Prenatal diagnostics of chromosomal aberrations, possibilities of prevention

Prenatal diagnostics of chromosomal aberrations is a broad group of different **examination methods** used for detection of chromosomal abnormalities (including both numerical and structural) prior to birth.

Non-invasive diagnostics

Noninvasive methods are usually performed as a part of the **screening programs**. The main advantage is that that those methods are quite easy to perform and are not dangerous to the fetus, on the other hands they are usually not able to 100% confirm the presence/absence of the chromosomal abnormality.

The main example is the **biochemical screening** of chromosomal abnormalities (in all its forms) and **ultrasound** screening/diagnostics. The new noninvasive diagnostics methods based on the analysis of free-cell fetal DNA circulating in maternal blood (commonly called as **NIPT** - noninvasive prenatal testing) are also usually considered to be screening methods, since they are not so precise as the genetic analysis of the sample obtained by invasive diagnostic procedures.

Invasive diagnostics

Invasive prenatal diagnostics methods are based on the **sampling of cells** which can be used for karyotyping (or another laboratory testing) of the fetus. The sample has to be obtained the **invasive way** - which brings small, but visible **risk of complications**, while the miscarriage is the worst of them (general risk of miscarriage following the invasive prenatal diagnostics is 0,5-1,0%). The main benefit is the level of the information you can obtain (diagnostic value).

- **Chorionic villus sampling** (**CVS**): performed from the 10th to the 14th week of pregnancy.
- **Amniocentesis** (amniotic fluid sampling, AMC): performed from the 16th week of pregnancy.
- **Cordocentesis** (umbilical cord blood sampling, CC): performed after 20th week of pregnancy.

Remember, the method itself is only used for obtaining cells sample. Those cells can be later used four routine cytogenetic examination (karyotyping), or any other type of laboratory genetic analysis (microarray, single gene testing etc.).

Prevention

Prevention of chromosomal aberrations is very difficult because they usually occur as "new and random". Basically there are two issues that can be mentioned:

1. Women should consider the pregnancy prior the "advanced age" (35th years of over) because of the increasing risk of chromosomal aneuploidies.
2. Couples with poor reproductive history (sterility, infertility) should be offered karyotyping in order to detect any possible chromosomal rearrangements, that can be potentially risky considering the numerical/structural chromosomal abnormalities in the fetus.