

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+)	<i>FY*01</i> or <i>FY*A</i>	c.125G	2		(1), PMID: 8248172 (2), PMID: 7705836 (3), PMID: 7833467	NG_011626.2 NM_002036	
FY:2 or Fy(b+)	<i>FY*02</i> or <i>FY*B</i>	c.125G>A	2	p.Gly42Asp	(1), PMID: 8248172 (2), PMID: 7705836 (3), PMID: 7833467	U01839 X85785 S76830	rs12075
FY:-2,3	<i>FY*02.01</i>	c.126T>G	2	p.Gly42Glu	(27), PMID: 30848497	MH733493	n.a.
Weak <i>FY*01</i> phenotypes							
Fy(a ^w)	<i>FY*01W.01</i>	c.265C>T	2	p.Arg89Cys	(17), PMID: 26829175	n.a.	rs34599082
Fy(a ^w)	<i>FY*01W.02</i>	c.265C>T c.298G>A	2	p.Arg89Cys p.Ala100Thr	(19), PMID: 25092430	KF784871	rs34599082 rs13962
Fy(a ^w)	<i>FY*01W.03</i>	c.680G>A	2	p.Gly227Glu	(23), Abstract	n.a.	rs758564844
Weak <i>FY*02</i> phenotypes							
Fy(b ^w), Fy ^x	<i>FY*02W.01</i>	c.265C>T c.298G>A	2	p.Arg89Cys p.Ala100Thr	(6), PMID: 9731074 (7), PMID: 9746760 (8), PMID: 9886340	AF055992	rs34599082 rs13962
Fy(b ^w), Fy ^x	<i>FY*02W.02</i>	c.145G>T c.265C>T c.298G>A	2	p.Ala49Ser p.Arg89Cys p.Ala100Thr	(12), PMID: 15569072	n.a.	rs1307925062 rs34599082 rs13962
Fy(b ^w)	<i>FY*02W.03</i>	c.266G>A	2	p.Arg89His	(14), Abstract	KY354073	rs371909350
Fy(b ^w)	<i>FY*02W.04</i>	c.901C>T	2	p.Pro301Ser	(14), Abstract	KY354074	rs753831902
Fy(b ^w)	FY*02W.05	c.976C>T	2	p.Ser326Phe	GenBank Accession number only	HE572751	n.a.
Null phenotypes, <i>FY*01</i> alleles							
Fy(a-b-) erythroid cells only	<i>FY*01N.01</i>	c.-67T>C	Promoter	p.0	(9), PMID: 10570183	AF100634	rs2814778

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Fy(a-b-)	<i>FY*01N.02</i>	c.286_299del	2	p.Trp96_Thrfs*22	(5), PMID: 7669660	KC924824	rs587776507
Fy(a-b-)	<i>FY*01N.03</i>	c.408G>A	2	p.Trp136Ter	(10), PMID: 10691880	n.a.	n.a.
Fy(a-b-)	<i>FY*01N.04</i>	c.287G>A	2	p.Trp96Ter	(10), PMID: 10691880 (20), PMID: 25900316	KC924825	rs750052723
Fy(a-b-)	<i>FY*01N.05</i>	c.327delC	2	p.Phe109Leufs*12	(11), Abstract	n.a.	n.a.
Fy(a-b-)	<i>FY*01N.06</i>	c.395G>A	2	p.Gly132Asp	(13), Abstract	KY799577	rs530992295
Fy(a-b-)	<i>FY*01N.07</i>	c.719delG	2	p.Gly240Alafs*4	(20), PMID: 25900316	KC924823	rs769160977
Fy(a-b-) erythroid cells only	<i>FY*01N.08</i>	c.-69T>C	Promoter	p.0	(18), PMID: 26173389	KP967558 LN715170	n.a.
Fy(a-b-)	<i>FY*01N.09</i>	c.296_496delinsAG GCCACTG	2	p.Leu99_Leu165 delinsGlnAlaThrAla	(22), Abstract	n.a.	n.a.
Fy(a-b-)	<i>FY*01N.10</i>	c.762G>A	2	p.Trp254Ter	(24), Abstract	n.a.	rs766558424
Fy(a-b-)	<i>FY*01N.11</i>	c.854delT	2	p.Leu285Argfs*2	GenBank Accession number only	KY799578	rs763701958
Null phenotypes, <i>FY*02</i> alleles							
Fy(a-b-) erythroid cells only	<i>FY*02N.01</i>	c.-67T>C	Promoter	p.0	(4), PMID: 7663520	X85785 MK813902	rs2814778
Fy(a-b-)	<i>FY*02N.02</i>	c.407G>A	2	p.Trp136Ter	(10), PMID: 10691880	n.a.	rs76819093
Fy(a-b-)	<i>FY*02N.03</i>	c.781G>A	2	p.Gly261Arg	(15), PMID: 24845979	HG512885	n.a.
Fy(a-b-)	<i>FY*02N.04</i>	c.179_180delCT	2	p.Ser60Cysfs*16	(16), Abstract	KY799579	n.a.
Fy(a-b-)	<i>FY*02N.05</i>	c.895G>A	2	p.Ala299Thr	(16), Abstract	KY799581	rs752428245
Fy(a-b-)	<i>FY*02N.06</i>	c.151delT	2	p.Cys51Alafs*24	(21), Abstract	LN875782	n.a.
Fy(a-b-)	<i>FY*02N.07</i>	c.124delG	2	p.Asp42Metfs*33	GenBank Accession number only	KX018789	n.a.

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
Fy(a-b-)	<i>FY*02N.08</i>	c.400delT	2	p.Cys134Valfs*14	(25), Abstract (26), Abstract	MH211118	n.a.
Fy(a-b-)	FY*02N.09	c.214G>C	2	p.Gly72Arg	GenBank Accession number only	KY799580	rs1054826033

References

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3. PMID: 7833467 Iwamoto S, Omi T, Kajii E, Ikemoto S. Genomic organization of the glycoprotein D gene: Duffy blood group Fya/Fyb alloantigen system is associated with a polymorphism at the 44-amino acid residue. *Blood* 1995; **85**(3):622-6.
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26. 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
27. 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

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	LRG_801
6		Reference allele changed to <i>FY*01</i> in agreement with Ref Seqs	NG_011626.2 (genomic) (NOTE this NG Ref Seq corresponds to a <i>FY*01</i> allele) NM_002036.4 (transcript) (NOTE this NM Ref Seq corresponds to a <i>FY*01</i> allele)
7	Intro	Reference allele line for encoded antigens moved from Allele Table to Intro and updated to <i>FY*01</i>	<i>FY*01</i> (shaded) Acceptable: <i>FY*A</i> or <i>Fy^a</i> if inferred by haemagglutination
8	Intro	Antithetical Antigens line created in Intro	Reference allele <i>FY*01</i> encodes FY1, FY3, FY5, FY6
9	Intro	Table column and header additions	Antithetical antigens: [FY1 FY2]
10	Allele Table	Text changed and Line moved to Intro	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	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 in the Allele Table as c.-67T>C has been reported previously as -33 and -46.

13 Allele Table	Allele added:	n.a.	<i>FY*02.01</i>
		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.	<i>FY*01N.10</i>
		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
15 Allele Table Allele Table	Allele added:	n.a.	<i>FY*02N.07</i>
		n.a.	<i>FY*B</i> silencing by variant <i>FY*B(124delG)</i> in an Arab family. Communication to the RCIBGT Working Party. Unpublished.
16 Allele Table Allele Table	Allele added:	n.a.	<i>FY*02N.08</i>
		n.a.	Abstract. Nogués N, González C. Boto N et al. Identification of a new <i>FY*02</i> 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 <i>FY*02</i> 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
17 Allele Table	Allele added:	n.a.	<i>FY*01N.11</i> provisional status, GenBank entry by Sunitha Vege in 2017
18 Allele Table	Allele added:	n.a.	<i>FY*02W.05</i> provisional status, GenBank entry by Andrea Doescher in 2011
19 References	References added	n.a.	Complete References provided for all alleles
		Only References for variation not included in the dbRBC were provided	All references from 1 to 27 added by chronological order of allele description
20 End Version		v4.1 160816	v5.0 25-FEB-2020

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.	<i>FY*02N.09</i> provisional status, GenBank entry by Sunitha Vege in 2017
6	End Version	v5.0 25-FEB-2020	v6.0 30-JUN-2021