Names for CTL2 (ISBT 039) Blood Group Alleles

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: not yet assigned
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
<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Allele name</th>
<th>Nucleotide change</th>
<th>Exon</th>
<th>Predicted amino acid change</th>
<th>(Reference No.) PMID</th>
<th>Accession number</th>
<th>rs number</th>
</tr>
</thead>
<tbody>
<tr>
<td>CTL2:1 or VER+</td>
<td>CTL2*01</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CTL2:–2 or RIF–</td>
<td>CTL2*01.-02</td>
<td>c.1192C&gt;A</td>
<td>14</td>
<td>p.Pro398Thre</td>
<td>WOS:000502826600026</td>
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<td>rs1401833882</td>
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</table>

**Null Phenotypes**

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Allele name</th>
<th>Nucleotide change</th>
<th>Exon</th>
<th>Predicted amino acid change</th>
<th>(Reference No.) PMID</th>
<th>Accession number</th>
<th>rs number</th>
</tr>
</thead>
<tbody>
<tr>
<td>CTL2:–1 or VER–</td>
<td>CTL2*01N.01</td>
<td>Deletion of exons 1 to 14 (37 kbp). Break-points with coordinates from chromosome 19:10,598,733-10,636,021</td>
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<td></td>
<td>WOS:000502826600026</td>
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</tr>
</tbody>
</table>

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 so far reported RIF– people originate from
References

General Document created

Intro Intro added

Allele Table Table created

Allele Table Alleles added:

References References added:

End of changes