\\Vm-ficheros\datos_genetica$\articles_IMO\SHILCA\Supplementary material\Figure S1.tif

**Figure S1.** Study of the expression of the *NMNAT1* cannonical isoform (isoform 1) in the father and the patient, in comparison to a control subject, using primers specifically designed to detect the expression of this isoform (Table S3). The expression of isoform 1 was detected both in the father and the patient, although the later with significant lower levels. Moreover, two other alternative transcripts were also detected: the skipping of exon 4 in the father, and the duplication of exon 4 and partly exon 5 in the patient. The right panel represents the exon composition of the three detected mRNAs, together with the location of the primer pair (arrows).