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Prenatal diagnosis of mosaic ring chromosome 16 - a rare event with uncertain prognosis

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Abstract:

Ring chromosomes are rare cytogenetic findings (prenatal frequency ~ 0.0075%) often associated with an abnormal phenotype, depending of the chromosomal origin, genetic content and the presence of a mosaic. Supernumerary ring chromosome 16 [r(16)] is rarely observed and mosaicism makes the genotype/phenotype correlation difficult.

We report a de novo mosaic r(16) detected after prenatal diagnosis in a woman referred for advanced maternal age. Multiplex ligation-dependent probe amplification (MLPA) for aneuploidy testing of chromosomes 13, 18, 21 and X was normal. Karyotype was 47,XX,+r[10]/46,XX[15]. Chromosomal microarray analysis (CMA) on DNA obtained from long-term cultured amniocytes did not detect any alterations. MLPA with a pericentromeric probe kit on an uncultured sample showed a chromosome 16 gain, encompassing 16p11.2 and 16q11.2 regions, including *TGFB111*, *AHSP*, *VPS35* and *ORC6* genes, leading to partial characterization of the r(16). Although no phenotype has been correlated with overexpression of these genes, the 16p11.2 region is associated with neurodevelopmental disorders. Nevertheless individuals with microduplication of 16p11.2 and normal development have been described.

The lack of a precise definition of genetic content of the r(16) and its mosaic form leads to uncertain prognosis of clinical outcome.

After genetic counseling the couple opted to continue the pregnancy. At birth no major malformations were observed and a lower level of mosaic r(16) was observed in peripheral blood.

The mosaicism, as well as limitations of CMA in those cases, prevent a refined characterization of these genomic imbalances and pose a challenge in genetic counseling.

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