splice_donor_variant	HIGH	0,0000150	11
TREH	11:118660604:G:C	rs533985931	346	P/R	missense_variant	pathogenic	0,0000164	12
TREH	11:118659953:G:A	-	372	Q/*	stop_gained	HIGH	0,0000164	12
TREH	11:118661483:C:A	rs541953573	215	R/L	missense_variant	pathogenic	0,0000232	17
TREH	11:118661636:C:T	rs782414721	-	-	splice_donor_variant	HIGH	0,0000287	21
TREH	11:118660668:G:A	rs574707283	325	R/C	missense_variant	pathogenic	0,0000300	22
TREH	11:118661911:C:T	rs782377538	168	R/H	missense_variant	pathogenic	0,0000327	24

TREH	11:118660673:G:A	rs1403604307	323	S/F	missense_variant	pathogenic	0,0000368	27
TREH	11:118659866:C:T	rs199814175	401	D/N	missense_variant	pathogenic	0,0000464	34
TREH	11:118661667:A:G	rs147473566	196	M/T	missense_variant	pathogenic	0,0000519	38
TREH	11:118658976:A:G	rs374204865	492	F/L	missense_variant	pathogenic	0,0000641	47
TREH	11:118659390:A:AG	rs782422052	471	L/PX	frameshift_variant	HIGH	0,0000846	62
TREH	11:118661912:G:A	rs781984077	168	R/C	missense_variant	pathogenic	0,0000914	67
TREH	11:118659447:G:A	rs782653438	452	P/L	missense_variant	pathogenic	0,0001023	75
TREH	11:118659854:CAA:C	rs1224861159	404	L/X	frameshift_variant	HIGH	0,0001405	103
TREH	11:118658904:C:A	rs377365396	-	-	splice_donor_variant	HIGH	0,0001542	112
TREH	11:118663404:A:G	rs200713103	42	V/A	missense_variant	pathogenic	0,0001706	125
TREH	11:118661453:G:A	rs782245431	225	P/L	missense_variant	pathogenic	0,0001910	140
TREH	11:118658915:A:G	rs556006762	512	M/T	missense_variant	pathogenic	0,0003084	226
TREH	11:118658925:T:A	rs201467126	509	K/*	stop_gained	HIGH	0,0017534	1284

**Gene:** gene symbol; **Genomic position and variation** (GRCh38): genomic coordinates (hg38 genome build); **dbSNP:** SNP ID (rsID); **aa position:** protein residue affected by the amino acid change; **aa change:** reference/hypomorphic amino acid; **Consequence:** functional consequence at the protein level; **VEP/AlphaMissense classification:** variant annotation as hypomorphic based on VEP HIGH impact rating and AlphaMissense pathogenic score; **UKBB AF:** allele frequency in UKBB; **Ncarriers:** number of carriers.