Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Case Report

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

F. Ashton[1], R. O'Connor[1], J.M. Love[1], E. Doherty[1], S. Aftimos[2], A. George[1] and D.R. Love[1,3]

[1]Diagnostic Genetics, Lab Plus, Auckland City Hospital, Auckland, New Zealand
[2]Northern Regional Genetic Service, Auckland City Hospital, Auckland, New Zealand
[3]School of Biological Sciences, University of Auckland, Auckland, New Zealand

Corresponding author: D.R. Love
E-mail: donaldl@adhb.govt.nz

Genet. Mol. Res. 9 (3): 1815-1823 (2010)
Received May 28, 2010
Accepted July 18, 2010
Published September 14, 2010
DOI 10.4238/vol9-3gmr896

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

