

Names for DO (ISBT 014) Blood Group Alleles

Intro

General description: The Dombrock blood group system consists of 10 antigens carried on a GPI-linked glycoprotein (DO, ART4, CD297) that consists of 314 amino acids. It has a leader sequence and a GPI motif, both of which are cleaved from the membrane bound protein. The *DO* gene consists of 3 exons distributed over 18 kb of gDNA.

Gene name: *ART4 (DO)*

Number of exons: 3

Initiation codon: Within exon 1

Stop codon: Within exon 3

Entrez Gene ID: 420

LRG: LRG_807

LRG sequence: NG_007477.2 (genomic)
NM_021071.4, ENST00000228936.6 (transcript)

Reference allele: *DO*02* (shaded)

Acceptable: *DO*B*, or *Do^b* if inferred by haemagglutination

Reference allele

*DO*02* encodes: DO2, DO3, DO4, DO5, DO6, DO7, DO8, DO9, DO10

Antithetical antigens: [DO1 DO2]

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
DO:1+ or Do(a+)	<i>DO*01</i> or <i>DO*A</i>	c.793G>A	2	p.Asp265Asn	PMID: 11552072	NM021071 AF290204	rs11276
DO:2 or Do(b+)	<i>DO*02</i> or <i>DO*B</i>	c.793G	2	p.Asp265	PMID: 11552072	NM021071 AF290204	rs11276
DO:-4 or Hy-	<i>DO*02.-04</i>	c.323G>T	2	p.Gly108Val	PMID: 11896313	AH011615 AH011616	rs28362797
DO:-5 or Jo(a-)	<i>DO*01.-05</i>	c.350C>T c.793G>A	2 2	p.Thr117Ile p.Asp265Asn	PMID: 11896313	AH011617	rs28362798 rs11276
DO:-6 or DOYA-	<i>DO*01.-06</i>	c.547T>G c.793G>A	2 2	p.Tyr183Asp p.Asp265Asn	PMID: 20088839	n.a.	n.a. rs11276
DO:-7 or DOMR-	<i>DO*02.-07</i>	c.431C>A c.432C>A	2 2	p.Ala144Glu	PMID: 20412531	GU724770	rs1355202105 rs1210078970
DO:-8 or DOLG-	<i>DO*01.-08</i>	c.674T>A c.793G>A	2 2	p.Leu225Gln p.Asp265Asn	(1), Abstract	n.a.	rs532592412 rs11276
DO:-9 or DOLC-	<i>DO*01.-09</i>	c.566C>T c.793G>A	2 2	p.Thr189Met p.Asp265Asn	(2), Abstract	n.a.	rs28362800 rs11276
DO:-10 or DODE-	<i>DO*01.-10</i>	c.405C>A c.793G>A	2 2	p.Asp135Glu p.Asp265Asn	(3), Abstract	n.a.	rs28362799 rs11276
Null phenotypes							
DO:-3 or Gy(a-)	<i>DO*01N.01</i>	c.442C>T c.793G>A	2 2	p.Gln148Ter† p.Asp265Asn	PMID: 11552072	AH011373	rs56340844 rs11276
DO:-3 or Gy(a-)	<i>DO*01N.02</i>	c.343_350del c.793G>A	2 2	p.Met115Hisfs*18 p.Asp265Asn	PMID: 11552072	AH011373	rs587777832 rs11276
DO:-3 or Gy(a-)	<i>DO*01N.03</i>	c.219delT c.793G>A	2 2	p.Val73Valfs*5 p.Asp265Asn	(4), Abstract	n.a.	n.a. rs11276
DO:-4 or Gy(a-)	<i>DO*01N.04</i>	c.730dupG (published as c.728_729insG) c.793G>A	2 2	p.Glu244Glyfs*8 p.Asp265Asn	PMID: 33190238	MN082686	rs769684528 rs11276
DO:-5 or Gy(a-)	<i>DO*01N.05</i>	c.93G>A c.370delT c.793G>A	1 2 2	p.Leu31Leu p.Leu124Cysfs*5 p.Asp265Asn	PMID: 33206405	MT747635	rs4106889023 rs2137544113 rs11276
DO:-6 or Gy(a-)	<i>DO*01N.06</i>	c.201delA c.793G>A	2 2	p.Gly68Alafs*10 p.Asp265Asn	(5), Abstract	n.a.	n.a. rs11276
DO:-3 or Gy(a-)	<i>DO*02N.01</i>	c.145-2A>G	i1	Aberrant splicing	PMID: 11724986	AY029516	rs587777831
DO:-3 or Gy(a-)	<i>DO*02N.02</i>	c.144+2T>C	i1	Aberrant splicing	PMID: 12028057	AH011372	rs587777833
DO:-3 or Gy(a-)	<i>DO*02N.03</i>	c.185T>C	2	p.Phe62Ser	PMID: 17655578	EF178609	rs150640567
DO:-3 or Gy(a-)	<i>DO*02N.04</i>	c.268C>T	2	p.Gln90Ter	PMID: 25865759	LC011479	rs759901596

References

- PMID 11552072 Rios M, Hue-Roye K, Lee AH, et al. DNA analysis for the Dombrock polymorphism. *Transfusion* 2001; 41:1143-1146
- PMID 11896313 Rios M, Hue-Roye K, Oyen R, et al. Insights into the Holley- and Joseph-phenotypes. *Transfusion* 2002; 42:52-58
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- PMID 20412531 Costa FPS, Hue-Roy K, Sausais L, et al. Absence of DOMR, a new antigen in Dombrock blood group system that weakens expression of Do^b, Gy^a, Hy, Jo^a, and DOYA antigens. *Transfusion* 2010; 50:2026-2031
- Abstract (1) Karamatic Crew V, Poole J, Marais I, et al. DOLG, a novel high incidence antigen in the Dombrock blood group system. *Vox Sang* 2011; 101 (Suppl. 1):263
- Abstract (2) Karamatic Crew V, Thornton N, Bullock T, et al. Serological and molecular characterization of DOLC, a novel high incidence antigen in the Dombrock blood group system. *Vox Sang* 2013; 105 (Suppl.1), 30
- Abstract (3) Shakarian G, Vege S, Hue-Roye K, et al. A Dombrock system antibody detects a new high-prevalence antigen, DODE. *Transfusion* 2015; 55 (Suppl), 35A-36A
- PMID 11552072 Rios M, Storry JR, Hue-Roye K, et al. Two molecular bases for the Dombrock null phenotype. *Br J Haematol* 2002; 117:765-767
- Abstract (4) Vrignaud C, Ramelet S, Laiguillon G, et al. Characterization of a novel DO*01 silent allele caused by a nucleotide deletion mechanism and responsible for a Gy(a-) phenotype in a patient of French European ancestry with anti-Gy^a. *Transfusion* 2019, 59 (Suppl), 18A
- PMID 33190238 Bub CB, Aravechia MG, Santos L, et al. A novel DO*01 silent allele associated with a nucleotide insertion in a Brazilian patient with anti-Gya. *Transfusion* 2020, online, DOI: 10.1111/trf.16190
- PMID 33206405 Morin,P.-A., Ethier,C., Lavoie,J., Robitaille,N. and Baillargeon,N. A novel variant DO*A allele with a c.370delT mutation leading to a DO null phenotype in a Syrian family. *Transfusion* 2020, online, DOI 10.1111/trf.16193

- Abstract (5) Lubenow N, Petersen B, Sandberg M, Claesson-Linder Y, Jöud M, Storry J. Novel single nucleotide deletion in ART4 accounts for the Gy(a-) phenotype in a woman of Lebanese origin. *Vox Sang* 2020; 115(Suppl)
- PMID 11724986 Lucien N, Celton J-L, Le Pennec P-Y, et al. Short deletion within the blood group Dombrock locus causing a Do_{null} phenotype. *Blood* 2002; 100:1063-1064
- PMID 12028057 Rios M, Hue-Roye K, Storry JR, et al. Molecular basis of the Dombrock null phenotype. *Transfusion* 2001; 41:1405-1407
- PMID 17655578 Westhoff C, Vege S, Yazdanbakhsh K, et al. A DOB allele encoding an amino acid substitution (Phe62Ser) resulting in a Dombrock null phenotype. *Transfusion* 2007; 47:1356-1362
- PMID 25865759 Onodera T, Tsuneyama H, Ogasawara K, et al. A novel DO null allele with a c.268C>T (p.Gln90Stop) mutation in Japanese. *Vox Sang* 2015, 109:191-3

Track of changes		from	to
1	Version	v6.0 30-NOV-2021	v6.2 12-JUL-2022
2	Author	created: Lilian Castilho, November 2021	Lilian Castilho, May 2022
3	Review	reviewed: Barbera Veldhuisen, November 2021	Barbera Veldhuisen, July 2022; C. Hyland, May 2022
4	Allele Table	changed alleles	Corrected wrong amino acid changes for <i>DO*01</i> from p.Asn265Asp to p.Asp265Asn in 12 alleles (according to reference-allele-switch performed in version v6.0)
5	Intro	changed	
6	Allele Table	Exons Introns	Recorrected wrong exon-intron numbering: all back to exon 2, except: <i>DO*01N.05</i> c.93G>A changed from exon 2 => exon 1 <i>DO*02N.01</i> changed i2 => i1 <i>DO*02N.02</i> changed i2 => i1
7	End Version	v6.0 30-NOV-2021	v6.2 12-JUL-2022

Track of changes		from	to
1	Version	v5.0 30-JUN-2021	v6.0 30-NOV-2021
2	Author	created: Lilian Castilho, June 2021	Lilian Castilho, November 2021
3	Review	reviewed: Barbera Veldhuisen, June 2021	Barbera Veldhuisen, November 2021
4	Intro	changed	Reference allele changed from <i>DO*01</i> to <i>DO*02</i>
5	Allele Table	All alleles changed	All alleles changed according correct reference allele (<i>DO*02</i>)
6	Allele Table	Exons	Corrected exon numbering - mistakenly.
7	Allele Table	Introns	Intron numbering added.
8	End Version	v5.0 30-JUN-2021	v6.0 30-NOV-2021

Track of changes			from	to
1	Version		v4.1	v5.0 30-JUN-2021
2	Author	created:	Lilian Castilho, v4.1	Lilian Castilho, June 2021
3	Review	reviewed:	n.a.	Barbera Veldhuisen, June 2021
4	General		Last word version publised on ISBT website	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	Text changed	The Dombrock blood group system consists of 10 antigens carried on a GPI-linked glycoprotein (DO, ART4, CD297) that consists of 314 amino acids. It has a leader sequence and a GPI motif, both of which are cleaved from the membrane bound protein.	The Dombrock blood group system consists of 10 antigens carried on a GPI-linked glycoprotein (DO, ART4, CD297) that consists of 314 amino acids. It has a leader sequence and a GPI motif, both of which are cleaved from the membrane bound protein. The DO gene consists of 3 exons distributed over 14 kbp of gDNA
6	Intro	LRG ID line added:	n.a.	LRG_807
7	Intro	Reference allele line moved from Allele Table to Intro:	n.a.	Reference allele <i>DO*01</i> encodes DO1, DO3, DO4, DO5, DO6, DO7, DO8, DO9, DO10
8	Intro	Antithetical Antigens line created in Intro:	n.a.	Antithetical antigens: [DO1 DO2]
9	Allele Table			Table columns "(Reference No.) PMID", "Accession number" and "rs-number" added, content added.
10	Allele Table	Text change: Line moved to Intro:	Reference allele <i>DO*01</i> encodes DO1, DO3, DO4, DO5, DO6, DO7, DO8, DO9, DO10	see above
11	Allele Table	Allele added:	n.a.	<i>DO*01N.03</i>
12	References	Abstract added	n.a.	Abstract. Vrignaud C, Ramelet S, Laiguillon G, et al. Characterization of a novel <i>DO*01</i> silent allele caused by a nucleotide deletion mechanism and responsible for a Gy(a-) phenotype in a patient of French European ancestry with anti-Gya. Transfusion 2019, 59 (Suppl), 18A

Track of changes		from	to
1	Version	v4.1	v5.0 30-JUN-2021
13	Allele Table Allele added:	n.a.	<i>DO*01N.04</i>
14	References PMID added	n.a.	PMID: 33190238. Bub CB, Aravechia MG, Santos L, et al. A novel DO*01 silent allele associated with a nucleotide insertion in a Brazilian patient with anti-Gya. Transfusion 2020, online , DOI: 10.1111/trf.16190
15	Allele Table Allele added:	n.a.	<i>DO*01N.05</i>
16	References PMID added	n.a.	PMID: 33206405. Morin,P.-A., Ethier,C., Lavoie,J., Robitaille,N. and Baillargeon,N. A novel variant DO*A allele with a c.370delT mutation leading to a DO null phenotype in a Syrian family. Transfusion 2020, online, DOI 10.1111/trf.16193
17	Allele Table Allele added:	n.a.	<i>DO*01N.06</i>
18	References Abstract added	n.a.	Abstract. Lubenow N, Petersen B, Sandberg M, Claesson-Linder Y, Jöud M, Storry J. Novel single nucleotide deletion in ART4 accounts for the Gy(a-) phenotype in a woman pf Lebanese origin. Vox Sang 2020; 115(Suppl)
19	References References new:	n.a.	All references from abstract (4) to PMID 25865759 added for the first time.
20	End Version	v4.1	v5.0 30-JUN-2021