

Figure 1

Delineation of the large heterozygous 4qTer deletion. **(a)** Systematic screening of heterozygous rearrangements at the *F11* locus by QFM-PCR. The QFM-PCR electropherogram of patient XXIV/41 is overlaid with that of a normal control following normalization against the reference diploid *F2* amplicon located on chromosome 11 (Ref). Another control amplicon located in the *F9* gene on chromosome X (control) was used to validate the quantitative analysis by determining the sex based on chromosome X dosage. All peaks corresponding to sequences in the *F11* gene show a two-fold reduction (**horizontal bars**) of their height, indicating a heterozygous deletion of the whole gene. **(b)** The large deletion of 4qTer revealed by arrayCGH. The \log_2 of the Cy3/Cy5 fluorescence intensity ratio is plotted against the chromosome 4 nucleotide position (hg18). **Grey dots** indicate copy number losses ($\log_2 \text{ ratio} \sim -1$), while the black dots indicate no copy number change. More than 7 Mb of sequences are involved in this deletion, and the *F11* gene (outlined in red) is located in the middle.