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	*GIL (AQP3)*
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	*GIL*01* (shaded)
	Acceptable: *GIL* , if inferred by haemagglutination
Reference allele *GIL*01* encodes:	GIL1
Antithetical antigens:	n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
GIL:1 or GIL+	*GIL*01*						
GIL:−1 or GIL−	*GIL*01N.01*	c.710+1G>A	i5	Aberrant splicing	PMID: 12239222	AJ493597	rs1347380973
GIL:−1 or GIL−	*GIL*01N.02*	c.277_283dupCTGGCTC	3	p.Arg95ProfsTer4	(1), Abstract		

References

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

Abstract (1)   Jones B, Karamatic Crew V, Lawrence C et al. Anti-GIL in an individual with a novel AQP3 inactivating mutation resulting in the rare GIL− phenotype; a serological and molecular case study with clinical outcome data, ISBT abstract 31 regional ISBT Congress, ISBT in Focus, 2021; https://doi.org/10.1111/vox.13117

Track of changes		from	to
1	**Version**	**v4.0 15th January 2020**	**v4.1 30-NOV-2021**
2	Author created	Peter Ligthart, December 2019	Peter Ligthart, November 2021
3	Reviewer checked	Christoph Gassner, January 2020	Christoph Gassner, November 2021
4	Allele Table added		new allel *GIL*01N.02* , no accession number and rs number known
5	**End Version**	**v4.0 15th January 2020**	**v4.1 30-NOV-2021**

	Track of changes		from	to
1	**Version**		**v3.0 160622**	**v4.0 15th January 2020**
2	Author	created	Peter Ligthart	Peter Ligthart, December 2019
3	Reviewer	checked	n.a.	Christoph Gassner, January 2020
4	General		n.a.	First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	LRG ID line added:	n.a.	LRG_813
6	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.
7	Allele table & References		re-numberd the references	top to bottom
8	**End Version**		**from v3.0 160622**	**to v4.0 15th January 2020**