Names for I (ISBT 027) Blood Group Alleles

General description: The I blood group system consists of one antigen, I, carried on branched carbohydrate chains in the RBC membrane. The glucosaminyl (N-acetyl) transferase 2 that synthesizes I antigen on red cells consists of 402 amino acids and is encoded by the *GCNT2* gene transcript containing exon 1C. The I– phenotype in adults is associated with cataracts.
Gene name: *GCNT2*
Number of exons: 3 (Exon 1 has 3 alternative forms: 1A, 1B, and 1C)
Initiation codon: Within exon 1
Stop codon: Within exon 3
Entrez Gene ID: 2651
LRG sequence 819: NG_007469.3 (genomic)
NM_145655.3 (transcript)
NP_663630.2 (protein)
Entrez Gene ID: 2651
Reference allele: *GCNT2*01* (shaded)
Acceptable: *I,* if inferred by haemagglutination

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change
I	*GCNT2*01*			
I	*GCNT2*02*	c.816G>C	1C	p.Glu272Asp
I+^{W}	*GCNT2*01W.01*	c.243T>A	1C	p.Asn81Lys
I+^{W}	*GCNT2*01W.02*	c.505G>A	1C	p.Ala169Thr
I+^{W}	*GCNT2*01W.03*	c.683G>A	1C	p.Arg228Gln
Null phenotypes				
I– (i adult)	*GCNT2*01N.01*	c.1049G>A	3	p.Gly350Glu
I– (i adult)	*GCNT2*01N.02*	c.1154G>A	3	p.Arg385His
I– (i adult)	*GCNT2*01N.05*	c.984G>A	2	p.Trp328Ter
I– (i adult)	*GCNT2*01N.06*	del exon 1B, 1C, 2, 3	1B, 1C, 2, 3	p.0
I– (i adult)	*GCNT2*01N.07*	c.651delA	1C	p.Val219Cysfs*26 [1]
I– (i adult)	*GCNT2*01N.08*	c.935G>A	2	p.Gly312Asp
I– (i adult)	*GCNT2*01N.09*	c.1163_1166delATCA	3	p.Asn390Argfs*20 [2]
I– (i adult)	*GCNT2*02N.01*	c.816G>C; c.1006G>A	1C 2	p.Glu272Asp; p.Gly336Arg

1. Onodera T. A new IGNT allele found in the adult i-negative in Japanese without congenital cataracts. Vox Sang. 2011;101 (Suppl.1):262.
2. Cheong S-S, Hull S, Jones B, Chana R, Thornton N, Plagnol V, Moore AT, Hardcastle AJ. Pleiotropic effect of a novel mutation in GCNT2 causing congenital cataract and a rare adult i blood group phenotype. Human Genome Variation (2017) 4, 17004; doi:10.1038/hgv.2017.4