Names for ATP11C (ISBT 046) Blood Group Alleles

Intro

General description:	The ATP11C blood group system currently consists of one high-prevalence antigen, LIL. The LIL antigen is carried on the ATP11C protein, a flippase responsible for the active transport of phosphatidylserine (PS) from the outer to the inner leaflet of the membrane. ATP11C is described as the major flippase at the red cell membrane . However, the ATP11C protein is required for B cell maturation and differentiation as well. ATP11C is expressed ubiquitously in humans and mice. This multi-pass protein consists of 1132 amino acids (NM_173694.5 and NP_775965.3) with predicted 10 transmembrane helices and three cytosolic functional domains. The ATP11C protein forms a heterocomplex with its chaperone protein, CDC50A . The protein is encoded by the *ATP11C* gene, consisting in 218,931 pair bases, on chromosome Xq27.1 (chrX:139,726,346-139,945,276) (GRCh38/hg38). LIL– is a null phenotype caused by an *ATP11C* full gene deletion, resulting in the absence of the protein expression. This null phenotype was shown to be associated with mild anemia. An X-linked congenital hemolytic anemia was shown to be caused by a hemizygous missense mutation in the *ATP11C* gene (p.Thr418Asn), in a Japanese patient (Arashiki, N. et al. PMID: 26944472). Recently, a novel missense variant in the *ATP11C* gene (p.Leu789Phe), associated with mild hereditary hemolytic anemia, was described as well (Van Dijk, MJ, et al, PMID 37671681). GenBank accessions in the ATP11C blood group allele table reference the genomic sequence NG_016550.3 (chrX:139,726,346-139,945,276) (GRCh38/hg38).
Gene name:	*ATP11C*
Number of exons:	30
Initiation codon:	Within exon 1
Stop codon:	Within exon 30
Entrez Gene ID:	286410
LRG:	no record
Sequence:	NG_016550.3 NM_173694.5 (transcript) NP_775965.3 (protein)
Reference allele:	*ATP11C*01*
Reference allele *ATP11C*01* encodes:	ATP11C.1 (LIL)
Antithetical antigens:	

Phenotype	Allele name	Nucleotide change [1)]	Exon Intron	Predicted amino acid change [2)]	(Reference No.) PMID	Accession number	rs number
ATP11C:1 or LIL+	***ATP11C*01***					NM_173694.5	
Null Phenotypes							
ATP11C:-1 or LIL-	***ATP11C*01N.01***	Whole *ATP11C* deletion		p.0	Pending		

References

PMID: Pending

Track of changes

1			**v1.0 30-SEP-2024**
2	Author	created by	Thierry Peyrard, September 2024
3	Reviewer	reviewed by	Slim Azouzi, September 2024
4	Allele	Allele added	*ATP11C*01N.01*
5	Intro	Antigens added	LIL
6	**End Version**		**v1.0 30-SEP-2024**