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**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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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.