Outcome of fetuses with increased nuchal translucency in the first trimester

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Objective

To study the outcome of fetuses with increased nuchal translucency at the 11-13+6 scan.

Methods

Retrospective study between August 2005 to May 2012 included 574 fetuses with a measurement of nuchal translucency above the 95th percentile and a CRL measurement between 45 and 84 mm. We excluded cystic hygromas. When the karyotype was normal, an early anomaly scan at 16 weeks and a detailed anomaly with fetal echocardiography scans were performed at 19–20 weeks followed by review ultrasound examination at 30-32 weeks. Clinical examination of the neonates was performed by a pediatrician.

Results

In the 684 pregnancies with increased NT, 78 (11. 4%) had associated structural anomalies. Of these, 15 (19. 2%) had an abnormal karyotype. In the increased NT group with no structural anomalies, 47/ 541 (after excluding lost to follow up and terminations and IUDs without karyotyping, 8. 7%) had karyotypic anomalies. 24/489 (4. 9%) were found to have structural anomalies in the second trimester after excluding IUDs in the early second trimester. Of the 460 fetuses with normal karyotype and normal structure in the second trimester, only 1 fetus had a structural anomaly detected in the third trimester Congenital diaphragmatic hernia) and 1 fetus died in utero. In the remaining 459 fetuses, which were all live births, there were 2 neonatal deaths (1 mother had gestational diabetes and the other cause was not known), 1 baby had VSD and ASD, 1 baby had VSD with ambiguous genitalia and one baby had Down syndrome. This was a “false negative” report on antenatal testing which was done by CVS in the first trimester. There were 454 live births, which constitutes about 91. 9% of the entire cohort of fetuses with increased NT. About (22 fetuses), 5% of the fetuses with nuchal translucency above the 95th centile – 3. 4mm has chromosomal abnormalities. About 82% of them were live births of which 4 of them were abnormal postnatally. Major number of chromosomal abnormalities (9 fetuses, 50%) were seen in the group 6 – 7. 9mm. 4 live births were seen in >8mm nuchal translucency. These babies were normal at birth. However these babies would require a long term neurological follow up.

Conclusion

Increased fetal nuchal translucency is associated with chromosomal abnormalities, structural defects and genetic syndromes. When an increased NT is associated with structural anomalies, the risk of karyotypic anomaly is about 5 times higher. When increased NT is detected, the couple must be counseled depending upon the NT group that they fall under to “individualize” the risks. Parents must be encouraged to investigate the pregnancy as this has implications in the future. Outcome of fetuses with increased NT can be optimized by adhering to strict protocols of investigations i. e, karyotyping, detailed scans and increased awareness in the majority of the cases. Fetal karyotype, detailed scans, fetal echocardiography, as well as genetic testing and infection screening can be completed by 20 weeks of gestation. This would distinguish between the pregnancies destined to result in adverse outcome and those leading to the delivery of infants without major defects.