Supplementary material (SM)

SM1 – Information about Sanger sequencing of *ACTG2* and *ACTA2*

SM2 – Information about whole-exome sequencing

SM3 – Figure 1. Molecular analysis (DNA chromatograms) with identified pathogenic variants in the *ACTG2* (A, B, C and D) and *ACTA2* (E) - A: patient 3 (c.532C>T - p.Arg178Cys), B: patient 4 (c.770G>A – p.Arg257His), C: patient 5 (c.533G>T – p.Arg178Leu), D: patient 6 (c.584C>T - p.Thr195Ile) and E: patient 7 (c.535C>T – p.Arg179Cys). The variants were not identified in the evaluated parents

SM4: Pathogenicity of c.584C>T variant found in *ACTG2* (patient 6) predicted by different softwares

SM5 - Information about the individuals with molecularly confirmed ACTG2-related disorders