

Description of Additional Supplementary Files

Supplementary Data 1.

Detailed clinical information for *ACTB*-AST patients

Supplementary Data 2.

Rare variants identified in whole exome sequencing

Supplementary Data 3.

Summary of clinical, cellular and structural data for P3, P4 and P5

Supplementary Data 4.

Protein coding genes significantly deregulated in *ACTB*-AST P4 vs. healthy control primary fibroblasts

Supplementary Data 5.

Protein coding genes significantly deregulated in *ACTB*-AST P5 vs. healthy control primary fibroblasts

Supplementary Data 6.

Protein coding genes significantly deregulated in both *ACTB*-AST patient fibroblasts vs. healthy control primary fibroblasts

Supplementary Data 7.

List of ABP assessed in *ACTB*-AST patient fibroblasts

Supplementary Data 8.

Significantly deregulated ABP in *ACTB*-AST fibroblasts