Names for DI (ISBT 010) Blood Group Alleles

Intro

General description:	The Diego blood group system consists of 23 antigens carried on a multipass membrane glycoprotein called band 3 (aka *AE1* ; *SLC4A1* ; CD233). It consists of 911 amino acids and both amino and carboxyl termini are predicted to be intracellular. The transmembrane domains of band 3 function as the RBC anion transporter while the long amino terminal region of the protein is critical to maintaining RBC shape integrity through its interaction with the cytoskeleton. With the introduction of the DIST antigen there are 23 antigens.
Gene name:	*DI*
Number of exons:	20
Initiation codon:	Within exon 1
Stop codon:	Within exon 20
Entrez Gene ID:	6521
LRG:	LRG_803
LRG sequence:	NG_007498.1 (genomic) NM_000342.4 (transcript)
Reference allele:	*DI*02* (shaded) Acceptable: *DI*B* , or *Di*^{b} if inferred by haemagglutination
Reference allele *DI*02* encodes:	DI*02 encodes Dib, Wrb, DISK
Antithetical antigens:	[DI1 DI2]; [DI3 DI4]; [DI9 DI22]; [DI11 DI12]; [DI15 DI16]; [DI17 DI18];

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
DI:1,–2 or Di(a+b–)	*DI*01 or DI*A*	c.2561C>T	19	p.Pro854Leu	PMID: 8206915		rs2285644
DI:–1,2 or Di(a–b+)	*DI*02 or DI*B*	c.2561C	19	p.Pro854	PMID: 8206915		
DI:3,–4 or Wr(a+b–)	*DI*02.03*	c.1972G>A	16	p.Glu658Lys	PMID: 7812009		rs75731670
DI:-3,4 or Wr(a-b+)	*DI*02.04*	c.1972G	16	p. Glu658	PMID: 7812009		
DI:5 or Wd(a+)	*DI*02.05*	c.1669G>A	14	p.Val557Met	PMID: 6941697 PMID: 8873423 PMID: 9191821		rs121912743
DI:6 or Rb(a+)	*DI*02.06*	c.1643C>T	14	p.Pro548Leu	PMID: 9191821		rs879202054
DI:7 or WARR+	*DI*02.07*	c.1655C>T	14	p.Thr552Ile	PMID: 7625077		
DI:8 or ELO+	*DI*02.08*	c.1294C>T	12	p.Arg432Trp	PMID: 9460189		rs373768879
DI:9,–22 or Wu+, DISK–	*DI*02.09*	c.1694G>C	14	p.Gly565Ala	PMID: 1471249		rs551784583
DI:10 or Bp(a+)	*DI*02.10*	c.1707C>A	14	p.Asn569Lys	PMID: 9460189 PMID: 9845551		
DI:11 or Mo(a+)	*DI*02.11*	c.1967G>A	16	p.Arg656His	PMID: 9460189		rs758868427
DI:12 or Hg(a+)	*DI*02.12*	c.1966C>T	16	p.Arg656Cys	PMID: 9460189 PMID: 9845551		rs372514760
DI:13 or Vg(a+)	*DI*02.13*	c.1663T>C	14	p.Tyr555His	PMID: 9460189 PMID: 9845551		
DI:14 or Sw(a+)	*DI*02.14.01*	c.1937G>A	16	p.Arg646Gln			
DI:14 or Sw(a+)	*DI*02.14.02*	c.1936C>T	16	p.Arg646Trp			
DI:15 or BOW+	*DI*02.15*	c.1681C>T	14	p.Pro561Ser	PMID: 1471249 PMID: 10738034		
DI:16 or NFLD+	*DI*02.16*	c.1287A>T c.1681C>G	12 14	p.Glu429Asp p.Pro561Ala	PMID: 1471249 PMID: 10738034		
DI:17 or Jn(a+)	*DI*02.17*	c.1696C>T	14	p.Pro566Ser	PMID: 15373634		rs1393742050

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
DI:18 or KREP+	*DI*02.18*	c.1696C>G	14	p.Pro566Ala	PMID: 15373634		
DI:19 or Tr(a+)	*DI*02.19*	c.1653G>C	14	p.Lys551Asn	PMID: 9191821		
DI:20 or Fr(a+)	*DI*02.20*	c.1438G>A	13	p.Glu480Lys	PMID: 3604156 PMID: 10738034		rs121912756
SW1 or DI:21	*DI*02.21*	c.1936C>T	16	p.Arg646Trp	PMID: 11155072 PMID: 24094240		rs121912758
DISK or DI:22	*DI*02.22*	c.1694G>C	14	p.Gly565Ala	(1), Abstract		
DIST or DI:23	*DI*02.23*	c.1447G>A	13	p.Gly483Ser	(2), Abstract		rs544557335
Null phenotypes							
Di(a–b–)	*DI*02N.01*	c.1462G>A	13	p.Val488Met	PMID: 10942416		

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Track of changes			from version	to version
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2	Author	created:	Silvano Wendel v3.0	Silvano Wendel v4.0, June 2021
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