FETAL NUCHAL TRANSLUCENCY AND NASAL BONE SCREENING IN MULTIPLE PREGNANCIES

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Multiple pregnancies, mostly if obtained by assisted reproduction techniques, are at higher risk for chromosomal defects, because of the advanced maternal age, but at same time, as these pregnancies are precious to the parents, and there is no resolute acceptance of the need of invasive prenatal diagnosis procedures, which are more difficult to perform and more hazardous for fetuses than in singleton pregnancies.

Therefore non- invasive screening test for chromosomal abnormalities in multiple pregnancies are more acceptable.

Maternal serum biochemistry for trisomy 21 in multiple pregnancies has been proposed but it may be difficult to interpretate because of the interference between the serum marker concentrations and assisted reproductive drugs, and because of the overlap of biochemical markers measures from each fetus.

Multizygotic pregnancies originate from the fertilization of two or more oocytes by two or more spermatozoa, and present multichorionic placentation, while monozygotic pregnancies present monochorionic placentation in about 2/3 of cases.

Prenatal diagnosis of chorionicity can be made by ultrasound in the first trimester, as at 5 weeks gestation the number of sacs indicates the number of placentas (monochorionic, dichorionic, trichorionic etc.). As the gestation proceeds, the septum between the sacs become thinner, and only a triangular structure, the "lambda sign" remains at 10 to 14 weeks, in multichorionic pregnancies, while in monochorionic pregnancy there is no chorionic tissue between the two-amniotic sacs, the "T sign".

Monochorionic pregnancies are at higher risk than dichorionic pregnancies of prenatal and perinatal problems, as they are monozygotic, and because of the occurrence of specific problems due to transplacental circulation imbalance (twin-to-twin transfusion syndrome).

For multiple pregnancies derived from multizygotic conception, discordant karyotypes are expected. Prenatal sonographic estimation of zygosity is fundamental in the process of risk calculation. In a monochorionic pregnancy monozygosity can be supposed, and an equal risk for trisomy 21 for both fetuses, similar to the maternal age- related risk, can be calculated. The risk for the pregnancy of having one fetus with trisomy 21 is calculated by the sum of the risk for each fetus.

Ultrasound screening for trisomy 21 by first trimester nuchal translucency measurement is valid in twin and multiples. The test gives an individual risk for each fetus, and its accuracy is comparable to singleton pregnancies. NT measurement can be used in high order multiple pregnancies, and in those cases obtained by assisted reproductive technologies, because no evident difference has been found in the rate of enlarged nuchal translucency than in spontaneous multichorionic pregnancies.

In multiple pregnancies, the absence of nasal bone visualization, which is another accurate soft marker for trisomy 21 in the first trimester, can be used with the same efficiency as in singleton pregnancies. About 70% of trisomies 21 and an important percentage of other chromosomal abnormalities as trisomy 18 and 13, 45, X0, can manifest the sign of the absent nasal bone at ultrasound examination performed at 11- 14 weeks.

A higher rate of enlarged nuchal translucency has been described in monochorionic pregnancies. In monochorionic diamniotic pregnancy, an enlarged nuchal translucency could be caused by placental circulation problems, rather than chromosomal abnormalities. This early imbalance of fluids between the fetuses can anticipate in the first trimester the inter-twin-to-twin transfusion, that manifest the most important diagnostic sign only after 16 weeks.

In multifetal pregnancies, when the number of fetuses is high, embryoreduction is usually performed in the first trimester. The choice as to which fetus to reduce is based fundamentally on technical considerations. It was proposed to offer invasive prenatal diagnosis for karyotype by chorionic villus sampling before performing embryoreduction. Because of the difficulties and costs of such invasive prenatal diagnosis in multiples, this approach has not be universally approved and it has been proposed to offer the nuchal translucency screening before embryo reduction. In fact the finding of an enlarged nuchal translucency, allowing the possibility to calculate the individual risk for trisomy in each fetus, can identify cases at higher risk for chromosomal abnormalities and for structural malformation in multiple pregnancies.

The presence of an enlarged NT is associated with an increased rate of chromosomal abnormalities, but moreover the risk for structural malformations and mostly cardiac defects in fetuses with normal karyotype is increased.

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