**SDC Table 3.** *ABCB11* individual participant gene mutation data.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient number** | **Zygosity** | **Nucleotide change** | **Predicted amino acid change** | **Reference SNP cluster ID** | **CADD score (version 1.3)** | **AF**  **(gnomAD, all)** | **AF (gnomAD, population with highest frequency)** | **Mutation previously reported in literature?** |
| **Including E297G (8 patients)** | | | | | | | | |
| 1 | homozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| 2 to 3 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.611+1G>A | splicing alteration | rs769134865 | 27.8 | <0.0001 | <0.0001 | Yes (2,6) |
| 4 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.1146\_1166del | p.F383\_A389del | not found | not applicable | not reported | not reported | Yes (2)^ |
| 5 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.1708G>A | p.A570T | rs886043807 | 34 | not reported | not reported | Yes (2,5,7) |
| 6 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.1763C>T | p.A588V | rs917981474 # | 32 | <0.0001 | <0.0001 | Yes (2,8) |
| 7 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.1966\_1967delTT | p.L656Afs\*9 | rs1064797270 | not applicable | <0.0001 | <0.0001 | No |
| 8 | heterozygous | c.890A>G | p.E297G | rs11568372 | 32 | 0.00023 | 0.00044 | Yes (1-5) |
| heterozygous | c.2343+1G>T | splicing alteration | rs774411820 | 27.2 | not reported | not reported | Yes (2,3,9) |
| **Including G766R (5 patients)** | | | | | | | | |
| 9 to 10 | homozygous | c.2296G>A | p.G766R | rs763782349 | 33 | <0.0001 | 0.00021 | Yes (2,5) |
| 11 to 12 | heterozygous | c.2296G>A | p.G766R | rs763782349 | 33 | <0.0001 | 0.00021 | Yes (2,5) |
| heterozygous | c.2944G>A | p.G982R | rs72549399 | 28.6 | <0.0001 | <0.0001 | Yes (1,2,10) |
| 13 | heterozygous | c.2296G>A | p.G766R | rs763782349 | 33 | <0.0001 | 0.00021 | Yes (2,5) |
| heterozygous | c.3692G>A | p.R1231Q | rs758069019 | 35 | <0.0001 | <0.0001 | Yes (2,3) |
| **Including R1153C (6 patients)** | | | | | | | | |
| 14 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | <0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.959\_960delTA | p.I320Sfs31\* | not found |  | not reported | not reported | No |
| 15 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | <0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.1146\_1166del | p.F383\_A389del | not found |  | not reported | not reported | Yes (2)^ |
| 16 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | <0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.1408C>T | p.R470\* | rs774824767 | 38 | <0.0001 | <0.0001 | Yes (5,15,16) |
| 17 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | 0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.1685G>A | p.G562D | not found | 33 | not reported | not reported | Yes (2) |
| 18 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | 0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.3476T>C | p.V1159A | not found | 27.5 | not reported | not reported | No |
| 19 | heterozygous | c.3457C>T | p.R1153C | rs72549395 | 31 | <0.0001 | 0.00015 | Yes (1,2,5,11) |
| heterozygous | c.3724C>A | p.L1242I | rs1295206443 | 26.9 | <0.0001 | <0.0001 | Yes (2) |
| **Including F383\_A389del (4 patients total, including 2 listed above)** | | | | | | | | |
| 20 | heterozygous | c.1146\_1166del | p.F383\_A389del | not found |  | not reported | not reported | Yes (2)^ |
| heterozygous | c.2319dupC | p.F774Lfs\*14 | not found | not applicable | not reported | not reported | No |
| 21 | heterozygous | c.1146\_1166del | p.F383\_A389del | not found |  | not reported | not reported | Yes (2)^ |
| heterozygous | c.3709C>A | p.P1237T | not found | 28.1 | not reported | not reported | No |
| **Including V481E (3 patients)** | | | | | | | | |
| 22 | homozygous | c.1442T>A | p.V481E | not found | 25 | not reported | not reported | Yes (2,5) |
| 23 to 24 | heterozygous | c.1442T>A | p.V481E | not found | 25 | not reported | not reported | Yes (2,5) |
| heterozygous | c.2787\_2788insGAGAT | p.K930Efs\*79 | rs752919965 |  | <0.0001 | 0.00016 | Yes (2) |
| **Including G982R (3 patients total, including 2 listed above)** | | | | | | | | |
| 25 | heterozygous | c.2944G>A | p.G982R | rs72549399 | 28.6 | <0.0001 | <0.0001 | Yes (1,2,10) |
| heterozygous | c.1460G>C | p.R487P | rs188824058 ## | 34 | not reported | not reported | Yes (2) |
| **Including D482G (2 patients)** | | | | | | | | |
| 26 | heterozygous | c.1445A>G | p.D482G | rs72549402 | 25.9 | <0.0001 | <0.0001 | Yes (1,2,4,8,12) |
| heterozygous | c.99G>C | p.R33S | rs1306338605 | 18 | <0.0001 | <0.0001 | No |
| 27 | heterozygous | c.1445A>G | p.D482G | rs72549402 | 25.9 | <0.0001 | <0.0001 | Yes (1,2,4,8,12) |
| heterozygous | c.3804delG | p.T1269Pfs\*26 | rs1231877314 | not applicable | <0.0001 | 0.00018 | No |
| **Other homozygotes (7 patients)** | | | | | | | | |
| 28 | homozygous | c.1381A>G | p.K461E | rs1274558905 | 29.2 | <0.0001 | <0.0001 | Yes (1) |
| 29 | homozygous | c.1460G>C | p.R487P | rs188824058 ## | 34 | not reported | not reported | Yes (2) |
| 30 | homozygous | c.1622T>C | p.I541T | rs753994013 | 28.9 | not reported | not reported | Yes (2) |
| 31 | homozygous | c.1639-2A>C | splicing alteration | not found | 24.2 | not reported | not reported | Yes (13) |
| 32 | homozygous | c.3213+1delG | splicing alteration | not found | not applicable | not reported | not reported | Yes (10,14) |
| 33 | homozygous | c.3586C>T | p.Q1196\* | not found | 44 | not reported | not reported | No |
| 34 | homozygous | c.3637G>A | p.G1213R | rs777001075 | 34 | <0.0001 | <0.0001 | No |
| **Other compound heterozygotes** | | | | | | | | |
| 35 | heterozygous | c.22C>T | p.R8\* | rs886043986 | 39 | <0.0001 | <0.0001 | No |
| heterozygous | c.1271delA | p.N424Mfs2\* | rs886043703 | not applicable | not reported | not reported | No |
| 36 to 37 | heterozygous | c.203G>A | p.C68Y | not found | 25.1 | not reported | not reported | Yes (17,18) |
| heterozygous | c.2495G>A | p.R832H | rs376255350 | 35 | <0.0001 | <0.0001 | Yes (17,18) |
| 38 | heterozygous | c.263A>T | p.D88V | not found | 28.4 | not reported | not reported | No |
| heterozygous | c.2319dupC | p.F774Lfs\*14 | not found | not applicable | not reported | not reported | No |
| 39 | heterozygous | c.319T>C | p.C107R | rs781013887 | 26.6 | <0.0001 | <0.0001 | Yes (19) |
| heterozygous | c.611+4A>G | splicing alteration | rs780248854 ## | 15.7 | not reported | not reported | No |
| 40 | heterozygous | c.389+5G>A | splicing alteration | not found | 14.2 | not reported | not reported | No |
| heterozygous | c.1409G>A | p.R470Q | rs1463057954 # | 35 | not reported | not reported | Yes (2) |
| 41 | heterozygous | c.409G>T | p.E137\* | rs1026511416 | 41 | not reported | not reported | No |
| heterozygous | c.2542delG | p.D848Mfs\*10 | not found | not applicable | not reported | not reported | No |
| 42 to 43 | heterozygous | c.470A>G | p.Y157C | not found | 26.3 | not reported | not reported | Yes (2) |
| heterozygous | c.3892G>A | p.G1298R | not found | 34 | not reported | not reported | Yes (2) |
| 44 | heterozygous | c.783G>C | p.L261L (splicing alteration) | not found | 17.5 | not reported | not reported | Yes (5) |
| heterozygous | c.1763C>T | p.A588V | rs917981474 # | 32 | <0.0001 | <0.0001 | Yes (2,8) |
| 45 | heterozygous | c.908+1G>A | splicing alteration | rs147649016 | 27.8 | <0.0001 | <0.0001 | Yes (2,3) |
| heterozygous | c.3692G>A | p.R1231Q | rs758069019 | 35 | <0.0001 | <0.0001 | Yes (2,3) |
| 46 | heterozygous | c.1177A>G | p.I393V | rs369484793 | 11 | <0.0001 | <0.0001 | No |
| heterozygous | c.1709C>T | p.A570V | not found | 31 | not reported | not reported | Yes (20) |
| 47 | heterozygous | c.1408C>T | p.R470\* | rs774824767 | 38 | <0.0001 | <0.0001 | Yes (5,15,16) |
| heterozygous | c.3945delC | p.T1316Lfs\*63 | rs886043366 | not applicable | not reported | not reported | No |
| 48 | heterozygous | c.1409G>A | p.R470Q | rs1463057954 # | 35 | not reported | not reported | Yes (2) |
| heterozygous | c.1708G>A | p.A570T | rs886043807 | 34 | not reported | not reported | Yes (2,5,7) |
| 49 | heterozygous | c.1558A>T | p.R520\* | rs1162621436 | 37 | not reported | not reported | Yes (2) |
| heterozygous | c.3482A>G | p.Q1161R | not found | 27.3 | not reported | not reported | No |
| 50 | heterozygous | c.1723C>T | p.R575\* | rs72549401 | 39 | <0.0001 | 0.00027 | Yes (2,5) |
| heterozygous | c.2178+1G>T | splicing alteration | rs1459273753 ## | 26.7 | not reported | not reported | Yes (2,5) |
| 51 | heterozygous | c.2809G>A | p.G937R | not found | 34 | not reported | not reported | Yes (5) |
| heterozygous | c.3239T>C | p.F1080S | not found | 32 | not reported | not reported | Yes (5) |
| 52 | heterozygous | c.2842C>T | p.R948C | not found | 34 | not reported | not reported | Yes (2) |
| heterozygous | c.3669G>C | p.E1223D | rs199649780 | 23.8 | <0.0001 | <0.0001 | Yes (21) |
| 53 | heterozygous | c.3491delT | p.V1164Gfs\*7 | rs755647308 | not applicable | not reported | not reported | Yes (2) |
| heterozygous | c.3527A>G | p.Y1176C | rs767218250 | 28.1 | <0.0001 | <0.0001 | No |

# = 2 or more variants at this base, including the identified mutation, sharing the same ‘rs’ ID. ‘rs’ IDs do not uniquely identify a mutation, but, rather, a location in the genome.

## = variant(s) at this base, different than the mutation reported here, have been assigned this ‘rs’ ID.

^The nomenclature used in the referenced paper is different by one nucleotide and one amino acid.

Abbreviations: AF, allele frequency; CADD, combined annotation dependent depletion; gnomAD, genome aggregation database (<http://gnomad.broadinstitute.org/>); SNP, single nucleotide polymorphism.

**SDC TABLE 3 REFERENCES**

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