

Names for JR (ISBT 032) Blood Group Alleles

General description: The JR blood group system consists of one antigen carried on a multipass membrane glycoprotein (a.k.a. ATP-binding cassette, sub-family G, member 2 [ABCG2]; breast cancer resistance protein [BCRP], CD338) of 655 amino acids. ABCG2 is an ATP-dependent transporter with a highly diverse range of substrates.

Gene name: *ABCG2*

Number of exons: 16

Initiation codon: Within exon 2

Stop codon: Within exon 16

Entrez Gene ID: 9429

LRG sequence: NG_032067.2 (genomic)
NM_004827.2 (transcript)

Reference allele: *ABCG2* (shaded)

Acceptable: *Jr^a* if inferred by haemagglutination

| Phenotype | Allele name | Molecular basis | Exons/Introns affected | Amino acid change | Reference |
|--------------|------------------------|------------------|------------------------|--------------------|-----------|
| Jr(a+) | <i>ABCG2*01</i> | | | | |
| Null alleles | | | | | |
| Jr(a-) | <i>ABCG2*01N.01</i> | c.376C>T | Exon 4 | p.Gln126X | [1, 2] |
| Jr(a-) | <i>ABCG2*01N.02.01</i> | c.706C>T | Exon 7 | p.Arg236X | [1, 2] |
| Jr(a-) | <i>ABCG2*01N.02.02</i> | c.[34G>A;706C>T] | Exon 2 Exon 7 | p.Val12Met Arg236X | [1] |
| Jr(a-) | <i>ABCG2*01N.03</i> | c.736C>T | Exon 7 | p.Arg246X | [1] |

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|--------|---------------------|-----------------------|----------------|--------------------------|-----|
| Jr(a-) | <i>ABCG2*01N.04</i> | c.337C>T | Exon 4 | p.Arg113X | [3] |
| Jr(a-) | <i>ABCG2*01N.05</i> | c.784G>T | Exon 7 | p.Gly262X | [3] |
| Jr(a-) | <i>ABCG2*01N.06</i> | c.[34G>A;1591C>T] | Exon 2 Exon 13 | p.[Val12Met;Gln531X] | [3] |
| Jr(a-) | <i>ABCG2*01N.07</i> | 187_197delATATTATCGAA | Exon 2 | p.Ile63TyrfX | [2] |
| Jr(a-) | <i>ABCG2*01N.08</i> | c.542_543insA | Exon 6 | p.Phe182ValfsX | [2] |
| Jr(a-) | <i>ABCG2*01N.09</i> | c.730C>T | Exon 7 | p.Gln244X | [2] |
| Jr(a-) | <i>ABCG2*01N.10</i> | c.791_792delTT | Exon 7 | p.Leu264HisfsX | [2] |
| Jr(a-) | <i>ABCG2*01N.11</i> | c.875_878dupACTT | Exon 8 | p.Phe293LeufsX | [2] |
| Jr(a-) | <i>ABCG2*01N.12</i> | c.1111_1112delAC | Exon 9 | p.Thr371LeufsX | [2] |
| Jr(a-) | <i>ABCG2*01N.13</i> | c.[34G>A;244_245insC] | Exon 2 Exon 3 | p.[Val12Met;Thr82HisfsX] | [3] |
| Jr(a-) | <i>ABCG2*01N.14</i> | c.1017_1019delCTC | Exon 9 | p.Ser340del | [4] |
| Jr(a-) | <i>ABCG2*01N.15</i> | c.420_421insA | Exon 5 | p.Gln141Thrfs*16 | [5] |
| Jr(a-) | <i>ABCG2*01N.16</i> | c.986_987delTA | Exon 9 | p.Ile329Argfs*19 | [5] |
| Jr(a-) | <i>ABCG2*01N.17</i> | c.263 + 1G >A | Intron 3 | r.spl? | [6] |

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|---------------------------------------|---------------------|-----------------------------------------------------------------------------------|----------------|------------------------|------|
| Jr(a-) | <i>ABCG2*01N.18</i> | c.289A > T | Exon 4 | p.Lys97Ter | [6] |
| Jr(a-) | <i>ABCG2*01N.19</i> | c.565_566del | Exon 6 | p.Gly189fs | [6] |
| Jr(a-) | <i>ABCG2*01N.20</i> | c.1515del | Exon 13 | p.Ala506fs | [6] |
| Jr(a-) | <i>ABCG2*01N.21</i> | c.1723C > T | Exon 14 | p.Arg575Ter | [6] |
| Jr(a-) | <i>ABCG2*01N.22</i> | c.1789_1790insT | Exon 15 | p.Ala597fs | [6] |
| Jr(a-) | <i>ABCG2*01N.23</i> | 27-kb deletion including noncoding Exon 1 and the promoter region of <i>ABCG2</i> | | | [7] |
| Jr(a-) | <i>ABCG2*01N.24</i> | c.2T > C | Exon 2 | p.0 | [8] |
| Jr(a-) | <i>ABCG2*01N.25</i> | c.[421C > A;1515del] | Exon 5 Exon 13 | p.[Gln141Lys;Ala506fs] | [8] |
| Jr(a-) | <i>ABCG2*01N.26</i> | c.439C>T | Exon 4 | p.Arg147Trp | [9] |
| Jr(a-) | <i>ABCG2*01N.27</i> | Deletion of exons 3 to 5 | Exon 3 to 5 | | [11] |
| Altered phenotypes | | | | | |
| Jr(a+w) | <i>ABCG2*01W.01</i> | c.421C>A | Exon 5 | p.Gln141Lys | [10] |
| Jr(a+w) | <i>ABCG2*01W.02</i> | c.1858G>A | Exon 16 | Asp620Asn | [10] |
| Jr ^a phenotype unconfirmed | | | | | |

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|---------|--|------------------------|----------------|-------------------------|------|
| Unclear | | c.[421C > A;440G > A] | Exon 5 | p.[Gln141Lys;Arg147Gln] | [6] |
| Unclear | | c.[421C > A;458C > T] | Exon 5 | p.[Gln141Lys;Thr153Met] | [6] |
| Unclear | | c.[455T > C;1819T > C] | Exon 5 Exon 16 | p.[Met152Thr;Cys608Arg] | [6] |
| Unclear | | c.1384G > A | Exon 12 | p.Gly462Arg | [6] |
| Unclear | | c.1819T > C | Exon 16 | p.Cys608Arg | [6] |
| Unclear | | c.1820 + 1g > a | Intron 15 | r.spl? | [6] |
| Unclear | | c.1841T > G | Exon 16 | p.Leu614Trp | [6] |
| Unclear | | c.1714A > C | Exon 14 | p.Ser572Arg | [10] |

References:

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