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Importance of markers in fetuses with chromosomal anomalies and the pattern of their distribution

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Objective

To determine the pattern of distribution of first trimester markers (increased nuchal translucency, absent nasal bone and tricuspid regurgitation) in aneuploid fetuses detected at the 11+0 – 13+6 weeks' scan and assess the importance of multiple markers in the same fetus.

Methods

125 (1 %) fetuses with aneuploidies 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. All had completed outcomes wrt chromosomal makeup. All the scans were performed according to the unit protocol by FMF certified operators for the study of fetal anatomy, Nuchal Translucency, Nasal bone and Tricuspid Doppler. 3 markers viz., increased nuchal translucency (NT), absent nasal bone (ANB) and tricuspid regurgitation (TR) were studied in all the fetuses according to the guidelines as per the FMF.

Results

52/ 125 (41. 6%) fetuses were detected with Trisomy 21, of which 28 (53. 8%) had increased NT, 15 (28. 8%) had ANB and 8 (15. 3%) had TR. Out of 52 fetuses with T21, 22 (42. 3%), 10 (19. 2%), 2 (3. 8%) and 18 (34. 6%) had 1, 2, 3 and no markers respectively. 24/ 125 (19. 2%) fetuses were detected with Trisomy 18, of which 13 (54. 2%) had increased NT, 12 (50%) had ANB and 3(12. 5%) had TR. Out of 24 fetuses with T18, 9(37. 5%), 7(29. 2%), 1(4. 2%) and 7(29. 2%) had 1, 2, 3 and no markers respectively. 9/ 125 (7. 2%) fetuses were detected with Trisomy 13, of which 6 (66. 7%) had increased NT, 4 (44. 4%) had ANB and 2 (22. 2%) had TR. Out of 9 fetuses with T13, 5(55. 6%), 1(11. 1%), 1(11. 1%) and 2(22. 2%) had 1, 2, 3 and no markers respectively. 17/ 125 (13. 6%) fetuses were detected with sex chromosomal anomalies, of which 12 (70. 6%) had increased NT, 1 (5. 9%) had ANB and 1 (5. 9%) had TR. Out of 17 fetuses with sex chromosomal anomalies, 9(52. 9%), 1 (5. 9%), 0 and 7 (41. 2%) had 1, 2, 3 and no markers respectively. 23/ 125 (18. 4%) fetuses were detected with other chromosomal anomalies, of which 5 (21. 7%) had increased NT, 5 (21. 7%) had ANB and 3 (13%) had TR. Out of 23 fetuses with other chromosomal anomalies, 7 (30. 4%), 3 (13%), 0 and 13 (56. 5%) had 1, 2, 3 and no markers respectively. 11960 (99%) 0f 12085 fetuses had normal karyotype, of which 410 (3. 4%) had increased NT, 286 (2. 4%) had ANB and 71 (0. 6%) had TR. Out of 11960, euploid fetuses, 544 (4. 5%), 510 (4. 3%), 9 (0. 1%) and 11356 (94. 9%) had 1, 2, 3 and no markers respectively. Of all 85 Trisomies, 47 (55. 3%), 31 (36. 5%) and 13 (15. 3%) had increased NT, ANB and TR respectively. 36 (42. 4%), 18 (21. 2%), 4 (4. 7%) and 27 (31. 8%) had 1, 2, 3 and no markers respectively.

Conclusion

Our study shows that Trisomy 21 is the commonest aneuploidy followed by Trisomy 18, other chromosomal anomalies, sex chromosomal anomalies and Trisomy 13. Increased NT is seen most commonly in all chromosomal anomalies, followed by absent NB and Tricuspid regurgitation. However, no markers are seen in about 31. 8% Trisomies and atleast 1 marker in 4. 5% euploid fetuses. This emphasizes the importance of additional screening methodologies ie assessment of fetal structure and serum biochemistry to assessment of markers at the 11+0 – 13+6 weeks' scan to improve detection rate and reduce the false positives and negatives.