

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

| RHD negative, i.e. null |                                                             |                                                                               |                     |                                                                                          |                              |                                                     |
|-------------------------|-------------------------------------------------------------|-------------------------------------------------------------------------------|---------------------|------------------------------------------------------------------------------------------|------------------------------|-----------------------------------------------------|
| 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><br><i>RHD*Pseudo-gene</i><br><i>RHD*Ψ</i> | 37- bp insertion<br>c.609G>A;<br>c.654G>C; c.667T>G;<br>c.674C>T;<br>c.807T>G | Intron 3<br>45<br>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-<br>C+ very weak      | <i>RHD*01N.06</i>                                           | CE exons 4-7 with<br>733C>G<br>1006G>T                                        | 4-7                 | Hybrid with<br>p.Leu245Val<br>p. Gly336Cys                                               | <i>RHD*D-CEVS(4-7)-D</i>     | (Part of r <sup>S</sup> Haplotype)<br>type 2 hybrid |
| D-<br>C+                | <i>RHD*03N.01</i>                                           | c.186G>T<br>c.410C>T<br>c.455A>C<br>CE exons 4-7 with<br>733C>G<br>1006G>T    | 2<br>3<br>4-7       | p.Leu62Phe<br>p. Ala137Val<br>p. Asn152Thr<br>hybrid with<br>p.Leu245Val<br>p. Gly336Cys | <i>RHD*DIlla-CEVS(4-7)-D</i> | (Part of r <sup>S</sup> haplotype)<br>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                                |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                                 |             |                  |                                                                |                                     |
|----|-------------------|-------------------------------------------------|-------------|------------------|----------------------------------------------------------------|-------------------------------------|
| D- | <i>RHD*01N.09</i> | c.121C>T;<br>c.643T>C;<br>c.646T>C;<br>c.988T>C | 1<br>5<br>7 | p.Gln41Ter       | <i>RHD*121T,643C,646C,988C</i>                                 |                                     |
| 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><br>Previous<br><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 |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                                      |               |                      |                                                                                                               |                                                                |
|----|-------------------|------------------------------------------------------|---------------|----------------------|---------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------|
| D- | <i>RHD*01N.24</i> | c.335+1G>A                                           | Intron 2      | Alternative splicing | <i>RHD*335+1A</i><br>Previous<br><i>RHD*IVS2+1G&gt;A</i>                                                      | splice site change                                             |
| D- | <i>RHD*01N.25</i> | c.336-1G>A                                           | Intron 2      | Alternative splicing | <i>RHD*336-1A</i><br>Previous<br><i>RHD*IVS2-1G&gt;A</i>                                                      | splice site change                                             |
| D- | <i>RHD*01N.26</i> | c.1153+1G>A                                          | Intron 8      | Alternative splicing | <i>RHD*1153+1A</i><br>Previous<br><i>RHD*IVS8+1G&gt;A</i>                                                     | splice site mutation                                           |
| D- | <i>RHD*01N.27</i> | c.905_906insTGGCT;<br>c.939+2_939+5delTAA<br>G       | 6<br>Intron 6 | p.Leu303Glyfs*3      | <i>RHD*905_906insTG<br/>GCT,</i><br><i>939+2_939+5del<br/>TAAG</i><br>Previous<br><i>RHD*906ins,IVS6</i>      | Frameshift and<br>premature stop codon &<br>splice site change |
| D- | <i>RHD*01N.28</i> | c.970_972delCAC;<br>c.976_991delTCCATC<br>ATGGGCTACA | 7             | p.His324Serfs*29     | <i>RHD*970_972delCA<br/>c,976_991delTCCAT<br/>CATGGGCTACA</i><br>Previous<br><i>RHD*970del3,<br/>976del16</i> | Frameshift and<br>premature stop codon                         |
| D- | <i>RHD*01N.29</i> | c.660delG                                            | 5             | p.Trp220Cysfs*9      | <i>RHD*660delG</i>                                                                                            | Frameshift and<br>premature stop codon                         |
| D- | <i>RHD*01N.30</i> | c.745_757del13                                       | 5             | p.Val249Serfs*35     | <i>RHD*745_757del</i><br>Previous<br><i>RHD*745del13</i>                                                      | Frameshift and<br>premature stop codon                         |
| D- | <i>RHD*01N.31</i> | c.93delC<br>(T at position 93)                       | 1             | fs at 31             | <i>RHD*93delC</i>                                                                                             | frameshift                                                     |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                             |          |                      |                                                                                                         |                                     |
|----|-------------------|---------------------------------------------|----------|----------------------|---------------------------------------------------------------------------------------------------------|-------------------------------------|
| D- | <i>RHD*01N.32</i> | c.78delC                                    | 1        | p.Leu27Serfs*12      | <i>RHD*78delC</i>                                                                                       | Frameshift and premature stop codon |
| 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><br>Previous<br><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><br>Previous<br><i>RHD*330delGT</i>                                              | Frameshift and premature stop codon |
| D- | <i>RHD*01N.36</i> | c.1080_1089delCTTC<br>CAGGTC                | 8        | p.Phe361Serfs*12     | <i>RHD*1080_1089delCTTC</i><br><i>CAGGTC</i><br>previous<br><i>RHD*1080del</i>                          | Frameshift and premature stop codon |
| D- | <i>RHD*01N.37</i> | c.297_319delCCAGTT<br>CCCTTCTGGGAAGG<br>TGG | 2        | p.Gln100Hisfs*52     | <i>RHD*297_319delCCAGTT</i><br><i>CCCTTCTGGGAAGG</i><br><i>TGG</i><br>Previous<br><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><br>Previous<br><i>RHD*IVS6+2T&gt;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_371delTTGTGCT<br>GTGCT                | 3        | p.Leu121Aspfs*35     | <i>RHD*361_371delTTGTGCT</i><br><i>GTGCT</i><br>Previous<br><i>RHD*361del11bp</i>                       | Frameshift and premature stop codon |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                            |          |                  |                                                                                            |                                     |
|----|-------------------|--------------------------------------------|----------|------------------|--------------------------------------------------------------------------------------------|-------------------------------------|
| 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<br>ATTTGGCTGTTGGA | Intron 9 | p.?              | <i>RHD*1228-1_1248delTTTCCTC<br/>ATTTGGCTGTTGGA<br/>A<br/>previous<br/>RHD*1228-2del21</i> | splice site change                  |
| D- | <i>RHD*01N.45</i> | c.216_217dupCA;<br>c.1195G>A               | 2        | p.Ser73Thrfs*27  | <i>RHD*216_217dupCA<br/>, 1195A<br/>Previous<br/>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<br/>Previous<br/>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<br/>Previous<br/>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 |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|           |                   |                                      |               |                      |                                                                                        |                                                                     |
|-----------|-------------------|--------------------------------------|---------------|----------------------|----------------------------------------------------------------------------------------|---------------------------------------------------------------------|
| D-        | <i>RHD*01N.50</i> | c.93dupT                             | 1             | p.Thr32Tyrfs*4       | <i>RHD*93dupT</i><br>Previous<br><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><br>Previous<br><i>RHD*IVS5+1G&gt;A</i>                               | splice site mutation                                                |
| D-        | <i>RHD*01N.55</i> | c.939+1G>A                           | Intron 6      | Alternative splicing | <i>RHD*939+1A</i><br>Previous<br><i>RHD*IVS6+1G&gt;A</i>                               | splice site mutation                                                |
| D-        | <i>RHD*01N.56</i> | c.1073+2T>C                          | Intron 7      | Alternative splicing | <i>RHD*1073+2C</i><br>Previous<br><i>RHD*IVS7+2T&gt;C</i>                              | splice site mutation                                                |
| D-<br>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;<br>c.1157T>A | Intron 5<br>9 | p.Leu386Ter          | <i>RHD*802-41_802-38deCTCTI, 1157A</i><br>Previous<br><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                                                |
| D-        | <i>RHD*01N.60</i> | c.1213C>T                            | 9             | p.Gln405Ter          | <i>RHD*1213T</i>                                                                       | premature stop codon                                                |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                                        |               |                      |                                                                                  |                      |
|----|-------------------|--------------------------------------------------------|---------------|----------------------|----------------------------------------------------------------------------------|----------------------|
| 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><br>Previous<br><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<br>(intron breakpoints<br>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;<br>c.1136C>T                               | Intron 4<br>8 | Alternative splicing | <i>RHD*634+1T, 1136T</i><br>Previous<br><i>RHD*IVS4+1G&gt;T,</i><br><i>1136T</i> | splice site change   |
| D- | <i>RHD*01N.70</i> | c.1073+1G>T                                            | Intron 7      | Alternative splicing | <i>RHD*1073+1T</i><br>Previous<br><i>RHD*IVS7+1G&gt;T</i>                        | splice site change   |
| D- | <i>RHD*01N.71</i> | c.1074-1G>A                                            | Intron 7      | Alternative splicing | <i>RHD*1074-1A</i><br>Previous<br><i>RHD*IVS7-1G&gt;A</i>                        | splice site change   |

Names for RHD (ISBT 004) negative null blood group alleles v3.1 170309

|    |                   |                                                                                        |                  |                                                                                                       |                                                            |                                     |
|----|-------------------|----------------------------------------------------------------------------------------|------------------|-------------------------------------------------------------------------------------------------------|------------------------------------------------------------|-------------------------------------|
| D- | <i>RHD*01N.72</i> | c.361T>A;<br>c.380T>C;<br>c.383A>G;<br>c.455A>C;<br>c.602C>G;<br>c.667T>G;<br>c.819G>A | 3<br>4<br>5<br>6 | p.Leu121Met;<br>p.Val127Ala;<br>p.Asp128Gly;<br>p.Asn152Thr;<br>p.Thr201Arg;<br>p.Phe223Leu<br>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><br>Previous<br><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><br>Previous<br><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> | <i>c.1074-2A&gt;C</i>                                                                  | <i>Intron 7</i>  | <i>Alternative splicing</i>                                                                           | <i>RHD*1074-2C</i><br>Previous<br><i>RHD*IVS7-2A&gt;C</i>  | <i>splice site change</i>           |