

Pre-conception prevention of heritable and inborn diseases

So called **primary prevention** of congenital anomalies and inborn diseases is actually a group of basic recommendations 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 to lower the risk of neural tube defects);

As for heritable - monogenic diseases, the prevention itself can be challenging. The carriers (heterozygotes) are usually asymptomatic, so the clinical examination can not identify those carriers. In some areas with high frequency of specific **recessive disease** - the population based **screening for heterozygotes** (who plan the family) can be considered. The genetic testing for population-specific mutations can be especially considered for couples with common ancestors (**inbreeding**) and for **gamete donors**. Couples with high risk of heritable diseases can consider preimplantation genetic diagnostics (**PGD**) as an option.