Case Report

Smallest critical region for microcephaly in a patient with mosaic ring chromosome 13

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ABSTRACT. A ring chromosome 13 or r(13) exhibits breakage and reunion at breakage points on the long and short arms of chromosome 13, with deletions of the chromosomal segments distal to the breakage points. The r(13) chromosome accounts for approximately 20% of ring chromosomes compatible with life. We describe a female patient with mental retardation, growth retardation, microcephaly, craniofacial dysmorphism, hearing impairment, and prolonged prothrombin time. Chromosomal analysis via GTG banding of peripheral blood lymphocytes revealed a karyotype of 46,XX,r(13)(p13q34)[71]/45,XX,-13[12]/46,XX,dic r(13;13)(p13q34;p13q34)[9]/46,XX,-13,+mar[5]/47,XX,+r(13)(p13q34)>2[2]/46,XX[1] at the age of 6 years and 46,XX,r(13)(p13q34)[82]/45,XX,-13[14]/46,XX,dic r(13;13)(p13q34; p13q34)[2]/46,XX,-13,+mar[2]. Array comparative genomic hybridization analysis of the blood demonstrated a 4.37-Mb deletion on chromosome 13q [arr
cgh 13q34q34(109,743,729-144,110,721)]. A cytogenetic study of peripheral blood revealed a rare chromosomal abnormality associated with different cell lines that included structural and numerical abnormalities of chromosome 13. This case, along with 14 previously reported cases, indicate that the smallest critical region for chromosome 13 microcephaly is 109,743,729-144,110,721.

**Key words:** Array comparative genomic hybridization; Microcephaly; Mosaic ring chromosome; Ring chromosome 13 duplication/deletion; Hearing impairment; Prolonged prothrombin time