Names for CD59 (ISBT 035) Blood Group Alleles

Intro

General description:	The CD59 blood group system consists of 1 antigen carried on a 20 kDa glycosylphosphatidylinositol (GPI) linked glycoprotein (CD59). It consists of 128 amino acids and has a signal sequence of 25 amino acids. Another 26 amino acids are removed from the C-terminal end of mature protein which consists of 77 amino acids.
Gene name:	*CD59*
Number of exons:	6
Initiation codon:	Beginning of exon 4
Stop codon:	Within exon 6
Entrez Gene ID:	966
LRG:	LRG_41
LRG sequence:	NG_008057.1 (genomic) NM_203330.2 (transcript) NP_976075.1 (protein)
Reference allele:	*CD59*01* (shaded)
Reference allele *CD59*01* encodes:	CD59.1

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
CD59:+1 or CD59.1+	*CD59*01*					NG_008057	
CD59:+1 or CD59.1+	*CD59*01.02*†	c.238A>G	6	p.Arg80Gly	(1), PMID: 30325039	MH165189	n.a.
Null phenotypes							
CD59:-1 or CD59.1-	*CD59*01N.01*	c.146delA	5	p.Asp49Valfs*31	(2), PMID: 24382084	n.a.	rs587777149
CD59:-1 or CD59.1-	*CD59*01N.02*	c.123delC c.361delG	5 6	p.Val42Serfs*38 not applicable	(3), PMID: 1382994	n.a.	rs577673753 rs1435725939
CD59:-1 or CD59.1-	*CD59*01N.03*	c.266G>A	6	p.Cys89Tyr	(4), PMID: 23149847	n.a.	rs397514767
CD59:-1 or CD59.1-	*CD59*01N.04*	c.146A>T	5	p.Asp49Val	(5), PMID: 25716358	n.a.	rs587777149

† Provisional number

References

1. PMID: 30325039 Li XF, Lin FQ, Li JP. Identification of c.238 A>G (p.Arg80Gly) of CD59 blood group gene. Transfusion (2018) 58(12), 3033-4.

2. PMID: 24382084 Höchsmann B, Dohna-Schwake C, Kyrieleis HA, et al. Targeted therapy with eculizumab for inherited CD59 deficiency. N Engl J Med (2014) 370(1), 90-2.

3. PMID: 1382994 Motoyama N, Okada N, Yamashina M, et al. Paroxysmal nocturnal hemoglobinuria due to hereditary nucleotide deletion in the HRF20 (CD59) gene. Eur J Immunol (1992) 22(10), 2669-73.

4. PMID: 23149847 Nevo Y, Ben-Zeev B, Tabib A, et al. CD59 deficiency is associated with chronic hemolysis and childhood relapsing immune-mediated polyneuropathy. Blood (2013) 121(1), 129-35.

5. PMID: 25716358 Haliloglu G, Maluenda J, Sayinbatur B, et al. Early-onset chronic axonal neuropathy, strokes, and hemolysis: inherited CD59 deficiency. Neurology (2015) 84(12), 1220-4.

Track of changes		**from v1.3 2020.01.02.**	**to v2.0 25-FEB-2020**
	created:	Christof Weinstock, 2nd of January 2020	Christof Weinstock, 31st of January 2020
	reviewed:	n.a.	
General			First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
Intro	LRG ID line added:	n.a.	LRG_41
Intro	Reference allele line moved from Allele Table to Intro	n.a.	Reference allele *CD59*01* encodes: CD59.1
Allele Table		n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
Allele Table	Text change: Line moved to Intro	Reference allele *CD59*01* encodes: CD59.1	moved to Intro, see above
Allele Table	Antigen/allele added:	n.a.	CD59*01.02 provisional
		n.a.	Li XF, Lin FQ, Li JP. Identification of c.238 A>G (p.Arg80Gly) of CD59 blood group gene. Transfusion (2018) 58(12), 3033-4.
Allele Table	Antigen/allele added:	n.a.	*CD59*01N.04*
		n.a.	Haliloglu G, Maluenda J, Sayinbatur B, et al. Early-onset chronic axonal neuropathy, strokes, and hemolysis: inherited CD59 deficiency. Neurology (2015) 84(12), 1220-4.
Allele Table	References added:	n.a.	All references added for the first time.
	Gene Bank accession no. added:	n.a.	All Gene Bank accession numbers added for the first time.
	rs no. added:	n.a.	All rs numbers numbers added for the first time.
Reference Table		n.a.	Table added
	References added:	n.a.	All references added for the first time.
End of changes		**from v1.3 2020.01.02.**	**to v2.0 25-FEB-2020**