

# Prenatal diagnostics of inborn errors of development, possibilities of prevention

Inborn errors of development (or congenital anomalies/malformation or birth defects) are specific structural defects - results of abnormal prenatal development. These defects can be nowadays diagnosed prenatally, using different diagnostic procedures. Ultrasound examination should be offered to all pregnancies as a type of screening examination, while other different (more detailed) methods can be offered to mothers with positive family history etc.

## Diagnostic possibilities

- **Ultrasound:** Ultrasound is possibly the most important method for the diagnostics of structural defects. The diagnostic ultrasound examination in the first trimester (round 13th GW) can identify usually only the most severe defects (like severe neural tube defects), while the detailed ultrasound examination in the second trimester (between 18th and 22th week of pregnancy) is most important for the examination of all organs (CNS, heart, kidneys etc.).
- **Fetal Echocardiography:** Detailed ultrasound examination of fetal heart should be offered when the routine ultrasound examination shows suspicious/pathological result.
- **Fetal MRI:** Fetal MRI is sometimes used in order to confirm the previous diagnostic of several defects of "soft tissues", like defects of heart, lungs or CNS. MRI examination (unlike CT or basic X-ray) is considered completely safe for the developing fetus.
- **Biochemical screening:** Biochemical screening itself is a method that is useful for the calculation of risk for selected chromosomal abnormalities (e.g. Down syndrome). Anyway, the AFP (alpha-fetoprotein) also serves as a marker of several fetal defects which are not covered by the fetal skin (like neural tube defects or abdominal wall defects). This suspicion shall be confirmed by detailed ultrasound examination.
- **Cytogenetic examination:** Invasive prenatal diagnostics is not crucial for the diagnosis of structural defect itself (the ultrasound examination is the one that actually can confirm the presence of particular structural defect), but the karyotype analysis is extremely useful, since it can confirm/exclude that the particular structural defect is actually isolated anomaly (better prognosis) or actually a part of complex phenotype in a fetus with chromosomal aberration (like Down or Edwards syndrome). The chromosomal microarray examination can be sometimes indicated in cases of severe fetal anomalies with normal karyotype.

## Prevention

Prevention of congenital anomalies is possible. So called **primary prevention** of congenital anomalies is actually a group of basic recommendation that shall lower the general risk of congenital anomaly. The crucial part of the recommendations is the **planned pregnancy**. The studies show, that the unwanted/unexpected pregnancies are more prone to the effects of the environmental teratogens (drugs, alcohol medication) since the mothers usually learn about their pregnancy already during the most crucial developmental part of the pregnancy.

### Recommendations in general

- No alcohol, no drugs;
- no harmful medications (pregnant women shall consult the specialist before taking any potentially harmful medication while pregnant);
- safe environment (applies especially for women working in industry/agriculture/power plants etc);
- folic acid intake (usually a daily dose of 0.4 mg is recommended in order the lower the risk of neural tube defects);

So called **secondary prevention** is a general term for the active population-based examination of pregnancies (ultrasound screening) where we are not able to prevent the formation of the defect itself, but we can identify the defects already during the pregnancy - so the mother can be informed about the defect itself and its possible prognosis for the health of the baby. In some countries all over the world (including the Czech Republic) it is legal to apply for termination of pregnancy if your fetus has a confirmed severe anomaly, which will interfere with normal postnatal life.

## Links

### External links

- <http://www.eurocat-network.eu/content/EUROCATEUROPLAN-Primary-Preventions-Reccomendations.pdf>