# 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)<br>NM_203330.2 (transcript)<br>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.01* | c.146delA | 5 | p.Asp49Valfs*31 | PMID: 24382084 | n.a. | rs587777149 |
| CD59:-1 or CD59.1- | *CD59*01N.02* | c.123delC c.361delG | 5<br>6 | p.Val42Serfs*38<br>not applicable | PMID: 1382994 | n.a. | rs577673753<br>rs1435725939 |
| 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 |

† Provisional number

**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** | | **v3.0 30-NOV-2021** | **v3.1 30-SEP-2023** |
| **2** | Author | created: | Christof Weinstock, November 2021 | Christof Weinstock, September 2023 |
| **3** | Reviewer | reviewed: | Christoph Gassner, November 2021 | Christoph Gassner, September 2023 |
| **4** | Allele Table | corrected | *CD59*01N.01* invisible line | *CD59*01N.01* is visible again |
| **5** | Allele Table | corrected | *CD59*01N.02* invisible line | *CD59*01N.02* is visible again |
| **6** | **End Version** | | **v3.0 30-NOV-2021** | **v3.1 30-SEP-2023** |

| | 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 Version** | | **from v1.3 2020.01.02.** | **to v2.0 25-FEB-2020** |