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 Intron	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	PMID: 30325039	MH165189	n.a.
Null phenotypes							
CD59:-1 or CD59.1-	***CD59*01N.03***	c.266G>A	6	p.Cys89Tyr	PMID: 23149847	n.a.	rs397514767
CD59:-1 or CD59.1-	***CD59*01N.04***	c.146A>T	5	p.Asp49Val	PMID: 25716358	n.a.	rs587777149
CD59:-1 or CD59.1-	***CD59*01N.05*** †	c.323C>A	6	p.Ser108Ter	PMID: 31752029	n.a.	rs749308157
CD59:-1 or CD59.1-	***CD59*01N.06*** †	c.85T>G	5	p.Tyr29Asp	PMID: 32612799	n.a.	rs1564972905

References

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.

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.

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.

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.

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.

PMID 31752029 Solmaz I, Aytekin ES, Çagdas D, Tan C, Tezcan I, Gocmen R et al: Recurrent Demyelinating Episodes as Sole Manifestation of Inherited CD59 Deficiency. Neuropediatrics 2020;51:206-10.

PMID 32612799 Javadi Parvaneh V, Ghasemi L, Rahmani K, Shiari R, Mesdaghi M, Chavoshzadeh Z et al: Recurrent angioedema, Guillain-Barré, and myelitis in a girl with systemic lupus erythematosus and CD59 deficiency syndrome. Auto Immun Highlights 2020;11:9.

	Track of changes		from	to
1	**Version**		**v2.0 25-FEB-2020**	**v3.0 30-NOV-2021**
2	Author	created:	Christof Weinstock, January 2020	Christof Weinstock, November 2021
3	Reviewer	reviewed:	n.a.	Christoph Gassner, November 2021
4	Allele Table	Antigen/allele added:	n.a.	*CD59*01N.05*
5	Allele Table	nucleotide change	n.a.	c.323C>A
6	Allele Table	exon added	n.a.	6
7	Allele Table	predicted amino acid change	n.a.	p.Ser108Ter
8	Allele Table	PMID	n.a.	PMID: 31752029
9	Allele Table	rs-number	n.a.	rs749308157
10	Allele Table	Antigen/allele added:	n.a.	*CD59*01N.06*†
11	Allele Table	nucleotide change	n.a.	c.85T>G
12	Allele Table	exon added	n.a.	5
13	Allele Table	predicted amino acid change	n.a.	p.Tyr29Asp
14	Allele Table	PMID	n.a.	PMID: 32612799
15	Allele Table	rs-number	n.a.	rs1564972905
16	Allele Table	Allele added	n.a.	*CD59*01N.05*†
17	Allele Table	Allele added	n.a.	*CD59*01N.06*†
18	Reference Table	References added	n.a.	PMID 31752029, PMID 32612799
20	**End of changes**		**to v2.0 25-FEB-2020**	**v3.0 30-NOV-2021**

	Track of changes		from	to
1	**Version**		**v1.3 2020.01.02.**	**v2.0 25-FEB-2020**
2	Author	created:	Christof Weinstock, January 2020	Christof Weinstock, January 2020
3	Reviewer	reviewed:	n.a.	n.a.
4	General			First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	LRG ID line added:	n.a.	LRG_41
6	Intro	Reference allele line moved from Allele Table to Intro	n.a.	Reference allele *CD59*01* encodes: CD59.1
7	Allele Table		n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
8	Allele Table	Text change: Line moved to Intro	Reference allele *CD59*01* encodes: CD59.1	moved to Intro, see above
9	Allele Table	Antigen/allele added:	n.a.	CD59*01.02 provisional
10			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.
11	Allele Table	Antigen/allele added:	n.a.	*CD59*01N.04*
12			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.
13	Allele Table	References added:	n.a.	All references added for the first time.
14		Gene Bank accession no. added:	n.a.	All Gene Bank accession numbers added for the first time.
15		rs no. added:	n.a.	All rs numbers numbers added for the first time.
16	Reference Table		n.a.	Table added
17		References added:	n.a.	All references added for the first time.
18	**End of changes**		**from v1.3 2020.01.02.**	**to v2.0 25-FEB-2020**