Hereditary immunodeficiencies

Immunodeficiencies are very broad and very difficult group of diagnoses – sharing one main feature – inherited or acquired **failure** of (some part of) immune system. Medical genetics deals mostly with the group of so called "**primary**" or **"hereditary" immunodeficiencies** – since those are usually monogenic diseases with certain type of inheritance.

The second group of acquired or secondary immunodeficiencies is very heterogenic, including huge group of pathologies, where the immune system failure can be the main symptom (AIDS) or secondary symptom (leukaemias etc.).

Common features

- Children with primary immunodeficiencies are usually very **prone to the infectious diseases**. Even the "normal" children tends to be "ill very often" since their immune system is in development, but children with primary immunodeficiencies tend to be prone even against **opportunity infections** like patients with AIDS).
- The main features of the diseases (types of recurrent infections) are depended on the part of immune system that is impaired (humoral immunity vs cellular immunity vs combined forms). For humoral immunodeficiencies it is important, that the infant is guarded by maternal antibodies - usually up to the 6th month of life.
- Numerous forms of inherited immunodeficiencies have X-linked recessive inheritance, therefore majority of affect individuals can be **boys**.
- Children with clinical suspicion of hereditary immunodeficiency **should NOT be vaccinated** (especially not with living vaccines).

Selected examples

- **Bruton agammaglobulinemia**: X-linked humoral immunodeficiency, most common of humoral immunodeficiencies.
- **SCID - Severe combined immunodeficiency**: SCID is a group of diagnoses of **severe** humoral+cellular (**combined**) immunodeficiencies, which lead to severe condition (repeated infections), often leading to the death of affected child.
- **Chronic granulomatous disease**: X-linked defect of NADPH-oxidase - leading to severe defect of phagocytosis. This defects leads to increased risk of infections caused by the germs that are usually destroyed in phagocytes.
- **Chromosomal instability syndromes**: like Bloom syndrome or Ataxia teleangiectatica also leads to the increased risk of infections.
- **DiGeorge syndrome**: Complex syndrome caused by 22q11 deletion also leads to congenital immunodeficiency caused by the thymic aplasia.