Postnatal screening of heritable diseases

Postnatal screening of heritable diseases is usually a population-based programme of examinations of (all born) individuals for specific heritable (genetic, monogenic) diagnoses. The precise program is usually different in different countries (organized an financed by the particular Ministry of Health) and reflects the specific population incidence of the heritable diseases in the particular geographic area (the most common diseases are the ones you want to screen in the general population).

Birth defect screening in newborns

The newborns are usually routinely examined by the specialists (neonatologist) on their day of birth. The examination includes:

- **Heart and lung** auscultation (identification of heart defects or birth defects of lungs/airways).
- **Eye red reflex** test (identification of congenital cataract).
- **Muscle tone** test (identification of congenital neuro/myopathies or other genetic conditions).
- **Defecation and urination** (identification of strictures/stenoses/atresias).
- **Otoacoustic emissions** (identification of congenital hearing loss).
- **Hip joint stability** (identification of congenital hip dysplasia).
- **Ultrasound examination** (usually only kidneys, or kidneys + heart).

Newborn screening of inherited metabolic disorders

Screening is usually performed on the level of metabolites obtained from he blood of the newborn that is 72hours old or older. The blood sample is usually obtained from the small heel prick. The list of tested diseases varies significantly among different coutnries. The following (18) dieases are now routinely tested during the newborn screening in the Czech Republic (more information here: http://www.novorozeneckyscreening.cz/en/):

- Endocrine Disorders
  - Congenital hypothyroidism
  - Congenital adrenal hyperplasia
- Disturbances of amino acid metabolism
  - Argininemia
  - Citrullinemia
  - Phenylketonuria
  - Glutaric aciduria, Type I (glutaryl-CoA dehydrogenase deficiency)
  - Homocystinuria from CBS deficiency, pyridoxine non-responsive
  - Homocystinuria from MTHFR deficiency
  - Isovaleryl-CoA dehydrogenase deficiency (Isovaleric acidemia)
  - Maple syrup urine disease
- Disorders of fatty acid oxidation
  - Carnitine uptake/transporter defects
  - Carnitine-acylcarnitine translocase deficiency
  - Carnitine palmitoyl transferase I deficiency (CPT I)
  - Carnitine palmitoyl transferase II deficiency (CPT II)
  - Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)
  - Long chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
  - Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Biotinidase deficiency
- Cystic fibrosis