

Wildtype allele

Primer F →
 1 gtgggagggc tataggaag aacaattagg atatggggca ttgagggggc aggcaagagc
 61 catgtcaaca cttctctctg acactagGTC CAGAGTCCAG CCAGACATTG CCCCAGAAT
 121 CCACGGCCAC AGATATCCTG GGGCTAAGGC CTGGAACCTC CTACCAGGTG GCTGTGTCAG
 181 CACTGCGAGG GAGAGAGGAG AGTCCCCCTG TGGTCATCGT GGCTCAAACC Ggtcagggtc
 241 tgacctagct ccttggctgg ctttctccag tggcccttta gaccctatg ctttccctct
 301 cagactctca ctttgcccca caacagtctc cttcttcaga ttcccatgcc ttctccctca
 361 gagtctgct ttctcctttg gcctccttt
 ← **Primer R**

Mutant allele

Primer F →
 1 gtgggagggc tataggaag aacaattagg atatggggca ttgagggggc aggcaagagc
 61 catgtcaaca cttctctctg acactagGTC CAGAGTCCAG CCAGACATTG CCCCAGAAT
 121 CCACGGCCAC AGATATCCTG GGGCTAAGGC CTGGAACCTC CTACCAGGTG GCTGTGTCAG
 181 CACTGCGAGG GAGAGAGGAG AGTCCCCCAT CGTGGCTCAA ACCGgtcagg gtctgacctg
 241 gctccttggc tggctttctc cagtggccct ttagaccct GTGTCAGCAC TGCGAGGGAG
 301 AGAGGAGAGT CCCCATCGT GGCTCAAACC Ggtcagggtc tgacctagct ccttggctgg
 361 ctttctccag tggcccttta gaccctatg ctttccctct cagactctca ctttgcccca
 421 caacagtctc cttcttcaga ttcccatgcc ttctccctca gagtctgct ttctcctttg
 481 gcctccttt
 ← **Primer R**

Figure S1. Genomic sequences of the wildtype and mutant alleles. The sequences correspond to the PCR amplicons used for genotyping the *COL7A1*:[c.2028_2034del; c.1993_2050+56dup] variant. The primer sequences are indicated in green. Intronic bases are given in lowercase letters, exonic bases are given in uppercase letters. The duplicated region is highlighted in grey and the 7 bp deletion within the duplicated region is underlined.