Names for CTL2 (ISBT 039) Blood Group Alleles

Intro

General description:	The CTL2 blood group system consists of two high-prevalence antigens, VER and RIF, carried on the Choline like transporter 2 (CTL2) protein, also known as SLC44A2 (solute carrier family 44 member 2). This multi-pass protein consists of 706 amino acids (isoform 1), with predicted 10 transmembrane domains and 5 extracellular loops. The protein is encoded by *SLC44A2,* 42,103 bases, chromosome *19p13.2* (*chr19:10,602,455-10,644,557*) (GRCh38/hg38). The rare VER– null phenotype is associated with hearing impairment in the upper frequency range.
Gene name:	*SLC44A2*
Number of exons:	22
Initiation codon:	Within exon 1
Stop codon:	Within exon 22
Entrez Gene ID:	57153
LRG:	no record
LRG sequence:	NC_000019.10 (genomic) NM_001145056.2 (transcript) NP_001138528.1 (protein)
Reference allele:	*CTL2*01* (shaded)
Reference allele *CTL2*01* encodes:	CTL2.1 (VER), CTL2.2 (RIF)
Antithetical antigens:	n/a

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
CTL2:1 or VER+	***CTL2*01***						
CTL2:–2 or RIF–	***CTL2*01.-02***	c.1192C>A	14	p.Pro398Thre	(1), Abstract		rs1401833882
Null Phenotypes							
CTL2:–1 or VER–	***CTL2*01N.01***	Deletion of exons 1 to 14 (37 kbp). Break-points with coordinates from chromosome 19:10,598,733-10,636,021		no protein product	(1), Abstract		

VER: name suggested after the name of the city of origin of the null proband, Verona in Italy

VER–: deletion of 37 kbp (exons 1 to 14 along with the 5' UTR region of *SLC44A2*)

RIF: name suggested after the name of the North Mediterranean coast of Morocco from where all the reported RIF– people originate to date.

References

Abstract (1) Vrignaud C., Mikdar M., Koehl B., Nair T.S., Yang L., Laiguillon G., El Kenz H., Cartron J.P., Colin Y., Detante O., Le Van Kim C., Carey T.E., Azouzi S., Peyrard T. (2019) Alloantibodies directed to the SLC44A2/CTL2 transporter define two new red cell antigens and a novel human blood group system. *Transfusion,* 59 (Suppl. S3), 18A[abstract].

Track of changes

			from	to
1	**Version**		**v1.0 30-OCT-2020**	**v1.1 31-MAR-2022**
2	Author	created by	Thierry Peyrard, Oct. 2020	Thierry Peyrard, April 2022
3	Reviewer	reviewed by	Slim Azouzi, Oct. 2020	Jill Storry Dec. 2021, Christof Weinstock April 2022
4	General	Document created	First version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.	Updated to new project-2-format
5	Intro	Intro added	General description, gene name, number of exons, initiation codon, stop codon, Entrez Gene ID and Reference allele information added.	
6	Allele Table	Table created	Table columns "Phenotype", "Allele name", "Nucleotide change", "Exon", "Predicted amino acid change", "(Reference No.) PMID/WOS", "Accession number" and "rs-number" created and content to table columns added.	
7	Allele Table	Alleles added:	*CTL2*01,CTL2*01N.01* and *CTL2*01.-02* added	Updated the consequence of the *CTL2*01N.01* mutation; completed the definition of RIF.
8	References	References added:	References (1)	WOS-number is now "(1), Abstract"
9	Intro	LRG		Checked that there was no LRG record
10	**End Version**		**v1.0 30-OCT-2020**	**v1.1 31-MAR-2022**