

Names for EMM (ISBT 042) Blood Group Alleles

Intro

General description: The EMM blood group system consists of one antigen, Emm, carried on the glycosylphosphatidylinositol (GPI) anchor to which all GPI-linked proteins are attached. The Emm antigen is thought to involve the ethanolamine phosphate (EtNP) on the second mannose (Man2) of the GPI-anchor. This EtNP is specifically added to Man2 by GPI-ethanolaminetransferase II enzymatic complex GPI-ETII, which is composed of two proteins PIGF and PIGG. The catalytic part of GPI-ETII is defined by PIGG, encoded by the *PIGG* gene. The presence or absence of the Emm antigen is defined by the activity of the *PIGG* gene, with loss of function variants leading to the Emm⁻ phenotype.

Gene name: *PIGG*

Number of exons: 13

Initiation codon: Beginning of exon 1

Stop codon: Within exon 13

Entrez Gene ID: 54872

LRG: none

Ref sequence: NG_051621.1 (genomic)

NM_001127178.3 (transcript)

Reference allele: Preferred: *EMM*01* (shaded)

Acceptable:

Reference allele Emm

*JMH*01* encodes:

Antithetical antigens: n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
EMM:1 or Emm+	<i>PIGG*01</i>					NG_051621.1	n.a.
EMM:-1 or Emm-	<i>PIGG*01N.01</i>	c.2624_2625delTA	12	p.Leu875Ter	PMID: pending	MW355842.1	rs771819481
EMM:-1 or Emm-	<i>PIGG*01N.02</i>	c.901+1delIG	i5	Splicing defect (Frameshift)	PMID: 33763700		rs782318668
EMM:-1 or Emm-	<i>PIGG*01N.03</i>	c.1640G>A	9	p.Trp547Ter	PMID: pending	pending	rs547951371
EMM:-1 or Emm-	<i>PIGG*01N.04</i>	c.640C>T	4	p.His214Tyr	PMID: 33763700		
EMM:-1 or Emm-	<i>PIGG*01N.05</i>	4 kb deletion including Exon 6 with a 21 bp insertion (GGGCGCGGTGGCTCATTTGTG) g.18342_22403delinsGGGCGCGGT GGCTCATTTGTG	i5-i6	p.?	PMID: 33763700		
EMM:-1 or Emm-	<i>PIGG*01N.06</i>	6 kb deletion including Exons 2-3 g.5982_11944del	i1-i3	p.?	PMID: pending	MW355843.1	n.a.
EMM:-1 or Emm-	<i>PIGG*01N.07</i>	6.3 kb deletion including Exons 7-9 g.24216_30561del	i6 7-i9	p.?	PMID: pending	MW355844.1	n.a.
EMM:-1 or Emm-	<i>PIGG*01N.08</i>	c.361-51_383delinsGACTT	i2-e3	p.?	PMID: pending	MW355845.1	n.a.

References

- PMID pending Lane WJ, Aeschlimann J, Vege S et al.
PIGG Defines the Emm Blood Group System. Submitted
- PMID 33763700 Romain Duval, Gaël Nicolas, Alexandra Willemetz et al.
Inherited glycosylphosphatidylinositol defects cause the rare Emm-negative
blood phenotype and developmental disorders.
Blood. 2021 Mar 24;blood.2020009810. Online ahead of print.

Track of changes	from version	to version
1	Version	v1.0
2	Author created:	William Lane, June 2021
3	Reviewer reviewed:	unreviewed
4	End Version	