**Supplementary Table 1. Mutations detected in the *LDLR* gene in Russia**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Exon No. | Nucleotide substitutionNM\_000527.4(LDLR) | HGVS nomenclature NG\_009060.1(LDLR)(LRG 274) | Genomic location GRCh38 (Chr19) p12 | Predicted effect on protein | HGMD Public Accession | No. of families | ClinVar estimate of pathogenicity, Accession | dbSNP rs number | Populations in Russia | References for Russia | Other populations in the world |
| **Nonsense mutations** |
| 2 | c.85A>TAGA>TGA Originally erroneously described c.85T>A | g.15860A>T | 11100240 | p.R29\*[p.Arg29Ter, R8X]Originally erroneously described as E8X | CM045798Disease causing mutation | 1 | PathogenicAccession: VCV 000251011.1 | [rs879254401](https://www.ncbi.nlm.nih.gov/snp/rs879254401) | Moscow | Meshkov et al., 2004, 2009 | None |
| 2 | с.97C>TCAG>TAG | g.15872C>T | 11100252 | p.Q33\*[p.Gln33Ter Q12X, FH Turkey] | CM920404Disease causing mutation | 1 | PathogenicAccession: VCV 000003683.4 | [rs121908024](https://www.ncbi.nlm.nih.gov/snp/rs121908024) | St. Petersburg | Zakharova et al., 2005, 2007 | France, Italy, Spain, Turkey |
| 3 | c.285C>ATGC>TGA | g.18378C>A | 11102758 | p.C95\*[p.Cys95Ter, C74X] | CM983442Disease causing mutation | 1 | PathogenicAccession: VCV 000251115.1 | [rs139400379](https://www.ncbi.nlm.nih.gov/snp/rs139400379) | St. Petersburg | Zakharova et al., 2001, 2007 | Japan, Korea |
| 4 | c.495G>ATGG>TGA | g.21021G>A | 11105401 | p.W165\*[p.Trp165Ter, W144X] | CM067692Disease causing mutation | 1 | Pathogenic.Accession: VCV 000251254.1 | [rs879254546](https://www.ncbi.nlm.nih.gov/snp/rs879254546) | Moscow | Meshkov et al., 2009 | China, Greece |
| 4 | c.639G>ATGC>TGA | g.21219C>A | 11105599 | p.C231\*[p.Cys231Ter, C210X] | CM930475Disease causing mutation | 1 | Pathogenic/ Likely pathogenic. Accession: VCV 000003730.4 | [rs121908035](https://www.ncbi.nlm.nih.gov/snp/rs121908035) | Moscow | Meshkov et al., 2009 | Australia, Czech Republic, Great Britain, French Canadian, Ireland, Northern Poland, etc. |
| 4 | c.682G>TGAG>TAG | g.21208G>T | 11105588 | p.E228\*[p.Glu228Ter, E207X, FH Morocco] | CM920425Disease causing mutation | 1 | Pathogenic /Likely pathogenicAccession:VCV 000226333.8 | [rs121908029](https://www.ncbi.nlm.nih.gov/snp/rs121908029) | St. Petersburg | Zakharova et al., 2005, 2007 | China, Czech Republic, Germany, Great Britain, Korea, Morocco, The Netherlands,Norway, Sweden, USA |
| 5 | c.718G>TGAA>TAA | g.22208G>T | 11106588 | p.E240\* [p.Glu240Ter] | CM055368Disease causing mutation | N/A | PathogenicAccession: VCV 000251422.1 | [rs768563000](https://www.ncbi.nlm.nih.gov/snp/rs768563000) | Novosibirsk | Voevoda et al., 2008 | The Netherlands, USA |
| 5 | c.810G>ATGC>TGA | g.22300G>A | 11106680 | p.C270\*[p.Cys270Ter, C249X] | CM051098Disease causing mutation | 2 | PathogenicAccession: VCV 000251465.1 | [rs773328511](https://www.ncbi.nlm.nih.gov/snp/rs773328511) | St. Petersburg, Moscow | Zakharova et al., 2005, 2007; Meshkov et al., 2009 | The Netherlands |
| 9 | с.1252G>TGAG>TAG | g.28963G>T | 11113343 | p.E418\*[p.Glu418Ter, E418X, E397X,FH St Petersburg] | CM983997Disease causing mutation | 1 | PathogenicAccession:VCV 000251755.1 | [rs869320651](https://www.ncbi.nlm.nih.gov/snp/rs869320651) | St. Petersburg | Chakir et al., 1998 | Northern Italy |
| 9 | с.1328G>ATAG>TGG | g.29039G>A | 11113419 | p.W443\*[p.Trp443Ter, W422X] | CM051099Disease causing mutation | 2 | PathogenicVCV 000251789.1 | [rs879254866](https://www.ncbi.nlm.nih.gov/snp/rs879254866) | St. Petersburg | Zakharova et al., 2005, 2007 | Poland |
| 12 | c.1731G>ATGG>TGA | g.32504G>A | 11116884 | p.W577\*[p.Trp577Ter, W556X, FH Moscow] | CM920462Disease causing mutation | 1 | PathogenicAccession: VCV 000226370.1 | [rs875989928](https://www.ncbi.nlm.nih.gov/snp/rs875989928) | Moscow | Hobbs et al., 1992 | France, China |
| 15 | c.2215C>T | g.38868C>T | 11123248 | p.Q739\*[p.Gln739Ter] | CM024745 Disease causing mutation | 1 | Likely pathogenic/PathogenicAccession:VCV000252258.5 | N/A | Moscow | Semenova et al., 2020 | Italy, Mexico, Japan, Taiwan  |
| **Frameshift mutations** |
| 3 | c.193\_201 delinsGACTTCA | g.18286\_18294delins GACTTCA | 11102666-11102674 | p.Ser65fs[p.Ser65Asp fs\*64, FsS44:D108X] | CX138996Disease causing mutation | 1 | PathogenicAccession:VCV 000251072.1 | [rs879254434](https://www.ncbi.nlm.nih.gov/snp/rs879254434) | Petrozavodsk | Komarova et al., 2013b,c; Korneva et al., 2013 | None |
| 3 | c.195dupT[c.195\_196insT] | g.18288dup | 11102668 | p.Val66fs[p.Val66Cysfs\*64),FsV45: D108X] | CI136948 Disease causing mutation | 1 | PathogenicAccession:VCV 000251075.1 | [rs879254435](https://www.ncbi.nlm.nih.gov/snp/rs879254435) | Petrozavodsk | Komarova  et al., 2013b,c | None |
| 3 | c.230dupG [c.230\_231insG] | g.18323dup | 11102703 | p.Arg78fs[p.Arg78Pro fs\*52, G56fsX53] | CI045722,Disease causing mutation | 1 | N/A | [rs879254440](https://www.ncbi.nlm.nih.gov/snp/rs879254440) | Moscow | Meshkov et al., 2004, 2009 | None |
| 4 | с.351\_352 insTTCCOriginally described as c.352\_355 insTTCC | g.20877\_20878 insTTCC | 11105258 | p.Asp118fs[p.Asp118Phe fs\*13, D97FfsX13] | CI099861Disease causing mutation | 1 | N/A | N/A | Moscow | Meshkov et al., 2009 | None |
| 4 | c.355\_356insTTCC | g.20881\_20882ins | 11105261\_11105261 | p.(Gly119Valfs\*)[p.(Gly119Valfs\*12)] | N/ALikely pathogenic | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 4 | c.670dupG | g.21196dup | 11105575 | p.(Asp224fs)[p.(Asp224Glyfs\*4),FsK202: S206X] | CI051351Disease causing mutation | 1 | PathogenicAccession: VCV 000251372.1 | [rs879254629](https://www.ncbi.nlm.nih.gov/snp/rs879254629) | St. Petersburg | Zakharova et al., 2005, 2007 | None |
| 6 | [c.925\_931del](https://www.ncbi.nlm.nih.gov/projects/sviewer/?id=NM_000527.4&search=NM_000527.4:c.925_931del&v=1:100&content=5)CCCATCA | g.23119\_23125del | 11107499-11107505 | p.(Pro309fs)[p.(Pro309Lysfs\*59,FsE287:V348X, FH North Karelia] | CD920880Disease causing mutation | 2 | PathogenicAccession:VCV 000003729.3 | [rs387906304](https://www.ncbi.nlm.nih.gov/snp/rs387906304) | St. Petersburg, Petrozavodsk | Zakharova et al., 2005, 2007; Komarova et al., 2013a,b,c | Finland, Norway, Sweden, USA. Typical for North Karelia in Finland |
| 6 | [c.939\_940+3del](https://www.ncbi.nlm.nih.gov/nuccore/NM_000527.4?report=graph&mk=939|NM_000527.4\\:c.939_940+3del|green" \t "_blank)[c.936-940del5] | g.23133\_23137del | 11107510-11107514 | p.(Glu291fs\*)[ FsE291: N309X] | CD051317Disease causing mutation | 1 | PathogenicAccession: VCV 000251536.1Considered as variant affecting splicing | [rs879254727](https://www.ncbi.nlm.nih.gov/snp/rs879254727) | St. Petersburg | Zakharova et al., 2005, 2007 | None |
| 9 | [c.1291\_1331 del](https://www.ncbi.nlm.nih.gov/nuccore/NM_000527.4?report=graph&mk=1291|NM_000527.4\:c.1291_1331del|green)41 | g.29002\_29042del | 11113377-11113422 | p.(Ala431fs)[p.Ala431Ter, p.(Val430\_Ala431insTer), FsV409:S423X] | CG052715Disease causing mutation | 1 | PathogenicAccession: VCV 000251769.1 | [rs879254854](https://www.ncbi.nlm.nih.gov/snp/rs879254854) | St. Petersburg | Zakharova et al., 2005, 2007 | None |
| 9 | c.1303delG | g.29014del | 11113393 | p.(Glu435fs)[p.(Glu435Argfs\*16), FsE414:M429X] | CD132146Disease causing mutation | 1 | PathogenicAccession:VCV 000251776.1 | [rs879254857](https://www.ncbi.nlm.nih.gov/snp/rs879254857) | St. Petersburg | Zakharova et al., 2005, 2007 | Germany |
| 11 | c.1618delG | g.31745del | 11116125 | p.(Ala540fs)[p.(Ala540Profs\*8), A519PfsX8] | CD099865Disease causing mutation | 1 | N/A | N/A | Moscow | Meshkov et al., 2009 | None |
| 11 | c.1686\_1693delinsT | g.31813\_31820delinsT | 11116193-11116200 | p.(Trp562fs)[p.(Trp562Cysfs\*5), FsW541:L547X] | CX1313040Disease causing mutation | 1 | PathogenicAccession: VCV 000251968.1 | [rs879254984](https://www.ncbi.nlm.nih.gov/snp/rs879254984) | Petrozavodsk | Komarova et al., 2013c | None |
| 13 | c.1855\_1856insA | g.35721\_ 35722insA | 11120101-11120102 | p.(Phe619fs)[p.(Phe619Tyrfs\*26)FsV597:A622X] | CI051352Disease causing mutation | 1 | PathogenicAccession: VCV 000252082.1 | [rs879255053](https://www.ncbi.nlm.nih.gov/snp/rs879255053) | St. Petersburg | Zakharova et al., 2005, 2007 | None |
| 15 | c.2191delG | g.38844del | 11123223 | p.(Val731fs)[p.(Val731Serfs\*6), FsV710:V715X] | CD136949Disease causing mutation | 1 | PathogenicAccession: VCV 000252253.1 | [rs879255161](https://www.ncbi.nlm.nih.gov/snp/rs879255161) | Petrozavodsk | Komarova et al., 2013 b,cKorneva et al., 2014 | None |
| 17 | c.2416dupG | [g.45159dup](https://www.ncbi.nlm.nih.gov/nuccore/NG_009060.1?report=graph&mk=45159|NG_009060.1\:g.45159dup|green) | 11129534-11129535 | p.(Val806fs)[=p.Val806Glyfs\*11)] | N/ALikely pathogenic | 1 | Conflicting interpretations of pathogenicity​Likely pathogenic Pathogenic;Uncertain significanceVCV000252330.11 | N/A | Moscow | Semenova et al., 2020 | USA, Europe |
| **Missense mutations** |
| 4 | c.326G>ATGC>TAC | g.20852G>A | 11105232 | p.(Cys109Tyr)[C88Y] | CM983629Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession: VCV000226319.3 | [rs121908042](https://www.ncbi.nlm.nih.gov/snp/rs121908042) | Moscow | Meshkov et al., 2009 | Great Britain, Poland |
| 4 | c.444T>GTGT>TGG | g.20970T>G | 11105350 | p.(Cys148Trp)[C127W] | CM981179Disease causing mutation | 1 | Likely pathogenicAccession:VCV000251228.1 | [rs879254528](https://www.ncbi.nlm.nih.gov/snp/rs879254528) | St. Petersburg | Chakir et al., 1998a | None |
| 4 | c.451G>CGCC>CCC | g.20977G>C | 11105357 | p.(Ala151Pro)[A130P] | CM014574Disease causing mutation | 1 | Variant of uncertain significanceAccession: VCV000251234.3 | [rs763233960](https://www.ncbi.nlm.nih.gov/snp/rs763233960) | St. Petersburg | Tatishcheva et al., 2001; Zakharova et al., 2007 | The Netherlands |
| 4 | c.478T>G TGC>GGC | g.21004T>G | 11105384 | p.(Cys160Gly)[=C160G, C139G] | CM981180Disease causing mutation | 4 | Likely pathogenic Accession: VCV000251248.1 | [rs879254540](https://www.ncbi.nlm.nih.gov/snp/rs879254540) | St. Petersburg, Novosibirsk, Moscow | Chakir et al., 1998а; Mandelshtam and Maslennikov, 2001; Meshkov et al., 2004, 2009 | None |
| 4 | c.499T>CTGC>CGC | g.21025T>C | 11105405 | p.(Cys167Arg)[C146R] | CM014575Disease causing mutation | 1 | Likely pathogenicAccession: VCV000251255.1 | [rs879254547](https://www.ncbi.nlm.nih.gov/snp/rs879254547) | St. Petersburg | Tatishcheva et al., 2001; Zakharova et al., 2007 | None |
| 4 | c.530C>TTCG>TTG | g.21056C>T | 11105436  | p.(Ser177Leu)[FH Puerto Rico] | CM890079Disease causing mutation | 1 | Pathogenic./Likely pathogenicAccession:VCV000003686.8 | N/A | Moscow | Semenova et al., 2020 | Europe, Latin America, USA |
| 4 | c.534T>GGAT>GAG | g.21060T>G | 11105440 | p.(Asp178Glu)[D178E] | CM003319Disease causing mutation | N/A | Pathogenic/Likely pathogenicAccession: VCV000251287.1 | [rs879254566](https://www.ncbi.nlm.nih.gov/snp/rs879254566) | Novosibirsk | Shakhtshneider et al., 2019a | Austria, Germany, France |
| 4 | c.542C>T CCG>CTG | g.21068C>T | 11105448 | p.(Pro181Leu) | CM150269Disease causing mutation | 1 | Likely pathogenicAccession:RCV000497065.3 | N/A | Moscow | Semenova et al., 2020 | Brazil |
| 4 | c.551G>ATGT>TAT | g.21077G>A | 11105457 | p.(Cys184Tyr) [C163Y] | CM981182Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession: VCV000003739.5 | [rs121908039](https://www.ncbi.nlm.nih.gov/snp/rs121908039) | Moscow | Meshkov et al., 2009 | Brazil, Canada, Great Britain, Ireland, Portugal, Scotland |
| 4 | c.601G>AGAG>AAG | g.21127G>A | 11105507 | p.(Glu201Lys) [E180K] | CM067689Disease causing mutation | 1 | Likely pathogenicAccession: VCV000251317.1 | [rs879254589](https://www.ncbi.nlm.nih.gov/snp/rs879254589) | Moscow | Meshkov et al., 2009 | Malaysia, Venezuela |
| 4 | c.622G>AGAG>AAG | g.21148G>A | 11105528 | p.(Glu208Lys)[E187K] | CM920419Disease causing mutation | 2 | Pathogenic/ Likely pathogenicAccession: VCV000251328.2 | [rs879254597](https://www.ncbi.nlm.nih.gov/snp/rs879254597) | St. Petersburg, Moscow | Tatishcheva et al., 2001; Zakharova et al., 2007; Averkova et al., 2018 | Czech Republic, Israel, The Netherlands, South Africa, Sweden, etc. |
| 4 | c.626G>ATGC>TAC | g.21152G>A | 11105532 | p.(Cys209Tyr)[C209Y, C188Y] | CM012621Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession:VCV000251332.1 | [rs879254600](https://www.ncbi.nlm.nih.gov/snp/rs879254600) | St. Petersburg | Tatishcheva et al., 2001; Zakharova et al., 2007 | Czech Republic |
| 4 | c.661G>TGAC>TAC | g.21187G>T | 11105567 | p.(Asp221Tyr)[D221Y, D200Y] | CM950754Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession: VCV000251356.1 | [rs875989906](https://www.ncbi.nlm.nih.gov/snp/rs875989906) | Moscow | Meshkov et al., 2009 | Germany, Finland, RSA Indians, Italy, Portugal, Spain, etc. |
| 4 | c.682G>AGAG>AAG | g.21208G>A | 11105588 | p.(Glu228Lys) [E228K, E207K] | CM900153Disease causing mutation | 1 | Pathogenic/ Likely pathogenic Accession: VCV000003691.7 | [rs121908029](https://www.ncbi.nlm.nih.gov/snp/rs121908029) | Moscow | Meshkov et al., 2009 | French Canadian, China, Israel, Korea, Mexico, The Netherlands, Portugal, etc. |
| 5 | c.782G>TTGC>TTC | g.22272G>T | 11106652 | p.(Cys261Phe)[C261F] | CM990795Disease causing mutation | N/A | Likely pathogenicAccession: VCV000003740.4 | [rs121908040](https://www.ncbi.nlm.nih.gov/snp/rs121908040) | Novosibirsk | Voevoda et al., 2008 | Denmark, Norway, Sweden |
| 5 | c.796G>AGAT>AAT | g.22286G>A | 11106666 | p.(Asp266Asn)[D266N]] | CM013562Disease causing mutation | N/A | Pathogenic/ Likely pathogenicAccession: VCV000226334.1 | [rs875989907](https://www.ncbi.nlm.nih.gov/snp/rs875989907) | Novosibirsk | Shakhtshneider et al., 2019a | Czech Republic, Denmark, Japan, The Netherlands, Norway, Tunis, etc. |
| 6 | c.862G>AGAA>AAA | g.23056G>A | 11107436 | p.(Glu288Lys)[E288K] | CM990796Disease causing mutation | N/A | Conflicting interpretations of pathogenicityAccession: VCV000161268.4 | [rs368657165](https://www.ncbi.nlm.nih.gov/snp/rs368657165) | Novosibirsk | Voevoda et al., 2008 | Germany Italy, The Netherlands, New Zealand, Portugal |
| 6 | [c.922G>A](https://www.ncbi.nlm.nih.gov/nuccore/NM_000527.4?report=graph&mk=922|NM_000527.4\:c.922G%3EA|green)GAA>AAA | [g.23116G>A](https://www.ncbi.nlm.nih.gov/nuccore/NG_009060.1?report=graph&mk=23116|NG_009060.1\:g.23116G%3EA|green) | 11107496  | p.(Glu308Lys) | CM066911Disease causing mutation | 1 | Likely pathogenicAccession:VCV000251528.1 | [rs879254721](https://www.ncbi.nlm.nih.gov/snp/rs879254721) | Moscow | Semenova et al., 2020 | Poland, the Netherlands |
| 7 | c.973T>CTGT>CGT | g.26304T>C | 11110684 | p.(Cys325Arg)[C325R] | N/A | N/A | Likely pathogenicAccession: VCV000627979.1 | [rs1568600328](https://www.ncbi.nlm.nih.gov/snp/rs1568600328) | Novosibirsk | Shakhtshneider et al., 2019a | None |
| 7 | c.986G>ATGC>TAC | g.26317G>A | 11110697 | p.(Cys329Tyr)[=C329Y, C308Y] | CM981186Disease causing mutation | 5 | Conflicting interpretations of pathogenicityAccession:VCV000226344.2 | [rs761954844](https://www.ncbi.nlm.nih.gov/snp/rs761954844) | St. Petersburg, Novosibirsk | Zakharova et al., 2005, 2007; Shakhtshneider et al., 2019a; Semenova et al., 2020 | Canada, Chinese, Czech Republic, Filipins, Poland, Taiwan, The Netherlands |
| 7 | c.1009G>AGAG>AAG | g.26340G>A | 11110720 | p.(Glu337Lys)[E337K] | CM182772Disease causing mutation? | 1 | Variant of uncertain significance Accession: VCV000523729.1 | [rs539080792](https://www.ncbi.nlm.nih.gov/snp/rs539080792) | Novosibirsk | Shakhtshneider et al., 2019a | Chile, Singapore |
| 7 | c.1019G>ATGC>TAC | g.26350G>A | 11110730 | p.(Cys340Tyr)[C340Y] | CM042078Disease causing mutation | 1 | Conflicting interpretations of pathogenicityAccession: VCV000251600.2 | [rs755757866](https://www.ncbi.nlm.nih.gov/snp/rs755757866) | Novosibirsk | Shakhtshneider et al., 2019a | Spain |
| 7 | c.1054T>CTGC>CGC | [g.26385T>C](https://www.ncbi.nlm.nih.gov/nuccore/NG_009060.1?report=graph&mk=26385|NG_009060.1\:g.26385T%3EC|green) | 11110765  | p.(Cys352Arg) | CM984574 | 2 | Likely pathogenicAccession:VCV000251618.1 | [rs879254769](https://www.ncbi.nlm.nih.gov/snp/rs879254769) | Moscow | Semenova et al., 2020 | Austria |
| 9 | c.1202T>ACTC>CAC | g.28913T>A | 11113293 | p.(Leu401His)[L401H, L380H, FH Pori; FH Finn-7] | CM950762Disease causing mutation | 2 | Likely pathogenicAccession:VCV000003735.1 | [rs121908038](https://www.ncbi.nlm.nih.gov/snp/rs121908038) | St. Petersburg, Novosibirsk | Zakharova et al., 2005; Shakhtshneider et al., 2019a | Finland, The Netherlands |
| 9 | c.1228A>GAGG>GGG | g.28939A>G | 11113319 | p.(Arg410Gly)[R410G] | N/ALikely pathogenic | 1 | N/A | N/A | Novosibirsk | Voevoda et al., 2008 | None |
| 9 | c.1234A>GATG>GTG | g.28945A>G | 11113325 | p.(Met412Val) [M412V] | N/ALikely pathogenic | 1 | N/A | [rs1225797407](https://www.ncbi.nlm.nih.gov/snp/rs1225797407)Allele G of this SNP not described | Novosibirsk | Voevoda et al., 2008 | None |
| 9 | c.1246C>T CGG>TGG | g.28957C>T | 11113337 | p.(Arg416Trp)[R416W, R395W] | CM970893Disease causing mutation | 4 | Pathogenic/ Likely pathogenicAccession: VCV000183110.4 | [rs570942190](https://www.ncbi.nlm.nih.gov/snp/rs570942190) | Moscow, Novosibirsk | Meshkov et al., 2009; Shakhtshneider et al., 2019a; Semenova et al., 2020 | Austria, Canada, Czech Republic Germany, Israel, Italy, Korea, The Netherlands, Norway, Poland, Spain, Taiwan, etc. |
| 9 | c.1253A>GGAG>GGG | g.28964A>G | 11113344 | p.(Glu418Gly) [E418G] | CM172606Disease causing mutation | N/A | N/A | N/A | Novosibirsk | Voevoda et al., 2008 | China |
| 9 | c.1295T>GCTG>CGG | g.29006T>G | 11113386 | p.(Leu432Arg) [L432R] | N/ALikely pathogenic | 1 | N/A | [rs879254855](https://www.ncbi.nlm.nih.gov/snp/rs879254855) Allele G of this SNP not described | Novosibirsk | Voevoda et al., 2008 | Other populations, no citations available |
| 9 | c.1299C>GGAC>GAG | g.29010C>G | 11113390 | p.(Asp433Glu) [D433E] | N/ALikely pathogenic | 1 | N/A | [rs778309692](https://www.ncbi.nlm.nih.gov/snp/rs778309692) | Novosibirsk | Voevoda et al., 2008 | Rare in other populations, no citations available. |
| 9 | c.1322T>AATC>AAC | g.29033T>A | 11113413 | p.(Ile441Asn)[I441N, I420N] | CM920454Disease causing mutation | 1 | Likely pathogenicAccession: VCV000251782.1 | [rs879254862](https://www.ncbi.nlm.nih.gov/snp/rs879254862) | Russia | Hobbs et al., 1992 | Greece |
| 9 | c.1327T>CTGG>CGG | g.29038T>C | 11113418 | p.(Trp443Arg) [W443R, W422R] | CM045800 Disease causing mutation | 2 | N/A | [rs773566855](https://www.ncbi.nlm.nih.gov/snp/rs773566855) | Moscow, Petrozavodsk | Meshkov et al., 2004, 2009; Korneva et al., 2017a,b; Semenova et al., 2020 | Rare in other populations, no citations available |
| 10 | c.1444G>T GAC>TAC | g.29240G>T | 11113620 | p.(Asp482Tyr)[D482Y, D461Y] | CM045709Disease causing mutation | 1 | Likely pathogenic Accession: VCV000251845.1 | [rs139624145](https://www.ncbi.nlm.nih.gov/snp/rs139624145) | Moscow | Krapivner et al., 2001; Meshkov et al., 2004, 2009 | None |
| 10 | c.1465T>ATAC>AAC | g.29261T>A | 11113641 | p.(Tyr489Asn) | N/ALikely pathogenic? | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 11 | c.1633G>AGGG>AGG | g.31760G>C | 11116140 | p.(Gly545Arg) | CM108002 | 1 | Likely pathogenicAccession:RCV000237535.3 | [rs879254965](https://www.ncbi.nlm.nih.gov/variation/tools/1000genomes/?chr=19&from=11226816&to=11226816&gts=rs879254965&mk=11226816:11226816|rs879254965) | Moscow | Semenova et al., 2020 | Brazil, France, Korea |
| 11 | c.1672G>AGAA>AAA | g.31799G>A | 11116179 | p.(Glu558Lys)[E558K] | N/ALikely pathogenic? | 1 | N/A | N/A | Novosibirsk | Voevoda et al., 2008 | None |
| 12 | c.1741A>CAAA>CAA | g.32514A>C | 11116894 | p.(Lys581Gln) | N/A | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 12 | c.1756T>C TCA>CCA | g.32529T>C | 11116909 | p.(Ser586Pro) | Likely pathogenic? | 2 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 12 | c.1775G>AGGG>GAG | g.32548G>A | 11116928 | p.(Gly592Glu)[G592E, G571E, FH Sicily;FH Foggia-1;FH Naples4;FH Sicilia-4] | CM920464Disease causing mutation | 8 | Pathogenic/ Likely pathogenicAccession: VCV000161271.6 | [rs137929307](https://www.ncbi.nlm.nih.gov/snp/rs137929307) | St. Petersburg, Novosibirsk | Zakharova et al., 2001, 2007; Voevoda et al., 2008, etc.; Semenova et al., 2020 | Austria, Belgium, Brazil, Canada, Greece, Czech Republic, Germany, Italy, The Netherlands, Norway, Poland, Portugal, Spain, USA, etc. |
| 12 | c.1813C>GCTG>GTG | g.32586C>G | 11116966 | p.(Leu605Val)[L605V] | N/A | 1 | N/A | [rs879255031](https://www.ncbi.nlm.nih.gov/snp/rs879255031)Allele G of this SNP not described | Novosibirsk | Voevoda et al., 2008 | None |
| 12 | c.1814T>GCTG>CGG | g.32587T>G | 11116967 | p.(Leu605Arg)[L605R] | N/ALikely pathogenic | 1 | N/A | [rs875989932](https://www.ncbi.nlm.nih.gov/snp/rs875989932)Allele G of this SNP not described | Novosibirsk | Voevoda et al., 2008 | None |
| 12 | c.1835C>GGCC>GGC | g.32608C>G | 11116988 | p.(Ala612Gly)A612G | N/ALikely pathogenic | 1 | N/A | [rs377449975](https://www.ncbi.nlm.nih.gov/snp/rs377449975)Allele G of this SNP not described | Novosibirsk | Voevoda et al., 2008 | None |
| 13 | c.1864G>AGAT>AAT | g.35730G>A | 11120110 | p.(Asp622Asn)[D622N, D601N] | CM051100Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession:VCV000252092.2 | [rs879255059](https://www.ncbi.nlm.nih.gov/snp/rs879255059) | St. Petersburg | Zakharova et al., 2005, 2007 | Czech Republic, China |
| 14 | c.1999T>ATGT>AGT | g.36001T>A | 11120381 | p.(Cys667Ser)[C667S, C646S] | CM051101Disease causing mutation | 1 | Likely pathogenicAccession: VCV000252162.1 | [rs150021927](https://www.ncbi.nlm.nih.gov/snp/rs150021927) | St. Petersburg | Zakharova et al., 2005, 2007 | None |
| 15 | c.2155G>CGTG>CTG | g.38808G>C | 11123188 | p.(Val719Leu)[V719L, V698L] | CM045799Disease causing mutation | 1 | Likely benignAccession: VCV000252243.1 | [rs879255151](https://www.ncbi.nlm.nih.gov/snp/rs879255151) | Moscow | Meshkov et al., 2004, 2009 | Unknown |
| 16 | c.2326G>TGCT>TCT | g.43642G>T | 11128022 | p.(Ala776Ser)[A776S] | N/A | 1 | N/AVariant of uncertain significance | N/A | Moscow | Averkova et al., 2018 | None |
| 16 | c.2374A>TATT>TTT | g.43690A>T | 11128070 | p.(Ile792Phe)[I792F, I771F, FH Russia-2] | CM920470Disease causing mutation | 1 | Conflicting interpretations of pathogenicityAccession: VCV000252294.1 | [rs761123215](https://www.ncbi.nlm.nih.gov/snp/rs761123215) | Russia | Hobbs et al., 1992 | Unknown |
| 16 | c.2389G>AGTG>ATG | g.43705G>A | 11128085 | p.(Val797Met)[V797M, V776M] | CM950772Disease causing mutation | 1 | Conflicting interpretations of pathogenicityAccession:VCV000226393.5 | [rs750518671](https://www.ncbi.nlm.nih.gov/snp/rs750518671) | St. Petersburg | Zakharova et al., 2005, 2007 | China, Cuba, Czech Republic, France, India, Italy, Japan, Norway, Mexico, The Netherlands, Portugal, Scotland South Africa |
| 17 | c.2479G>AGTC>ATC | g.45222G>A | 11129602 | p.(Val827Ile)[V827I, V806I, FH New York-5] | CM920471Disease causing mutation? | N/A | Conflicting interpretations of pathogenicityAccession: VCV000036462.8 | [rs137853964](https://www.ncbi.nlm.nih.gov/snp/rs137853964) | St. Petersburg, Novosibirsk | Zakharova et al., 2005, 2007; Shakhtshneider et al., 2019a | Czech Republic, Denmark, Germany, Israel, Italy, The Netherlands, Norway, Slovak Republic, USA, etc. |
| **In-frame deletions and duplications** |
| 3 | c.191\_313del | g.18254\_18376del | 11102634\_11102656 | p.(Leu64\_Pro105delinsSer) | N/A | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | France, The Netherlands |
| 4 | c.347\_349del | g.20873\_20875del | 11105253-11105255 | p.(Cys116\_His117delinsTyr) [347delGCC] | CD984000Disease causing mutation | 1 | Likely pathogenicAccession: VCV000251164.1 | [rs879254483](https://www.ncbi.nlm.nih.gov/snp/rs879254483) | St. Petersburg | Chakir et al., 1998b | None |
| 4 | c.654-656 delTGGCited also as 652-654delGGT or 651-653del3 | g.21177\_21179delTGG | 11105560-11105562 | p.(Gly219del)[G219del,G218del, G197del, deltaG197, dG197, FH Lithuania, FH Piscataway] | CD910540Disease causing mutation | 2 | Pathogenic/ Likely pathogenicAccession: VCV000226329.6 | [rs121908027](https://www.ncbi.nlm.nih.gov/snp/rs121908027) | St. Petersburg, Novosibirsk | Mandelshtam et al., 1998; Mandelshtam and Maslennikov, 2001; Zakharova et al., 2005, 2007 | Czech Republic, Germany, Great Britain, Israel, The Netherlands, Poland, South Africa, USA, usually in Ashkenazi Jews |
| 4 | c.658\_663del | g.21184\_21189del | 11105564-11105569 | p.(Pro220\_Asp221del)[P199\_D200del] | CD099863Disease causing mutation | 2 | Pathogenic Accession: VCV000440589.1 | [rs1555803409](https://www.ncbi.nlm.nih.gov/snp/rs1555803409) | Moscow | Meshkov et al., 2009; Semenova et al., 2020 | None |
| 4 | c.673\_681dup | g.21199\_21207dup | 11105579-11105587 | p.(Lys225\_Asp227dup)[D203S205dup] | CI099862Disease causing mutation | 1 | Likely pathogenic Accession: VCV000251377.1 | [rs1555803425](https://www.ncbi.nlm.nih.gov/snp/rs1555803425) | Moscow | Meshkov et al., 2004, 2009 | France |
| 4 | c.672\_686del Originally described as c.670\_684del | g.21198\_21212del | 11105578-11105592 | p.(Asp224\_ Glu228del)[D203\_E207del] | CD099864Disease causing mutation | 1 | Likely pathogenic Accession: VCV000441189.1 | [rs1555803439](https://www.ncbi.nlm.nih.gov/snp/rs1555803439) | Moscow | Meshkov et al., 2009 | None |
| 11 | N/A | N/A | Originally described as chr19: 11226838-11226855GRCh37 | Originally described as 17 bp deletion. | N/ADisease causing mutation? | 1 | N/A | N/A | Novosibirsk | Shakhtshneider et al., 2019aDescription in abstract ambiguous. | None |
| **Splice site mutations** |
| 3i | c.313+1G>A | g.18407G>A | 11102787 | Exon 3 skip[IVS3+1 G>A, FH-Elverum,FH Olbia] | CS941506Disease causing mutation | 2 | Conflicting interpretations of pathogenicityAccession: VCV000003736.6 | [rs112029328](https://www.ncbi.nlm.nih.gov/snp/rs112029328) | St. Petersburg | Zakharova et al., 2005, 2007 | Austria, Belgium, Denmark, Germany, Great Britain, Italy, Korea, The Netherlands, Norway, South Africa (black), Spain, Sweden |
| 3i | c.313+2T>G | g.18408T>G | 11102788 | Intron splicing defect [IVS3+2T>G] | N/A | 1 | N/A | [rs793888517](https://www.ncbi.nlm.nih.gov/snp/rs793888517)Allele G not described | Petrozavodsk | Korneva et al., 2016; Korneva et al., 2017b | None |
| 6i | c.940+3\_940+6delGAGT | g.23137\_23140del | 11107517-11107520 | Intron splicing defect [IVS6+3\_6del] | N/A | 2 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 9i | c.1358+1G>A | g.29070G>A | 11113450 | Intron splicing defect [IVS9+1G>A] | CS930843Disease causing mutation | 1 | Pathogenic/ Likely pathogenicAccession: VCV 000251802.1 | [rs775924858](https://www.ncbi.nlm.nih.gov/snp/rs775924858) | St. Petersburg | Zakharova et al., 2005, 2007 | The Netherlands |
| 12i | c.1846-3T>G | g.35709T>G | 11117089 | Intron splicing defect [IVS12-3T>G] | N/ALikely pathogenic | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | None |
| 16i | c.2389+5G>A | g.43710G>A | 11128090 | Intron splicing defect[IVS9+5G>A] | CS067833Disease causing mutation | 3 | Conflicting interpretations of pathogenicity Accession: VCV 000252306.2 | [rs879255191](https://www.ncbi.nlm.nih.gov/snp/rs879255191) | Moscow, Novosibirsk | Meshkov et al., 2009; Shakhtshneider et al., 2017; Semenova et al., 2020 | Italy, Poland |
| **Gross insertions and deletions** |
| 3i\_6i | Not yet available | N/A | 5 kb deletion characterized at genomic level | Not yet available | CG931314Disease causing mutation | 1 | N/A | N/A | St. Petersburg | [Mandelshtam et al., 1993](http://www.ncbi.nlm.nih.gov/pubmed/08401534) | None |
| 4-8 | c.314-?\_1186 + ?dup | g.20840-?\_27259+? | N/A | p.(Pro106\_Val395dup)Inframe duplication of exons 4 to 8 | N/A | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | Poland, Czech Republic, The Netherlands |
| 5i\_6i | c.817+303\_940+943del | g.22610\_24077del | 11106990-11108457 | p.(Val273\_Cys313del)In frame 1468 bp deletion of exon 6 | N/ADisease causing mutation? | 1 | N/A | N/A | Moscow, identified by target sequencing | Averkova et al., 2018 | None |
| 14i-16i | c.2141-966\_2390-330del | g.37256\_44815del | 111121636-111129195 | p.(Glu714\_Ile796del) Deletion of exons 15 and 16 | N/ADisease causing mutation? | 1 | N/A | N/A | Moscow | Semenova et al., 2020 | Japan, Brazil |