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*03* (shaded)
Reference allele *C4B*03* 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*03* (shaded)
Reference allele *C4A*03* encodes:	Reference allele *C4A*03* 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*07*	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	to
1	**Version**		**v1.0 30-JUN-2021**	**v1.1 30-SEP-2023**
2	Author	created:	Ji Yanli, June 2021	Ji Yanli, September 2023
3	Review	reviewed:	Peter Ligthart, June 2021	Peter Ligthart, (Christoph Gassner,) September 2023
4	Allele	renamed	*C4A*03* existed wrongly 2 times	*C4A*03* and *C4A*07* are different allele but had the same name. WH-variant of the *C4A* -gene is now *C4A*07*
5	Intro	corrected	Reference Allele *C4A*03* was wrongly named *C4B*03.*	2nd Reference Allele correctly named *C4A*03*
6	Intro	corrected	Reference Allele *C4B*3* was missing a '0'	corrected to *C4B*03*
7	Intro	corrected	Reference Allele *C4A*3* was missing a '0'	corrected to *C4A*03*
8	**End Version**		**v1.0 30-JUN-2021**	**v1.1 30-SEP-2023**

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			from	to
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**