Postnatal prevention and therapy of heritable and inborn diseases

Possibilities of postnatal preventions of inborn diseases are **limited** - when the baby is already born with particular inborn disease it can be prevented, however early diagnostics and therapy may prevent constant worsening of the initial status.

The therapy can be **causal**, where we directly repair the original defect (mutation) or **symptomatic**, where we are treating the complications of the disease without influencing the (genetic) cause. The current possibilities of causal therapy of genetic diseases - **gene therapy** - are still **very limited**. Although the gene therapy is very promising in the long-term course, it is still not a widely used method for routine therapy. Therefore we have to still mainly consider the symptomatic therapy.

Examples of symptomatic therapy:

- **Surgery** (reconstructive surgery e.g. for orofacial clefts, abdominal wall defects, congenital hearth anomalies).
- **Transplantation** (kidney transplantation for polycystic kidney disease, haematopoietic stem cells transplantation for some storage diseases).
- **Rehabilitation**, early stimulation (for children with muscle tone abnormalities, psychomotor retardation).
- **Diet** (for example for some inherited metabolic disorders like diet with no phenylalanine in phenylketonuria).
- **Chelation therapy** (for example Penicillamine in Wilson disease).
- **Enzyme replacement therapy** (can be used to treat some metabolic disorders like Gaucher disease).
- **Targeted protein activators/re-activators** (used to enhance function of selected mutated proteins - e.g. ivacaftor for cystic fibrosis).
- **Special prevention** (extreme sun protection in individuals with Xeroderma pigmentosum).
- **Bioengineering** (prosthesis in individuals with reduction limb defects, ICD implantation in individuals with cardiomyopathies etc.)