# Names for PEL (ISBT 040) Blood Group Alleles

## Intro

| | |
|---|---|
| General description: | The PEL blood group system consists of one high-prevalence antigen, PEL, initially included in the 901 series (see PMID 8928488). It is carried on the ABCC4 protein (ATP-binding cassette sub-family C member 4), member of the superfamily of ATP-binding cassette transporters. ABCC4 is also known as the multidrug resistance-associated protein 4 (MRP4). ABC proteins transport various molecules across extra- and intra-cellular membranes. This multi-pass protein consists of 1325 amino acids, with predicted 12 transmembrane domains and 6 extracellular loops. The protein is encoded by *ABCC4,* 281,641 bases, chromosome *13q32.1* (*chr13:95,019,835-95,301,475* ) (GRCh38/hg38). The rare PEL–null phenotype is associated with a moderately impaired platelet aggregation. |
| Gene name: | *ABCC4* |
| Number of exons: | 31 |
| Initiation codon: | Within exon 1 |
| Stop codon: | Within exon 31 |
| Entrez Gene ID: | 10257 |
| LRG: | LRG_1183 |
| LRG sequence: | NC_000013.11 (genomic)<br>NM_005845.5 (transcript)<br>NP_005836.2 (protein) |
| Reference allele: | *ABCC4*01* (shaded) |
| Reference allele *ABCC4*01* encodes: | PEL |
| Antithetical antigens: | n/a |

| Phenotype | Allele name | Nucleotide change | Exon Intron | Predicted amino acid change | (Reference No.) PMID | Accession number | rs number | Comments |
|---|---|---|---|---|---|---|---|---|
| PEL:1 or PEL+ | *ABCC4*01* | | | | | | | |
| PEL:1 or PEL+w | *ABCC4*01.01W* | c.559G>T | 5 | p.Gly187Trp | PMID: 31826245 | | rs11568658 | Reduces PEL expr. by 16% |
| PEL:1 or PEL+w | *ABCC4*01.02W* | c.912G>T | 8 | p.Lys304Asn | PMID: 31826245 | | rs2274407 | Reduces PEL expr. by 30% |
| PEL:1 or PEL+w | *ABCC4*01.03W* | c.2269G>A | 18 | p.Glu757Lys | PMID: 31826245 | | rs3765534 | Reduces PEL expr. by 33% |
| | | | | | | | | |
| PEL:–1 or PEL– | *ABCC4*01N.01* | Deletion of the last 10 exons and part of downstream 3'UTR (67,528-bp deletion). Breakpoint:chromosome 13:95 018 454 to 95 085 982 ((GRCh38/hg38 assembly). Deletion accompanied by the insertion of an intervening 18-bp sequence that corresponds to an intronic sequence repeated 62 times in the *ABCC4* gene | del21-31 | no protein produced | PMID: 31826245 | | | |

## References

PMID 8928488 Daniels GL, Simard H, Goldman M, Taliano V, Fleury M, Decary F, Spurll G, Garcia C, Ouellet P. PEL, a 'new' high-frequency red cell surface antigen. *Vox Sanguinis* 1996;70(1):31-3.

PMID 31826245 Azouzi S, Mikdar M, Hermand P, Gautier E.F, Salnot V, Willemetz A, Nicolas G, Vrignaud C, Raneri A, Mayeux P, Bole-Feysot C, Nitschké P, Cartron J.P, Colin ,Y Hermine O, Jedlitschky G, Cloutier M, Constanzo-Yanez J, Ethier C, Robitaille N, St-Louis M, Le Van Kim C. & Peyrard T. (2020) Lack of the multidrug transporter MRP4/ABCC4 defines the PEL-negative blood group and impairs platelet aggregation. Blood 135 441-448.

## Track of changes

| | | | from<br>v1.1 31-MAR-2022 | to<br>v1.2 30-SEP-2022 |
|---|---|---|---|---|
| **1** | Author | created by | | |
| **2** | Review | reviewed by | Jill Storry Feb 2022, Ji Yanli March 2022 | Jill Storry Feb 2022, Ji Yanli March 2022 |
| **3** | Allele | corrected | | *ABCC4*01.02W*: nucleotide change corrected from p.Lys304Arg to p.Lys304Asn |
| **4** | **End Version** | | **v1.1 31-MAR-2022** | **v1.2 30-SEP-2022** |

## Track of changes

| | | | from<br>v1.0 30-OCT-2020 | to<br>v1.1 31-MAR-2022 |
|---|---|---|---|---|
| **1** | Author | created by | Thierry Peyrard | |
| **2** | Review | reviewed by | Slim Azouzi | Jill Storry Feb 2022, Ji Yanli March 2022 |
| **3** | General | Document created | First version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created. | |
| **4** | Intro | Intro added | Generel description, gene name, number of exons, initiation codon, stop codon, Entrez Gene ID and Reference allele information added. | |
| **5** | Allele Table | Table created | Table columns "Phenotype", "Allele name", "Nucleotide change", "Exon", "Predicted amino acid change", "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added. | |
| **6** | Allele Table | Alleles added | *ABCC4*01* and *ABCC4*01N.01* added | |
| **7** | Allele Table | Alleles added | | Added 3 alleles that express the PEL antigen weakly, based on expression data from reference 2: *ABCC4*01.01W, ABCC4*01.02W, ABCC4*01.03W* |
| **8** | References | numbering changed | | (1), (2) replaced by PMIDs |
| **9** | References | References added | | PMID 8928488 |
| **10** | References | References added | | Reference added to introduction: PMID 8928488 |
| **11** | **End Version** | | **v1.0 30-OCT-2020** | **v1.1 31-MAR-2022** |