Incidence of chromosomal abnormalities in fetuses with ventriculomegaly

BCNatal - Fetal Medicina Research Center (Hospital Clínic and Hospital Sant Joan de Déu), Barcelona, Spain

Objective
To evaluate the incidence of chromosomal abnormalities according to the characteristics of fetal ventriculomegaly (VM) by chromosomal microarray analysis (CMA).

Methods
Retrospective study including all cases of VM who underwent invasive prenatal diagnosis from May 2014 to February 2022 at our tertiary centre. VM was classified as mild (10-11.9 mm), moderate (12-14.9 mm), or severe (≥15 mm). Data recorded included maternal characteristics, ventricular width, laterality, associated anomalies including intra- or extra-cranial findings, CMA results and pregnancy outcomes. We excluded cases with confirmed CMV infection.

Results
Of 167 patients included, there were 100 (59.9%) cases of mild VM, 28 (16.8%) of moderate VM, and 36 (23.3%) fetuses with severe VM. CMA identified 14 cases (8.4%) of chromosomal abnormalities, of those 50% would be undetectable by standard karyotype. The incidence of abnormal CMA in mild, moderate and severe VM was 7%, 14.3%, and 7.7%, respectively. Non-isolated VM was present in 46.1% of cases, showing increased prevalence of chromosomal anomalies when compared with isolated VM (14.5% vs. 3.4%, p= 0.011), especially those with central nervous system anomalies. Isolated mild ventriculomegaly was present in 66 patients and abnormal CMA was identified in 4.5% of cases. Unilateral VM was diagnosed in 72 of cases (43.1%), with no significant differences in chromosomal abnormality rate when compared with bilateral (6.9% vs 9.5%, p=0.559, respectively). Pregnancy was terminated in 42.9% of patients with chromosomal abnormalities. Among women with normal CMA, 18.3% underwent termination of pregnancy due to severe anomalies.

Conclusion
Chromosomal abnormalities were present in 8.4% of VM in our series. Despite abnormal CMA was more frequent in moderate and severe VM, also mild isolated cases present a significant proportion of abnormalities (4.5%). Our results confirm previous data and provide valuable information for clinical management of VM.