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Pattern of aneuploidies in fetuses with structural defects detected at the 11-13 weeks' scan

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Objective

To determine the prevalence and pattern of aneuploidies in fetuses with structural defects detected at the 11+0 – 13+6 weeks' scan.

Methods

This is a retrospective analysis of singleton pregnancies found to have structural defects at the 11+0 – 13+6 weeks' scan. All the scans were performed according to protocol by the Fetal Medicine Foundation (FMF) certified operators for the study of fetal anatomy and Nuchal Translucency.

Results

356 (2. 9 %) fetuses with structural defects were detected at the 11+0 – 13+6 weeks' scan in 12, 085 singleton pregnancies, during the study period from July 2004 to December 2017. These were divided into two groups, based on the number of systems affected, as "isolated", when only one system was affected and as "multiple" when two or more systems were affected. Isolated defects were present in 278 (78. 1%) fetuses and multiple defects in 78 (21. 9%) fetuses. In the entire study group, 87/ 356 (24. 4%) fetuses had karyotyping. In the isolated defects' group, 69/278 (24. 8%) pregnancies had karyotyping. The prevalence of chromosomal anomalies was 28 /69 (40. 5 %) of which there were 9 (32. 1%), 8 (28. 5%), 4 (14. 2%), 2 (7. 1%) and 5 (17. 8%) fetuses with Trisomy 18, 21, 13, Turner Syndrome and other chromosomal anomalies respectively. In the multiple defects' group, 18/78 (23%) pregnancies had karyotyping. The prevalence of chromosomal anomalies was 11 /18 (61. 1 %) of which there were 5 (45. 4%), 4 (46. 3%), 1 (9%), 1 (9%) and 0 fetuses with Trisomy 13, 18, Turner Syndrome, other chromosomal anomalies and Trisomy 21 respectively. 116/278 in the isolated defects' group had at least one marker of which 35 had karyotyping. 17/35(48. 5%) had an abnormal karyotype. 162/278 (58. 2%) did not show any marker of which 34 had karyotyping and 12/34 (35. 2%) had an abnormal karyotype. 44/78 in multiple defect group had at least one marker of which 15 had karyotyping. 11/15 (73. 3%) had abnormal karyotype. 34/44 (77. 2%) did not show any marker of which 3 had karyotyping and none had an abnormal karyotype.

Conclusion

Our study shows that although the rate of karyotyping in fetuses with structural defects is only 1: 4, the prevalence of aneuploidies is significantly high both, in multiple and in isolated fetal defects. Trisomy 13 and 18 are most frequently associated with multiple and isolated fetal defects respectively. Even in the absence of markers, the prevalence of these aneuploidies is significantly high even in the isolated group. Hence, karyotyping is recommended when fetal defects are detected, irrespective of the presence of markers as this will help to assess for the recurrence of such defects.