

ISSN 0972-3757

International Journal of

HUMAN GENETICS

Special Volume

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PRINT: ISSN 0972-3757 ONLINE: 2456-6360

Int J Hum Genet, 10(1-3): 77-80 (2010)

DOI: 10.31901/24566330.2010/10.01-3.10

45,X/47,X,i(X)(q10),i(X)(q10)/46,X,i(X)(q10)

Isochromosome Xq in Mosaic Turner syndrome

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KEYWORDS Primary Amenorrhea. Short Stature. Poor Secondary Sexual Characters. Mosaic Isochromosome Xq. Turner Syndrome. Genetic Counseling

ABSTRACT A 17-year old female has been referred for karyotyping and genetic counseling. Proband had primary amenorrhea, short stature and poorly developed secondary sexual characteristics. Ultrasound scanning showed hypoplastic uterus and gonadal dysgenesis. Chromosomal analysis revealed the mosaic status for the isochromosome formation in the long arm of X, i(Xq). Proband had 3 cell lines. Her karyotype: 45,X(4%)/47,X,i(X)(q10), i(X)(q10)(8%)/46,X,i(X)(q10)(88%). Proband has expressed X numerical anomaly for the constitutional X structural anomaly. Proband and family were counseled about education, career, appropriate medical management and hormonal therapy.