Names for PEL (ISBT 040) Blood Group Alleles

General description:	The PEL blood group system consists of one high-prevalence antigen, PEL, initially included in the 901 series. 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) NM_005845.5 (transcript) 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	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
PEL:1 or PEL+	*ABCC4*01*						
PEL:–1 or PEL–	*ABCC4*01N.01*	Deletion of the last 10 exons and part of downstream 3'UTR (67,528-bp deletion). Breakpoints of the deletion between intron 21 and the intergenic region, with coordinates from chromosome 13:95 018 454 to 95 085 982. Deletion accompanied by the insertion of an intervening 18-bp sequence that corresponds to an intronic sequence repeated 62 times in the *ABCC4* gene			PMID: 31826245		

PEL was the name already assigned in the 901 series, after the proband's name (Daniels GL, Simard H, Goldman M, et al.

PEL, a “new” high-frequency red cell surface antigen. Vox Sang. 1996;70(1):31-33)

References

1. 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		**v1.0 30-OCT-2020**	**to**
	created by	Thierry Peyrard	
	reviewed by	Slim Azouzi	
General	Document created	First version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.	
Intro	Intro added	Generel description, gene name, number of exons, initiation codon, stop codon, Entrez Gene ID and Reference allele information added.	
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.	
Allele Table	Alleles added:	*ABCC4*01* and *ABCC4*01N.01* added	
References	References added:	References (1)	
End of changes		**v1.0 30-OCT-2020**	