Names for CH/RG (ISBT 017) Blood Group Alleles

Intro

General description:	The Chido/Rodgers blood group system consists of nine antigens, which located on the C4d region of the fourth component of complement (C4). C4d is a tryptic fragment of C4, which become bound to the red cells from the plasma. The encoding gene C4A and C4B are two highly homologous genes, which are remarkable polymorphic distributed in the populations combined with other factors including variant gene copy number and long form/short form gene contribute to the diverse expression of C4A and C4B proteins.
Gene name:	*CH (C4B)*
Number of exons:	41
Initiation codon:	Within exon 1
Stop codon:	Within exon 41
Entrez Gene ID:	721
LRG:	LRG_138
LRG sequence:	NG_011639.1 (genomic) NM_001002029.4 (Encoding complement C4B preproprotein, 1744 amino acids)
Reference allele:	*C4B*3* (shaded)
Reference allele *C4B*3* encodes:	Ch1 (CH1), CH2, CH3, CH4, CH5, and CH6
Antithetical antigens:	None

Gene name:	*RG (C4A)*
Number of exons:	41
Initiation codon:	Within exon 1
Stop codon:	Within exon 41
Entrez Gene ID:	720
LRG:	LRG_137
LRG sequence:	NG_011638.1 (genomic)
	NM_007293.3 (transcript variant 1, complement C4A isoform 1 preproprotein, 1744 amino acids)
Reference allele:	*C4A*3* (shaded)
Reference allele *C4B*3* encodes:	Reference allele *C4A*3* encodes: Rg1 (RG1) and RG2
Antithetical antigens:	None

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number	comment
Ch+Rg– or CH:1,2,3,4,5,6 RG:–1,–2	*C4B*03*							
Ch+Rg– or CH:1,2,–3,4,5,–6 RG:–1,–2	*C4B*01*	c.3527G>A	28	p.Ser1176Asn	PMID: 2453459	n.a.	rs2746414	
Ch+Rg– or CH:1,–2,3,4,–5,6 RG:–1,–2	*C4B*02*	c.3218G>A	25	p.Gly1073Asp	PMID: 2453459	n.a.	rs2258218	
Ch+Rg+WH+ or CH:–1,–2,–3,4,–5,6 RG:1,–2 WH+	*C4B*05*	c.3218G>A c.3620C>T c.3629G>T c.3630G>C	25 28 28 28	p.Gly1073Asp p.Ala1207Val p.Arg1210Leu	PMID: 2444535	n.a.	rs2258218 rs2229403 rs2229409 rs1138376	
Null phenotypes								
Ch- or CH:–1,–2,–3, –4,–5, –6	*C4B*N.01*	c.1623delC	13	p.Phe541PhefsTer587	PMID: 12133986	n.a.	n.a.	Known as *C4BQ0*
Ch- or CH:–1,–2,–3, –4,–5, –6	*C4B*N.02*	c.3695_3696dupTC	29	p.Ser1232SerfsTer1307	PMID: 10092831	AF092085.1	rs367709216	Known as *C4BQ0*
Ch- or CH:–1,–2,–3, –4,–5, –6	*C4B*N.03*	IVS28+1g>a	i28	Aberrant splicing	PMID: 15294999	n.a.	rs771378213	Known as *C4BQ0*

Phenotype	Allele name	Nucleotide change	Exon Intron	Predicted amino acid change	(Reference No.) PMID	Accession number	rs number	comment
Ch–Rg+ or CH:–1,–2,–3,–4,–5,–6 RG:1,2	*C4A*03*							
Ch+Rg– or CH:1,–2,3,–4,5,6 RG:–1,–2	*C4A*01*	c.3218A>G c.3527A>G c.3620T>C c.3629T>G c.3630C>G	25 28 28 28 28	p.Asp1073Gly p.Asn1176Ser p.Val1207Ala p.Leu1210Arg	PMID: 2453459	n.a.	rs147162052 rs17874654 rs28357075 rs28357076 rs28357077	Hybrid *C4*B-A-B* gene
Ch–Rg+WH+ or CH:–1,–2,–3, –4,–5,6 RG:1,–2 WH+	*C4A*03*	c.3527A>G	28	p.Asn1176Ser	PMID: 2444535	n.a.	rs17874654	
Null phenotypes								
Rg- or RG: –1,–2	*C4A*N.01*	C4A gene deletion	1-41	Deletion of 28-30 kb including C4A and 21-OHA	PMID: 15794202 PMID: 2996881	n.a.	n.a.	Known as *C4AQ0*
Rg- or RG: –1,–2	*C4A*N.02*	c.3695_3696dupTC	29	p.Ser1232SerfsTer1307	PMID: 12133986 PMID: 8473511	n.a.	rs760602547	Known as *C4AQ0*
Rg- or RG: –1,–2	*C4A*N.03*	c.2490delC	20	p.Phe830PhefsTer863	PMID: 9796739	n.a.	n.a.	Known as *C4AQ0*
Rg- or RG: –1,–2	*C4A*N.04*	c.1546_1547delGT	13	p.Val516ValfsTer625	PMID: 15294999	n.a.	n.a.	Known as *C4AQ0*

References

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	Track of changes		from version	to version
1	**Version**			**v1.0 30-JUN-2021**
2	Author	created:		Ji Yanli, June 2021
3	Review	reviewed:		Peter Ligthart, June 2021
4	General			First Excel map version. Spread-sheets "Intro", "Allele Table", "References", and "Versioning" created.
5	References	All references new		All references until PMID 9796739 added for the first time.
6	**End Version**			**v1.0 30-JUN-2021**