Names for FY (ISBT 008) Blood Group Alleles

Intro

General description:	The Duffy blood group system consists of five antigens carried on a multipass membrane glycoprotein called the Atypical Chemokine Receptor 1 (ACKR1), CD234 (previously known as Duffy Antigen Receptor for Chemokines: DARC). It consists of 336 (major) and 338 (minor) amino acids. The major transcript is derived from exon 1 and exon 2 of *ACKR1* ; the minor product is a transcript initiated at the beginning of exon 2. The amino terminus is predicted to be extracellular and the carboxyl terminus intracellular.
HUGO Gene name:	*ACKR1*
ISBT Gene Name:	*FY*
Number of exons:	2
Initiation codon:	Beginning of exon 1 (major) and beginning exon 2 (minor)
Stop codon:	End of exon 2
Entrez Gene ID:	2532
LRG:	LRG_801
LRG sequence:	NG_011626.2 (genomic) (NOTE this NG Ref Seq corresponds to a *FY*01* allele) NM_002036.4 (transcript) (NOTE this NM Ref Seq corresponds to a *FY*01* allele)
Reference allele:	*FY*01 (* shaded) Acceptable: *FY*A* or Fy^a if inferred by haemagglutination
Reference allele *FY*01* encodes:	FY1, FY3, FY5, FY6
Antithetical antigens:	[FY1 FY2]

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
FY:1 or Fy(a+)	***FY*01 or FY*A***	c.125G	2		PMID: 8248172 PMID: 7705836 PMID: 7833467	NG_011626.2 NM_002036	
FY:2 or Fy(b+)	***FY*02 or FY*B***	c.125G>A	2	p.Gly42Asp	PMID: 8248172 PMID: 7705836 PMID: 7833467	U01839 X85785 S76830	rs12075
FY:-2,3	***FY*02.01***	c.125G>A c.126T>G	2	p.Gly42Asp p.Gly42Glu	PMID: 30848497	MH733493	rs12075
Weak *FY*01* phenotypes							
Fy(a+^{w})	***FY*01W.01***	c.265C>T	2	p.Arg89Cys	PMID: 26829175	n.a.	rs34599082
Fy(a+^{w})	***FY*01W.02***	c.265C>T c.298G>A	2	p.Arg89Cys p.Ala100Thr	PMID: 25092430	KF784871	rs34599082 rs13962
Fy(a+^{w})	***FY*01W.03***	c.680G>A	2	p.Gly227Glu	(5), Abstract	n.a.	rs758564844
Weak *FY*02* phenotypes							
Fy(b+^{w}), Fy^{x}	***FY*02W.01***	c.125G>A c.265C>T c.298G>A	2	p.Gly42Asp p.Arg89Cys p.Ala100Thr	PMID: 9731074 PMID: 9746760 PMID: 9886340	AF055992	rs12075 rs34599082 rs13962
Fy(b+^{w}), Fy^{x}	***FY*02W.02***	c.125G>A c.145G>T c.265C>T c.298G>A	2	p.Gly42Asp p.Ala49Ser p.Arg89Cys p.Ala100Thr	PMID: 15569072	n.a.	rs12075 rs1307925062 rs34599082 rs13962
Fy(b+^{w})	***FY*02W.03***	c.125G>A c.266G>A	2	p.Gly42Asp p.Arg89His	(2), Abstract	KY354073	rs12075 rs371909350
Fy(b+^{w})	***FY*02W.04***	c.125G>A c.901C>T	2	p.Gly42Asp p.Pro301Ser	(2), Abstract	KY354074	rs12075 rs753831902
Fy(b+^{w})	***FY*02W.05***	c.125G>A c.976C>T	2	p.Gly42Asp p.Ser326Phe	GenBank Accession number only	HE572751	rs12075 n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Null phenotypes, *FY*01* alleles							
Fy(a−b−) erythroid cells only	***FY*01N.01***	c.-67T>C	Promoter	p.0	PMID: 10570183	AF100634	rs2814778
Fy(a−b−)	***FY*01N.02***	c.286_299del	2	p.Trp96_Thrfs*22	PMID: 7669660	KC924824	rs587776507
Fy(a−b−)	***FY*01N.03***	c.408G>A	2	p.Trp136Ter	PMID: 10691880	n.a.	n.a.
Fy(a−b−)	***FY*01N.04***	c.287G>A	2	p.Trp96Ter	PMID: 10691880 PMID: 25900316	KC924825	rs750052723
Fy(a−b−)	***FY*01N.05***	c.327delC	2	p.Phe109Leufs*12	(1), Abstract	n.a.	n.a.
Fy(a−b−)	***FY*01N.06***	c.395G>A	2	p.Gly132Asp	PMID: 34570912	MZ448627	rs530992295
Fy(a−b−)	***FY*01N.07***	c.719delG	2	p.Gly240Alafs*4	PMID: 25900316	KC924823	rs769160977
Fy(a−b−) erythroid cells only	***FY*01N.08***	c.-69T>C	Promoter	p.0	PMID: 26173389	KP967558 LN715170	n.a.
Fy(a−b−)	***FY*01N.09***	c.296_496delinsAGGCCACTG	2	p.Leu99_Leu165delinsGlnAlaThrAla	(4), Abstract	n.a.	n.a.
Fy(a−b−)	***FY*01N.10***	c.762G>A	2	p.Trp254Ter	(6), Abstract	n.a.	rs766558424
Fy(a−b−)	***FY*01N.11***	c.854delT	2	p.Leu285Argfs*2	PMID: 34570912	MZ448628	rs763701958
Null phenotypes, *FY*02* alleles							
Fy(a−b−) erythroid cells only	***FY*02N.01***	c.-67T>C c.125G>A	Promoter 2	p.0 p.Gly42Asp	PMID: 7663520	X85785 MK813902	rs2814778 rs12075
Fy(a−b−)	***FY*02N.02***	c.125G>A c.407G>A	2	p.Gly42Asp p.Trp136Ter	PMID: 10691880	n.a.	rs12075 rs76819093
Fy(a−b−)	***FY*02N.03***	c.125G>A c.781G>A	2	p.Gly42Asp p.Gly261Arg	PMID: 24845979	HG512885	rs12075 n.a.
Fy(a−b−)	***FY*02N.04***	c.125G>A c.179_180delCT	2	p.Gly42Asp p.Ser60Cysfs*16	PMID: 34570912	MZ448629	rs12075 n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Fy(a−b−)	***FY*02N.05***	c.125G>A c.895G>A	2	p.Gly42Asp p.Ala299Thr	PMID: 34570912	MZ448631	rs12075 rs752428245
Fy(a−b−)	***FY*02N.06***	c.125G>A c.151delT	2	p.Gly42Asp p.Cys51Alafs*24	(3), Abstract	LN875782	rs12075 n.a.
Fy(a−b−)	***FY*02N.07***	c.124delG c.125G>A	2	p.Asp42Metfs*33 p.fs	GenBank Accession number only	KX018789	n.a. rs12075
Fy(a−b−)	***FY*02N.08***	c.125G>A c.400delT	2	p.Gly42Asp p.Cys134Valfs*14	(7), Abstract (8), Abstract	MH211118	rs12075 n.a.
Fy(a−b−)	***FY*02N.09***	c.125G>A c.214G>C	2	p.Gly42Asp p.Gly72Arg	PMID: 34570912	MZ448630	rs12075 rs1054826033

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Abstract (7) Nogués N, González C. Boto N et al. Identification of a new FY*02 null allele in a Caucasian blood donor with a Fy(a-b-) phenotype. *Vox Sang* 2017; 112, Suppl. 1:230, P530

Abstract (8) Babinet J, Ramelet S, Laiguillon G et al. A novel FY*02 silent allele caused by a nucleotide deletion mechanism and responsible of a FY null phenotype in an Algerian patient with strong anti-Fy3 immunization. *Vox Sang* 2018; 113, Suppl. 1:248, P541

PMID 30848497 Weinstock C, Mytilineos J, Bugert P et al. A novel allele of the atypical chemokine receptor 1 (ACKR1) gene containing the nucleotide change c.126 T>G (p.42Glu) encodes a third Duffy blood group protein sequence antithetical to that encoding Fya and Fyb antigens. *Transfusion* 2019; 59: 2158-2159. doi:10.1111/trf.15232

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Track of changes			from version	to version
1	**Version**		**v6.1 30-NOV-2021**	**v6.2 30-SEP-2024**
2	Author	created:	Núria Nogués, November 2021	Núria Nogués, September 2024
3	Review	reviewed:	Christoph Gassner, November 2021	Christoph Gassner, September 2024
4	Allele Table	added mutation		added c.125G>A mutation, amino-acid-exchange p.Gly42Asp and rs12075 for *FY*02.01* allele where it was wrongly absent.
5	**End Version**		**v6.1 30-NOV-2021**	**v6.2 30-SEP-2024**

	Track of changes		from version	to version
1	**Version**		**v6.0 30-JUN-2021**	**v6.1 30-NOV-2021**
2	Author	created:	Núria Nogués, June 2021	Núria Nogués, November 2021
3	Review	reviewed:	n.a.	Christoph Gassner, November 2021
4	Allele Table	added mutation	n.a.	added c.125G>A mutation, amino-acid-exchange p.Gly42Asp and rs12075 for all FY*02 alleles where wrongly absent.
5	Allele Table	updated reference	n.a.	added PMID 34570912 as reference for alleles *FY*01N.06, FY*01N.11, FY*02N.04, FY*02N.05, FY*02N.09*
6	Allele Table	changed GenBank Accession Numbers for alleles *FY*01N.06, FY*01N.11, FY*02N.04, FY*02N.05, FY*02N.09*	n.a.	for *FY*01N.06* KY799578 changed to MZ448627 for *FY*01N.11* KY799577 changed to MZ448628 for *FY*02N.04* KY799579 changed to MZ448629 for *FY*02N.05* KY799581 changed to MZ448631 for *FY*02N.09* KY799580 changed to MZ448630
7	References			changed increasing numbers to PMID only instead
8	References			added PMID 34570912 as reference for alleles *FY*01N.06, FY*01N.11, FY*02N.04, FY*02N.05, FY*02N.09*
9	References			Abstracts (2) and (4) from previous version have been deleted
10	References			renumbered Abstract (11) to Abstract (1)
11	References			renumbered Abstract (14) to Abstract (2)
12	References			renumbered Abstract (21) to Abstract (3)
13	References			renumbered Abstract (22) to Abstract (4)
14	References			renumbered Abstract (23) to Abstract (5)
15	References			renumbered Abstract (24) to Abstract (6)
16	References			renumbered Abstract (25) to Abstract (7)
17	References			renumbered Abstract (26) to Abstract (8)
18	**End Version**		**v6.0 30-JUN-2021**	**v6.1 30-NOV-2021**

	Track of changes		from version	to version
1	**Version**		**v5.0 25-FEB-2020**	**v6.0 30-JUN-2021**
2	Author	created:	Núria Nogués, December 2019	Núria Nogués, June 2021
3	Review	reviewed:	Greg Denomme, January 2020	
4	Allele Table	Format of the "w" used for weak expression on phenotype changed to superscript. The same format change has been applied to Fyx	n.a.	Fy(a+w) changed to Fy(a+^{w}) Fy(b+w) changed to Fy(b+^{w}) Fyx changed to Fy^{x}
5	Allele Table	Allele added:	n.a.	*FY*02N.09* provisional status, GenBank entry by Sunitha Vege in 2017
6	**End Version**		**v5.0 25-FEB-2020**	**v6.0 30-JUN-2021**

	Track of changes		from version	to version
1	**Version**		**v4.1 160816**	**v5.0 25-FEB-2020**
2	Author	created:	Núria Nogués, August 2016	Núria Nogués, December 2019
3	Review	reviewed:	n.a.	Greg Denomme, January 2020
4	General	LRG ID line added		First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	Intro	Note added to LRG sequence line	n.a.	LRG_801
6		Reference allele changed to *FY*01* in agreement with Ref Seqs	NG_011626.2 (genomic) NM_002036.3 (transcript)	NG_011626.2 (genomic) (NOTE this NG Ref Seq corresponds to a *FY*01* allele) NM_002036.4 (transcript) (NOTE this NM Ref Seq corresponds to a *FY*01* allele)
7	Intro	Reference allele line for encoded antigens moved from Allele Table to Intro and updated to *FY*01*	***FY*02*** (shaded) Acceptable: *FY*B* or Fy^b if inferred by haemagglutination	***FY*01*** (shaded) Acceptable: *FY*A* or Fy^a if inferred by haemagglutination
8	Intro	Antithetical Antigens line created in Intro	n.a.	Reference allele *FY*01* encodes FY1, FY3, FY5, FY6

Track of changes			from version	to version
1	**Version**		**v4.1 160816**	**v5.0 25-FEB-2020**
9	Intro	Table column and header additions	n.a.	Antithetical antigens: [FY1 FY2]
10	Allele Table	Text changed and Line moved to Intro	n.a.	Table columns "(Reference No.) PMID", "Accession number" and "rs-number" created and content to table columns added.
11	Allele Table	Note on nucleotide numbering within the transcript moved to Versioning	Reference allele *FY*02* encodes FY3, FY5, FY6	see above
12	Allele Table	Format of the "w" used for weak expression on phenotype changed to superscript. The same format change has been applied to Fyx	Nucleotide positions within the transcript are numbered according to the major transcript. The GATA-1 mutation listed here as c.-67T>C has been reported previously as -33 and -46.	Nucleotide positions within the transcript are numbered according to the major transcript. The GATA-1 mutation listed in the Allele Table as c.-67T>C has been reported previously as -33 and -46.
13	Allele Table	Allele added:	n.a.	*FY*02.01*

Track of changes			from version	to version
1	**Version**		**v4.1 160816**	**v5.0 25-FEB-2020**
			n.a.	**PMID: 30848497.** Weinstock C, Mytilineos J, Bugert P et al. A novel allele of the atypical chemokine receptor 1 (ACKR1) gene containing the nucleotide change c.126 T>G (p.42Glu) encodes a third Duffy blood group protein sequence antithetical to that encoding Fya and Fyb antigens. Transfusion 2019; 59: 2158-2159.
14	Allele Table	Allele added:	n.a.	*FY*01N.10*
15			n.a.	**Abstract.** Guglieri L, Lorenzi M. Salafia M. et al. A novel mutation in an apparent Fy(a-b-) phenotype.Vox Sang 2017; 112, Suppl. 1:230, P531
16	Allele Table	Allele added:	n.a.	*FY*02N.07*
	Allele Table		n.a.	*FY*B* silencing by variant *FY*B(124delG)* in an Arab family. Communication to the RCIBGT Working Party. Unpublished.
17	Allele Table	Allele added:	n.a.	*FY*02N.08*
18	Allele Table		n.a.	**Abstract.** Nogués N, González C. Boto N et al. Identification of a new *FY*02* null allele in a Caucasian blood donor with a Fy(a-b-) phenotype. Vox Sang 2017; 112, Suppl. 1:230, P530. **Abstract.** Babinet J, Ramelet S, Laiguillon G et al. A novel *FY*02* silent allele caused by a nucleotide deletion mechanism and responsible of a FY null phenotype in an Algerian patient with strong anti-Fy3 immunization. Vox Sang 2018; 113, Suppl. 1:248, P541

Track of changes			from version	to version
1	**Version**		**v4.1 160816**	**v5.0 25-FEB-2020**
19	Allele Table	Allele added:	n.a.	*FY*01N.11* provisional status, GenBank entry by Sunitha Vege in 2017
20	Allele Table	Allele added:	n.a.	*FY*02W.05* provisional status, GenBank entry by Andrea Doescher in 2011
21	References	References added	n.a.	Complete References provided for all alleles
22			Only References for variation not included in the dbRBC were provided	All references from 1 to 27 added by chronological order of allele description
23	**End Version**		**v4.1 160816**	**v5.0 25-FEB-2020**