CVS IN MULTIPLE PREGNANCIES

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The frequency of multiple pregnancies is about 2% of all gestation. While monozygotic pregnancies are relatively constant in number, multizygotic multiples reveal an increased rate in recent years, due to the widespread use of infertility treatments.

The aged maternal age at pregnancy, some genetic indications related to infertility problems, and the increased individual risk of aneuploidies in women carrying multiples, lead to an increased demand for invasive prenatal diagnosis in these pregnancies. Ultrasound screening by first trimester nuchal translucency, can indicate in multiple pregnancies, cases at higher risk for aneuploidies.

The development of ultrasound equipments allows less traumatic access to obtain fetal and placental tissues, reduction of procedure- related risks, and earlier sampling. At the same time, great developments of laboratory techniques in prenatal diagnosis of mendelian and chromosomal diseases has given greater opportunities for diagnostic accuracy.

In multiple pregnancies, before genetic counselling, a very accurate ultrasound examination to establish the chorionicity, is mandatory. The lambda-sign at 10- 14 weeks indicates dichorionic placentation, while the T- sign is suggestive of monochorionic diamniotic pregnancy. Ultrasound examination of chorionicity is highly indicative at first trimester ultrasound, but it become less accurate at second trimester scan.

During the genetic counselling, the assessment of the genetic risk, the possibility to perform a further or different sampling (first trimester chorionic villi sampling and second trimester amniocentesis), the likelihood of sampling all the sacs, the accuracy of genetic analysis (sampling contamination, false positives, false negatives, misdiagnosis), the eventuality of one abnormal fetus and therapeutic options (selective fetocide), and the risk of early and late pregnancy loss related to the procedure, are the issues to be discussed.

Chorionic villi sampling (CVS) is the technique of choice for diagnosis of beta-thalassemia and other monogenic diseases at first and second trimester, while for karyotype analysis, CVS and amniocentesis are the procedures most frequently performed.

First trimester CVS can be performed either transcervically (by catheter or rigid biopsy forceps) or transabdominally (by double coaxial needle or by single needle) and in a few cases by the two techniques combined.

The transabdominal free- hand CVS technique is performed using a 20 gauge spinal needle connected to a syringe for aspiration under continue ultrasonic monitoring.

Second trimester CVS is performed only by transabdominal approach, using 19 gauge from13 to 15 weeks, and 18 gauge spinal needle after 15 weeks.

A very accurate ultrasound examination should be carried out before invasive procedure to evaluate vitality, gestational age, number of fetuses, chorionicity and presence of fetal malformations.

Multiple sagittal and trasverse scanning plane should be carried out in order to obtain a three dimensional map of the uterine content and relationship with the maternal surface. The placental position and umbilical cord insertions must be identified.

In multichorionic pregnancies, all placentas should be sampled by separated needles.

In monochorionic placentas, a single sample could be taken, but also two samples are indicated (samples obtained from a point in the placenta very close to the cord insertion).
Fetal loss following CVS in multiples is the same as reported for second trimester amniocentesis in multiples (2- 3%). Sampling success is very high in expert hands and large series (>98%).

In the case of like- sex results or concordant genetic results, chromosome polymorphism and DNA analysis or highly variable polymorphic sites studies should be more informative.

The risk of entire pregnancy loss following selective fetocide in cases of one or more fetuses resulting abnormal is higher if termination is performed in the second trimester than in the first trimester.

First trimester CVS is the technique of choice in multiple pregnancies at high genetic risk, since the earlier diagnosis offers an advantage in cases of selective termination.

Prenatal diagnosis in multiple pregnancies is safe and accurate, but obstetric risks and diagnostic errors are likely to be increased and great skill and experience are requested.

It is indicated to perform multiple pregnancy sampling and analysis for prenatal diagnosis in centers of large expertise.

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