

objective :

To describe the role of ultrasound signs in detection of chromosomal aberration even in the third trimester.

Material and methods :

30-year-old patient with no notable pathological history, GIPII, non-consanguineous marriage was referred to our outpatient clinic at 28 weeks of amenorrhea, for a suspicion of a mega big cistern.

Results:

Ultrasound in our department at 28 weeks of gestation showed a significantly enlarged cisterna magna, absence of the vermis between the two cerebellar hemispheres, and a dilated fourth ventricle appearing fused with the posterior cisterna. A fetal echocardiogram at 31 weeks of gestation revealed cardiomegaly with left heart hypoplasia and valve alignment abnormalities. Fetal MRI indicated an elevated tentorium cerebelli, an enlarged posterior fossa due to cystic dilation of the large cisterna magna communicating with the fourth ventricle, and evident vermian hypoplasia.

A chromosomal aberration was suspected in association with syndromic conditions (Dandy-Walker syndrome and complex congenital heart disease). A therapeutic termination of pregnancy was performed at the parents' request. Histopathological examination revealed the following findings: male fetus at 31-32 weeks of gestation, with craniofacial dysmorphia, Dandy-Walker syndrome, cardiac abnormalities (ventricular septal defect, valve alignment, left heart hypoplasia), and urogenital anomaly (pelvic dilation, cryptorchidism). These findings suggested trisomy 18. The postmortem fetal karyotype confirmed the diagnosis.

Conclusion:

The detailed analysis of fetal morphology and the classification of anomalies into syndromes can lead directly to the diagnosis of a well-defined chromosomal aberration, facilitating the option of early therapeutic termination of pregnancy.

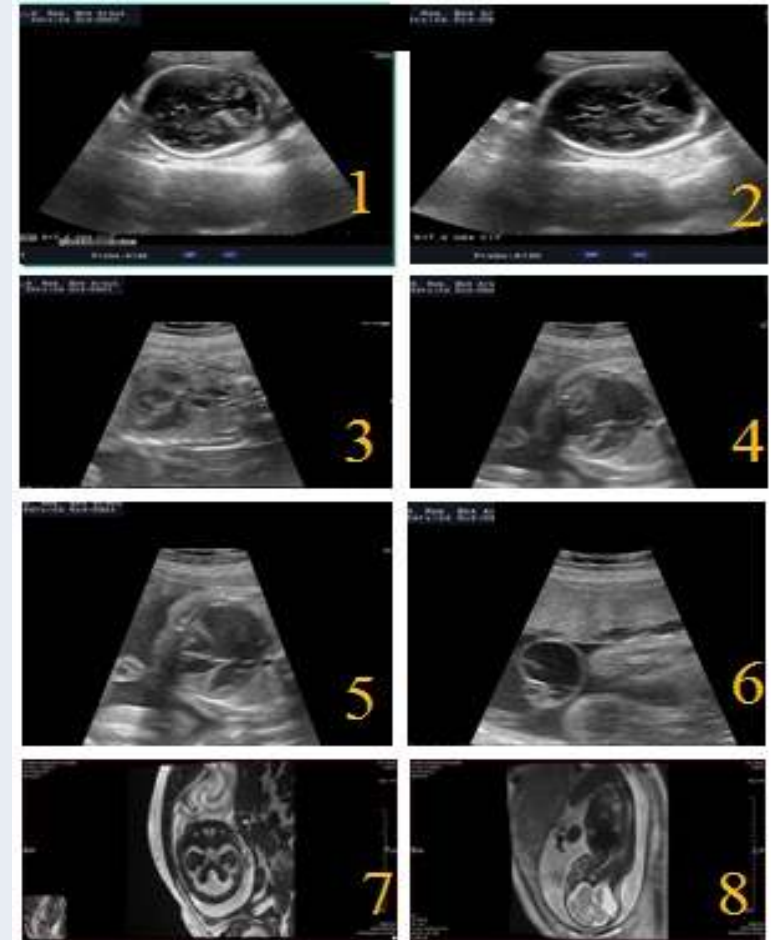


Figure 1-2: Brain morphology

Figure 3-4-5: Cardiac morphology

Figure 6: Ultrasound appearance of the cord cyst

Figure 7-:8 Fetal MRI