

## Names for GLOB (ISBT 028) Blood Group Alleles

General description: The GLOB system was acknowledged in 2002 when the P or globoside antigen was moved from the 209 collection. The P antigen is the most common neutral glycosphingolipid in the red cell membrane, belongs to the globoseries and has the following structure: GalNAc $\beta$ 3Gal $\alpha$ 4Gal $\beta$ 4Glc $\beta$ 1 ceramide, also known as globoside (Gb4Cer). The *B3GALT3* gene was first reported in 1998 by Amado *et al.* to be a member of the  $\beta$ 1,3-galactosyltransferase gene family and its product given the name  $\beta$ 3Gal-T3. It was later shown by Okajima *et al.* to possess UDP-*N*-acetyl galactosamine:globotriaosylceramide 3- $\beta$ -*N*-acetylgalactosaminyl-transferase or globoside synthase activity and the gene name changed to *B3GALNT1* and its product renamed  $\beta$ 3GalNAc-T1. This enzyme is responsible for the final step in the synthesis of the P antigen, the transfer of GalNAc to the terminal Gal of the P<sup>k</sup> antigen. The final proof of this was the identification by Hellberg *et al.* of critical mutations in the *B3GALNT1* gene as the genetic basis of P<sub>1</sub><sup>k</sup> and P<sub>2</sub><sup>k</sup>, the rare globoside-deficient null phenotypes of the GLOB system.

Gene name: *GLOB (B3GALNT1)*  
 Number of exons: 5  
 Initiation codon: Exon 5  
 Stop codon: Exon 5  
 Entrez Gene ID: 26879  
 LRG sequence: NG\_007854.1 (genomic)  
 NM\_033169.2 (transcript)  
 Reference allele: *GLOB\*01 (B3GALNT1\*01)*  
 Acceptable: P if inferred by haemagglutination

Reference allele <i>GLOB*01</i> encodes P				
Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
GLOB:1 (P+)	<i>GLOB*01</i>			
GLOB:1 (P+)	<i>GLOB*02</i>	c.376G>A	5	p.Asp126Asn
Null phenotypes <sup>†</sup>				
GLOB:-1 (P-)	<i>GLOB*01N.01</i>	c.202C>T	5	p.Arg68Ter
GLOB:-1 (P-)	<i>GLOB*01N.02</i>	c.292_293insA	5	p.Arg98Lysfs*6
GLOB:-1 (P-)	<i>GLOB*01N.03</i>	c.433C>T	5	p.Arg145Ter
GLOB:-1 (P-)	<i>GLOB*01N.04</i>	c.537_538insA	5	p.Asp180Argfs*3
GLOB:-1 (P-)	<i>GLOB*01N.05</i>	c.648A>C	5	p.Arg216Ser

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GLOB:-1 (P-)	<i>GLOB*01N.06</i>	c.797A>C	5	p.Glu266Ala
GLOB:-1 (P-)	<i>GLOB*01N.07</i>	c.811G>A	5	p.Gly271Arg
GLOB:-1 (P-)	<i>GLOB*01N.08</i>	c.959G>A	5	p.Trp320Ter
GLOB:-1 (P-)	<i>GLOB*01N.09</i>	c.203delG	5	p.Arg68Glnfs*17
GLOB:-1 (P-)	<i>GLOB*01N.11</i>	c.456T>G	5	p.Tyr152Ter
GLOB:-1 (P-)	<i>GLOB*01N.12</i>	c.449A>G	5	p.Asp150Gly
GLOB:-1 (P-)	<i>GLOB*02N.01</i>	c.376G>A; c.598delT	5	p.Asp126Asn; p.Ser200Glnfs*10

†The null phenotype caused by these alleles can either be P1+ or P1-, i.e. P<sub>1</sub><sup>k</sup> or P<sub>2</sub><sup>k</sup>