# Names for JR (ISBT 032) Blood Group Alleles

## Intro

| | |
|---|---|
| General description: | The JR blood group system consists of one antigen carried on a multipass membrane glycoprotein, ATP binding cassette subfamily G member 2 (ABCG2), also known as breast cancer resistance protein (BCRP) or CD338. ABCG2 consists of 655 amino acids, with a function of an ATP-dependent transporter with a highly diverse range of substrates. The glycoprotein is encoded by the *ABCG2* gene, located on chromosome 4 (chr4:88,090,150-88,231,628; GRCh38/hg38). |
| Gene name: | *ABCG2* |
| Number of exons: | 16 |
| Initiation codon: | Within exon 2 |
| Stop codon: | Within exon 16 |
| Entrez Gene ID: | 9429 |
| LRG: | LRG_823 |
| LRG sequence: | NG_032067.2 (genomic)<br>NM_004827.3 (transcript)<br>NP_004818.2 (protein) |
| Reference allele: | *ABCG2*01* (shaded)<br>Acceptable: $Jr^a$ if inferred by haemagglutination |
| Antithetical antigens: | N/A |

| Phenotype | Allele name | Nucleotide change | Exon Intron | Predicted amino acid change | (Reference No.) PMID | Accession number | rs number |
|---|---|---|---|---|---|---|---|
| Jr(a+) | *ABCG2*01* | | | | | | |
| Null phenotypes | | | | | | | |
| Jr(a−) | *ABCG2*01N.01* | c.376C>T | 4 | p.Gln126Ter | PMID: 22246507<br>PMID: 22246505 | N/A | rs72552713 |
| Jr(a−) | *ABCG2*01N.02.01* | c.706C>T | 7 | p.Arg236Ter | PMID: 22246505 | N/A | rs140207606 |
| Jr(a−) | *ABCG2*01N.02.02* | c.34G>A<br>c.706C>T | 2<br>7 | p.Val12Met<br>Arg236Ter | PMID: 22246507 | N/A | rs2231137<br>rs140207606 |
| Jr(a−) | *ABCG2*01N.03* | c.736C>T | 7 | p.Arg246Ter | PMID: 22246507 | N/A | rs200190472 |
| Jr(a−) | *ABCG2*01N.04* | c.337C>T | 4 | p.Arg113Ter | PMID: 23066723 | N/A | rs201121511 |
| Jr(a−) | *ABCG2*01N.05* | c.784G>T | 7 | p.Gly262Ter | PMID: 23066723 | N/A | rs200473953 |
| Jr(a−) | *ABCG2*01N.06* | c.34G>A<br>c.1591C>T | 2<br>13 | p.Val12Met<br>p.Gln531Ter | PMID: 23066723 | N/A | rs2231137<br>rs201584210 |
| Jr(a−) | *ABCG2*01N.07* | 187_197delATATTATCGAA | 2 | p.Ile63TyrfsTer54 | PMID: 22246505 | N/A | rs565722112 |
| Jr(a−) | *ABCG2*01N.08* | c.542dupA | 6 | p.Phe182ValfsTer14 | PMID: 22246505 | N/A | rs1445054262 |
| Jr(a−) | *ABCG2*01N.09* | c.730C>T | 7 | p.Gln244Ter | PMID: 22246505 | N/A | N/A |
| Jr(a−) | *ABCG2*01N.10* | c.791_792delTT | 7 | p.Leu264HisfsTer14 | PMID: 22246505 | N/A | rs387906870 |
| Jr(a−) | *ABCG2*01N.11* | c.875_878dupACTT | 8 | p.Phe293LeufsTer8 | PMID: 22246505 | N/A | rs1560674481 |
| Jr(a−) | *ABCG2*01N.12* | c.1111_1112delAC | 9 | p.Thr371LeufsTer20 | PMID: 22246505 | N/A | rs387906869 |
| Jr(a−) | *ABCG2*01N.13* | c.34G>A<br>c.243dupC | 2<br>3 | p.Val12Met<br>p.Thr82HisfsTer39 | PMID: 23066723 | N/A | rs2231137<br>rs1212687042 |
| Jr(a−) | *ABCG2*01N.14* | c.1017_1019delCTC | 9 | p.Ser340del | PMID: 23438071 | N/A | rs755318857 |
| Jr(a−) | *ABCG2*01N.15* | c.420dupA | 5 | p.Gln141ThrfsTer16 | PMID: 28836283 | KY581280 | rs1560695576 |
| Jr(a−) | *ABCG2*01N.16* | c.986_987delTA | 9 | p.Ile329ArgfsTer19 | PMID: 28836283 | KY581281 | rs781465213 |

| Phenotype | Allele name | Nucleotide change | Exon Intron | Predicted amino acid change | (Reference No.) PMID | Accession number | rs number |
|---|---|---|---|---|---|---|---|
| Jr(a−) | *ABCG2*01N.17* | c.263+1g>a | i3 | Altered splicing | (1), Abstract | N/A | rs767710822 |
| Jr(a−) | *ABCG2*01N.18* | c.289A>T*<br>*see note in v5.0 | 4 | p.Lys97Ter | (1), Abstract | N/A | N/A |
| Jr(a−) | *ABCG2*01N.19* | c.566delG | 6 | p.Gly189GlufsTer8 | (1), Abstract | N/A | N/A |
| Jr(a−) | *ABCG2*01N.20* | c.1515delC | 13 | p.Phe506SerfsTer4 | (1), Abstract | N/A | rs868217328 |
| Jr(a−) | *ABCG2*01N.21* | c.1723C>T | 14 | p.Arg575Ter | (1), Abstract | N/A | rs548254708 |
| Jr(a−) | *ABCG2*01N.22* | c.1789_1790insT | 15 | p.Ala597ValfsTer8 | (1), Abstract | N/A | N/A |
| Jr(a−) | *ABCG2*01N.23* | c.-273-10904_-19-2432<br>(27097* bp deletion)<br>*see note in v5.0 | 5' to i1 | p.0 | PMID: 25522810 | N/A | N/A |
| Jr(a−) | *ABCG2*01N.24* | c.2T>C | 2 | p.Met1Thr | PMID: 23713577 | N/A | rs765550029 |
| Jr(a−) | *ABCG2*01N.25* | c.421C>A<br>c.1515delC | 5<br>13 | p.Gln141Lys<br>p.Phe506SerfsTer4 | PMID: 23713577 | N/A | rs2231142<br>rs868217328 |
| Jr(a−) | *ABCG2*01N.26* | c.439C>T | 4 | p.Arg147Trp | PMID: 29106709 | N/A | rs372192400 |
| Jr(a–) | *ABCG2*01N.27* | c.204-193_531+13delinCATTTCAGTGGTCTCCC | 3 to 5 | p.0 | PMID: 26173500 | N/A | N/A |
| Jr(a–) | *ABCG2*01N.28* | c.1820+1g>a | i15 | Altered splicing | (1), Abstract | N/A | rs199897813 |
| Altered phenotypes | | | | | | | |
| Jr(a+$^{w}$) | *ABCG2*01W.01* | c.421C>A | 5 | p.Gln141Lys | PMID: 23438071 | N/A | rs2231142 |
| Jr(a+$^{w}$) | *ABCG2*01W.02* | c.1858G>A | 16 | Asp620Asn | PMID: 23438071 | N/A | rs34783571 |
| Jr(a+$^{w}$) | *ABCG2*01W.03* | c.1714A>C | 14 | p.Ser572Arg | PMID: 23438071 | N/A | rs200894058 |

| Phenotype | Allele name | Nucleotide change | Exon Intron | Predicted amino acid change | (Reference No.) PMID | Accession number | rs number |
|---|---|---|---|---|---|---|---|
| $Jr^a$ phenotype unconfirmed | | | | | | | |
| Unclear $Jr^a$ phnotype | N/A | c.34G>A | 2 | p.Val12Met | PMID: 22246507 | N/A | rs2231137 |
| Unclear $Jr^a$ phnotype | N/A | c.421C>A<br>c.440G>A | 5 | p.Gln141Lys<br>p.Arg147Gln | (1), Abstract | N/A | rs2231142<br>rs780594297 |
| Unclear $Jr^a$ phnotype | N/A | c.421C>A<br>c.458C>T | 5 | p.Gln141Lys<br>p.Thr153Met | (1), Abstract | N/A | rs2231142<br>rs753759474 |
| Unclear $Jr^a$ phnotype | N/A | c.455T>C<br>c.1822T>C | 5<br>16 | p.Met152Thr<br>p.Cys608Arg | (1), Abstract | N/A | rs199753603<br>rs200933122 |
| Unclear $Jr^a$ phnotype | N/A | c.1841T>G | 16 | p.Leu614Trp | (1), Abstract | N/A | rs1721693678 |

## References


PMID 22246507 Zelinski T, Coghlan G, Liu XQ, et al.: ABCG2 null alleles define the Jr(a-) blood group phenotype. Nat Genet 2012; 44: 131-2. doi: 10.1038/ng.1075.

PMID 22246505 Saison C, Helias V, Ballif BA, et al.: Null alleles of ABCG2 encoding the breast cancer resistance protein define the new blood group system Junior. Nat Genet 2012; 44: 174-7. doi: 10.1038/ng.1070.

PMID 23066723 Hue-Roye K, Lomas-Francis C, Coghlan G, et al.: The JR blood group system (ISBT 032): molecular characterization of three new null alleles. Transfusion 2013; 53: 1575-9. doi: 10.1111/j.1537-2995.2012.03930.x.

PMID 23438071 Hue-Roye K, Zelinski T, Cobaugh A, et al.: The JR blood group system: identification of alleles that alter expression. Transfusion 2013; 53;2710-4. doi: 10.1111/trf.12118.

PMID 28836283 Berardi P, Cote J, Vege S, et al.: Two novel ABCG2 alleles resulting in a Jr(a-) phenotype. Transfusion 2017; 57: 2811-2. doi: 10.1111/trf.14302.

Abstract (1) Tobita R, Kato S, Osabe T, et al.: Genetic analysis of the Jr(a–) in Japanese people. Vox Sanguinis 2013; 105: 230.

PMID 25522810 Ogasawara K, Osabe T, Suzuki Y, et al.: A new ABCG2 null allele with a 27-kb deletion including the promoter region causing the Jr(a-) phenotype. Transfusion 2015; 55: 1467-71. doi: 10.1111/trf.12969.

PMID 23713577 Tanaka M, Kamada I, Takahashi J, et al.: Defining the Jr(a-) phenotype in the Japanese population. Transfusion 2014; 54: 412-7. doi: 10.1111/trf.12277.

PMID 29106709 Wieckhusen C, Rink G, Scharberg EA, et al.: A new genetic background for the Jr(a-) blood group phenotype caused by the ABCG2*439T allele encoding a p.Arg147Trp change. Transfusion 2017; 57: 3063-4. doi: 10.1111/trf.14375.

PMID 26173500 Saison C, Cartron JP, Arnaud L. Deletion of Exons 3 through 5 of ABCG2 causes the Jr(a-) phenotype in a West African woman. Transfusion 2015;55:2766-7. doi: 10.1111/trf.13223.

## Track of changes

| | | | from | to |
|---|---|---|---|---|
| **1** | **Version** | | **v4.0 08-APR-2019** | **v5.0 31-JUL-2023** |
| **2** | Author | created: | Thierry Peyrard, April 2019 | Vanja Crew, supp. Louise Tilley, July 2023 |
| **3** | Review | reviewed: | Slim Azouzi, April 2019 | Silvano Wendel, supp. Mayra Altobelli Brito, July 2023 |
| **4** | General | All | v4.0 Word-document | First Excel map version. Spread-sheets "Intro", "Allele Table", "References", "Versioning (v5.0)" created. |
| **5** | Introduction | Intro updated | | The JR blood group system consists of one antigen carried on a multipass membrane glycoprotein, ATP binding cassette subfamily G member 2 (ABCG2), also known as breast cancer resistance protein (BCRP) or CD338. ABCG2 consists of 655 amino acids, with a function of an ATP-dependent transporter with a highly diverse range of substrates. The glycoprotein is encoded by the *ABCG2* gene, located on chromosome 4 (chr4:88,090,150-88,231,628; GRCh38/hg38). |
| **6** | LRG | LRG ID and reference sequences updated | | LRG_823 added, transcript reference sequence updated from NM_004827.2 to NM_004827.3, protein reference NP_004818.2 added |
| **7** | Allele table | Table updated | | Nucleotide change and predicted amino acid change sections updated for a number of variants to reflect current nomenclature. References changed from numbers to PMID. GenBank accession numbers and dbSNP rs numbers added where available. |
| **8** | Allele table | *ABCG2*01N.08* updated | | Changed c.542_543insA to c.542dupA |
| **9** | Allele table | *ABCG2*01N.13* updated | | Changed c.244_245insC to c.243dupC |
| **10** | Allele table | *ABCG2*01N.15* updated | | Changed c.420_421insA to c.420dupA |

| | | | | |
|---|---|---|---|---|
| **11** | Allele table | *Note for *ABCG2*01N.18* | | Note: reference sequence NM_004827.3 encodes c.289_291AAA (p.Lys97). In the original abstract (1), Jr(a-) sample was erroneously described to have c.289A>G mutation (K97Ter); c.289A>G would encode p.Lys97Glu, not Lys97Ter. In subsequent databases and publications, c.289A>G was corrected to c.289A>T (for example, ISBT JR blood group allele table v4.0 and Haer-Wigman et al, Transfusion 2014; 54: 1836-46. |
| **12** | Allele table | *Note for *ABCG2*01N.23* | | Note: Ogasawara et al. (PMID 26173500) used BAC clone RP11-368G2 (GenBank no. AC084732.1; 09/11/2000) as a contemporary reference sequence for *ABCG2* and the published deletion was described as 27,094-bp deletion ranging from 9732 to 36,825 (GenBank no. AB973570.1). Current *ABCG2* references are NG_032067.2 (genomic) and NM_004827.3 (transcript). Using the HGVS variant nomenclature and the current reference, the deletion is c.-273-10904_--19-2432. The deletion is 27,097-bp long, reflecting the 3-bp difference between the AC084732.1 and the current NG_032067.2. |
| **13** | Allele table | Alleles added | | *ABCG2*01N.28, ABCG2*01W.03* |
| **14** | Allele table | Jra phenotype unconfirmed section updated | | Moved [c.1714A>C] allele to altered phenotype section as *ABCG2*01W.03* . Moved [c.1820+1g>a] allele to Jr(a-) section as *ABCG2*01N.28.* Removed [c.1384G>A] and [c.1822T>C] alleles because of no evidence of their conection to JR. |
| **15** | References | PMID | | Added PMIDs. Added doi. |
| **16** | References | Abstracts | | Changed Reference (6) to Abstract (1). |
| **17** | **End Version** | | **v4.0 08-APR-2019** | **v5.0 31-JUL-2023** |