Prenatal diagnosis of a partial trisomy 13q (q14→qter): phenotype, cytogenetics and molecular characterization by spectral karyotyping and array comparative genomic hybridization

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Received November 30, 2009
Accepted January 4, 2010
Published March 16, 2010

ABSTRACT. Partial trisomy 13q is an uncommon chromosomal abnormality with variable phenotypic expression. We report prenatal diagnosis of partial trisomy 13q in a fetus with partial agenesis of the cerebellar vermis, partial agenesis of the corpus callosum, hydrops and polyhydramnios. G-banding karyotyping, spectral karyotyping and array comparative genomic hybridization (aCGH) analysis of fetal blood were performed. Cytogenetic analysis of fetal blood displayed 46,XX,add(4)(q28). The parental karyotypes were normal. A girl was delivered at 34 weeks gestation; she died within 2 h. Autopsy confirmed all the prenatal findings and also showed agenesis of the diaphragm. Spectral
Karyotyping identified the additional material’s origin as chromosome 13. aCGH was carried out and showed amplification of distal regions of the long arm of chromosome 13 from region 13q14 to qter. This is the first report of a fetus with molecular characterization of a partial trisomy 13q (q14→qter), present as a de novo unbalanced translocation at chromosome 4q. This case demonstrates the usefulness of molecular characterization of malformed fetuses for prenatal diagnosis and counseling.

**Key words:** Partial trisomy 13q; Prenatal diagnosis; SKY; Array comparative genomic hybridization; Corpus callosum agenesis; Dandy-Walker malformation