

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. ABCG2 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

Reference allele <i>ABCB6*01</i> encodes LAN1 (Lan)				
Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
Lan+	<i>ABCB6*01</i>			
Null phenotypes				
Lan-	<i>ABCB6*01N.01</i>	c.197_198insG	1	p.Gly67Argfs*95 [1]
Lan-	<i>ABCB6*01N.02</i>	c.717G>A	3	p.Trp239Ter [1]
Lan-	<i>ABCB6*01N.03</i>	c.953_956delGTGG	4	p.Gly318Alafs*8 [1]
Lan-	<i>ABCB6*01N.04</i>	c.1533_1543dupCGGCTC CCTGC	9	p.Leu515Profs*43 [1]
Lan-	<i>ABCB6*01N.05</i>	c.1709_1710delAG	11	p.Glu570Glyfs*21 [1]
Lan-	<i>ABCB6*01N.06</i>	c.1690_1691delAT	11	p.Met564Valfs*2 [1]
Lan-	<i>ABCB6*01N.07</i>	c.1867delinsAACAGGTGA	14	p.Gly623Asnfs*3 [1]
Lan-	<i>ABCB6*01N.08</i>	c.1942C>T	14	p.Arg648Ter [1]
Lan-	<i>ABCB6*01N.09</i>	c.1985_1986delTC	15	p.Leu662Profs*15 [1]
Lan-	<i>ABCB6*01N.10</i>	c.2256+2T>G	Intron 16	Alternative splicing [1]
Lan-	<i>ABCB6*01N.11</i>	c.1236G>A	6	p.Trp412Ter [2,4]
Lan-	<i>ABCB6*01N.12</i>	c.1558_1559insT	9	p.Thr521Hisfs*2 [2,4]
Lan-	<i>ABCB6*01N.13</i>	c.574C>T	2	p.Arg192Trp [2,3,4]

Lan- ^	ABCB6*01N.14	c.85_87delTTC	1	p.Phe29del [3]
Lan- ^	ABCB6*01N.15	c.376delIG	1	p.Val126Serfs*124 [2,4]
Lan-	???	c.459delIC	1	p.Leu154Serfs*97
Lan-	???	c.2256+1G>A	Intron 16	Alternative splicing
Lan-	???	c.301dupG	1	p.Ala101Glyfs*61
Lan-	???	c.718C>T	3	p.Arg240Ter
Lan-	???	c.1199_1210delTTGGCAT CATCT	6	p.Ile400_Tyr404delinsA sn
Lan-	???	c.2383_2385delCTC	18	p.Leu795del
Lan-	???	c.20A>G; c.403C>A; c.459delIC	1	p.Tyr7Cys; p.Arg135Ser; p.Leu154Serfs*97
Lan-	???	c.301dupG; c.459delIC	1	(p.Ala101Glyfs*61; p.Leu154Serfs*97) ???
Lan-	<i>Identical to c.459delIC on protein level?</i>	c.459delIC; (c.881_884delCTGA)	1 4	p.Leu154Serfs*97; (p.Thr294Argfs*31)
Lan-	<i>Identical to c.459delIC on protein level?</i>	c.459delIC; (c.1617delIG)	1 10	p.Leu154Serfs*97; (p.Gly539Hisfs*15)
Lan-	<i>Identical to c.459delIC on protein level?</i>	c.459delIC; (c.2256+1G>A)	1 Intron 16	p.Leu154Serfs*97; (alternative splicing)
Altered phenotypes				
Lan+ ^w /-	ABCB6*01W.01	c.826C>T	3	p.Arg276Trp [2,3,4]
Lan+ ^w	ABCB6*01W.02	c.1028G>A	5	p.Arg343Gln [2,4]
Lan+ ^w	ABCB6*01W.03	c.1762G>A	12	p.Gly588Ser [2,3,4]
Lan+ ^w /-	ABCB6*01W.04	c.2216G>A	16	p.Arg739His [2,4]

^ = presumed

References

1. Helias V, Saison C, Ballif BA, Peyrard T, Takahashi J, Takahashi H, Tanaka M, Deybach JC, Puy H, Le Gall M, Sureau C, Pham BN, Le Pennec PY, Tani Y, Cartron JP, Arnaud L. ABCB6 is dispensable for erythropoiesis and specifies the new blood group system Langereis. *Nature Genetics* 2012;44: 170-3.

2. Zelinski T, Hue-Roye K, Huang A, Coghlan G, Lomas-Francis C, Westhoff CM, Reid ME. Molecular characterization of alleles of the Lan blood group system (Abstract). *Transfusion* 2012;52 (Suppl): 42A-3A.
3. Saison C, Helias V, Peyrard T, Merad L, Cartron J-P, Arnaud L The ABCB6 mutation Arg192Trp is a recessive mutation causing the Lan⁻ blood type Vox Sanguinis 2013;104:159-165.
4. Reid ME, Hue-Roye K, Huang A, Velliquette RW, Tani Y, Westhoff CW, Lomas-Francis C, Zelinski T. Alleles of the LAN blood group system: molecular and serological investigations. *Transfusion* 2013, submitted.