Names for P1PK (ISBT 003) Blood Group Alleles

General description: The gene *A4GALT* encodes 4-α-galactosyltransferase, the enzyme that synthesizes P1 and P^k antigens. P1 antigen is synthesized on paragloboside and P^k antigen on lactosylceramide. Changes in exon 2a distinguish P_1 and P_2 phenotypes. Differences from reference allele *A4GALT*P1.01* (accession number GU902278) are given below. A nucleotide change of C>T (ACG>ATG) in exon 2a of *A4GALT* introduces an open reading frame in transcripts that include exons 1 and 2a. This is associated with fewer enzyme-encoding transcripts (comprising exon 3) in the presence of the P^2 allele but it is unknown how this transcriptional regulation occurs.

Gene name: *A4GALT*
Number of exons: 4
Initiation codon: Beginning of exon 4
Stop codon: Within exon 4
Entrez Gene ID: 53947
LRG sequence: NG_007495.1 (genomic)
NM_017436.4 (transcript)
Reference allele: *A4GALT*P1.01* (shaded)

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
P1/P2 determining transcript (GU902278)				
P1+ P^k+ (P_1)	*A4GALT*P1.01*			
P1– P^k+ (P_2)	*A4GALT*P2.01*	c.42C>T	2a	Potential start codon introduced
P1– P^k+ (P_2)	*A4GALT*P2.02*	c.42C>T; c.122T>G	2a	p.Gly28Trp Potential start codon introduced
4-α-galactosyltransferaseencoding transcript (NM_017436.4)				
P1+/–, P^k+	*A4GALT*01*†			
P1+/–, P^k+	*A4GALT*02*†	c.109A>G	3	p.Met37Val
NOR+, P1+, P^k+	*A4GALT*04*‡	c.631C>G	3	p.Gln211Glu
Null phenotypes				
p	*A4GALT*01N.01.01*	c.241_243delTTC; c.903C>G	3	p.Phe81del
p	*A4GALT*01N.01.02*	c.241_243delTTC	3	p.Phe81del

p	*A4GALT*01N.02*	c.287G>A	3	p.Cys96Tyr
p	*A4GALT*01N.03.01*	c.299C>T; c.903C>G	3	p.Ser100Leu
p	*A4GALT*01N.03.02*	c.299C>T	3	p.Ser100Leu
p	*A4GALT*01N.04*	c.301delG	3	p.Ala101Profs*13
p	*A4GALT*01N.05*	c.418_428delinsTGGACCTGCTGGACCTGCTGGACCTGCTGGAACA	3	p.Gln140Trpfs*73
p	*A4GALT*01N.06*	c.470_496delinsCGTACCCGAC	3	p.Asp157Alafs*120
p	*A4GALT*01N.07*	c.473G>A	3	p.Trp158Ter
p	*A4GALT*01N.08*	c.504dupC; c.914C>T	3	p.Tyr169Leufs*114
p	*A4GALT*01N.09.01*	c.548T>A	3	p.Met183Lys
p	*A4GALT*01N.09.02*	c.548T>A; c.987G>A	3	p.Met183Lys
p	*A4GALT*01N.10*	c.559G>C	3	p.Gly187Arg
p	*A4GALT*01N.11*	c.560G>A	3	p.Gly187Asp
p	*A4GALT*01N.12*	c.656C>T	3	p.Ala219Val
p	*A4GALT*01N.13*	c.657delG	3	p.Phe220Serfs*130
p	*A4GALT*01N.14*	c.732dupG	3	p.Ile245Aspfs*38
p	*A4GALT*01N.15*	c.751C>T	3	p.Pro251Ser
p	*A4GALT*01N.16*	c.752C>T	3	p.Pro251Leu
p	*A4GALT*01N.17*	c.769delG	3	p.Val257Serfs*93
p	*A4GALT*01N.18*	c.783G>A	3	p.Trp261Ter
p	*A4GALT*01N.19*	c.972_997del	3	p.Arg325Alafs*113
p	*A4GALT*01N.20*	c.1029dupC	3	p.Thr344Hisfs*103
p	*A4GALT*01N.21*	c.201dupC	3	p.Thr68Hisfs*215
p	*A4GALT*01N.22*	c.418C>T	3	p.Gln140Ter
p	*A4GALT*01N.23*	c.498G>A	3	p.Trp166Ter
p	*A4GALT*02N.01*	c.68dupT	3	p.Phe24Valfs*31
p	*A4GALT*02N.02*	c.290C>T	3	p.Ser97Leu
p	*A4GALT*02N.03*	c.752C>T	3	p.Pro251Leu

p	*A4GALT*02N.04*	c.902delC	3	p.Glu302Argfs*48
p	*A4GALT*02N.05*	c.972_997del	3	p.Arg325Alafs*113
p	*A4GALT*02N.06*	c.388dupA	3	p.Ile130Asnfs*153
p	*A4GALT*02N.07*	c.367T>C; c.547_548delAT	3	p.Ser123Pro; p.Met183Valfs*99
p	*A4GALT*02N.08*	c.480_495dupGGCCG TGCAGGGGCGC	3	p.Trp166Glyfs*122
p	*A4GALT*N.01*	NC_000022.10:g.4309 7156_43129968del (Exon 1 deleted)		p.0
p	*A4GALT*N.02*	NC_000022.10:g.4310 3896_43124759del (Exon 1 deleted)		p.0
p	*A4GALT*N.03*	NC_000022.10:g.4309 5125_43120758del (Exon 1 deleted)		p.0

† Can be *in cis* to P^1 or P^2 polymorphism in exon 2a, i.e. can travel with either P_1 or P_2 phenotype.

‡ *In cis* to P^1 allele, i.e. travels with the P_1 phenotype.