Prenatal diagnosis of idic(9)

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Tetrasomy of the short arm of chromosome 9 is a rare chromosome imbalance that may result from a supernumerary isochromosome 9 with the most recurrent breakpoints being 9p10, 9q12 and 9q13.
On ultrasound, it usually presents with intrauterine growth restriction (IUGR), abnormal facial profile and ventriculomegaly.
However, few reports establish a correlation between fetal features and the size of isochromosome or the presence of isodicentric 9.

We report the clinical case of a 32-year-old pregnant woman, G2P1, underwent amniocentesis at 13 weeks of gestation with fetal increased nuchal translucency (7mm). The fetus also presented IUGR, cystic higroma, generalized subcutaneous edema, cardiac malformations, facial anomalies and fetal death.
The karyotype was performed by standard *in situ* methods. Fluorescence *in situ* hybridization (FISH) was performed using centromeric probe CEP9.

Conventional cytogenetic and FISH analyses revealed a supernumerary chromosome idic(9)(q12) in all cells examined.

After counseling the couple opted for termination of pregnancy. The post-mortem analysis revealed a single umbilical arteria, IUGR, cystic higroma, facial dysmorphism with cleft lip and palate, hypertelorism and low set ears. These findings are in accordance with other reports.
Nevertheless, the hypertelorism is not commonly described and such an early detection of a cardiac anomaly is uncommon. Additionaly the fetal death occurred early than in the most cases described in literature.
Although breakpoint position effect on the severity on the phenotype is not consensual it has proposed that cases presenting with breakpoints on p10, on q12 or on q13 show a similar phenotype. However, cardiac defects seem more frequent on cases in which the abnormality includes 9q material.
This work aims to contribute to a better karyotype-phenotype correlation in cases with tetrasomy 9p and isodicentric chromosomes idic(9).