

## Ring chromosome instability evaluation in six patients with autosomal rings

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**ABSTRACT.** Ring chromosomes are often associated with abnormal phenotypes due to loss of genomic material and also because of ring instability at mitosis after sister chromatid exchange events. We investigated ring chromosome instability in six patients with ring chromosomes 4, 14, 15, and 18 by examining 48- and 72-h lymphocyte cultures at the first, second and subsequent cell divisions after bromodeoxyuridine incorporation. Although most cells from all patients showed only one monocentric ring chromosome, ring chromosome loss and secondary aberrations were observed both in 48- and 72-h lymphocyte cultures and in

metaphase cells of the different cell generations. We found no clear-cut correlation between ring size and ring instability; we also did not find differences between apparently complete rings and rings with genetic material loss. The cytogenetic findings revealed secondary aberrations in all ring chromosome patients. We concluded that cells with ring chromosome instability can multiply and survive *in vivo*, and that they can influence the patient's phenotype.

**Key words:** Ring chromosome; Chromosome instability; Sister chromatid differentiation