

Interphase fluorescence in situ hybridization in a Turner syndrome variant with IsoXq. A case report.

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BACKGROUND: Performing fluorescence in situ hybridization (FISH) on uncultured amniotic fluid cells has been known to produce rapid diagnoses of major chromosomal aberrations. However, if the aberration involves a structural chromosomal anomaly, the FISH result may be not only uninformative but misleading. **CASE:** FISH with alpha-satellite probes of chromosomes X and Y was performed on the uncultured amniocytes of a prenatal amniotic fluid specimen. Eighty-five percent of the hybridized interphase nuclei displayed two signals when probing with X; no significant hybridization was found when probing with Y. This FISH result was interpreted as normal, disomic for chromosome X. Cytogenetic analysis later, however, disclosed the fetal karyotype to be 46,X,i(Xq). **CONCLUSION:** Interphase FISH with an alpha-satellite probe (or probe made of repeat centromeric sequences) may be useful in the detection of a numerical anomaly of a chromosome but not of a structural anomaly within the chromosome itself.