



Supplementary figure 2. Results of multiplex PCR in the present case (**lanes 1**), which reveal integrity of the *SRY* gene, the 22 sequence-tagged sites (STS) of the AZFa, AZFb, AZFc regions (amplicons size and location of each STS is indicated in the table), including Yq12. The marker ZFX/Y amplifies both in XY and XX individuals, and serves as an internal control amplicon to ensure proper genomic DNA quality. Lanes **MWM**: DNA molecular weight marker, ladder from 100 bp to 1 kb; **NL**: healthy male control; **DEL**: affected male control with a known Yq microdeletion that extends from AZFb to Yq12 region; **(-)**: negative control.