Names for JMH (ISBT 026) Blood Group Alleles

Intro

General description:	The JMH blood group system consists of eight antigens carried on a GPI-linked glycoprotein (CD108, Semaphorin, SEMA7A) that consists of 656 amino acids. It has a leader sequence of 44 amino acids and a GPI motif of 19 amino acids, both of which are cleaved from the membrane bound protein.
Gene name:	*SEMA7A*
Number of exons:	14
Initiation codon:	Beginning of exon 1
Stop codon:	Within exon 14
Entrez Gene ID:	8482
LRG:	LRG_818
LRG sequence:	NG_011733.1 (genomic) NM_003612.3 (transcript)
Reference allele:	Preferred: *JMH*01* (shaded) Acceptable: *JMH* if inferred by haemagglutination
Reference allele *JMH*01* encodes:	JMH1, JMH2, JMH3, JMH4, JMH5, JMH6, JMH7, JMH8
Antithetical antigens:	n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
JMH:1 or JMH+	*JMH*01*				PMID: 11449960	NG_011733.1	n.a.
JMH:–2 or JMHK–	*JMH*01.–02*	c.619C>T c.1545A>G	6 12	p.Arg207Trp p.Gln515Gln	PMID: 17207242	AM180447	rs56367230 rs741761
JMH:–3 or JMHL–	*JMH*01.–03*	c.620G>A	6	p.Arg207Gln	PMID: 17207242	AM180445	rs55637216
JMH:–4 or JMHG–	*JMH*01.–04*	c.1379G>A	11	p.Arg460His	PMID: 17207242	AM180448	rs56204206
JMH:–5 or JMHM–	*JMH*01.–05*	c.1381C>T	11	p.Arg461Cys	PMID: 17207242	AM180446	rs56001514
JMH: –6 or JMHQ–	*JMH*01.–06*	c.1040G>T	9	p.Arg347Leu	PMID: 20854351	n.a.	rs387907241
JMH: –7 or JMHN–	*JMH*01.–07*	c.709G>A c.1545A>G c.1865G>A	7 12 14	p.Asp237Asn p.Gln515Gln p.Arg622His	(1), Abstract	n.a.	rs140707085 rs741761 rs140128092
JMH: –8 or JMHA–	*JMH*01.–08*	c.507C>T c.556G>A c.1545A>G	5 6 12	p.Tyr169Tyr p.Glu186Lys p.Gln515Gln	(2), Abstract	MT017654	rs2075589 rs572867366 rs741761

N of "JMHN", stands for "North Africa".

A of "JMHA", stands for "Arabia".

Not a blood group antigen. Linkage shown to progressive familial intrahepatic cholestasis.	*no blood group allele JMH*Arg148Trp*	c.442C>T	4	p.Arg148Trp	PMID: 34585848	n.a.	rs200895370

Additional well documented GenBank Accession Numbers with SNVs not causing any amino-acid exchanges (silent mutations) are: AM180449 to AM180451

References

PMID 11449960 Daniels GL, Anstee DJ, Cartron JP et al.: International Society of Blood Transfusion Working Party on Terminology for Red Cell Surface Antigens. Vox Sang. 2001 Apr;80(3):193-7.

PMID 17207242 Seltsam A, Strigens S, Levene C, et al.: The molecular diversity of Sema7A, the semaphorin that carries the JMH blood group antigens. Transfusion. 2007 Jan;47(1):133-46.

PMID 20854351 Richard M, St-Laurent J, Perreault J, Long A, St-Louis M.: A new SEMA7A variant found in Native Americans with alloantibody. Vox Sang. 2011 Apr;100(3):322-6.

Abstract (1) Vrignaud C. Ramelet S, Herb A, et al.: Characterization of a Novel High-Prevalence Antigen in the JMH Blood Group System. Vox Sanguinis 2019, 114 (Suppl. 1), 52, Abstract 4C-S20-05

Abstract (2) Henny C, Thornton N, Laundy R, Borowski A, Stettler J, Lejon Crottet S, Daskalakis M, Hustinx H. An Antibody Against a Novel High Prevalence Antigen in the JMH Blood Group System. Vox Sanguinis (2020) 115 (Suppl. s1), 3–387 (P-450)

PMID 34585848 Qiong Pan, Gang Luo , Jiaquan Qu, Sheng Chen, Xiaoxun Zhang, et al. A homozygous R148W mutation in Semaphorin 7A causes progressive familial intrahepatic cholestasis. EMBO Mol Med. 2021 Nov 8;13(11):e14563. doi: 10.15252/emmm.202114563. Epub 2021 Sep 29.

Track of changes			from	to
1	**Version**		**v4.0 25-FEB-2020**	**v5.0 30-NOV-2021**
2	Author	created	Christoph Gassner, January 2020	Christoph Gassner, November 2021
3	Review	reviewed	Thierry Peyrard, January 2020	Cristof Weinstock, November 2021
4	Intro	changed	numbers of antigens: 7	numbers of antigens: 8
5	Reference	changed	Reference allele encodes for: JMH1, JMH2, JMH3, JMH4, JMH5, JMH6, JMH7	Reference allele encodes for: JMH1, JMH2, JMH3, JMH4, JMH5, JMH6, JMH7, JMH8
6	Allele Table	Antigen/allele added		JMH: –8
7	Allele Table	Disease allele added (not a blood group antigen)		*JMH*Arg148Trp* (c.442C>T)
8	References	renumbered		renumbered Abstract (4) to Abstract (1)
9	References	renumbered		renumbered Abstract (5) to Abstract (2)
10	**End Version**		**to v4.0 25-FEB-2020**	**v5.0 30-NOV-2021**

	Track of changes		from	to
1	**Version**		**v3.0 160622**	**v4.0 25-FEB-2020**
2	Author	created:	n.a. v3.0 160622	Christoph Gassner, January 2020
	Review	reviewed:	n.a.	Thierry Peyrard, January 2020
3				
4	General			First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	Text changed	The JMH blood group system consists of six antigens carried on a GPI-linked glycoprotein	The JMH blood group system consists of seven antigens carried on a GPI-linked glycoprotein
6	Intro	LRG ID line added:	n.a.	LRG_818
7	Intro	Reference allele line moved from Allele Table to Intro and updated:	n.a.	Reference allele JMH*01 encodes: JMH1, JMH2, JMH3, JMH4, JMH5, JMH6, JMH7
8	Intro	Antithetical Antigens line created in Intro:	n.a.	n.a.
9	Allele Table	Table column and header additions	n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
10	Allele Table	Text change: Line moved to Intro:	Reference allele JMH*01 encodes JMH1, JMH2, JMH3, JMH4, JMH5, JMH6	see above
11	Allele Table	Silent mutation added:	n.a.	JMH:–2 or JMHK– , *JMH*01.–02*
12			n.a.	Silent mutation: c.1545A>G, p.Gln515Gln, added to *JMH*01.–02* .
13	Allele Table	Antigen/allele added:	n.a.	JMH: –7 or JMHN–, *JMH*01.–07*
14	References	Abstract added	n.a.	Abstract (1 (former 4)), Vrignaud C. Ramelet S, Herb A, et al.: Characterization of a Novel High-Prevalence Antigen in the JMH Blood Group System. Vox Sanguinis 2019, 114 (Suppl. 1), 52, Abstract 4C-S20-05
15	References	All references new:	n.a.	All references (1) to (4) added for the first time.
16	**End Version**		**v3.0 160622**	**v4.0 25-FEB-2020**