Names for LAN (ISBT 033) Blood Group Alleles

General description: The LAN blood group system consists of one antigen carried on a multipass membrane protein (a.k.a. ATP-binding cassette, sub- family B, member 6 [ABCB6]) of 842 amino acids. ABCB6 is an ATP-dependent transporter of porphyrins (including heme) and is localized in the golgi apparatus, lysosomes, and plasma membranes. The biologically active protein is a homodimer. ABCB6 is up regulated during erythroid maturation.
Gene name: *ABCB6*
Number of exons: 19
Initiation codon: Within exon 1
Stop codon: Within exon 19
Entrez Gene ID: 10058
LRG sequence: NG_032110.1 (genomic)
NM_005689.2 (transcript)
Reference allele: *ABCB6* (shaded)
Acceptable: Lan, if inferred by haemagglutination

Phenotype	Allele name	Molecular basis	Exons/Introns affected	Amino acid change	Reference
Lan+	*ABCB6*01*				
Null alleles					
Lan-	*ABCB6*01N.01*	c.197_198insG	Exon 1	p.Ala66Gly fs stop	[1]
Lan-	*ABCB6*01N.02*	c.717G>A	Exon 3	p.Gln239X	[1]
Lan-	*ABCB6*01N.03*	c.953_956delGTGG	Exon 4	p.Gly318fsX	[1]
Lan-	*ABCB6*01N.04*	c.1533_1543dupCGGCTCCCTGC	Exon 9	p.Leu515fsX	[1]

Phenotype	Allele name	Molecular basis	Exons/Introns affected	Amino acid change	Reference
Lan-	*ABCB6*01N.05*	c.1709_1710delAG	Exon 11	p.Glu570fsX	[1]
Lan-	*ABCB6*01N.06*	c.1690_1691delAT	Exon 11	p.Met564fsX	[1]
Lan-	*ABCB6*01N.07*	c.1867delinsAACAGGTGA	Exon 14	p.Gly623fsX	[1]
Lan-	*ABCB6*01N.08*	c.1942C>T	Exon 14	p.Arg648X	[1]
Lan-	*ABCB6*01N.09*	c.1985_1986delTC	Exon 15	p.Leu662fsX	[1]
Lan-	*ABCB6*01N.10*	c.2256+2t>g	Intron 16	p.Splicing defect	[1]
Lan-	*ABCB6*01N.11*	c.1236G>A	Exon 6	p.Trp412X	[2]
Lan-	*ABCB6*01N.12*	c.1558_1559insT	Exon 9	p.Val520fsX	[2]
Lan-	*ABCB6*01N.13*	c.574C>T	Exon 2	p.Arg192Trp	[3]
Lan-	*ABCB6*01N.14*	c.85_87delTTC	Exon 1	p.Phe29del	[3]
Lan-	*ABCB6*01N.15*	c.376delG	Exon 1	p.Val126fsX	[2]
Lan-	*ABCB6*01N.16*	c.459delC	Exon 1	p.Leu154SerfsX97	[4]
Lan-	*ABCB6*01N.17*	c.IVS16+1g>a	Intron 16	Splicing defect	[4]
Lan-	*ABCB6*01N.18*	c.296_301insG	Exon 1	p.Ala101GlyfsX61	[4]
Lan-	*ABCB6*01N.19*	c.718C>T	Exon 3	p.Arg240X	[4]

Phenotype	Allele name	Molecular basis	Exons/Introns affected	Amino acid change	Reference
Lan-	*ABCB6*01N.20*	c.IVS3−2a>g	Intron 3	Splicing defect	[4]
Lan-	*ABCB6*01N.21*	c.1199_1210delTTGGCATCATCT	Exon 6	p.Ile400_Gly401_Ile402_Ile403_Tyr404delinsAsn	[4]
Lan-	*ABCB6*01N.22*	c.2383_2385delCTC	Exon 18	p.Leu795del	[4]
Lan-	*ABCB6*01N.23*	c.20A>G, c.403C>A, c.459delC	Exon 1	p.Tyr7Cys, p.Arg135Ser, p.Leu154SerfsX97	[4]
Lan-	*ABCB6*01N.24*	c.296_301insG, c.459delC	Exon 2	p.Ala101GlyfsX61, p.Leu154SerfsX97	[4]
Lan-	*ABCB6*01N.25*	c.459delC, c.881_884delCTGA	Exon 1 Exon 4	p.Leu154SerfsX97, p.Thr294ArgfsX31	[4]
Lan-	*ABCB6*01N.26*	c.459delC, c.1617delG	Exon 1 Exon 10	p.Leu154SerfsX97, p.Gly539HisfsX15	[4]
Lan-	*ABCB6*01N.27*	c.459delC, IVS16+1g>a	Exon 1 Intron 16	p.Leu154SerfsX97, Splicing defect	[4]
Lan-	*ABCB6*01N.28*	c.1A > C	Exon 1	p.0	[5]
Lan-	*ABCB6*01N.29*	c.827G > A	Exon 3	p.Arg276Glu	[5]
Lan-	*ABCB6*01N.30*	c.971-1g > a	Intron 4	r.spl?	[5]
Lan-	*ABCB6*01N.31*	c.1825G > A	Exon 13	p.Val609Met	[5]
Lan-	*ABCB6*01N.32*	c.1912C > T	Exon 14	p.Arg638Cys	[5]
Lan-	*ABCB6*01N.33*	c.2155C > T	Exon 16	p.Glu719Ter	[5]
Lan-	*ABCB6*01N.35*	c.2351 + 1g > a	Intron 17	r.spl?	[5]

Phenotype	Allele name	Molecular basis	Exons/Introns affected	Amino acid change	Reference
Altered phenotypes					
Lan weak	*ABCB6*01W.01*	c.826C>T	Exon 3	p.Arg276Trp	[2],[3]
Lan weak	*ABCB6*01W.02*	c.1028G>A	Exon 5	p.Arg343Gln	[2]
Lan weak	*ABCB6*01W.03*	c.1762G>A	Exon 12	p.Gly588Ser	[2],[3]
Lan weak	*ABCB6*01W.04*	c.2216G>A	Exon 16	p.Arg739His	[2]
Lan weak	*ABCB6*01W.05*	c.317A > G	Exon 1	p.Tyr106Cys	[6]
Lan weak	*ABCB6*01W.06*	c.2206G > C	Exon 16	p.Ala736Pro	[6]
Lan weak	*ABCB6*01W.07*	c.403C > A	Exon 1	p.Arg135Ser	[6]
Lan phenotype unconfirmed					
Lan ?		c.20A > G	Exon 1	p.Tyr7Cys	[7]
Lan ?		c.55 A>T	Exon 1	p.Met19Leu	[5]
Lan ?		c.869-2A > G	Intron 3	r.spl?	[7]
Lan ?		c.1199_1210del	Exon 6	p.Ile400_Tyr404del	[7]
Lan ?		c.2256 + 1G > A	Intron 16	r.spl?	[7]
Lan ?		c.2383_2385del	Exon18	p.Leu795del	[7]

References

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