**Supplementary Table 1.** Summary of variants in *CHST6* gene.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Nucleotide change (NM\_021615.4)** | **Amino acid change** | **Genomic coordinates (GRCh37/hg19）** | **Remarks** | **Nature of effect** | **gnomAD frequency** | **Number of families**  **/individuals** | **ACMG classification** | **Reference** |
| c.1A>T | M1L | 16:75513726 | rs755563003 | missense | 5.09E-05 | 14 | VUS | (Klintworth et al., 2006; Gruenauer-Kloevekorn et al., 2008; Liskova et al., 2008; Dudakova et al., 2014; Nowinska et al., 2014; Yaylacioglu Tuncay et al., 2016) |
| **c.6G>A** | W2X | 16:75513721 |  | nonsense | 4.67E-05 | 2 | P | (Sultana et al., 2005; Sultana et al., 2009) |
| c.7C>A | L3M | 16:75513720 |  | missense | 4.67E-05 | 2 | VUS | (Sultana et al., 2005; Sultana et al., 2009) |
| c.13C>T | R5C | 16:75513714 |  | missense | **-** | **1** | VUS | (Dudakova et al., 2014) |
| **-** | V6fs | 16:75513712 |  | frameshift | 1.29E-05 | 7 | P | (Liu et al., 2000; Sultana et al., 2003; Warren et al., 2003; Sultana et al., 2005; Liu et al., 2006) |
| c.44T>C | L15P | 16:75513683 |  | missense | **-** | **1** | VUS | (Niel et al., 2003) |
| c.51delG | Q18fs | 16:75513676 |  | frameshift | 9.84E-06 | 1 | P | (Klintworth et al., 2006) |
| c.52C>T | Q18X | 16:75513675 |  | nonsense | **-** | **2** | P | (Sultana et al., 2003; Sultana et al., 2009) |
| c.61-62insA, 62T>G | F20fs | 16:75513666 |  | frameshift | **-** | **3** | P | (Liu et al., 2010; Wang et al., 2017) |
| c.65T>C | L22P | 16:75513666 |  | missense | **-** | **1** | LP | (El-Ashry et al., 2010) |
| c.65T>G | L22R | 16:75513662 |  | missense | **-** | **2** | P | (Warren et al., 2003; Paliwal et al., 2012) |
| c.91C>T | P31S | 16:75513636 |  | missense | **-** | **3** | LP | (El-Ashry et al., 2002; Klintworth et al., 2006) |
| c.92C>T | P31L | 16:75513635 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| c.97-100delTCCC | P31fs | 16:75513627-75513630 |  | frameshift | **-** | **1** | P | (Sultana et al., 2003) |
| c.95C>A | S32X | 16:75513632 |  | nonsense | **-** | **2** | P | (Liu et al., 2010; Park et al., 2015) |
| - | S32fs | 16:75513632 |  | frameshift | **-** | **1** | P | (Sultana et al., 2009) |
| c.124C>T | H42Y | 16:75513603 |  | missense | **-** | **1** | LP | (Warren et al., 2003) |
| c.137T>C | L46P | 16:75513590 |  | missense | **-** | **1** | LP | (Klintworth et al., 2006) |
| c.143C>A | S48X | 16:75513584 |  | nonsense | **-** | **1** | P | (Birgani et al., 2009) |
| c.148C>T | R50C | 16:75513579 |  | missense | **-** | **3** | P | (Akama et al., 2000; Warren et al., 2003) |
| c.148C>A | R50S | 16:75513579 |  | missense | **-** | **1** | LP | (El-Ashry et al., 2010) |
| C.149G>T | R50L | 16:75513578 | rs778668750 | missense | 7.31E-06 | 2 | LP | (Warren et al., 2003) |
| C.152C>T | S51L | 16:75513575 | rs370335460 | missense | 7.31E-06 | 2 | LP | (Ha et al., 2003b; Aldave et al., 2004) |
| C.152C>A | S51X | 16:75513575 |  | nonsense | **-** | **1** | P | (Huo et al., 2011) |
| C.155G>A | G52D | 16:75513572 |  | missense | **-** | **4** | LP | (Sultana et al., 2003; Sultana et al., 2009) |
| c.158C>T | S53L | 16:75513569 |  | missense | **-** | **9** | LP | (Sultana et al., 2003; Warren et al., 2003; Klintworth et al., 2006; Sultana et al., 2009) |
| c.161C>A | S54Y | 16:75513566 |  | missense | **-** | **1** | LP | (Paliwal et al., 2012) |
| c.161C>T | S54F | 16:75513566 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.164 T>C | F55S | 16:75513563 |  | missense | **-** | **1** | LP | (Yaylacioglu Tuncay et al., 2016) |
| c.165C>A | F55L | 16:75513562 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| c.166G>A;c.167T>G | V56R | 16:75513560-75513561 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.172C>T | Q58X | 16:75513555 | rs756036451 | nosense | 1.23E-05 | 2 | P | (Niel et al., 2003; Paliwal et al., 2012) |
| c.173A>G | Q58R | 16:75513554 |  | missense | **-** | **1** | LP | (Paliwal et al., 2012) |
| C.176T>C | L59P | 16:75513551 |  | missense | **-** | **2** | P | (Ha et al., 2003b) |
| c.176T>A | L59H | 16:75513551 |  | missense | **-** | **1** | LP | (Paliwal et al., 2012) |
| c.180delC | F60fs | 16:75513547 |  | frameshift | **-** | **6** | P | (Sultana et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| C.182A>C | N61T | 16:75513545 |  | missense | **-** | **1** | P | (Niel et al., 2003) |
| C.183C>A | N61K | 16:75513544 |  | missense | **-** | **2** | LP | (Liu et al., 2010) |
| c.189C>G | H63Q | 16:75513538 | rs150479531 | missense | 2.05E-05 | 1 | LP | (Klintworth et al., 2006) |
| c.191C>T | P64L | 16:75513536 |  | missense | 8.20E-06 | 2 | LP | (Nowinska et al., 2014; Rubinstein et al., 2016) |
| C.196G>C | V66L | 16:75513531 |  | missense | **-** | **1** | P | (Ha et al., 2003b) |
| C.196G>T | V66F | 16:75513531 | rs72547547 | missense | 8.20E-06 | 1 | LP | (Gulias-Canizo et al., 2006) |
| C.198delC | V66fs | 16:75513529 |  | frameshift | **-** | **7** | P | (Sultana et al., 2003; Warren et al., 2003) |
| - | F67Sfs | 16:75513526 |  | frameshift | **-** | **1** | P | (Sultana et al., 2009) |
| c.202T>C | Y68H | 16:75513525 | rs775742450 | missense | 8.20E-06 | 1 | LP | (Niel et al., 2003) |
| c.208A>T | M70L | 16:75513519 |  | missense | **-** | **1** | LP | (Niel et al., 2003) |
| c.211G>C | E71Q | 16:75513516 |  | missense | **-** | **1** | LP | (Carstens et al., 2016) |
| c.214C>T | P72S | 16:75513513 | rs377617168 | missense | 4.10E-06 | 2 | LP | (El-Ashry et al., 2002; Aldave et al., 2004) |
| c.217G>C | A73P | 16:75513510 |  | missense | **-** | **1** | LP | (Klintworth et al., 2006) |
| c.217G>A | A73T | 16:75513510 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.218C>T | A73V | 16:75513509 | rs776750339 | missense | 4.10E-06 | 2 | LP | (Liu et al., 2010) |
| c.226G>A | V76M | 16:75513501 |  | missense | **-** | **1** | LP | (Ha et al., 2003b) |
| c.231G>A | W77X | 16:75513496 |  | missense | 1.23E-05 | 1 | P | (Klintworth et al., 2006) |
| c.231G>C | W77C | 16:75513496 |  | missense | 3.23E-05 | 1 | LP | (Aldave et al., 2004) |
| c.244C>T | Q82X | 16:75513483 |  | nosense | **-** | **3** | P | (Ha et al., 2003a; Ha et al., 2003b; Niel et al., 2003) |
| c.271\_273delGCTinsA | A91fs | 16:75513454-75513456 | rs776082747 | frameshift | 7.26E-06 | 2 | P | (Niel et al., 2003; Klintworth et al., 2006) |
| c.274G>C | V92L | 16:75513453 |  | missense | 4.09E-06 | 1 | LP | (Klintworth et al., 2006) |
| c.277C>A | R93S | 16:75513450 |  | missense | **-** | **3** | LP | (Aldave et al., 2004; Klintworth et al., 2006) |
| c.278G>A | R93H | 16:75513449 |  | missense | **-** | **4** | P | (Warren et al., 2003; Paliwal et al., 2012) |
| c.289C>T | R97C | 16:75513438 |  | missense | 4.09E-06 | 1 | LP | (Dudakova et al., 2014) |
| c.290G>C | R97P | 16:75513437 |  | missense | **-** | **2** | P | (Warren et al., 2003; Paliwal et al., 2012) |
| c.290-291 ins G | S98fs | 16:75513436-75513437 |  | frameshift | **-** | **1** | P | (Wang et al., 2017) |
| c.293C>G;c.294C>G | S98W | 16:75513433-75513434 |  | missense | **-** | **2** | LP | (Sultana et al., 2003; Sultana et al., 2009) |
| c.293C>T;c.294C>G | S98L | 16:75513433-75513434 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.304T>G | C102G | 16:75513423 | rs121917822 | missense | 1.45E-05 | 4 | LP | (Niel et al., 2003; Abbruzzese et al., 2004; Aldave et al., 2004) |
| c.305G>A | C102Y | 16:75513422 |  | missense | **-** | **2** | P | (Warren et al., 2003) |
| c.310A>G | M104V | 16:75513417 |  | missense | **-** | **1** | LP | (Aldave et al., 2004) |
| c.320T>C | F107S | 16:75513407 | rs72547545 | missense | 2.86E-05 | 7 | LP | (El-Ashry et al., 2002; Sultana et al., 2003; Sultana et al., 2009) |
| c.329A>G | Y110C | 16:75513398 | rs72547544 | missense | 4.09E-06 | 4 | LP | (Aldave et al., 2004; Gonzalez-Rodriguez et al., 2014; Nowinska et al., 2014) |
| c.340C>T | R114C | 16:75513387 | rs146440274 | missense | 3.69E-05 | 1 | VUS | (Klintworth et al., 2006) |
| c.353C>T | S118F | 16:75513374 |  | missense | **-** | **1** | LP | (Park et al., 2015) |
| c.362T>C | F121S | 16:75513365 |  | missense | **-** | **1** | LP | (Sultana et al., 2009) |
| c.363\_364insC | F121fs | 16:75513363-75513364 |  | frameshift | **-** | **1** | P | (Niel et al., 2003) |
| c.363C>G | F121L | 16:75513364 |  | missense | 8.19E-06 | 2 | P | (Sultana et al., 2003; Klintworth et al., 2006) |
| c.365A>C | Q122P | 16:75513362 | rs758105699 | missense | 4.10E-06 | 3 | LP | (Aldave et al., 2004; Klintworth et al., 2006) |
| c.369G>A | W123X | 16:75513358 |  | nonsense | **-** | **5** | P | (Sultana et al., 2003; Sultana et al., 2005; Sultana et al., 2009; Paliwal et al., 2012) |
| c.375\_376ins(GGCCGTG) | V125fs | 16:75513351-75513352 |  | frameshift | **-** | **4** | P | (Ha et al., 2003a; Ha et al., 2003b) |
| c.379C>T | R127C | 16:75513348 |  | missense | **-** | **4** | LP | (Warren et al., 2003; Birgani et al., 2009) |
| c.382G>A | A128T | 16:75513345 |  | missense | **-** | **1** | LP | (Wang et al., 2017) |
| c.383C>T | A128V | 16:75513344 |  | missense | **-** | **14** | P | (Liu et al., 2006) |
| c.391T>C | S131P | 16:75513336 |  | missense | **-** | **3** | P | (Niel et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| c.392C>T | S131L | 16:75513335 | rs375059043 | missense | 4.01E-05 | 3 | LP | (Klintworth et al., 2006) |
| c.395C>T | P132L | 16:75513332 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| c.401C>A | A134D | 16:75513326 |  | missense | 4.10E-06 | 1 | LP | (Liu et al., 2010) |
| c.406A>G | S136G | 16:75513321 |  | missense | **-** | **1** | VUS | (Birgani et al., 2009) |
| c.414insTT | A137fs | 16:75513313 |  | frameshift | **-** | **1** | P | (Akama et al., 2000) |
| c.418C>T | R140X | 16:75513309 | rs757912368 | nonsense | 8.21E-06 | 7 | P | (El-Ashry et al., 2005; Liu et al., 2005; Liu et al., 2010; Wang et al., 2017) |
| c.430A>C | S144R | 16:75513297 |  | missense | **-** | **2** | LP | (Liu et al., 2010; Wang et al., 2017) |
| c.445T>G | C149G | 16:75513281 |  | missense | **-** | **1** | LP | (Liu et al., 2010) |
| c.446G>A | C149Y | 16:75513281 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| - | C149fs | 16:75513280 |  | frameshift | 4.13E-06 | 1 | P | (Gruenauer-Kloevekorn et al., 2008) |
| c.455T>C | L152P | 16:75513272 |  | missense | **-** | **1** | LP | (Niel et al., 2003) |
| c.459C>A | C153X | 16:75513268 |  | nonsense | **-** | **3** | P | (Sultana et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| c.463-464 del CG | R155Afs | 16:75513262 | rs141905571 | frameshift | 3.23E-05 | 2 | P | (Yaylacioglu Tuncay et al., 2016; Wang et al., 2017) |
| c.494G>A | C165Y | 16:75513233 |  | missense | **-** | **2** | LP | (Liskova et al., 2008; Birgani et al., 2009) |
| c.495C>G | C165W | 16:75513232 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.494G>C;c.495C>T | C165S | 16:75513232-75513233 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.497G>C | R166P | 16:75513230 |  | missense | **-** | **3** | LP | (Liu et al., 2000; Niel et al., 2003) |
| c.500C>T | S167F | 16:75513227 | rs756399261 | missense | 8.37E-05 | 2 | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.517C>T | L173F | 16:75513210 | rs764287174 | missense | 4.09E-06 | 1 | LP | (Gruenauer-Kloevekorn et al., 2008) |
| c.518T>C | L173P | 16:75513209 | rs763075517 | missense | 1.23E-05 | 1 | LP | (Yellore et al., 2007) |
| c.521A>G | K174R | 16:75513206 |  | missense | **-** | **4** | LP | (Akama et al., 2000; Park et al., 2015) |
| c.526G>A | V176M | 16:75513201 |  | missense | **-** | **3** | LP | (Yaylacioglu Tuncay et al., 2016) |
| c.529C>T | R177C | 16:75513198 | rs760489234 | missense | 2.86E-05 | 2 | LP | (Klintworth et al., 2006) |
| c.529C>G | R177G | 16:75513198 |  | missense | **-** | **1** | LP | (Patel et al., 2011) |
| c.530G>A | R177H | 16:75513197 |  | missense | **-** | **1** | P | (Iida-Hasegawa et al., 2003) |
| c.533T>G | F178C | 16:75513194 |  | missense | **-** | **1** | LP | (Sultana et al., 2005) |
| c.545delA | Q182fs | 16:75513182 |  | frameshift | **-** | **14** | P | (Warren et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| c.557C>G | P186R | 16:75513170 | rs376162109 | missense | 2.55E-05 | 2 | LP | (Patel et al., 2011; Park et al., 2015) |
| c.573\_574insC | A192fs | 16:75513153 | rs570082634 | frameshift | 7.30E-06 | 1 | P | (Klintworth et al., 2006) |
| c.578T>C | L193P | 16:75513149 |  | missense | **-** | **2** | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.581ACCTAC>GGT | 194 Asn Leu Arg 196; 194 Arg Cys 195 | 16:75513146 |  | replacement mutation | **-** | **2** | VUS | (Warren et al., 2003) |
| c.587insACG | R195-196ins | 16:75513140 |  | Insertation | **-** | **2** | VUS | (Sultana et al., 2003; Sultana et al., 2009) |
| c.593T>A | V198E | 16:75513134 |  | missense | **-** | **1** | LP | (Abbruzzese et al., 2004) |
| c.599T>G | L200R | 16:75513128 | rs28937879 | missense | 0.0002205 | 37 | LP | (El-Ashry et al., 2002; Niel et al., 2003; Abbruzzese et al., 2004; Aldave et al., 2004; El-Ashry et al., 2005; Klintworth et al., 2006; Gruenauer-Kloevekorn et al., 2008; Liskova et al., 2008; Dudakova et al., 2014; Nowinska et al., 2014) |
| c.601\_621delGTGCGCGACCCGCGGGCCGTG | deletion from aa201-207 | 16:75513106-75513126 |  | deletion | **-** | **1** | VUS | (Gulias-Canizo et al., 2006) |
| c.604C>A | R202S | 16:75513123 |  | missense | **-** | **2** | P | (Sultana et al., 2003; Sultana et al., 2005) |
| c.606\_607delGC | R202fs | 16:75513120-75513121 |  | frameshift | **-** | **1** | P | (El-Ashry et al., 2010) |
| c.607G>A | D203N | 16:75513120 |  | missense | **-** | **2** | LP | (Klintworth et al., 2006) |
| c.607G>T | D203Y | 16:75513120 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| c.609C>A | D203E | 16:75513118 |  | missense | **-** | **2** | P | (Akama et al., 2000) |
| c.610C>T | P204S | 16:75513117 |  | missense | **-** | **2** | LP | (Yaylacioglu Tuncay et al., 2016) |
| - | P204G | 16:75513116 |  | missense |  | 1 | LP | (Niel et al., 2003) |
| c.611C>A | P204Q | 16:75513116 | rs759870075 | missense | 3.23E-05 | 3 | P | (Iida-Hasegawa et al., 2003; Sultana et al., 2003; Sultana et al., 2009) |
| c.611C>G | P204R | 16:75513116 | rs759870075 | missense | 3.23E-05 | 2 | LP | (Sultana et al., 2005; Sultana et al., 2009) |
| c.612\_614delGCG insAT | P204fs | 16:75513113-75513115 |  | frameshift |  | 2 | P | (Sultana et al., 2003; Warren et al., 2003) |
| c.613C>T | R205W | 16:75513114 | rs750219546 | missense | 8.38E-06 | 8 | LP | (Liu et al., 2010; Park et al., 2015) |
| c.614G>A | R205Q | 16:75513113 | rs377706989 | missense | 1.26E-05 | 3 | P | (Warren et al., 2003; Liskova et al., 2008) |
| c.614G>T | R205L | 16:75513113 |  | missense | **-** | **1** | LP | (Iida-Hasegawa et al., 2003) |
| - | R205fs | 16:75513113 |  | frameshift | **-** | **1** | P | (Sultana et al., 2009) |
| c.616G>A | A206T | 16:75513111 | rs374493344 | missense | 8.39E-06 | 1 | LP | (Warren et al., 2003) |
| c.617C>T | A206V | 16:75513110 |  | missense | **-** | **1** | LP | (El-Ashry et al., 2002) |
| c.629C>T | S210F | 16:75513098 | rs745571211 | missense | 1.68E-05 | 2 | LP | (Sultana et al., 2003; Sultana et al., 2009) |
| c.631C>G | R211G | 16:75513096 |  | missense | **-** | **1** | LP | (Liu et al., 2010) |
| c.631C>T | R211W | 16:75513096 |  | missense | **-** | **8** | P | (Akama et al., 2000; Iida-Hasegawa et al., 2003; Yaylacioglu Tuncay et al., 2016) |
| c.632G>A | R211Q | 16:75513095 | rs771397083 | missense | 4.21E-06 | 12 | LP | (Ha et al., 2003a; Ha et al., 2003b; Gruenauer-Kloevekorn et al., 2008; Patel et al., 2011) |
| c.649G>A | A217T | 16:75513078 | rs752785520 | missense | 4.21E-06 | 2 | LP | (Iida-Hasegawa et al., 2003; Kobayashi et al., 2007) |
| c.656insCTG | W219-220ins | 16:75513071 |  | Insertion | **-** | **10** | VUS | (Sultana et al., 2003; Sultana et al., 2009) |
| c.661G>T | D221Y | 16:75513066 |  | missense | **-** | **6** | LP | (Sultana et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| c.663C>G | D221E | 16:75513064 |  | missense | **-** | **10** | P | (Sultana et al., 2003; Sultana et al., 2005; Sultana et al., 2009) |
| c.668G>A | G223D | 16:75513059 |  | missense | **-** | **1** | LP | (Liu et al., 2005) |
| c.682A>G; 683C>A | T228D | 16:75513044-75513045 |  | missense | **-** | **1** | VUS | (Klintworth et al., 2006) |
| c.696G>A | W232X | 16:75513031 |  | nonsense | **-** | **3** | P | (Ha et al., 2003b; Huo et al., 2011) |
| c.730G>T | E244X | 16:75512997 |  | nonsense | **-** | **3** | P | (Liu et al., 2010; Wang et al., 2017) |
| c.738C>G | C246W | 16:75512989 |  | missense | 3.24E-05 | 2 | LP | (Klintworth et al., 2006; Yaylacioglu Tuncay et al., 2016) |
| c.740delG | A247fs | 16:75512987 |  | frameshift | **-** | **1** | P | (Klintworth et al., 2006) |
| c.744C>G | S248R | 16:75512983 | rs760287510 | missense | 4.29E-06 | 1 | LP | (Klintworth et al., 2006) |
| c.746A>C | H249P | 16:75512981 | rs72547540 | missense | 3.24E-05 | 1 | P | (Warren et al., 2003) |
| c.746A>G | H249R | 16:75512981 |  | missense | **-** | **1** | LP | (Birgani et al., 2009) |
| c.760G>T | E254X | 16:75512967 |  | nonsense | **-** | **1** | P | (Liu et al., 2010) |
| c.786delC | L264fs | 16:75512941 |  | frameshift | **-** | **1** | P | (Park et al., 2015) |
| c.803A>G | Y268C | 16:75512924 | rs72547539 | missense | 6.64E-05 | 4 | LP | (Ha et al., 2003b; Liu et al., 2010) |
| c.814C>A | R272S | 16:75512913 | rs769833756 | missense | 4.15E-06 | 4 | LP | (Sultana et al., 2005; Sultana et al., 2009; Liu et al., 2010) |
| c.815G>A | R272H | 16:75512912 | rs745966069 | missense | 4.15E-06 | 1 | LP | (Klintworth et al., 2006) |
| c.820G>A | E274K | 16:75512907 | rs72547538 | missense | 4.14E-06 | 4 | P | (Akama et al., 2000; Warren et al., 2003; El-Ashry et al., 2010; Park et al., 2015) |
| c.820G>C | E274Q | 16:75512907 |  | missense | **-** | **1** | LP | (Paliwal et al., 2012) |
| c.827T>C | L276P | 16:75512900 | rs121917824 | missense | 5.49E-05 | 5 | LP | (Aldave et al., 2004; El-Ashry et al., 2005; Sultana et al., 2005; Sultana et al., 2009) |
| c.877C>T | L293F | 16:75512850 |  | missense | **-** | **2** | LP | (Paliwal et al., 2012) |
| c.891-892insC | P297fs | 16:75512835-75512836 |  | frameshift | **-** | **1** | P | (Birgani et al., 2009) |
| c.892C>T | Q298X | 16:75512835 |  | nonsense | **-** | **9** | P | (Dang et al., 2009; Liu et al., 2010; Wang et al., 2017) |
| c.894\_895 insG | Q298fs | 16:75512832-16:75512833 |  | frameshift | **-** | **5** | P | (Yaylacioglu Tuncay et al., 2016) |
| c.922C>T | H308Y | 16:75512805 |  | missense | **-** | **1** | LP | (Park et al., 2015) |
| c.925G>T | G309X | 16:75512802 |  | nonsense | **-** | **2** | LP | (Sultana et al., 2003; Sultana et al., 2009) |
| c.985G>C | V329L | 16:75512742 |  | missense | **-** | **4** | LP | (Liu et al., 2006) |
| c.991C>T | Q331X | 16:75512736 |  | nonsense | **-** | **1** | LP | (Aldave et al., 2004) |
| c.993G>T | Q331H | 16:75512734 | rs140699573 | missense | 0.003734 | 1 | LP | (Liu et al., 2005) |
| c.997T>G | W333G | 16:75512730 |  | missense | **-** | **1** | LP | (Liu et al., 2010) |
| c.1000C>T | R334C | 16:75512727 |  | missense | 4.07E-06 | 6 | LP | (Sultana et al., 2005; Gulias-Canizo et al., 2006; Klintworth et al., 2006; Sultana et al., 2009) |
| - | H335fs | 16:75512724 |  | frameshift | **-** | **2** | VUS | (Sultana et al., 2009) |
| c.1039G>T | E347X | 16:75512688 |  | nonsense | **-** | **3** | LP | (Sultana et al., 2003; Sultana et al., 2009) |
| c.1043\_1044delTG | C349fs | 16:75512683-75512684 |  | frameshift | **-** | **1** | VUS | (Gruenauer-Kloevekorn et al., 2008) |
| c.1045T>A | C349S | 16:75512682 |  | missense | **-** | **2** | LP | (Liu et al., 2010) |
| c.1046G>A | C349Y | 16:75512681 | rs747897373 | missense | 1.22E-05 | 4 | LP | (Klintworth et al., 2006; Dudakova et al., 2014) |
| c.1044\_1051dupGTGCGCTG | L353fs | 16:75512676-75512683 |  | frameshift | **-** | **1** | VUS | (Aldave et al., 2004) |
| c.1056\_1078delGCTGCAGCTGCTGGGCTACCGGC | A352fs | 16:75512649-75512671 |  | frameshift | **-** | **1** | VUS | (Warren et al., 2003) |
| c.1072T>C | Y358H | 16:75512655 |  | missense | **-** | **11** | LP | (Dang et al., 2009; Liu et al., 2010; Park et al., 2015; Wang et al., 2017) |
| c.1072T>G | Y358D | 16:75512655 |  | missense | **-** | **1** | P | (El-Ashry et al., 2005) |
| c.1089\_1090delTG | E364fs | 16:75512637-75512638 |  | frameshift | **-** | **1** | VUS | (Rubinstein et al., 2016) |
| Deletion Including ORF |  |  |  | entire |  | 7 | P | (Akama et al., 2000; Warren et al., 2003; Klintworth et al., 2006; Young et al., 2007; El-Ashry et al., 2010) |
| Replacement of 5'region |  |  |  | entire |  | 3 | P | (Akama et al., 2000) |
| Deletion of 5'region |  |  |  | entire |  | 3 | P | (Abbruzzese et al., 2004) |

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