

## Names for GIL (ISBT 029) Blood Group Alleles

Description	The GIL blood group system consists of one antigen carried on a multipass membrane glycoprotein aquaporin 3 (AQP3), an aquaglyceroporin. It consists of 292 amino acids and both amino and carboxyl termini are predicted to be intracellular.
Gene name	<i>GIL (AQP3)</i>
Number of exons	6
initiation codon:	Within exon 1
Stop codon:	Within exon 6
Entrez Gene ID	360
LRG:	LRG_821
LRG sequence	NG_007476.1 (genomic) NM_004925.4 (transcript)
Reference allele	<i>GIL*01</i> (shaded) Acceptable: <i>GIL</i> , if inferred by haemagglutination
Reference allele	GIL1
<i>GIL*01</i> encodes:	
Antithetical antigens:	n.a.

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
GIL:1 or GIL+	<i>GIL*01</i>						
GIL:-1 or GIL-	<i>GIL*01N.01</i>	c.710+1G>A	Intron 5	Aberrant splicing	[1]; PMID: 12239222	AJ493597	rs1347380973

## References

- 1      PMID:      Roudier N, Ripoche P, Gane P et al. AQP3 deficiency in humans and the molecular  
12239222      basis of a novel blood group system, GIL. J Biol Chem 2002; 277(48), 45854-9

<b>Track of changes</b>		<b>from v3.0 160622</b>	<b>to v4.0 25-FEB-2020</b>
	created	Peter Ligthart	Peter Ligthart, December 2019
	checked	n.a.	Christoph Gassner, January 2020
General		n.a.	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
Intro	LRG ID line added:	n.a.	LRG_813
Allele Table	Table column and header additions	n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
Allele table	n.a.	subdivided <i>GE*01*-02</i> into 4 groups	new alleles
	n.a.	subdivided <i>GE*01*-03</i> into 3 groups	new alleles
References		reference [3]; PMID: 18407531	original publication of allele
Allele table & References		re-numberd the references	top to bottom
<b>End of changes</b>		<b>from v3.0 160622</b>	<b>to v4.0 25-FEB-2020</b>