**SDC Table 4.** *ABCB4* 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?** |
| **Homozygotes** |
| 1 | homozygous | c.475C>T | p.R159\* | rs377160065 | 36 | <0.0001 | <0.0001 | Yes (1,2) |
| 2 | homozygous | c.2860G>A | p.G954S | rs779829759  | 27.4 | <0.0001 | <0.0001 | Yes (1) |
| 3 - 5 | homozygous | c.3170T>C | p.L1057P | rs371486074 | 28.5 | <0.0001 | <0.0001 | No |
| **Compound Heterozygotes** |
| 6 | heterozygous | c.490T>G | p.W164G | not found | 28.8 | Not reported | Not reported | Yes (3) |
| heterozygous | c.3081+1G>C | splicing alteration | rs750829010  | 27 | Not reported | Not reported | No |
| 7 | heterozygous | c.526C>T | p.R176W | rs754287486 | 32 | <0.0001 | <0.0001 | Yes (4) |
| heterozygous | c.1666C>G | p.L556V | rs1458045354 | 26.6 | <0.0001 | <0.0001 | No |
| 8 | heterozygous | c.526C>T | p.R176W | rs754287486 | 32 | <0.0001 | <0.0001 | Yes (4) |
| heterozygous | c.1637C>A | p.A546D | rs121918441 | 32 | Not reported | Not reported | Yes (5) |
| 9 | heterozygous | c.527G>A | p.R176Q | rs778380044 | 35 | <0.0001 | <0.0001 | No |
| heterozygous | c.754G>C | p.A252P | not found | 33 | Not reported | Not reported | No |
| 10 | heterozygous | c.760G>A | p.A254T | rs147998447 | 29.1 | <0.0001 | 0.00083 | No |
| heterozygous | c.2318G>T | p.G773V | not found | 33 | Not reported | Not reported | Yes (6) |
| 11 | heterozygous | c.911C>A | p.A304D | rs374836252 | 27.8 | <0.0001 | <0.0001 | No |
| heterozygous | c.2834A>G | p.Q945R | rs1357391684     | 25 | Not reported | Not reported | No |
| 12 | heterozygous | c.1283T>C | p.V428A | not found | 33 | Not reported | Not reported | No |
| heterozygous | c.2563C>T | p.Q855\* | not found | 41 | Not reported | Not reported | No |
| 13 | heterozygous | c.1327C>T | p.Q443\* | not found | 40 | Not reported | Not reported | No |
| heterozygous | c.3279+5G>A | splicing alteration | not found | 20.5 | Not reported | Not reported | No |
| 14 | heterozygous | c.2318G>T | p.G773V | not found | 33 | Not reported | Not reported | Yes (6) |
| heterozygous | c.2800G>A | p.A934T | rs61730509 | 34 | 0.0012 (1 homozygote) | 0.014 | Yes (7,8) |
| 15 | heterozygous | c.2833C>T | p.Q945\* | rs886042562 | 41 | <0.0001 | <0.0001 | Yes (9) |
| heterozygous | c.3491A>G^^ | p.Y1164C^^ | not found | 26.7 | Not reported | Not reported | Yes (10) |
| **Heterozygotes** |
| 16 | heterozygous | c.787A>G | p.I263V | rs45547936  | 20.3 | <0.0001 | <0.0001 | No |
| 17 | heterozygous | c.3230C>T | p.T1077M | rs754565782 | 33 | <0.0001 | 0.00013 | No |
| 18 | heterozygous | c.3272G>T | p.G1091V | not found | 33 | Not reported | Not reported | No |
| 19 | heterozygous | c.3486+5G>A^^ | splicing alteration | not found | 12.6 | Not reported | Not reported | Yes (11) |

There are 2 MDR3 isoforms, differing modestly in amino acid length, the sequence of which can be used for mutation notation. Here, we use NM\_000443.3 as the basis for mutation notation. The 2 mutations indicated by '^^' are 3' of the position where the isoforms begin to differ, so their notations on the longer isoform would be different.

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

**SDC TABLE 4 REFERENCES**

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