

Postnatal prevention of hereditary diseases

Postnatal prevention - is focused on screening of genetically at-risk persons and families and the targeted influence of the development of unfavorable dispositions and dispositions. It can only be implemented in close cooperation between geneticists and doctors from other fields.

Population Screening

It is rational only for relatively frequent disabilities with the possibility of successful prevention or treatment. At low costs, it must ensure high specificity and sensitivity.

Ex. screening of Rh negative pregnant women or newborns with PKU:

- if antiD gammaglobulin was not administered to Rh- mothers of Rh+ newborns within 72 hours after delivery, the fetus could be at risk of the mother's antiD antibodies in the next pregnancy;
- similarly, if homozygotes for PKU were not started on a low-phenylalanine diet as soon as possible after birth, irreversible CNS damage would develop;

Regular medical check-ups of infant and toddler are important. In adulthood, population-wide screening is focused mainly on diseases of the circulatory system and some types of tumors.

 For more information see *Newborn screening*.

Genealogical screening of genetically at-risk families

Effective in dominantly inherited (AD) and polygenically inherited diseases and birth defects:

- eg polycystic kidney disease or hypercholesterolemia.

With early detection of heterozygotes or persons with a genetic disposition, the development of the disease can be delayed. Diet, increased activity and possibly drugs for hypercholesterolemia - atherosclerotic vascular changes then do not occur or only at a significantly older age.

Links

Related Articles

- Preconception prevention of hereditary diseases and defects
- Prenatal diagnosis
- Newborn Screening

Source

- ŠTEFÁNEK, Jiří. *Medicína, nemoci, studium na 1. LF UK* [online]. [cit. 2009]. <<http://www.stefajir.cz>>.