Names for FY (ISBT 008) Blood Group Alleles

General description: The Duffy blood group system consists of five antigens carried on a multipass membrane glycoprotein called the Duffy Antigen Receptor for Chemokines (aka DARC, CD234). It consists of 336 (major) and 338 (minor) amino acids. The major transcript is derived from exon 1 and exon 2 of *FY*; 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 sequence: NG_011626.2 (genomic)
NM_002036.3 (transcript)
NP_002027.2 (protein-isoform b; 336 aa)

Reference allele: *FY*02* (shaded)
Acceptable: *FY*B*

Reference allele *FY*02* encodes FY3, FY5, FY6				
Phenotype	**Allele name**	**Nucleotide change**†	**Exon**	**Predicted amino acid change**
FY:1 or Fy(a+)	*FY*01* or *FY*A*	c.125A>G	2	p.Asp42Gly
FY:2 or Fy(b+)	*FY*02* or *FY*B*			
Null phenotypes				
Fy(a−b−) erythroid cells only‡	*FY*01N.01*	c.-67T>C	Promoter	p.0
Fy(a−b−)	*FY*01N.02*	c.281_295del	2	p.Pro94_Val98del
Fy(a−b−)	*FY*01N.03*	c.408G>A	2	p.Trp136Ter
Fy(a−b−)	*FY*01N.04*	c.287G>A	2	p.Trp96Ter
Fy(a−b−)	*FY*01N.05*	c.327delC	2	p.Phe109Leufs*12 [1]
Fy(a−b−)	*FY*01N.06*	c.395G>A	2	p.Gly132Asp [2]
Fy(a−b−)	*FY*01N.07*	c.719delG	2	p.Gly240Alafs*4

Fy(a−b−) erythroid cells only	*FY*01N.08*	c.-69T>C	Promoter	p.0 [6]
Fy(a−b−)	*FY*01N.09*	c.296_496delinsAGGCCACTG	2	p.Leu99_Leu165delins GlnAlaThrAla ? [8]
Fy(a−b−) erythroid cells only	*FY*02N.01*	c.-67T>C	Promoter	p.0
Fy(a−b−)	*FY*02N.02*	c.407G>A	2	p.Trp136Ter
Fy(a−b−)	*FY*02N.03*	c.781G>A	2	p.Gly261Arg
Fy(a−b−)	*FY*02N.04*	c.179_180delCT	2	p.Ser60Cysfs*16 [5]
Fy(a−b−)	*FY*02N.05*	c.895G>A	2	p.Ala299Thr [5]
Fy(a−b−)	*FY*02N.06*	c.151delT	2	p.Cys51Alafs*24 [7]
Weak phenotypes				
Fy(a+^{w})	*FY*01W.01*	c.265C>T	2	p.Arg89Cys [3]
Fy(a+^{w})	*FY*01W.02*	c.265C>T; c.298G>A	2	p.Arg89Cys; p.Ala100Thr
Fy(a+^{w})	*FY*01W.03*	c.680G>A	2	p.Gly227Glu [9]
Fy(b+^{w}), Fy^{x}	*FY*02W.01*	c.265C>T; c.298G>A	2	p.Arg89Cys; p.Ala100Thr
Fy(b+^{w}), Fy^{x}	*FY*02W.02*	c.145G>T; c.265C>T; c.298G>A	2	p.Ala49Ser; p.Arg89Cys; p.Ala100Thr
Fy(b+^{w})	*FY*02W.03*	c.266G>A	2	p.Arg89His [4]
Fy(b+^{w})	*FY*02W.04*	c.901C>T	2	p.Pro301Ser [4]

† Nucleotide numbering within the transcript is numbered according to the major transcript. The GATA-1 mutation listed here as c.-67T>C has been reported previously as -33 and -46.

‡ This allele is rarely found. The common allele in Africans with the same promoter mutation is *FY*02N.01*.

References for variation not in the dbRBC:

1. Tsuneyama H, et al. Transfusion 2000;40(Suppl.):116S
2. Vege S , et al. Transfusion 2013;53(Suppl):164A
3. Ardnt P, et al. Immunohematology 2015;31(3):103-7.

4. Gauthier E , et al. Transfusion 2013;53(Suppl):165A
5. Westhoff CM, et al. Vox Sanguinis 2014; 107, Suppl. 1: 195, P433.
6. Pisacka M, et al. Transfusion 2015; 55:2616-2619.
7. Henny C, et al. Vox Sanguinis 2015;109, Suppl. 1:284, P-578
8. Kupatawintu P, et al. Vox Sanguinis 2015; 109, Suppl. 1:287, P589
9. Tilley LA, et al. Vox Sanguinis 2015; 109, Suppl. 1:297, P620