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

Case Report

Identification of a *de novo* inv dup(X) (pter→q22) by multicolor banding in a girl with Turner syndrome

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Genet. Mol. Res. 9 (2): 780-784 (2010)
Received January 12, 2010
Accepted February 12, 2010
Published April 27, 2010
DOI 10.4238/vol9-2gmr777

ABSTRACT. We report on a 23-year-old girl with short stature, short and wide neck, low posterior hairline, hypogonadism, underdeveloped breasts, infantile uterus, ovaries not visualized, and primary amenorrhea. Cytogenetic G-banding analysis revealed a mosaic karyotype of 46,X,dup(X)(q22)[35]/45,X[15], confirming the clinical suspicion of Turner syndrome. Molecular cytogenetics using a multicolor banding probe set for the X-chromosome characterized an inverted dup(X). The

karyotype of the patient was therefore interpreted as 46,X,inv dup(X) (pter→q22::q22→pter). This patient had a mosaic Turner syndrome with a cell line comprising partial trisomy Xpter to Xq22 and partial monosomy Xq22 to Xqter.

Key words: Inverted duplication; Partial monosomy; Partial trisomy; Multicolor banding; Mosaicism

