**Figure 2 – figure supplement 1. RNA splicing effect of the *FBXW11* *de novo* splice site variant (c.1468-2A>G).** Panel A shows the agarose gel on cDNA PCR products of patient and control Epstein–Barr virus (EBV)-transformed lymphoblastoid cell lines (EBV-LCLs) treated with or without cycloheximide (CHX). Three distinct bands were identified and are indicated by arrows next to a 100bp ladder (L). Both the wildtype allele of the patient and the control show a smear, possibly indicating the presence of multiple *FBXW11* isoforms. Panel B shows traces of the three bands from the agarose gel that were cut out and sent for Sanger sequencing. As the splice site variant in the patient was expected to lead to skipping of exon 12, the boundaries between exons 11, 12 and 13 were shown. The second band confirms skipping of exon 12 that results in a shorter transcript of the mutated *FBXW11* allele. After CHX treatment, this band increased in size, indicating that the mutated allele undergoes nonsense mediated decay, but incomplete. Furthermore, a smaller transcript is formed in the patient, which is shown to contain part of exon 12, but not exon 13.