

Cytogenetic findings in Serbian patients with Turner's syndrome stigmata

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ABSTRACT. Cytogenetic findings are reported for 31 female patients with Turner's syndrome. Chromosome studies were made from lymphocyte cultures. Non-mosaicism 45,X was demonstrated in 15 of these patients, whereas only three were apparently mosaic. Eight patients showed non-mosaic and four patients showed mosaic structural aberrations of the X-chromosome. One non-mosaic case displayed a karyotype containing a small marker chromosome. Conventional cytogenetics was supplemented by fluorescence *in situ* hybridization (FISH) with an X-specific probe to identify the chromosomal origin of the ring and a 1q12-specific DNA probe to identify *de novo* balanced translocation (1;9) in one patient. To our knowledge, this is the first finding of karyotype 45,X,t(1;9)(cen;cen)/46,X,r(X),t(1;9)(cen;cen) in Turner's syndrome. The same X-specific probe was also used to identify a derivative chromosome in one patient.

Key words: Chromosomal abnormalities; Turner's syndrome