Names for AUG (ISBT 036) Blood Group Alleles

Intro

General description: The Augustine (AUG) blood group system consists of four antigens carried

on a multipass membrane glycoprotein equilibrative nucleoside transporter

1 (ENT1). It consists of 456 amino acids with an intracellular amino

terminus and extracellular carboxyl terminus.

Gene name: SLC29A1

Number of exons: 13

Initiation codon: within exon 1 Stop codon: within exon 13

Entrez Gene ID: 2030

LRG: LRG_1027

LRG sequence: NG 042893.1 (genomic)

NM 001304463.1 (transcript)

Reference allele: AUG*01

Reference allele AUG1, At^a, ATAM

AUG*01 encodes:

Phenotype	Allele name	Nucleofide change			l` '	Accession number	rs number	
AUG1+, At(a+), ATML-, ATAM+ AUG:1,2,-3,4	AUG*01							
At(a-) AUG:-2	AUG*02	c.1171G>A	12	p.Glu391Lys	PMID: 25896650		rs45458701	
ATML+ AUG:3	AUG*03	c.1159A>C	12	p.Thr387Pro	PMID: 29504136	MF034879	rs1131690802	
ATAM- AUG:-4	AUG*04	c.242A>G	4	p. Asn81Ser	(1), Abstract		rs72555353	
Null phenotype								
AUG1- AUG:-1	AUG*01N	c.589+1G>C	i6-i7	p.Ser197fs	PMID: 25896650			

References

- PMID 25896650 Daniels G, Ballif B, Helias V, et al. Lack of the nucleoside transporter ENT1 results in the Augustine-null blood type and ectopic mineralization. *Blood* 2015:125(23):3651-4.
- PMID 29504136 Millard GM, McGowan EC, Wilson B et al. A proposed new low-frequency antigen in the Augustine blood group system associated with a severe case of hemolytic disease of the fetus and newborn. *Transfusion* . 2018;58:1320-1322
- Abstract (1) Vrignaud C, Mikdar M, Raneri A, et al. Characterization of a novel highprevalence red blood cell antigen in the Augustine blood group system. *Vox* Sang 2018;113(Suppl.1):64-5 [Abstract]

Track of changes

		,	from	to
1	Version		v1.0 08-APR-2019	v2.0 31-MAR-2022
2	Author	created:	Thierry Peyrard, April 2019	Jill Storry, January 2022
3	Review	reviewed:	Slim Azouzi, April 2019	Nicole Thornton, February 2022
4	General	document	v4.0 Word-document	First Excel map version. Spread-sheets 'Intro', 'Allele Table', 'References', 'Versioning (v1.1)' created.
5	Intro	update to reference alelle information		ATAM added to antigens encoded by reference-allele $AUG*01$ encodes:
6	Table	update to the reference-allele		Complete phenotype information added for reference-allele AUG*01
7	Table	formatting		Format changed for phenotype entry of $AUG*03$, to bring in line with other table entries
8	Table	correction		Phenotype information for <i>AUG*04</i> allele changed from ATAM+, AUG:4 to ATAM-, AUG:-4. Also format change as above.
9	Table	update to allele entry		Alternative phenotype nomenclautre (AUG1+) added to $AUG*01N$ entry
10	10 End Version		v1.0 08-APR-2019	v2.0 31-MAR-2022