

**Appendix S1 Materials and methods: Direct diagnostic test of the *LAMA3* c.7825C>T mutation.**

A 5' exonuclease assay was developed to genotype the *LAMA3* c.7825C>T mutation, using 5'- TGA TTG GTG TTG ACA GGA GAA AAG A -3' and 5'- GTC TGA ATC GTC TGT CCA AAG GT - 3' as PCR primers, and 5'- AGT TGG AAC TC[G] AGC ATA G - 3' (wild-type G allele) and 5' - AGT TGG AAC TC[A] AGC ATA G - 3' (mutant A allele) as probes (Taqman, Applied Biosystems, Fosters City, CA). Reactions were carried out on an ABI7900HT instrument (Applied Biosystems, Fosters City, CA) using standard procedures.