

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]

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]

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]
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					
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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- 3 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.
- 4 Hue-Roye K, Zelinski T, Coughan A, et al.: The JR blood group system: identification of alleles that alter expression. *Transfusion* 2013.
- 5 Berardi P, Cote J, Vege S, et al.: Two novel ABCG2 alleles resulting in a Jr(a-) phenotype. *Transfusion* 2017; 57: 2811-2.
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- 10 Hue-Roye K, Zelinski T, Coughan A, et al.: The JR blood group system: identification of alleles that alter expression. *Transfusion* 2013; 53: 2710-4.