Names for RHAG (ISBT 030) Blood Group Alleles

General description: The RHAG blood group system consists of three antigens carried on a multipass membrane glycoprotein called RhAG (Rh-associated glycoprotein; aka CD241). It consists of 409 amino acids and both amino and carboxyl termini are predicted to be intracellular.
Gene name: *RHAG*
Number of exons: 10
Initiation codon: Beginning of exon 1
Stop codon: End of exon 10
Entrez Gene ID: 6005
LRG Sequence: NG_011704.1 (genomic)
NM_000324.2 (transcript)
Reference allele: *RHAG*01* (shaded)

Reference allele *RHAG*01* encodes RHAG1, RHAG3				
Phenotype	**Allele name**	**Nucleotide change**	**Exon**	**Predicted amino acid change**
RHAG:1 or Duclos+	*RHAG*01*			
RHAG:−1 or Duclos−	*RHAG*−01*	c.316C>G	2	p.Gln106Glu
RHAG:2 or Ol(a+)	*RHAG*02*	c.680C>T	5	p.Ser227Leu
RHAG:−3 or DSLK−	*RHAG*−03*	c.490A>C	3	p.Lys164Gln
RHAG:4	*RHAG*04*	c.808G>A	6	p.Val270Ile
Null phenotypes				
Rh_{null}	*RHAG*01N.01*	c.154_157delinsGA	2	p.Pro52Aspfs*57
Rh_{null}	*RHAG*01N.02*	c.1086delA	8	p.Ala363Leufs*15
Rh_{null}	*RHAG*01N.03*	c.157+1G>A	Intron 1	Alternative splicing
Rh_{null}	*RHAG*01N.04*	c.945+1G>A	Intron 6	Alternative splicing [1]
Rh_{null}	*RHAG*01N.05*	c.946-1G>A	Intron 6	Alternative splicing
Rh_{null}	*RHAG*01N.06*	c.946-1G>T	Intron 6	Alternative splicing
Rh_{null}	*RHAG*01N.07*	c.1067+1G>A	Intron 7	Alternative splicing
Rh_{null}	*RHAG*01N.08*	c.808G>A; c.838G>A	6	p.Val270Ile; p.Gly280Arg
Rh_{null}	*RHAG*01N.09*	c.836G>A	6	p.Gly279Glu
Rh_{null}	*RHAG*01N.10*	c.1094T>G	8	p.Leu365Arg [1]

Rh_{null}	*RHAG*01N.11*	c.1139G>T	9	p.Gly380Val; Alternative splicing
Rh_{null}	*RHAG*01N.12*	c.353C>T	3	p.Ala118Glu [5]
Rh_{null}	*RHAG*01N.13*	c.1003G>A	7	p.Gly335Ser
Rh_{null}	*RHAG*01N.14*	c.946-2A>G	Intron 6	Alternative splicing [7]
Rh_{null}	*RHAG*01N.15*	c.(?_-62)_(*638_?)del Gene deletion	1-10	p.0 [8]
Rh_{null}	*RHAG*01N.16*	c.310C>T	2	p.Gln104Ter [9]
Mod phenotypes				
Rh_{mod}	*RHAG*01M.01*	c.1183delA	9	p.Asn395Thrfs*68
Rh_{mod}	*RHAG*01M.02*	c.3G>T	1	p.Arg2_Met8del
Rh_{mod}	*RHAG*01M.03*	c.236G>A	2	p.Ser79Asn
Rh_{mod}	*RHAG*01M.04*	c.269G>T	2	p.Gly90Val [2]
Rh_{mod}	*RHAG*01M.05*	c.398T>C	3	p.Leu133Pro [3]
Rh_{mod}	*RHAG*01M.06*	c.560G>A	4	p.Gly187Asp [2]
Rh_{mod}	*RHAG*01M.07*	c.1195G>T	9	p.Asp399Tyr
Rh_{mod}	*RHAG*01M.08*	c.182T>G	2	p.Ile61Arg
Rh_{mod}	*RHAG*01M.09*	c.194T>C	2	p.Phe65Ser
Rh_{mod}	*RHAG*01M.10*	c.572G>A	4	p.Arg191Gln [6]

RHAG3 assigned provisionally.
Assignment of null (*N*) and mod (*M*) alleles has been made according to the phenotypic expression of RhD and RhCE antigens.

1. Tsuneyama H, et al. Transfusion 2005;45(suppl):130A (abstract).
2. Scharberg A, et al. Vox Sang 2006;91(suppl 3):129 (abstract).
3. Tsuneyama H, et al. Transfusion 2008;48(suppl):194A-195A (abstract).
4. Poole J, et al. A novel RHAG blood group antigen associated with severe HDFN. Vox Sang 2011;101 (Suppl. 1):70 (abstract).
5. Grimsley S, Poole J, Thornton N, et al. Novel mutations in RHAG causing two new examples of the regulator type of Rhnull. Transfus Med 2012;22(Suppl.1):21 (abstract).
6. Tsuneyama H, Isa K, Ogasawara K, Yabe R, Uchikawa M, Minami M. Identification of a mutation in the *RHAG* gene of Japanese with weak D phenotype. Transfusion 2014;53 (Suppl.):167A (abstract).
7. Arsenovic MG, *et al*. *Vox Sang* 2014;107(suppl.1):192 (abstract).
8. Gómez-Torreiro, *et al*. *Transfusion* 2015;55:197-198.
9. Arnoni CP, *et al*. Submitted to *Transfusion*.