Molecular characterisation of a der(Y)t(Xp;Yp) with Xp functional disomy and sex reversal

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ABSTRACT. Sex reversal due to duplication of the Xp21 dosage-sensitive sex reversal locus results in XY females with gonadal dysgenesis. Pure Xp disomy (without a concurrent loss of genetic material) can occur by translocation or interstitial duplication. The case reported here is the rare form with a t(Xp;Yp). The combination of conventional clinical cytogenetic techniques, microsatellite analysis and high-density microarrays identified the X-chromosome breakpoint as centromeric of the NR0B1 gene and its control elements. Cytogenetics and array technology complemented each other in characterizing the translocation event and the extent of the dosage-sensitive sex reversal critical region on the derivative Y-chromosome. The implications of this analysis also lie in genetic counseling that highlight the likely de novo nature of a paternal meiotic event.

Key words: Xp functional disomy; Duplication Xp; Sex reversal; Translocation X;Y; Microarray

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