Appendix 1

Table A1.1. Antigens of the Rh Blood Group System in Four Nomenclatures

NUMERIC	FISHER-RACE	WEINER	ISBT NUMBER	OTHER NAMES OR COMMENT
Rh1	D	Rh_0	004001	
Rh2	C	rh′	004002	
Rh3	E	rh″	004003	
Rh4	c	hr′	004004	
Rh5	e	hr″	004005	
Rh6	ce	hr	004006	f
Rh7	Ce	rh_i	004007	
Rh8	C^w	rh^{w1}	004008	
Rh9	C^x	rh^x	004009	
Rh10	V	hr^v	004010	ce^S
Rh11	E^w	rh^{w2}	004011	
Rh12	G	rh^G	004012	
Rh13†		Rh^A	004013	
Rh14†		Rh^B	004014	
Rh15†		Rh^C	004015	
Rh16†		Rh^D	004016	
Rh17		Hr_0	004017	
Rh18		Hr	004018	Hr^S (High prevalence)
Rh19		hr^s	004019	
Rh20	VS	e^s	004020	
Rh21	C^G		004021	
Rh22	CE	Rh	004022	Jarvis
Rh23	D^W		004023	Wiel
Rh24†	E^T		004024	
Rh25*/†			004025	
Rh26	c-like		004026	Deal
Rh27	cE	rh_i	004027	
Rh28		hr^H	004028	Hernandez
Rh29			004029	Total Rh
Rh30	D^{cor}		004030	Go^a (low prevalence) DIVa
Rh31		hr^B	004031	
Rh32		$\overline{R^N}$	004032	Troll (low prevalence)
Rh33		R_0^{Har}	004033	D^{HAR} (low prevalence)
Rh34		Hr^B	004034	Bastiaan
Rh35			004035	(low prevalence)
Rh36			004036	Be^a (Berrens; low prevalence)

Table A1.1. Antigens of the Rh Blood Group System in Four Nomenclatures-continued.

NUMERIC	FISHER-RACE	WEINER	ISBT NUMBER	OTHER NAMES OR COMMENT
Rh37			004037	Evans (low prevalence)
Rh38†				Formerly Duclos
Rh39	C-like		004039	
Rh40	Tar		004040	Targett (low prevalence)
Rh41	Ce-like		004041	
Rh42	Ce^S, Cce^S	rh_i^s	004042	Thornton
Rh43			004043	Crawford (low prevalence)
Rh44			004044	Nou (high prevalence)
Rh45			004045	Riv
Rh46			004046	Sec (high prevalence)
Rh47	"Allelic"	to $\overline{R^N}$	004047	Dav (high prevalence)
Rh48			004048	JAL (low prevalence)
Rh49			004049	Stem
Rh50			004050	FPTT (low prevalence)
Rh51			004051	MAR (high prevalence)
Rh52			004052	BARC (low prevalence)
Rh53			004053	JAHK (low prevalence)
Rh54			004054	DAK (low prevalence)
Rh55			004055	LOCR (low prevalence)
Rh56			004056	CENR (low prevalence)
Rh57			004057	CEST

Adapted from (Harmening, 2012)

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RHCE    GCCTTGCAGCCTGAGATAAGGCCTTTGGCGGGTGTCTCCCCTATCGCTCCCTCAAGCCCT   60
RHD     ------------------------------------------------------------    0

RHCE    CAAGTAGGTGTTGGAGAGAGGGGTGATGCCTGGTGCTGGTGGAACCCCTGCACAGAGACG   120
RHD     ----------------------------CTGGTGCTGGTGGAACCCCTGCACAGAGACG   31
                                     ********************************

RHCE    GACACAGGATGAGCTCTAAGTACCCGCGGTCTGTCCGGCGCTGCCTGCCCCTCTGCGCCC   180
RHD     GACACAGGATGAGCTCTAAGTACCCGCGGTCTGTCCGGCGCTGCCTGCCCCTCTGGGCCC   91
        ************************************************** ****

RHCE    TAACACTGGAAGCAGCTCTCATTCTCCTCTTCTATTTTTTTACCCACTATGACGCTTCCT   240
RHD     TAACACTGGAAGCAGCTCTCATTCTCCTCTTCTATTTTTTTACCCACTATGACGCTTCCT   151
        ************************************************************

RHCE    TAGAGGATCAAAAGGGGCTCGTGGCATCCTATCAAGTCGGCCAAGATCTGACCGTGATGG   300
RHD     TAGAGGATCAAAAGGGGCTCGTGGCATCCTATCAAGTTGGCCAAGATCTGACCGTGATGG   211
        ************************************ **********************

RHCE    CGGCCCTTGGCTTGGGCTTCCTCACCTCAAATTTCCGGAGACACAGCTGGAGCAGTGTGG   360
RHD     CGGCCATTGGCTTGGGCTTCCTCACCTCGAGTTTCCGGAGACACAGCTGGAGCAGTGTGG   271
        ***** ********************** * ****************************

RHCE    CCTTCAACCTCTTCATGCTGGCGCTTGGTGTGCAGTGGGCAATCCTGCTGGACGGCTTCC   420
RHD     CCTTCAACCTCTTCATGCTGGCGCTTGGTGTGCAGTGGGCAATCCTGCTGGACGGCTTCC   331
        ************************************************************

RHCE    TGAGCCAGTTCCCTCCTGGGAAGGTGGTCATCACACTGTTCAGTATTCGGCTGGCCACCA   480
RHD     TGAGCCAGTTCCCTTCTGGGAAGGTGGTCATCACACTGTTCAGTATTCGGCTGGCCACCA   391
        ************** ********************************************

RHCE    TGAGTGCTATGTCGGTGCTGATCTCAGCGGGTGCTGTCTTGGGGAAGGTCAACTTGGCGC   540
RHD     TGAGTGCTTTGTCGGTGCTGATCTCAGTGGATGCTGTCTTGGGGAAGGTCAACTTGGCGC   451
        ******** ****************** ** ****************************

RHCE    AGTTGGTGGTGATGGTGCTGGTGGAGGTGACAGCTTTAGGCACCCTGAGGATGGTCATCA   600
RHD     AGTTGGTGGTGATGGTGCTGGTGGAGGTGACAGCTTTAGGCAACCTGAGGATGGTCATCA   511
        ****************************************** *****************

RHCE    GTAATATCTTCAACACAGACTACCACATGAACCTGAGGCACTTCTACGTGTTCGCAGCCT   660
RHD     GTAATATCTTCAACACAGACTACCACATGAACATGATGCACATCTACGTGTTCGCAGCCT   571
        ********************************* *** **** ******************

RHCE    ATTTTGGGCTGACTGTGGCCTGGTGCCTGCCAAAGCCTCTACCCAAGGGAACGGAGGATA   720
RHD     ATTTTGGGCTGTCTGTGGCCTGGTGCCTGCCAAAGCCTCTACCCGAGGGAACGGAGGATA   631
        *********** ********************************** ***************

RHCE    ATGATCAGAGAGCAACGATACCCAGTTTGTCTGCCATGCTGGGCGCCCTCTTCTTGTGGA   780
RHD     AAGATCAGACAGCAACGATACCCAGTTTGTCTGCCATGCTGGGCGCCCTCTTCTTGTGGA   691
        * ******* ************************************************

RHCE    TGTTCTGGCCAAGTGTCAACTCTGCTCTGCTGAGAAGTCCAATCCAAAGGAAGAATGCCA   840
RHD     TGTTCTGGCCAAGTTTCAACTCTGCTCTGCTGAGAAGTCCAATCGAAAGGAAGAATGCCG   751
        ************** ***************************** **************

RHCE    TGTTCAACACCTACTATGCTCTAGCAGTCAGTGTGGTGACAGCCATCTCAGGGTCATCCT   900
RHD     TGTTCAACACCTACTATGCTGTAGCAGTCAGCGTGGTGACAGCCATCTCAGGGTCATCCT   811
        ******************** ********** ****************************

RHCE    TGGCTCACCCCCAAAGGAAGATCAGCATGACTTATGTGCACAGTGCGGTGTTGGCAGGAG   960
RHD     TGGCTCACCCCCAAGGGAAGATCAGCAAGACTTATGTGCACAGTGCGGTGTTGGCAGGAG   871
        ************** ************ ********************************

RHCE    GCGTGGCTGTGGGTACCTCGTGTCACCTGATCCCTTCTCCGTGGCTTGCCATGGTGCTGG   1020
RHD     GCGTGGCTGTGGGTACCTCGTGTCACCTGATCCCTTCTCCGTGGCTTGCCATGGTGCTGG   931
        ************************************************************

RHCE    GTCTTGTGGCTGGGCTGATCTCCATCGGGGGAGCCAAGTGCCTGCCGGTGTGTTGTAACC   1080
RHD     GTCTTGTGGCTGGGCTGATCTCCGTCGGGGGAGCCAAGTACCTGCCGGGGTGTTGTAACC   991
        ********************** *************** ******** ************
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RHCE    GAGTGCTGGGGATTCACCACATCTCCGTCATGCACTCCATCTTCAGCTTGCTGGGTCTGC  1140
RHD     GAGTGCTGGGGATTCCCCACAGCTCCATCATGGGCTACAACTTCAGCTTGCTGGGTCTGC  1051
        *************** ***** **** *****  ** ** ********************

RHCE    TTGGAGAGATCACCTACATTGTGCTGCTGGTGCTTCATACTGTCTGGAACGGCAATGGCA  1200
RHD     TTGGAGAGATCATCTACATTGTGCTGCTGGTGCTTGATACCGTCGGAGCCGGCAATGGCA  1111
        ************ ********************** **** *** *   ***********

RHCE    TGATTGGCTTCCAGGTCCTCCTCAGCATTGGGGAACTCAGCTTGGCCATCGTGATAGCTC  1260
RHD     TGATTGGCTTCCAGGTCCTCCTCAGCATTGGGGAACTCAGCTTGGCCATCGTGATAGCTC  1171
        ************************************************************

RHCE    TCACGTCTGGTCTCCTGACAGGTTTGCTCCTAAATCTCAAAATATGGAAAGCACCTCATG  1320
RHD     TCATGTCTGGTCTCCTGACAGGTTTGCTCCTAAATCTTAAAATATGGAAAGCACCTCATG  1231
        *** ********************************* **********************

RHCE    TGGCTAAATATTTTGATGACCAAGTTTTCTGGAAGTTTCCTCATTTGGCTGTTGGATTTT  1380
RHD     AGGCTAAATATTTTGATGACCAAGTTTTCTGGAAGTTTCCTCATTTGGCTGTTGGATTTT  1291
          ************************************************************

RHCE    AAGCAAAAGCATCCAAGAAAAACAAGGCCTGTTCAAAAACAAGACAACTTCCTCTCACTG  1440
RHD     AAGCAAAAGCATCCAAGAAAAACAAGGCCTGTTCAAAAACAAGACAACTTCCTCTCACTG  1351
        ************************************************************

RHCE    TTGCCTGCATTTGTACGTGAGAAACGCTCATGACAGCAAAGTCTCCTTATGTATAATGAA  1500
RHD     TTGCCTGCATTTGTACGTGAGAAACGCTCATGACAGCAAAGTCTCCAATGTTCGCGCAGG  1411
        ********************************************     *

RHCE    ACAAGGTCAGAGACAGATTTGATATTAAAAAATTAAAGACTAAAAACTTA----------  1550
RHD     CACTGGAGTCAGAGAAAATGGAGTTGAATCCTTTCTCTGCCACTCTTTGAGGAGAATCTC  1471
             **    *** * * * **  * **    **     * *     * *

RHCE    ---GTTTAAGAGTCAATTTAAT----AAGTTTAAAATAAATGTT---------------T  1588
RHD     ACCATTTATTATGCACTGTAGAATACAACAATAAAATACAGCCATGTACCACATAACAAC  1531
            ****  *  ** * **      **   ******* *

RHCE    AGTTTCATTAGGATGATGCTATCAATATTTTCTTGGTTA-----------CAGACACAT  1636
RHD     ATCTTGGTAAACAACAGACTGCATATATGATGGTGGTCATCCAGTAAGCTAAGGTTAATT  1591
        *  **  * *  *  *  **    ****  *  **** *              *  *  *

RHCE    TATTAAAGTTTTGGGTTAATTTTA-----------------------------------  1660
RHD     TATTATTATTCCTTGTTTTTTTTTTTTTTTTTTTTTTTTTGAGATGTAGTCTTACTCTGT  1651
        *****   **    ***  ****

RHCE    ------------------------------------------------------------  1660
RHD     CACCCAGGCTAGAGTGCAATGGCACCATCTTGGCTCACTGCAACCTCTACCTCCTGGGTT  1711

RHCE    ------------------------------------------------------------  1660
RHD     CAAGCAAATCTCCTGCCTCAGCCTCCAAAGTAGCTGGGATTACAGGCACCCACCACATCT  1771

RHCE    ------------------------------------------------------------  1660
RHD     GGCTAATTTTTTGTATTTTTAGTAAAGATGGGGTTTCACCATGTTGGCCAGGCTGATCTC  1831

RHCE    ------------------------------------------------------------  1660
RHD     AAACTCCTGACCTCAAGTGATCTGCCCGCCTCGGCCTCCCAAAGTGCTGGAACCACAGGC  1891

RHCE    ------------------------------------------------------------  1660
RHD     CTGAGCCACTGTGCCCAGCCTTGTTTGCTTTTTTAACAGATAACAGTGTGCTCATAGAAA  1951

RHCE    ------------------------------------------------------------  1660
RHD     CTGCTTTGACATGACTGCAATCATGTGCTTCATAGAAACTTAATTAGATTATACCACTAG  2011

RHCE    ------------------------------------------------------------  1660
RHD     AGTCTTCAGATTTTTATACTTTTTTTTTTTGAAACGGAGTCTCACTCTGTCACCAGGCTG  2071

RHCE    ------------------------------------------------------------  1660
RHD     GAGTGCAGTGCCGCAATCTCGGCTCACTGCAACCTCCGCCTCCCAGGTTCAAGCAATTCT  2131
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RHCE        ------------------------------------------------------------  1660
RHD         CCTGCCTCAGCCTCCCGAGTAGCTGGAATTACAAGTGCGCACTACCACACCCAGCTAATT  2191

RHCE        ------------------------------------------------------------  1660
RHD         TTTGCATTTTTACTTGACAGGGTTTCACCATGTTGGCTAGGATAGTTTCACCAGGATCTC  2251

RHCE        ------------------------------------------------------------  1660
RHD         TTGGCCTCATGATCAGCCTGCCTCGGCCTCCCAAAGTGCTGGGATTACAGGTGTGAGCCA  2311

RHCE        ------------------------------------------------------------  1660
RHD         CCGTGCCCAGCCTATACTTCCCTTTTTGAATACCATTTGGTGTTTTGAAGAATTAACAGC  2371

RHCE        ------------------------------------------------------------  1660
RHD         TTTGTGAACGTGGCAGTGCTTGTGATTCAGGCTTCCATTGAGACCAAGGGGAGAACCTGG  2431

RHCE        ------------------------------------------------------------  1660
RHD         TTGCAGGACAAACAGACGGACAGCGTGTGGCAGTGTTTAAATGCTCTTCTGAAGGCTGAT  2491

RHCE        ------------------------------------------------------------  1660
RHD         ACGACAGCTCTCTGTGCACTGATTGCATATGCATCCCAAGATTATATTATTGTTTTCTAC  2551

RHCE        ------------------------------------------------------------  1660
RHD         TGCTATGTGTCACACTTTGCCAAACAGGATGTGGAAAATGAATAAGCGGTTTTCTTAGGC  2611

RHCE        ------------------------------------------------------------  1660
RHD         ACTTCTTAACAGACAATTGGTCAAAATGAACTCCATTGCTTAAGAAACACATAAACACCA  2671


RHCE        ------------------------------------------------------------  1660
RHD         TTTAGTCACTGAACATAGCTATATGTATGGTTGTTACTATGGGAAATCTTGTTTTGCCAA  2731

RHCE        ------------------------------------------------------------  1660
RHD         TTTTCTTTGAAAATTCTGGCAGACCAAGGTTCTTTTTGTTTACATAATACTTGAAAAATA  2791

RHCE        ------------------------        1660
RHD         AAAATGAACAAGCTAACAAACTA         2814
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Figure A1.1 cDNA sequence of *RHD* and *RHCE* genes as displayed in Ensembl genome browser.
The alignment was created using CLUSTAL O(1.2.4) multiple sequence alignment online software. The yellow highlight indicates start codon and start of coding sequence. The blue highlight indicates stop codon and end of coding sequence.

Table A1.2. List of alleles that cause RhD negative

Designation	ISBT name	Cluster	Haplotype	Mechanism	Alterations	First mention	Definitive publication
Ccdes-1	RHD*03N.01	DIVa cluster	Ccde	Complex changes	1006G>T (G336C) 186G>T (L62F) 410C>T (A137V) 455A>C (N152T) Hybrid RHCE[245V](4-7)	2004	2004
Ccdes-2	RHD*01N.06	DIVa cluster	Ccde	hybrid allele	1006G>T (G336C) Hybrid RHCE[245V](4-7)	2009	2009
RHCE(1)-D(6)-CE(7-10)	RHD*01N.42	no RHD	Cde	large deletion		2002	2002
RHCE(1-3)-RHD(4-10)	RHD*01N.43	Eurasian D cluster	cDE	hybrid allele	Hybrid RHCE(1-3)	2004	2009
RHCE(1-9)-RHD	RHD*01N.02	Eurasian D cluster	cDE	hybrid allele	Hybrid RHCE(1-9)	2001	2001
RHCE-D(3[361del11]-10)		Eurasian D cluster	not reported	Complex changes	361delTTGTCGGTGCT Hybrid RHCE(1-2)		
RHD deletion	RHD*01N.01	no RHD	cde	large deletion	RHD deletion	1991	1991
RHD psi	RHD*08N.01 RHD*Pseudogene	weak D type 4	cDe	Complex changes	609G>A 654G>C (M218I)	2000	2000

		cluster			667T>G (F223V) 674C>T (S225F) 807T>G (Y269X) IVS3-19 dupl 37		
RHD(1026C>T)		Eurasian D cluster	not reported	Silent mutation	1026C>T (I342I)	2015	2015
RHD(1080del10)	RHD*01N.36	Eurasian D cluster	not reported	Short deletion	1080del10	2010	
RHD(1228-2del21)	RHD*01N.44	Eurasian D cluster	not reported	Short deletion	IVS9-2del21	2014	
RHD(142delM)	RHD*01N.74 RHD*424_426delATG	Eurasian D cluster	not reported	In frame deletion	424delATG (142delM)		
RHD(208delinsTG)		Eurasian D cluster	not reported	Short insertion	208delinsTG	2017	
RHD(216dupCA,1195G>A)	RHD*01N.45	Eurasian D cluster	CDe	Complex changes	1195G>A (A399T) 216dupCA	2012	
RHD(297del23)	RHD*01N.37	Eurasian D cluster	not reported	Short deletion	297del23	2013	
RHD(325delA)	RHD*01N.11	Eurasia	CDe	Short	325del A	2007	2007

		n D cluster		deletion			
RHD(330delGT)	RHD*01N.35	Eurasian D cluster	not reported	Short deletion	330delGT	2007	2007
RHD(343del C)	RHD*01N.23	Eurasian D cluster	CDe	Short deletion	343del C	2004	2009
RHD(361del11)	RHD*01N.41	Eurasian D cluster	CDe	Short deletion	361delTTGTCGGTGCT	2014	2014
RHD(449del T)	RHD*01N.12	Eurasian D cluster	CDe	Short deletion	449del T	2004	
RHD(489delAGAC)	RHD*01N.13	Eurasian D cluster	CDe	Short deletion	487del ACAG	1998	1998
RHD(520G>A,1080_1989del)		Eurasian D cluster	CDe	Complex changes	1080del10 520G>A (V174M)	2014	2014
RHD(545delCTGT)	RHD*01N.46	Eurasian D cluster	cDe	Short deletion	545delCTGT	2012	2012
RHD(615delCA)	RHD*01N.34	Eurasian D cluster	CDe	Short deletion	615delCA	2009	2012

RHD(652delA 653T>G)	RHD*01N.17	Eurasian D cluster	not reported	Substitution with frameshift	652delA 653T>G	2006	
RHD(660delG)	RHD*01N.29 RHD*660delG RHD*01N.78	Eurasian D cluster	CDe	Short deletion	660delG	2008	2009
RHD(697delG)	RHD*01N.82 RHD*697delG	Eurasian D cluster	not reported	Short deletion	697delG		
RHD(702delG)	RHD*01N.83 RHD*702delG	Eurasian D cluster	not reported	Short deletion	702delG	2017	
RHD(711del C)	RHD*01N.16	Eurasian D cluster	cDE	Short deletion	711del C	2002	2002
RHD(712delG)	RHD*01N.33	Eurasian D cluster	CDe	Short deletion	712delG	2008	2009
RHD(745del13)	RHD*01N.47	Eurasian D cluster	CDe	Short deletion	745delGTGGTGACAGCCA 758TC>AG	2008	
RHD(786del A)	RHD*01EL.13 RHD*DEL13	Eurasian D cluster	CDe	Short deletion	786del A	2004	2009
RHD(78delC)	RHD*01N.32	Eurasian D	CDe	Short deletion	78delC	2009	2010

		cluster					
RHD(822delG)	RHD*01N.48	Eurasian D cluster	not reported	Short deletion	822delG	2014	
RHD(909ins TGGCT, IVS6+2del TAAG)	RHD*01N.27	Eurasian D cluster	CDe	Complex changes	909ins TGGCT IVS6+2del TAAG	2002	2002
RHD(915delC)	RHD*01N.49	Eurasian D cluster	not reported	Short deletion	915delC	2014	2014
RHD(93insT)	RHD*01EL.18 RHD*DEL18 RHD*01N.50	Eurasian D cluster	CDe	Short insertion	93insT	2008	2008
RHD(950delA)	RHD*01N.51	Eurasian D cluster	not reported	Short deletion	950delA	2012	
RHD(970delCAC,976delTCCATCATGGGCTACA)	RHD*01N.28	Eurasian D cluster	CDe	Short deletion	970delCAC 976delTCCATCATGGGCTACA	2008	2009
RHD(delEx1)	RHD*01N.67	Eurasian D cluster	not reported	large deletion	delEx1		
RHD(G212V)	RHD*01N.15	Eurasian D cluster	CDe	Missense (splice site affected)	635G>T (G212V)	2001	2001
RHD(G308X)	RHD*01EL.15	Eurasia	CDe	Nonsense	922G>T (G308X)	2010	

	RHD*DEL15 RHD*01N.52	n D cluster		mutation			
RHD(G314V)	RHD*01N.20	Eurasian D cluster	CDe	Missense (splice site affected)	941G>T (G314V)	1997	1997
RHD(G336D)	RHD*01N.80 RHD*1007A	Eurasian D cluster	not reported	Single missense mutation	1007G>A (G336D)	2011	
RHD(G385D)	RHD*01N.53	Eurasian D cluster	not reported	Missense (splice site affected)	1154G>A (G385D)	2014	2014
RHD(IVS2+1G>A)	RHD*01N.24	Eurasian D cluster	not reported	Splice site mutation	IVS2+1G>A	2007	2007
RHD(IVS2-1G>A)	RHD*01N.25	Eurasian D cluster	CDe	Splice site mutation	IVS2-1G>A	2005	2005
RHD(IVS3+1G>A)	RHD*01EL.08 RHD*DEL8	Eurasian D cluster	CDe	Splice site mutation	IVS3+1G>A	2001	2001
RHD(IVS3+2T>A)	RHD*01EL.09 RHD*DEL9	Eurasian D cluster	cDE	Splice site mutation	IVS3+2T>A	2008	2009
RHD(IVS4+1G>T,1136C>T)	RHD*01N.69	DAU cluster	not reported	Splice site mutation	1136C>T (T379M) IVS4+1G>T		
RHD(IVS5+1G>A)	RHD*01N.54	Eurasia	not	Splice site	IVS5+1G>A	2012	

		n D cluster	reported	mutation			
RHD(IVS6+1G>A)	RHD*01N.55	Eurasian D cluster	CDe	Splice site mutation	IVS6+1G>A	2014	2014
RHD(IVS6+2T>A)	RHD*01N.38	Eurasian D cluster	not reported	Splice site mutation	IVS6+2T>A	2013	
RHD(IVS7+1G>T)	RHD*01N.70	Eurasian D cluster	not reported	Splice site mutation	IVS7+1G>T		
RHD(IVS7+2T>C)	RHD*01N.56	Eurasian D cluster	not reported	Splice site mutation	IVS7+2T>C	2012	
RHD(IVS8+1G>A)	RHD*01N.26	Eurasian D cluster	CDe	Splice site mutation	IVS8+1G>A	2001	2001
RHD(L337R)	RHD*01EL.38 RHD*DEL38 RHD*01N.57	Eurasian D cluster	CDe	Single missense mutation	1010T>G (L337R)	2014	2014
RHD(L386X)		Eurasian D cluster	not reported	Single missense mutation	1157T>A (L386X) IVS5-41delCTCT	2014	2014
RHD(M218I, F223V, S225F,Y269X)		weak D type 4 cluster	not reported	Complex changes	609G>A 654G>C (M218I) 667T>G (F223V)	2009	

					674C>T (S225F) 807T>G (Y269X)		
RHD(Q200X)	RHD*01N.59	Eurasian D cluster	not reported	Nonsense mutation	598C>T (Q200X)	2011	
RHD(Q362X)	RHD*01N.64	Eurasian D cluster	not reported	Nonsense mutation	1084C>T (Q362X)		
RHD(Q405X)	RHD*01N.60	Eurasian D cluster	not reported	Nonsense mutation	1213C>T (Q405X)	2015	
RHD(Q41X)	RHD*01N.09	Eurasian D cluster	CDe	Nonsense mutation	121C>T (Q41X) 643T>C (F215L) 646T>C 988T>C (Y330H)	1997	1997
RHD(R318X)	RHD*01N.61	Eurasian D cluster	CDe	Nonsense mutation	952C>T (R318X)	2008	2009
RHD(S254X)	RHD*01N.62	Eurasian D cluster	CDe	Nonsense mutation	761C>G (S254X)	2015	2015
RHD(S256X)	RHD*01N.39	Eurasian D cluster	CDe	Nonsense mutation	767C>G (S256X)	2012	2013
RHD(S68T)-RHCe(3-9)-RHD	RHD*01N.04	Eurasian D	CDe	hybrid allele	203G>C (S68T) Hybrid RHCE(3-9)	2005	2005

		cluster					
RHD(T148R)	RHD*01N.73 RHD*443G	Eurasian D cluster	not reported	Single missense mutation	443C>G (T148R)	2012	2012
RHD(V56M,W90X)		Eurasian D cluster	CDe	Nonsense mutation	166G>A (V56M) 270G>A (W90X)	2008	2009
RHD(W16X)	RHD*01N.08	Eurasian D cluster	CDe	Nonsense mutation	48G>A (W16X)	2001	2001
RHD(W185X)	RHD*01N.14	Eurasian D cluster	CDe	Nonsense mutation	554G>A (W185X)	2005	2005
RHD(W185X) [c,555G>A]		Eurasian D cluster	not reported	Nonsense mutation	555G>A (W185X)	2015	2015
RHD(W90X)	RHD*01N.10	Eurasian D cluster	CDe	Nonsense mutation	270G>A (W90X)	2002	2002
RHD(Y269X)	RHD*01N.18	Eurasian D cluster	CDe	Nonsense mutation	807T>G (Y269X)	2004	2009
RHD(Y311X)[761G]	RHD*01N.63	Eurasian D cluster	not reported	Nonsense mutation	933C>G (Y311X)		
RHD(Y311X)[933A]	RHD*01N.19	Eurasia	CDe	Nonsense	933C>A (Y311X)	2005	2005

		n D cluster		mutation			
RHD(Y330X)	RHD*01N.21	Eurasian D cluster	CDe	Nonsense mutation	990C>G (Y330X)	2001	2001
RHD(Y343X)	RHD*01N.40	Eurasian D cluster	cDE	Nonsense mutation	1029C>A (Y343X)	2012	2013
RHD(Y401X)	RHD*01N.22 RHD*DEL17 RHD*01EL.17	Eurasian D cluster	cDE	Nonsense mutation	1203T>A (Y401X)	2004	2005
RHD*745_757del	RHD*01N.30	Eurasian D cluster	not reported	Short deletion	745delGTGGTGACAGCCA		
RHD-RHCE(2-10)		no RHD	Cde	hybrid allele	Hybrid RHCE(2-10)	2004	2004
RHD-RHCe(2-7)-RHD	RHD*01N.05	Eurasian D cluster	CDe	hybrid allele	Hybrid RHCE(3-7)	2001	2001
RHD-RHCe(2-9)-RHD	RHD*01N.03	Eurasian D cluster	CDe	hybrid allele	Hybrid RHCE(3-9)	1996	1996
RHD-RHCE(3)--weak D type 4.0	RHD*01N.72	weak D type 4 cluster	not reported	hybrid allele	602C>G (T201R) 667T>G (F223V) 819G>A Hybrid RHCE(3)		

RHD-RHCE(4-7)-RHD	RHD*01N.07	Eurasian D cluster	cDE	hybrid allele	Hybrid RHCE(4-7)	1996	1996
RHD-RHCE(4-7)-RHD1	RHD*01N.07	Eurasian D cluster	cDE	hybrid allele	Hybrid RHCE(4-7)		
RHD-RHCE(4-7)-RHD2	RHD*01N.07	Eurasian D cluster	cDE	hybrid allele	Hybrid RHCE(4-7)		
RHD-RHCE(4-8)-RHD	RHD*01N.07	Eurasian D cluster	CDe	hybrid allele	Hybrid RHCE(4-7)	2005	2005
RHD-RHCE(8-9)-RHD		Eurasian D cluster	CDe	hybrid allele	Hybrid RHCE(8-9)	1997	1997
RHDex10del type 2		Eurasian D cluster	not reported	large deletion	del1227-2108_1254+1317	2017	2017

Appendix 2: Forms

استمارة موافقة المتبرع على المشاركة بالبحث

أسم الباحث: مجتبى بن علي بن ابراهيم اللواتي و عامر بن علي الحارثي
عنوان الباحث: معهد العلوم الصحية – قسم المختبرات الطبية
مكان إجراء البحث: معهد العلوم الصحية ومختلف المستشفيات المرجعية في السلطنة
أنت مدعو للمشاركة ببحث علمي في فصائل الدم من نوع الريسيسي الموجب الضعيف . يرجى أن تأخذ الوقت المناسب لقراءة المعلومات الآتية بتأنٍ قبل أن تقرر إذا ما كنت راغباً بالمشاركة أم لا. وبإمكانك طلب مزيداً من الإيضاحات أو المعلومات الإضافية عن أي أمر مذكور بالاستمارة أو عن الدراسة من المختصين في مختبر بنك الدم.

1- **وصف مشروع البحث وأهدافه ومساره: هدف المشروع هو معرفة تسلسل الجيني لفصيلة الريسيسي الموجبة جزئيا**
2- **الفوائد الايجابية المحتملة للمشارك التي قد تنتج من هذا البحث: يمنح المشروع احتمالية النجاح في معرفة التسلسل الجيني الذي قد يكون جديدا في عالم الأبحاث**
3- **التأثيرات السلبية أو الأعراض الجانبية المحتملة التي يتعرض لها المشارك: لا توجد**

وفي حال موافقتك على المشاركة في هذه الدراسة سيبقى أسمك قيد الكتمان. ولا يسمح لأي شخص "ما لم ينص عليه القانون" حق الاطلاع على ذلك باستثناء الباحث عن الدراسة ومعاونيه ولجان الأخلاق المهنية المستقلة.

وثيقة الموافقة التحريرية
لقد أوضحت للمشارك (.....)بالتفصيل البحث وطبيعته ومجرياته وفوائده المحتملة وسلبياته المحتملة أيضاً. وأجبت عن كل استفساراته وأسئلته بوضوح على أفضل ما استطيع. وسأعلم المشارك بأي تغييرات في مجريات البحث أو فوائده أو سلبياته حال حصولها في أثناء البحث.

أسم الباحث أو الشخص المخول بذلك التوقيع التاريخ / /

موافقة المشارك
لقد اطلعت على استمارة الموافقة وأدركت مضمونها وتمت الإجابة عن جميع الأسئلة التي تجول في ذهني. وبناءً عليه فأني حراً ومختاراً أوافق على المشاركة بالبحث و نشر نتائجه في المجلات الطبية. وفهمت أن الباحث / (مجتبى بن علي بن ابراهيم اللواتي) وزملاؤه ومساعديه سيكونون مستعدين للإجابة عن أسئلتي المستقبلية. وباستطاعتي الاتصال بهم على رقم الهاتف (99777341).. كما أعلم تماماً بأنني حر في الانسحاب من هذا البحث متى شئت ولو بعد الموافقة التحريرية ومصادقتها.

أسم المشارك : التوقيع:
التاريخ: / /

Figure A2.1 Template of the Arabic consent letter given to the donor before sample collection.

Appendix 3: Calculations

Calculation of Rh haplotypes frequency:

$p^2 + 2pq + 2pr + q^2 + 2qr + r^2 = 1$

(r is *r* haplotype, p is *r´* haplotype and q is *r´´* haplotype, square is diplotype)

dce (r) phenotype = *r* haplotype = r in equation

rr = $r^2 = 0.773$

Therefore, frequency of r = $\sqrt{0.773} = 0.879$

dce haplotype frequency is 0.879

To calculate dCe (*r´*) haplotype (p in the equation),

$p^2 + 2pr = 0.197$ (total frequency of *r´r´* + *rr´* which is 0.01 + 0.187)

$p^2 + (2p \times 0.879) - 0.197 = 0$

$p^2 + 1.758p - 0.197 = 0$ (Quadratic equation)

(p-0.106) (p+1.86) = 0

Therefore, p = 0.106 , $p^2 = 0.011$

To calculate dcE (*r´´*) haplotype (q in the equation),

$q^2 + 2qr = 0.03$

$q^2 + (2p \times 0.879) - 0.03 = 0$

$p^2 + 1.758p - 0.03 = 0$ (Quadratic equation)

(p-0.016) (p+1.77) = 0

Therefore, p = 0.016 , $p^2 = 0.0002$

Thus, r + p + q = 1

0.879 + 0.105 + 0.016 = 1

Calculation of Genotype and allele frequencies of inactive RHD genes and RHD deletion using Bernstein's equation.

Two patterns that contributed to true serological D negative are, absence of hybrid (*RHD-CE-D*^{s}) and pseudogene (*del/del*) denoted as [r] and presence of pseudogene and absence of (*RHD-CE-D*^{s}) hybrid (*RHDΨ / RHDΨ* or *RHDΨ /del*) denoted as [q], where q + r = 1. The *del/del* genotype was counted to be 179 (94.709%), whereas *RHDΨ / RHDΨ* or *RHDΨ /del* was counted to be 10 (5.291%) with total of 189 true D negative.

Using Bernstein's equation

r (*del/del*) = $(F[del])^{½}$ or r = $\sqrt{0.94709}$ = 0.9732

q (*RHDΨ / RHDΨ or RHDΨ /del*) = 1 – $(F[del])^{½}$ = 0.0268

Deviation (D) = 1 - (q + r)

Since, only two alleles responsible for the serological D negative phenotype, deviation value (D) is 0 as we subtracted 0.9732 (*del/del*) from the 1 to get the only second allele (*RHDΨ / RHDΨ or RHDΨ /del*) frequency.