Names for I (ISBT 027) Blood Group Alleles

Intro

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:	LRG_819
LRG sequence:	NG_007469.3 (genomic) NM_145655.4 (transcript) NP_663630.2 (protein)
Reference allele:	*GCNT2*01* (shaded) Acceptable: *I*, if inferred by haemagglutination
Reference allele *GCNT2*01* encodes:	Glucosaminyl (N-acetyl) transferase 2 that synthesizes I antigen
Antithetical antigens:	i [see Ii collection (207)]

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number
I:1 or I+	*GCNT2*01*				PMID: 8449405	NG_007469.3	not applicable
I:1 or I+	*GCNT2*01.02 (old: GCNT2*02)*	c.816G>C	1C	p.Glu272Asp	PMID: 12468428		rs539351
Weak phenotypes							
I:1weak or I+^{W}	*GCNT2*01W.01*	c.243T>A	1C	p.Asn81Lys	PMID: 17076854		rs201291494
I:1weak or I+^{W}	*GCNT2*01W.02*	c.505G>A	1C	p.Ala169Thr	PMID: 12424189		rs56106312
I:1weak or I+^{W}	*GCNT2*01W.03*	c.683G>A	1C	p.Arg228Gln	PMID: 12424189		rs55795227
I:1weak or I+^{W}	*GCNT2*01W.04*	c.1054G>A c.1184C>T	3	p.Gly352Arg p.Ala395Val	(2), Abstract		rs369770528 rs371217806
Null phenotypes							
I:-1 or I– (i adult)	*GCNT2*01N.01*	c.1049G>A	3	p.Gly350Glu	PMID: 12468428 PMID: 12424189 PMID: 11739194	AF401652	rs56141211
I:-1 or I– (i adult)	*GCNT2*01N.02*	c.1154G>A	3	p.Arg385His	PMID: 12468428 PMID: 12424189 PMID: 11739194	AF401653	rs55940927
I:-1 or I– (i adult)	*GCNT2*01N.05*	c.983G>A	2	p.Trp328Ter	PMID: 15161861		n.a.
I:-1 or I– (i adult)	*GCNT2*01N.06*	del exon 1B, 1C, 2, 3	1B, 1C, 2, 3	p.0	PMID: 12424189 PMID: 11739194 PMID: 21761136		not available
I:-1 or I– (i adult)	*GCNT2*01N.07*	c.651delA	1C	p.Val219Cysfs*26	(1), Abstract		rs755005507
I:-1 or I– (i adult)	*GCNT2*01N.08*	c.935G>A	2	p.Gly312Asp	PMID: 21541272		rs777441702
I:-1 or I– (i adult)	*GCNT2*01N.09*	c.1169_1172delATCA	3	p.Asn390Argfs*20	PMID: 28224043		rs1114167314
I:-1 or I– (i adult)	*GCNT2*01N.10 (old GCNT2*02N.01)*	c.816G>C c.1006G>A	1C 2	p.Glu272Asp p.Gly336Arg	PMID: 12468428		rs539351 rs774740944

References

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Pras E, Raz J, Yahalom V, Frydman M, Garzozi HJ, Pras E, Fielding Hejtmancik J. A nonsense mutation in the glucosaminyl (N-acetyl) transferase 2 gene (*GCNT2*): association with autosomal recessive congenital cataracts. *Invest Ophthalmol Vis Sci* 2004; 45 (6): 1940-1945. DOI: 10.1167/iovs.03-1117

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PMID 21541272 Wussuki-Lior O, Abu-Horowitz A, Netzer I, Almer Z, Morad Y, Goldich Y, Yahalom V, Pras El, Pras Er. Hematologic biomarkers in childhood cataracts. *Mol Vis* 2011; 17: 1011-1015 .

PMID 28224043 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

References

Abstract (1) Onodera T. A new IGNT allele found in the adult i-negative in Japanese without congenital cataracts. *Vox Sang* 2011; 101 (Suppl.1): 262-263. [Abstract].

Abstract (2) Karamatic Crew V, Poole J, Thornton N, Gray A, Needs M, Daniels G. A novel *GCNT2* allele with two unique heterozygous mutations found in a donor with the I weak i negative phenotype. *Transfus Med* 2012; 22 (Suppl. 1): 53. [Abstract].

Track of changes

			from	to
1	**Version**		**v4.0 8th April 2019**	**v5.0 30-JUN-2022**
2	Author	created	Nicole Thornton, 08 April 2019	Nicole Thornton/Vanja Karamatic Crew, 21st April 2022
3	Review	reviewed	Vanja Karamatic Crew	Åsa Hellberg, 22nd June 2022
4	General		Word version	First Excel map version. Spread-sheets 'Intro', 'Allele Table', 'References', and 'Versioning' created.
5	References	moved		Moved reference's texts to tab-sheet 'References'
6	References	changed		changed reference (1) and (2) to Abstracts
7	References	added PMIDs		8449405, 12468428, 17076854, 12424189, 11739194, 15161861, 21761136, 21541272, 28224043
8	Allele	naming	*GCNT2*02*	changed to *GCNT2*01.02* , to follow agreed new allele nomenclature
9	Allele	naming	*GCNT2*02N.01*	changed to *GCNT2*01N.10* , to follow agreed new allele nomenclature
10	Allele	added		Line added for new *GCNT2.01W.04* allele
11	References	added		Reference: Karamatic Crew V, Poole J, Thornton N, Gray A, Needs M, Daniels G. A novel GCNT2 allele with two unique heterozygous mutations found in a donor with the I weak i negative phenotype. Transfus Med 2012; 22 (Suppl. 1): 53. [Abstract].
12	Intro	LRG sequence	NM_145655.3	NM_145655.4
13	Intro	Reference allele encodes' changed to		glucosaminyl (N-acetyl) transferase 2 that synthesizes I antigen
14	Intro	Antithetical antigens		i [see Ii collection (207)]
15	Allele	changed		*GCNT2*01N.05* corrected nucleotide to c.983G>A
16	Allele	added rsnumbers		
17	Allele	changed		*GCNT2*01N.09* changed c.1163_1166delATCA to c.1169_1172delATCA
18	**End Version**		**v4.0 8th April 2019**	**v5.0 30-JUN-2022**

Track of changes

			from	to
1	**Version**		**v3.0 160622**	**v4.0 8th April 2019**
2	Author	created	Nicole Thornton, 22nd of June 2016	Nicole Thornton, 8th of April 2019
3	Review	reviewed	n.a.	n.a.
4	General		Word version	Word version
5	Intro	General description		changed text
6	Intro	Gene name	*I*	changed Gene name from *I* to *GCNT2*
7	Intro	LRG sequence	NG_007469.2	changed to NG_007469.3
8	Allele Table	Allele added		added *GCNT2*01N.09*
9	**End Version**		**v3.0 160622**	**v4.0 8th April 2019**