Prenatal diagnosis of a large de novo terminal deletion of chromosome 11q

Abstract

Objective: To describe the prenatal phenotype of the 11q deletion syndrome (Jacobsen syndrome) and present the molecular characterization of the deletion in the case presented.

Case: Ultrasound at 18 and 20 weeks of gestation, on a 34-year-old woman who presented for amniocentesis, revealed slow movements, oligohydramnios and dilatation of the cerebral ventricles in the fetus. Maternal and paternal ages were 34 and 38 years, respectively.

Results: Prenatal karyotyping of cultured amniotic fluid cells revealed an 11q terminal deletion, 46,XX,del(11)(q23) (Jacobsen syndrome). Real-time quantitative PCR analysis was used to identify and map the breakpoint physically to a 45-kb region located 14.5 Mb from the 11q telomere. Polymorphic DNA marker analysis showed that DNA sequences on the paternally derived chromosome are deleted. At autopsy, facial dysmorphism without major malformations was recorded. Examination of the internal organs disclosed the following abnormalities: a Meckels' diverticulum of 4-mm length, adhesion between the gall bladder and the transverse colon, and bilaterally bilobed lungs without further situs anomalies.

Conclusion: Our case demonstrates significant phenotypic variability of Jacobsen syndrome at midtrimester pregnancy; the syndrome may be manifested at this stage only by mild to moderate ventriculomegaly of the brain.

Boehm D, Laccone F, Burfeind P, Herold S, Schubert C, Zoll B, Männer J, Pauer HU, Bartels I. Prenatal diagnosis of a large de novo terminal deletion of chromosome 11q. Prenat Diagn. 2006 Mar;26(3):286-90. doi: 10.1002/pd.1408. PMID: 16506277.