

## Names for MAM (ISBT 041) Blood Group Alleles

**General description:** The MAM blood group system consists of one antigen, MAM, carried on epithelial membrane protein 3 (EMP3). It consists of 163 amino acids, with predicted four transmembrane domains and two N-linked glycosylation sites. The protein is encoded by *EMP3*. There is a known association between EMP3 and CD44 (Indian blood group system), whereby MAM-negative individuals have reduced levels of CD44 and therefore express Indian blood group system antigens only very weakly.

**Gene name:** *EMP3*

**Number of exons:** 5

**Initiation codon:** Within exon 2

**Stop codon:** Within exon 5

**Entrez Gene ID:** 2014

**LRG:** not yet assigned

**LRG sequence:** NC\_000019.10 (genomic)

NM\_001425.3 (transcript)

NP\_001416.1 (protein)

**Reference allele:** *MAM\*01* (shaded)

Acceptable: *MAM*, if inferred by haemagglutination

**Reference allele**

*EMP3\*01* encodes: MAM; epithelial membrane protein 3 (EMP3)

**Antithetical antigens:** n/a

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	Reference (PMID)	Accession number	rs number
MAM:1 or MAM+	<i>MAM*01</i>					NM_001425.3	
Null phenotypes							
MAM:-1 or MAM-	<i>MAM*01N.01</i>	c.123C>G c.373A>G	3 5	p.Tyr41Ter p.p.Ile125Val	(1) PMID: 32678083	MN121937	rs201392469 rs4893
MAM:-1 or MAM-	<i>MAM*01N.02</i>	c.182-186_322+418del (745bp deletion)	4	p.Trp62_Ser108del	(1) PMID: 32678083	MN164486	N/A
MAM:-1 or MAM-	<i>MAM*01N.03</i>	c.323-231_492+338del (822bp deletion)	5	p.Val109_Ter164del	(1) PMID: 32678083	MN164487	N/A
MAM:-1 or MAM-	<i>MAM*01N.04</i>	c.1-3513_492+1379del (8518bp deletion)	1 to 5	p.Met1_Ter164del	(1) PMID: 32678083	MN175569	N/A
MAM:-1 or MAM-	<i>MAM*01N.05</i>	c.1-3532_492+1361del (8519bp deletion)	1 to 5	p.Met1_Ter164del	(1) PMID: 32678083	MN175570	N/A

## References

1. PMID: 32678083 Thornton N, Karamatic Crew V, Tilley L, Green CA, Tay CL, Griffiths RE, Singleton BK, Spring F, Walser P, Alattar AG, Jones B, Laundry R, Storry JR, Möller M, Wall L, Charlewood R, Westhoff CM, Lomas-Francis C, Yahalom V, Feick U, Seltsam A, Mayer B, Olsson ML, Anstee DJ. Disruption of the tumour-associated EMP3 enhances erythroid proliferation and causes the MAM-negative phenotype. *Nat Commun.* 2020 Jul 16;11(1):3569. doi: 10.1038/s41467-020-17060-4. PMID: 32678083; PMCID: PMC7366909.

<b>Track of changes</b>		<b>v1.0 30-OCT-2020</b>	<b>to</b>
	created by	Nicole Thornton	
	reviewed by	Vanja Crew	
General	Document created	First version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.	
Intro	Intro added	General description, gene name, number of exons, initiation codon, stop codon, Entrez Gene ID and Reference allele information added.	
Allele Table	Table created	Table columns "Phenotype", "Allele name", "Nucleotide change", "Exon", "Predicted amino acid change", "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.	
Allele Table	Alleles added:	<i>MAM*01</i> and <i>MAM*01N.01</i> to <i>MAM*01N.05</i> added	
References	References added:	References (1)	
<b>End of changes</b>		<b>v1.0 30-OCT-2020</b>	