

Figure S1. Genotyping of *CGB5* p.Val56Leu, *CGB8* p.Arg8Trp and p.Pro73Arg mutations in Danish RM cases and fertile controls using restriction fragment length polymorphism (RFLP) analysis. (A) RFLP analysis of *CGB5* p.Val56Leu (g.1178G>C, position on genomic sequence relative to mRNA start site; rs72556325). Amplified *CGB5* PCR product (1757 bp) was digested with FastDigest® *AlwNI*. Substitution G>C at position g.1178 introduces an additional restriction site and gives rise to a mutation-specific fragment of 543 bp (indicated by arrow). Lane 1 – DNA size (bp) marker Mass Ruler Low Range (Fermentas UAB, Lithuania); lane 2 – control individual with no g.1178G>C mutation (*g.1178GG* genotype); lane 3 - heterozygous p.Val56Leu mutation carrier (*g.1178GC* genotype). (B) Mutual RFLP analysis of *CGB8* p.Arg8Trp (g.806C>T; rs72556341) and p.Pro73Arg (g.1237C>G; rs72556345) mutations. Amplified *CGB8* nested-PCR product (2544 bp) was subjected to triple-digestion with *NcoI*, *PdiI* and *DraI*. Substitution C>T at position g.806 leads to a mutation-specific fragment of 498 bp in size, whereas substitution C>G at g.1237 generates a 442 bp index fragment (indicated by arrows). Lane 1 – DNA size (bp) marker GeneRuler™ 100 bp Plus DNA Ladder (Fermentas UAB, Lithuania). Lane 2 - heterozygous g.806C>T mutation carrier (genotypes *g.806CT*; *g.1237CC*). Lane 3 - heterozygous g.1237C>G mutation carrier (genotypes *g.806CC*; *g.1237CG*). Lane 4 - control individual with no g.806C>T or g.1237C>G mutations (genotypes *g.806CC*; *g.1237CC*). RFLP products were resolved on 2% TAE agarose gel.

