Names for SID (ISBT 038) Blood Group Alleles

Intro

General description: Sd^a or SID1, the only antigen of the SID blood group system, was discovered in 1967. About 90% of the European population carries the antigen on their red blood cells (RBCs). Although there are 10% missing the antigen on their RBCs only 4% lack expression in all tissues and fluids and thereby have the true null phenotype, Sd(a–). The antibodies against Sd^a form a characteristic RBC agglutination pattern of small agglutinates surrounded by numerous free cells. The *B4GALNT2* -encoded transferase synthesizes the Sd^a antigen by the addition of an *N* -acetylgalactosamine to its precursors, which can be glycans on glycoproteins or glycosphingolipid conjugates (in the neolacto synthetic pathway). Alterations in *B4GALNT2* that abolish transferase activity cause the Sd(a–) phenotype. Another phenotype, the rare Cad or Sd(a++) or Super-SID, describes RBCs that are more strongly agglutinated by anti-Sd^a. If this trait is quantitative or qualitative is to date not fully understood, nor is its genetic background. Differences from reference allele *SID*01* (accession number AJ517770) are given in the table.

Gene name:	*B4GALNT2*
Number of exons:	12†
Initiation codon:	Within exon 1-long (accession number AJ517770) or 1-short (AJ517771) †
Stop codon:	Within exon 11
Entrez Gene ID:	124872
LRG:	Not assigned
LRG sequence:	Not assigned
Reference allele:	*SID*01* (shaded)
Reference allele *SID*01* encodes:	*SID1*
Antithetical antigens:	n.a.

† Two transcripts have been experimentally associated with the gene. The enzymes translated from the different transcripts differ in the lengths of the N-terminal cytosolic domains, encoded by different exon 1 (long or short). Thereby there are in total 12 exons associated with the gene, although each transcript only utilizes 11 exons.

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
SID1 or Sd(a+)	*SID*01*					AJ517770	
Null Phenotypes							
Sd(a–)	*SID*01N.01*‡	c.1396T>C	10	p.Cys466Arg	(1) PMID: 31367682, (2) Abstract	MK765047	rs7224888
Sd(a–)	*SID*01N.02*	c.1134+5G>A	(Intron 8)	Splice-site defect predicted	(1) PMID: 31367682, (2) Abstract	MK797056	rs72835417
Sd(a–)	*SID*01N.03*	c.1307A>G	10	p.Glu436Arg	(1) PMID: 31367682	MK765048	rs148441237
Sd(a–)	*SID*01N.04*	c.1567C>T	11	p.Arg523Trp	(1) PMID: 31367682	MK765049	rs61743617

‡This is by far the most predominant allele associated with the Sd(a-) phenotype. It is also the only allele confirmed to abolish Sd^a synthesis in in vitro experiments

References

1. PMID: 31367682 Stenfelt L, Hellberg Å, Möller M, Thornton N, Larson G, Olsson ML. Missense mutations in the C-terminal portion of the *B4GALNT2* -encoded glycosyltransferase underlying the Sd(a-) phenotype. Biochem Biophys Rep. (2019) 17(19), 100659.

2. Abstract Veldhuisen B, Ligthart P, Mark-Zoet J van der, Javadi A, Tissoudali A, Dengerink I, Folman C, van der Schoot CE. Identification of a single homozygous mutation in the B4GALNT2 gene in individuals lacking the Sd(a) (SID) antigen on red blood cells. Vox Sang. (2019) 114 (S1), 5-240

Track of changes v1.0 30-OCT-2020

	created:	Linn Stenfelt, January 2020
	reviewed:	Åsa Hellberg, January 2020 Martin L. Olsson, January 2020
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:	*SID*01* and *SID*01N.01-04*
References	References added:	References (1) and (2).

End of changes v1.0 30-OCT-2020