

## 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.

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  
 GenBank #: NM\_002036  
 Entrez Gene ID: 2532  
 Reference allele:  
     Accession number: U01839  
     Preferred: *FY\*02*  
     Acceptable: *FY\*B* or *Fy<sup>b</sup>* if inferred by haemagglutination

Reference allele <i>FY*02</i> encodes FY3, FY5, FY6				
Phenotype	Allele name	Nucleotide	Exon	Amino acid
FY:1 or Fy(a+)	<i>FY*01</i> or <i>FY*A</i>	c.125A>G	2	p.Asp42Gly
FY:2 or Fy(b+)	<i>FY*02</i> or <i>FY*B</i>	c.125A	2	p.Asp42
Null phenotypes				
Fy(a-b-) erythroid cells only#	<i>FY*01N.01</i>	c.1-67T>C <sup>†</sup>	Promoter	p.0
Fy(a-b-)	<i>FY*01N.02</i>	c281_295del	2	p.Pro94fs
Fy(a-b-)	<i>FY*01N.03</i>	c.408G>A	2	p.Trp136Ter
Fy(a-b-)	<i>FY*01N.04</i>	c.287G>A	2	p.Trp96Ter
Fy(a-b-)	<i>FY*01N.05</i>	c.327delC	2	p.Phe109fs [1]
Fy(a-b-)	<i>FY*01N.06</i>	c.395G>A	2	p.Gly132Asp [2]
<i>Fy(a-b-)</i>	<i>FY*01N.07</i>	<i>c.719delG</i>	<i>2</i>	<i>p.Gly240Ala</i>
Fy(a-b-) erythroid cells only	<i>FY*02N.01</i>	c.1-67T>C	Promoter	p.0
Fy(a-b-)	<i>FY*02N.02</i>	c.407G>A	2	p.Trp136Ter
<i>Fy(a-b-)</i>	<i>FY*02N.03</i>	<i>c.781G&gt;A</i>	<i>2</i>	<i>p.Gly261Arg</i>

Weak phenotypes				
Fy(a <sup>w</sup> )	<i>FY*01W.01</i>	c.265C>T	2	p.Arg89Cys [3]
Fy(a <sup>w</sup> )	<i>FY*01W.02</i>	c.265C>T, 298G>A	2	p.Arg89Cys, Ala100Thr
Fy(b <sup>w</sup> ), Fy <sup>x</sup>	<i>FY*02W.01</i>	c.265C>T, 298G>A	2	p.Arg89Cys, Ala100Thr
Fy(b <sup>w</sup> ), Fy <sup>x</sup>	<i>FY*02W.02</i>	c.145G>T, 265C>T, 298G>A	2	p.Ala49Ser, Arg89Cys, Ala100Thr
Fy(b <sup>w</sup> )	<i>FY*02W.03</i>	c.266G>A	2	p.Arg89His [4]
Fy(b <sup>w</sup> )	<i>FY*02W.04</i>	c.901C>T	2	p.Pro303Ala [4]

†Nucleotide numbering within the transcript is numbered according to the major transcript. The GATA-1 mutation listed here as –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. Transfusion 2013;53(Suppl):39A
- [4] Gauthier E, et al. Transfusion 2013;53(Suppl):165A