

Names for RHD (ISBT 004) negative null blood group alleles v3.0 160713

Phenotype	Allele name	Nucleotide change	Exon	Predicted amino acid change	Allele name detail	Comments
D-	<i>RHD*01N.01</i>	<i>RHD</i> deletion	1-10	p.0		
D-	<i>RHD*08N.01</i> <i>RHD*Pseudo-gene</i> <i>RHD* Ψ</i>	37- bp insertion c.609G>A; c.654G>C; c.667T>G; c.674C>T; c.807T>G	Intron 3 45 6	p.0		
D-	<i>RHD*01N.02</i>	CE exons 1-9	1-9	Hybrid	<i>RHD*CE(1-9)-D</i>	
D-	<i>RHD*01N.03</i>	CE exons 2-9	2-9	Hybrid	<i>RHD*D-CE(2-9)-D</i>	Ce in trans
D-	<i>RHD*01N.04</i>	CE exons 3-9	3-9	Hybrid	<i>RHD*D-CE(3-9)-D</i>	
D-	<i>RHD*01N.05</i>	CE exons 2-7	2-7	Hybrid	<i>RHD*D-CE(2-7)-D</i>	
D- C+ very weak	<i>RHD*01N.06</i>	CE exons 4-7 with 733C>G 1006G>T	4-7	Hybrid with p.Leu245Val p. Gly336Cys	<i>RHD*D-CEVS(4-7)-D</i>	(Part of r ^S Haplotype) type 2 hybrid
D- C+	<i>RHD*03N.01</i>	c.186G>T c.410C>T c.455A>C CE exons 4-7 with 733C>G 1006G>T	2 3 4-7	p.Leu62Phe p. Ala137Val p. Asn152Thr hybrid with p.Leu245Val p. Gly336Cys	<i>RHD*DIlla-CEVS(4-7)-D</i>	(Part of r ^S haplotype) type 1 hybrid
D-G+	<i>RHD*01N.07</i>	CE exons 4-7	4-7	Hybrid	<i>RHD*D-CE(4-7)-D</i>	in cis to cE (r'G) or Ce
D-	<i>RHD*01N.08</i>	c.48G>A	1	p.Trp16Ter	<i>RHD*48A</i>	premature stop codon
D-	<i>RHD*01N.09</i>	c.121C>T; c.643T>C; c.646T>C; c.988T>C	1 5 7	p.Gln41Ter	<i>RHD*121T,643C,646C,988C</i>	

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D-	<i>RHD*01N.10</i>	c.270G>A	2	p.Trp90Ter	<i>RHD*270A</i>	premature stop codon
D-	<i>RHD*01N.11</i>	c.325delA	2	p.Thr109Hisfs*10	<i>RHD*325delA</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.12</i>	c.449delT	3	p.Leu150Ter	<i>RHD*449delT</i>	Premature stop codon
D-	<i>RHD*01N.13</i>	c.487_490delACAG	4	p.Asp164Thrfs*3	<i>RHD*487_490delACAG</i> Previous <i>RHD*487delACAG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.14</i>	c.554G>A	4	p.Trp185Ter	<i>RHD*554A</i>	premature stop codon
D-	<i>RHD*01N.15</i>	c.635G>T	5	p.Gly212Val	<i>RHD*635T</i>	splice site change
D-	<i>RHD*01N.16</i>	c.711delC	5	p.Val238Cysfs*8	<i>RHD*711delC</i>	frameshift and premature stop codon
D-	<i>RHD*01N.17</i>	c.652delA; c.653T>G	5	p.Met218Glyfs*11	<i>RHD*652delA,653G</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.18</i>	c.807T>G	6	p.Tyr269Ter	<i>RHD*807G</i>	premature stop codon
D-	<i>RHD*01N.19</i>	c.933C>A	6	p.Tyr311Ter	<i>RHD*933A</i>	premature stop codon
D-	<i>RHD*01N.20</i>	c.941G>T	7	p.Gly314Val	<i>RHD*941T</i>	splice site change
D-	<i>RHD*01N.21</i>	c.990C>G	7	p.Tyr330Ter	<i>RHD*990G</i>	premature stop-codon
D-	<i>RHD*01N.22</i>	c.1203T>A	9	p.Tyr401Ter	<i>RHD*1203A</i>	premature stop-codon
D-	<i>RHD*01N.23</i>	c.343delC	3	p.Leu115Trpfs*4	<i>RHD*343delC</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.24</i>	c.335+1G>A	Intron 2	Alternative splicing	<i>RHD*335+1A</i> Previous <i>RHD*IVS2+1G>A</i>	splice site change

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D-	<i>RHD*01N.25</i>	c.336-1G>A	Intron 2	Alternative splicing	<i>RHD*336-1A</i> Previous <i>RHD*IVS2-1G>A</i>	splice site change
D-	<i>RHD*01N.26</i>	c.1153+1G>A	Intron 8	Alternative splicing	<i>RHD*1153+1A</i> Previous <i>RHD*IVS8+1G>A</i>	splice site mutation
D-	<i>RHD*01N.27</i>	c.905_906insTGGCT; c.939+2_939+5delTAA G	6 Intron 6	p.Leu303Glyfs*3	<i>RHD*905_906insTG GCT,</i> <i>939+2_939+5del TAAG</i> Previous <i>RHD*906ins,IVS6</i>	Frameshift and premature stop codon & splice site change
D-	<i>RHD*01N.28</i>	c.970_972delCAC; c.976_991delTCCATC ATGGGCTACA	7	p.His324Serfs*29	<i>RHD*970_972delCA c,976_991delTCCAT CATGGGCTACA</i> Previous <i>RHD*970del3, 976del16</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.29</i>	c.660delG	5	p.Trp220Cysfs*9	<i>RHD*660delG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.30</i>	c.745_757del13	5	p.Val249Serfs*35	<i>RHD*745_757del</i> Previous <i>RHD*745del13</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.31</i>	c.93delC (T at position 93)	1	fs at 31	<i>RHD*93delC</i>	frameshift
D-	<i>RHD*01N.32</i>	c.78delC	1	p.Leu27Serfs*12	<i>RHD*78delC</i>	Frameshift and premature stop codon

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D-	<i>RHD*01N.33</i>	c.712delG	5	p.Val238Cysfs*8	<i>RHD*712delG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.34</i>	c.615_616delCA	4	p.Ser206Phefs*111	<i>RHD*615_616delCA</i> Previous <i>RHD*615delCA</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.35</i>	c.330_331delGT	3	p.Phe111Glnfs*48	<i>RHD*330_331delGT</i> Previous <i>RHD*330delGT</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.36</i>	c.1080_1089delCTTC CAGGTC	8	p.Phe361Serfs*12	<i>RHD*1080_1089delCTTC</i> <i>CAGGTC</i> previous <i>RHD*1080del</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.37</i>	c.297_319delCCAGTT CCCTTCTGGGAAGG TGG	2	p.Gln100Hisfs*52	<i>RHD*297_319delCCAGTT</i> <i>CCCTTCTGGGAAGGTGG</i> Previous <i>RHD*297del23bp</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.38</i>	c.939+2T>A	Intron 6	Alternative splicing	<i>RHD*939+2A</i> Previous <i>RHD*IVS6+2T>A</i>	splice site change
D-	<i>RHD*01N.39</i>	c.767C>G	5	p.Ser256Ter	<i>RHD*767G</i>	premature stop codon
D-	<i>RHD*01N.40</i>	c.1029C>A	7	p.Tyr343Ter	<i>RHD*1029A</i>	premature stop codon
D-	<i>RHD*01N.41</i>	c.361_371delTTGTCG GTGCT	3	p.Leu121Aspfs*35	<i>RHD*361_371delTTGTCG</i> <i>GTGCT</i> Previous <i>RHD*361del11bp</i>	Frameshift and premature stop codon

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D-	<i>RHD*01N.42</i>	CE exons1, 7-10	1, 7-10	Hybrid	<i>RHD*CE(1)-D(6)-CE(7-10)</i>	
D-	<i>RHD*01N.43</i>	CE exons 1-3	1, 2, 3	Hybrid	<i>RHD*CE(1-3)- D</i>	
D-	<i>RHD*01N.44</i>	c.1228-1_1248delgTTTCCTC ATTTGGCTGTTGGA	Intron 9	p.?	<i>RHD*1228-1_1248delTTTCCTC ATTTGGCTGTTGG A previous RHD*1228-2del21</i>	splice site change
D-	<i>RHD*01N.45</i>	c.216_217dupCA; c.1195G>A	2	p.Ser73Thrfs*27	<i>RHD*216_217dupCA , 1195A Previous RHD*216dupCA,1195A</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.46</i>	c.545_548delCTGT	4	p.Ser182Trpfs*46	<i>RHD*545_548delCTGT Previous RHD*545delCTGT</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.47</i>	c.745_759delinsAG	5	p.Val249Serfs*35	<i>RHD*745_759delinsAG Previous RHD*745del13bp,758AG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.48</i>	c.822delG	6	p.Leu275Trpfs*13	<i>RHD*822delG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.49</i>	c.915delC	6	p.Val306Serfs*53	<i>RHD*915delC</i>	Frameshift and premature stop codon

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D-	<i>RHD*01N.50</i>	c.93dupT	1	p.Thr32Tyrfs*4	<i>RHD*93dupT</i> Previous <i>RHD*93insT</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.51</i>	c.950delA	7	p.Asn317Thrfs*42	<i>RHD*950delA</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.52</i>	c.922G>T	6	p.Gly308Ter	<i>RHD*922T</i>	premature stop-codon
D-	<i>RHD*01N.53</i>	c.1154G>A	9	p.Gly385Asp	<i>RHD*1154A</i>	Splice site change
D-	<i>RHD*01N.54</i>	c.801+1G>A	Intron 5	Alternative splicing	<i>RHD*801+1A</i> Previous <i>RHD*IVS5+1G>A</i>	splice site mutation
D-	<i>RHD*01N.55</i>	c.939+1G>A	Intron 6	Alternative splicing	<i>RHD*939+1A</i> Previous <i>RHD*IVS6+1G>A</i>	splice site mutation
D-	<i>RHD*01N.56</i>	c.1073+2T>C	Intron 7	Alternative splicing	<i>RHD*1073+2C</i> Previous <i>RHD*IVS7+2T>C</i>	splice site mutation
D- Del	<i>RHD*01N.57</i>	c.1010T>G	7	p.Leu337Arg	<i>RHD*1010G</i>	Could be DEL per publication as no adsorption elution was performed
D-	<i>RHD*01N.58</i>	c.802-41_802-38delCTCT; c.1157T>A	Intron 5 9	p.Leu386Ter	<i>RHD*802-41_802-38delCTCTI, 1157A</i> Previous <i>RHD*IVS5-41delCTCT, 1157A</i>	premature stop-codon
D-	<i>RHD*01N.59</i>	c.598C>T	4	p.Gln200Ter	<i>RHD*598T</i>	premature stop codon

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D-	<i>RHD*01N.60</i>	c.1213C>T	9	p.Gln405Ter	<i>RHD*1213T</i>	premature stop codon
D-	<i>RHD*01N.61</i>	c.952C>T	7	p.Arg318Ter	<i>RHD*952T</i>	premature stop codon
D-	<i>RHD*01N.62</i>	c.761C>G	5	p.Ser254Ter	<i>RHD*761G</i>	premature stop codon
D-	<i>RHD*01N.63</i>	c.933C>G	6	p.Tyr311Ter	<i>RHD*933G</i>	premature stop codon
D-	<i>RHD*01N.64</i>	c.1084C>T	8	p.Gln362Ter	<i>RHD*1084T</i>	
D-	<i>RHD*01N.65</i>	c.124_125delAA	1	p.Lys42Glyfs*117	<i>RHD*124_125delAA</i> Previous <i>RHD*124delAA</i>	frameshift
D-	<i>RHD*01N.66</i>	c.1174delA	9	p.Ile392Tyrfs*25	<i>RHD*1174delA</i>	frameshift
D-	<i>RHD*01N.67</i>	RHD exon 1 deletion (intron breakpoints unknown)	1	p.0	<i>RHD*(2-10)</i>	
D-	<i>RHD*01N.68</i>	c.335G>T	2	p.Ser112Ile	<i>RHD*335T</i>	splice site change
D-	<i>RHD*01N.69</i>	c.634+1G>T; c.1136C>T	Intron 4 8	Alternative splicing	<i>RHD*634+1T, 1136T</i> Previous <i>RHD*IVS4+1G>T, 1136T</i>	splice site change
D-	<i>RHD*01N.70</i>	c.1073+1G>T	Intron 7	Alternative splicing	<i>RHD*1073+1T</i> Previous <i>RHD*IVS7+1G>T</i>	splice site change
D-	<i>RHD*01N.71</i>	c.1074-1G>A	Intron 7	Alternative splicing	<i>RHD*1074-1A</i> Previous <i>RHD*IVS7-1G>A</i>	splice site change

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D-	<i>RHD*01N.72</i>	c.361T>A; c.380T>C; c.383A>G; c.455A>C; c.602C>G; c.667T>G; c.819G>A	3 4 5 6	p.Leu121Met; p.Val127Ala; p.Asp128Gly; p.Asn152Thr; p.Thr201Arg; p.Phe223Leu silent	<i>RHD*361A, 380C, 383G, 455C, 602G, 667G, 819A</i>	
D-	<i>RHD*01N.73</i>	c.443C>G	3	p.Thr148Arg	<i>RHD*443G</i>	
D-	<i>RHD*01N.74</i>	c.424_426delATG	3	p.Met142del	<i>RHD*424_426delATG</i> Previous <i>RHD*424del3</i>	In-frame deletion
D-	<i>RHD*01N.75</i>	c.581_582insG	4	p.Thr195Asnfs*4	<i>RHD*581_582insG</i> Previous <i>RHD*581insG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.76</i>	c.1179G>A	9	p.Trp393Ter	<i>RHD*1179A</i>	premature stop codon
D-	<i>RHD*01N.77</i>	c.1228-1G>A	Intron 9	Alternative splicing	<i>RHD*1228-1A</i>	splice site change
D-	<i>RHD*01N.78</i>	c.659delG	5	p.Trp220Cysfs*9	<i>RHD*659delG</i>	Frameshift and premature stop codon
D-	<i>RHD*01N.79</i>	c.896T>C	6	p.Leu299Pro	<i>RHD*896C</i>	
D-	<i>RHD*01N.80</i>	c.1007G>A	7	p.Gly336Asp	<i>RHD*1007A</i>	
D-	<i>RHD*01N.81</i>	c.53delT	1	p.Leu18Glnfs*21	<i>RHD*53delT</i>	Frameshift and premature stop codon