

Names for I (ISBT 027) Blood Group Alleles

General description: The I blood group system consists of one antigen, carried branched carbohydrate chains in the RBC membrane. The glycosyl-transferase that synthesizes I antigen on red cells consists of 402 amino acids and is encoded by the transcript containing exon 1C. The I⁻ phenotype in adults is associated with cataracts.

Gene name: *I*
 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: NG_007469.2 (genomic)
 NM_145655.3 (transcript)
 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 | <i>GCNT2*01</i> | | | |
| I | <i>GCNT2*02</i> | c.816G>C | 1C | p.Glu272Asp |
| I ^{+W} | <i>GCNT2*01W.01</i> | c.243T>A | 1C | p.Asn81Lys |
| I ^{+W} | <i>GCNT2*01W.02</i> | c.505G>A | 1C | p.Ala169Thr |
| I ^{+W} | <i>GCNT2*01W.03</i> | c.683G>A | 1C | p.Arg228Gln |
| Null phenotypes | | | | |
| I ⁻ (i adult) | <i>GCNT2*01N.01</i> | c.1049G>A | 3 | p.Gly350Glu |
| I ⁻ (i adult) | <i>GCNT2*01N.02</i> | c.1154G>A | 3 | p.Arg385His |
| I ⁻ (i adult) | <i>GCNT2*01N.05</i> | c.984G>A | 2 | p.Trp328Ter |
| I ⁻ (i adult) | <i>GCNT2*01N.06</i> | del exon 1B, 1C, 2, 3 | 1B, 1C, 2, 3 | p.0 |
| I ⁻ (i adult) | <i>GCNT2*01N.07</i> | c.651delA | 1C | p.Val219Cysfs*26 [1] |
| I ⁻ (i adult) | <i>GCNT2*01N.08</i> | c.935G>A | 2 | p.Gly312Asp |
| I ⁻ (i adult) | <i>GCNT2*02N.01</i> | 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.