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Prenatal series of 462 cases of hydrops fetalis diagnosed between 11-14 weeks in a tertiary referral unit in South India

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Objective

To analyse a series of 462 cases of prenatally diagnosed fetal hydrops between 11-14 weeks of gestation with respect to etiology and associated structural anomalies.

Methods

This was a retrospective study done at Mediscan, Prenatal Diagnosis & Fetal Therapy centre a tertiary referral unit in South India. A search query of the “sonocare” database was done in the first trimester using the search expression “hydrops“ between the 2010 and 2017. A total of 462 pregnancies were taken into the study. Karyotyping and fetal autopsy was offered to all pregnancies diagnosed with hydrops. The data was analysed to evaluate the etiology on the basis of karyotyping, pathological examination, ultrasound findings and further genetic analysis.

Results

Of the 462 cases, 128 (28%) had structural abnormalities which were detected on ultrasound scan which could explain the presence of hydrops - 88 (69%) cardiac defects, 12 (9. 3%) skeletal dysplasias, 26 (20%) musculoskeletal abnormalities, 2 (1. 5%) other syndromes. In 111 cases (24%) patients opted for karyotyping. In 52 (47%) cases karyotype was abnormal and in 59 (53%) was normal. The abnormal results included trisomy 21 - 17 (33%), trisomy 13 - 5 (10%), trisomy 18 - 14 (27%), Turners syndrome - 12 (23%), trisomy 9 - 1(1. 9%), trisomy 20 – 1 (1. 9%), triploidy 1-(1. 9%) and 46+c chromosome 1 (1. 9%). In the remaining group of patients who declined karyotyping, ultrasound scan was able to identify a structural abnormalities explaining the presence of hydrops in 124 (35%) of the cases (46 (37%) cardiac, 10 (8%) skeletal, 2 (1. 6%) syndromic), of which 28 (87%) were also confirmed during the autopsy study. In a subgroup of patients with no abnormality explaining the presence of hydrops diagnosed on an ultrasound scan, an autopsy allowed to determine the cause in 28 (12. 3%) cases.

Conclusion

Nearly one half of the cases undergoing karyotype had a chromosomal abnormality of which 92% had the four common problems (trisomy 13, 18, 21 and Turners Syndrome). This emphasises the importance of a karyotyping as a baseline investigation. A detailed ultrasound evaluation focusing on fetal activity, cardiac and skeletal system assessment should form a part of a checklist when fetal hydrops is detected in the first trimester, as this would increase the diagnostic yield. Perinatal pathology is an invaluable tool, especially in diagnosis considering that the prenatal detection rates of structural malformations in the first trimester is generally low in the population. A systematic approach involving a detailed ultrasound scan, karyotype and an autopsy study as 3 levels of investigations alone, would provide an answer in 72% of cases.