



Fig. S1. Molecular genetic findings. Next generation sequencing (NGS) representation of the novel, nonsense mutation identified in the STS gene (NM_000351.4): the green column shows the G>A transversion (c.1116G>A, p.Trp372*) in exon 8 of the STS gene in our patient (a); DNA sequence control analysis of the region spanning the homozygous c.1116G>A mutation (arrow) in the patient (b, upper panel) compared to the normal control individual (b, lower panel); MLPA study demonstrating the absence of STS deletion (c).