17th World Congress in Fetal Medicine

Normal karyotype. . . time to celebrate?

Pafilis I, Trapali C, Tarasidou E
Centre of Fetal Medicine Athens, Athens, Greece

Objective

To present two cases in which even though the karyotypic analysis of the fetuses was normal, further ultrasound assessment and subsequent microarray analysis provided important diagnostic information.

Methods

This is a case report.

Results

A 38-year-old pregnant woman G1 and a 36-year-old pregnant woman G2P1 had amniocentesis at 16 and 18 weeks respectively, due to maternal anxiety, that showed normal karyotypic analysis (46XX) in both fetuses. When we performed the anatomy examination (second trimester scan) on the first fetus, we diagnosed a right aortic arch and subsequently requested microarray analysis on the amniotic fluid sample. The analysis showed Di George syndrome. On the second fetus, we observed a small penis and subsequent microarray analysis showed 46XX/46XY chimera.

Conclusion

It is apparent that in fetal medicine, like every other field of medicine, meticulous clinical examination should dictate further investigations and not vice versa.