Names for RAPH (ISBT 025) Blood Group Alleles

Intro

General description: The Raph blood group system consists of one antigen (MER2) carried on a

multipass membrane glycoprotein CD151 of the tetraspanin family. It consists of 253 amino acids and both amino and carboxyl termini are predicted to be intracellular. The glycoprotein is encoded by *CD151* (or

RAPH, if analysis is to predict a blood group antigen). The null phenotype is associated with renal failure.

Gene name: CD151 (RAPH)

Number of exons: 9

Initiation codon: Within exon 3 Stop codon: Within exon 9

Entrez Gene ID: 977 LRG: 817

LRG sequence: NG_007478.1 (genomic)

NM_004357.4 (transcript) NP_004348.2 (protein)

Reference allele: RAPH*01 (shaded)

Acceptable: RAPH*1 or MER2 if inferred by haemagglutination

Reference allele

RAPH*01 encodes for: RAPH1

Antithetical antigens n.a.

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number	
RAPH:1 or MER2+	RAPH*01				(1), PMID:15265795	NG_007478		
RAPH:-1 or MER2-	RAPH*0101.01	c.511C>T c.579A>G	. /	p.Arg171Cys p.Gly193	(2), PMID:18522704	MN535383	rs139042921 rs1130663	
RAPH:-1 or MER2-	RAPH*0101.02	c.533G>A	7	p.Arg178His	(1), PMID:15265795	MN535382	rs779114765	
RAPH:-1 or MER2-	RAPH*0101.03	c.494G>A	7	p.Arg165Gln	(3), Abstract	MN535384	rs949958895	
Null phenotypes								
RAPH _{null}	RAPH*01N.01	c.383_384insG	6	p.Asp128Glufs*13	(1), PMID:15265795	MN339517	n.a.	

References

- 1. PMID:
 15265795

 Karamatic Crew V, Burton N, Kagan A et al. CD151, the first member of the tetraspanin (TM4) superfamily detected on erythrocytes, is essential for the correct assembly of human basement membranes in kidney and skin. *Blood* 2004;104:2217-23.
- 2. PMID: Karamatic Crew V, Poole J, Long S et al. Two MER2-negative individuals with the same novel CD151 mutation and evidence for clinical significance of anti-MER2. *Transfusion* 2008; 48:1912-6.
- 3. Abstract Karamatic Crew V, Poole J, Bullock T et al. A new case and a novel molecular background in a MER2-negative (RAPH:-1) individual with anti-MER2. *Vox Sang* 2012;103(s1): 210-211 (P-452).

Track of changes		from v4.0 160662	to v5.0 01-MAR-2020		
	Created: Reviewed:		Vered Yahalom, December 2019 Vanja Karamatic Crew		
			Nicole Thornton, January 2020		
General			First Excel map version. Spread-sheets "Intro", "Allele Table", "References" and "Versioning" created.		
Intro	Antithetical alleles line added	n.a.	n.a.		
Allele Table	Table column and header additions	n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rsnumber" created and content to table columns added.		
Allele Table	Antigen/allele added:	n.a.	RAPH*0101.03 provisional allele added		
		n.a.	Karamatic Crew V, Poole J, Bullock T et al:		
			A new case and a novel molecular background in a MER2-negative		
			(RAPH:-1) individual with anti-MER2.		
- 0			Vox Sang 2012; 103(s1) : 210-211 (P-452).		
References	All references new:	n.a.	All references (1) to (3) added for the first time.		
End of changes		from v4.0 160662	to v5.0 01-MAR-2020		
Track of ch	anges	from v5.0 01-MAR-2020	to v6.0 30-OCT-2020		
Allele Table	Allele renamed	RAPH*-01.01	RAPH*0101.01		
	Allele renamed	RAPH*-01.02	RAPH*0101.02		
	Allele renamed	RAPH*-01.03	RAPH*0101.03		
End of chai	nges	from v5.0 01-MAR-2020	to v6.0 30-OCT-2020		