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

I.N. Machado[1,2], J.K. Heinrich[1], C. Campanhol[1], R.M. Rodrigues-Peres[1], F.M. Oliveira[3] and R. Barini[2]

[1]Laboratório de Cultivo Celular e Citogenética,
Centro de Atenção Integral à Saúde da Mulher,
Universidade Estadual de Campinas, Campinas, SP, Brasil
[2]Departamento de Tocoginecologia, Programa de Medicina Fetal da Divisão de Obstetrícia, Faculdade de Ciências Médicas,
Universidade Estadual de Campinas, Campinas, SP, Brasil
[3]Departamento de Clínica Médica,
Instituto Nacional de Ciência e Tecnologia em Células-Tronco e Terapia Celular,
Faculdade de Medicina de Ribeirão Preto,
Universidade de São Paulo, Ribeirão Preto, SP, Brasil

Corresponding author: I.N. Machado
E-mail: imachado@fcm.unicamp.br

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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

