ISSN 0972-3757
INTERNATIONAL JOURNALOF HUMAN GENETICS

 Int J Hum Genet, 4(3): 197-200 (2004)
PRINT: ISSN 0972-3757 ONLINE: 2456-6360

Ring Chromosome 18: Clinical, Cytogenetic and Molecular Genetic Studies on Four Patients

Matild Dobos[1], György Fekete[1], Ruth Raff[2], Regine Schubert[2], Judit Szabó[1], Zita Halász[1], Valeria Lukács[3], Thomas Eggermann[4] and Gesa Schwanitz[2]

1. Semmelweis University, II. Department of Pediatrics, Budapest, Hungary
2. Institute of Human Genetics, University of Bonn, Germany
3. Borsod Country Hospital, Miskolc, Hungary
4. Institute of Human Genetics, Technical University of Aachen, Germany

KEYWORDS Parental origin of ring chromosome 18; microsatellite typing; ring chromosome formation

ABSTRACT Ring chromosomes are a rare chromosomal aberration but have meanwhile been reported for nearly all human chromosomes. We describe four de novo carriers (1 boy and 3 girls) of ring chromosome 18 (r(18)): while three patients had a non-mosaic 46,r(18) karyotype, the fourth was a mosaic: mos 46,XX,r(18)/46,XX,der(18). Phenotypically, the boy showed only minor anomalies, but the female probands presented several clinical features, among them microcephaly, a moderate to severe muscular hypotonia, psychomotoric retardation and short stature. Major malformations were heart defects, cleft lip and palate and atresia of the external auditory canal. In one girl with very short stature, we found a hypothalamic growth hormone deficiency. By investigating the children over 2,5 years it could be demonstrated that the ring chromosomes were passed regularly through mitosis. The parental origin of the ring was determined in three cases indicating a postzygotic mitotic error.

Home Back