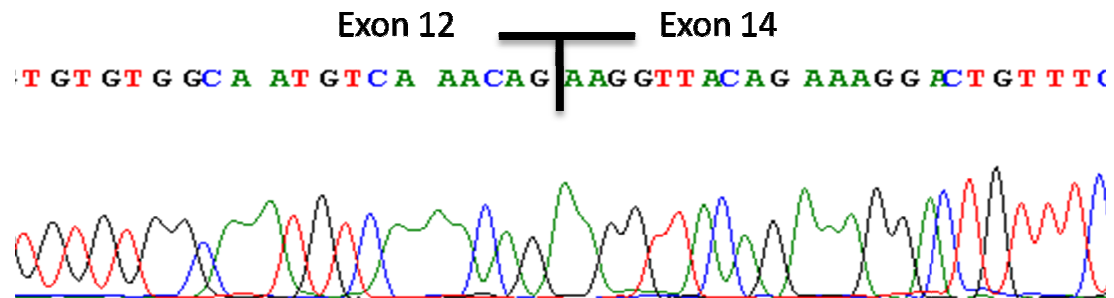


## Supplementary data

**Figure S4** – cDNA studies performed in a XLMTM patient with an AluYa5 insertion.



**Legend:** Partial *MTM1* cDNA sequencing result showing skipping of exon 13 in the patient.

## Material and Methods

Total RNA was extracted from the patient's affected muscle and reverse transcribed with random primers. Amplification was performed using a forward primer located in exon 12 (G1099A) 5' AAAACCCACACCGATGCTGAC 3' and a reverse primer located in exon 14 (G1099B) 5' TCAGTGACCATAAAGAAACAGTCC 3'. PCR products were purified on Microcon YM-100 (Millipore) and sequenced on an ABI 3130 genetic analyzer (Applied Biosystems) with primers used for the PCR amplification using BigDye® Terminator v3.1 Cycle Sequencing Kit (Applied Biosystems).