

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

Reference allele <i>ABCG2*01</i> encodes Jr1 (Jr ^a)				
Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
Jr(a+)	<i>ABCG2*01</i>			
Null phenotypes				
Jr(a-)	<i>ABCG2*01N.01</i>	c.376C>T	4	p.Gln126Ter [1,2]
Jr(a-)	<i>ABCG2*01N.02.01</i>	c.706C>T	7	p.Arg236Ter [1,2]
Jr(a-)	<i>ABCG2*01N.02.02</i>	c.34G>A; c.706C>T	2 7	p.Val12Met; p.Arg236Ter [1]
Jr(a-)	<i>ABCG2*01N.03</i>	c.736C>T	7	p.Arg246Ter [1]
Jr(a-)	<i>ABCG2*01N.04</i>	c.337C>T	4	p.Arg113Ter [3]
Jr(a-)	<i>ABCG2*01N.05</i>	c.784G>T	7	p.Gly262Ter [3]
Jr(a-)	<i>ABCG2*01N.06</i>	c.34G>A; c.1591C>T	2 13	p.Val12Met; p.Gln531Ter [3]
Jr(a-)	<i>ABCG2*01N.07</i>	c.187_197delATATTATC GAA	2	p.Ile63Tyrf*54 [2]
Jr(a-)	<i>ABCG2*01N.08</i>	c.542_543insA	6	p.Phe182Valfs*14 [2]
Jr(a-)	<i>ABCG2*01N.09</i>	c.730C>T	7	p.Gln244Ter [2]
Jr(a-)	<i>ABCG2*01N.10</i>	c.791_792delTT	7	p.Leu264Hisfs*14 [2]
Jr(a-)	<i>ABCG2*01N.11</i>	c.875_878dupACTT	8	p.Phe293Leufs*8 [2]

Jr(a-)	<i>ABCG2*01N.12</i>	c.1111_1112delAC	9	p.Thr371Leufs*20 [2]
Jr(a-)	<i>ABCG2*01N.13</i>	c.34G>A; c.244_245insC	2 3	p.Val12Met; p.Gly83Argfs*38 [3]
Jr(a-) ^	<i>ABCG2*01N.14</i>	c.1017_1019delCTC	9	p.Ser340del [4]
Altered phenotypes				
Jr(a+w)	<i>ABCG2*01W.01</i>	c.421C>A	5	p.Gln141Lys [4]
Jr(a+w)	<i>ABCG2*01W.02</i>	c.1858G>A	16	p.Asp620Asn [4]

^ Presumed

References:

1. Zelinski T, Coghlan G, Liu X-Q, Reid ME. *ABCG2* null alleles define the Jr(a-) phenotype. *Nat Genet* 2012;44:131-132.
2. Saison C, Helias V, Ballif BA, Peyrard T, Puy H, Miyazaki T, Perrot S, Vayssier-Taussat M, Waldner M, Le Pennec PY, Cartron JP, Arnaud L. Null alleles of *ABCG2* encoding the breast cancer resistance protein define the new blood group system Junior. *Nat Genet* 2012;44:174-177.
3. Hue-Roye K, Lomas-Francis C, Coghlan G, Zelinski T, Reid ME. The JR blood group system (ISBT 032): molecular characterization of three new null alleles. *Transfusion* doi:10.1111/j.1537-2995.2012.03930.x
4. Hue-Roye K, Zelinski T, Coughan A, Lomas-Francis C, Miyazaki T, Tani Y, Westhoff CM, Reid ME. The JR Blood group system: identification of alleles that alter expression. *Transfusion* doi:10.1111/trf12118