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First Trimester Screening on Aneuploidies

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A finding of increased nuchal translucency (NT) thickness above the 95th percentile increases the maternal age risk for chromosomal abnormalities and trisomy 21. The evaluation of the NT can be combined with the biochemical test for the first trimester (free- beta hCG and PAPP-A), in the combined test, which appears to be the most accurate test for predicting the risk for trisomy 21. The combined test in turn can be integrated with a second level ultrasound test (ductus venosus, nasal bone, tricuspid regurgitation, FMF angle), according to a program which includes the provision of subsequent tests only to cases that are positive to the first test (contingent program), or to a program that involves an universal provision. Integration with subsequent tests provides a more accurate estimate of individual risk, reducing false positives, and is useful for the choice of whether or not to execute an invasive prenatal diagnosis. In the case of increased risk for combined test, if the woman's decision is for invasive prenatal diagnosis, the choice of chorionic villus sampling technique seems most appropriate for the early result of the analysis of karyotype. Other fetal problems than abnormal karyotype (poor outcome of pregnancy) are expected at around 20% in cases of increased NT thickness, with a range that can vary from 8 %, when the NT and between the 95th and 99th percentile and 80 NT% for very large (> 6.5 mm). The greatest risk, such as frequency, is that for congenital heart disease, with rates of risk that are, according to recent meta-analysis, 3% for NT between 3.5 mm and 4.4 mm, 7% for NT between 4.5 mm and 5.4 mm, 20% for NT between 5.5 and 6.4 mm and 30% for NT of 6.5 mm or more. Therefore in cases with increased NT thickness, is recommended fetal echocardiography, which can be performed in experienced centers with an initial examination around 14 weeks, to get an early diagnosis or early reassurance for patients already at that age, but which must then be confirmed by echocardiography performed after 20 weeks. More than 60 rare diseases and syndromes have been reported that occur in fetuses with increased NT, some of which can only be suspected in the prenatal period or the presence of a family history or the presence of subtle and minor sonographic signs. Using three-dimensional ultrasound, can help to highlight the often subtle morphological differences in the case of syndromes. A more detailed diagnosis through molecular genetic studies on material taken at the chorionic villus sampling, and ultrasound examination for specific diseases may needed in case with enlarged NT and normal karyotype. An association between increased NT thickness and infections of the TORCH group has not been proved. In the case of normal karyotype and ultrasound performed at 20 weeks, and no particular sonographic obvious sign and normal anatomy, the neonatal and children outcome of fetuses that showed in the first trimester increased NT, is good in about 96 % of cases, coming very close to that expected in the general population. Studies in children observed until school age, who had a NT thickness showed no increased incidence of neuropsychiatric development issues significantly more frequent than normally expected. The aim of prenatal screening should be to provide information to pregnant women closer to reality on its individual condition of the risk of chromosomal anomaly. The information must be fully understood by the woman and should be clear all the negative and positive sides of different options. It has been shown that patients understand the test results of prenatal screening for trisomy 21, especially when expressed in the form of numerical risk, and that, in most cases, are focusing their choices based on this result, which is current and individual, and no longer act as generalized estimates only based on maternal age (greater or less than 35 years).

Prenatal screening then becomes a valuable tool in the hands of the woman to guide individual choices in the field of prenatal diagnosis. The task of the physician should be to suggest the tests more scientifically valid and accurate as possible.

Key points: Screening for chromosomal abnormalities, Nuchal translucency, Combined screening, Ultrasound markers for aneuploidies.