Screening for hereditary diseases

Genetic screening is part of health care programs today. Originally, it was about identifying newborns with a genetically determined disease that is treatable in case of early diagnosis, e.g. PKU. The main role of genetic screening is to improve the health of society.

Newborn Screening Programs

🔎 *For more information see Newborn Screening.*

Principles of an adequate screening-test:

- the disease state must not only be clearly defined, but also treatable and its incidence in the monitored population must be significant
- the examination itself must be done quickly and easily on a large number of samples
- the test should have few false positives and, if possible, zero false negatives
- completion of diagnosis and initiation of treatment must be well organized and prompt
- if these conditions are not met, complications will occur

Screening of heterozygotess

'*Conditions for enabling the screening test:*

- significant occurrence of the disease in a specific population group
- availability of a test suitable for mass screening
- possibility of prenatal diagnosis
- the following diseases meet these criteria:

1. Tay-Sachs disease

- often occurs in Ashkenazi Jews
- this is one of several different forms of familial amaurotic idiocy (amaurotic = unclear)
- the enzyme disorder in this disease is a significant deficiency of hexosaminidase A (hex A), in a wide range of tissues; the role of hex A is to cleave the N-acetyl-galactosamine residue from the polysaccharide chain of the ganglioside molecule. In case of its deficiency, ganglioside accumulates, mainly in brain tissue
- heterozygotess can be detected by screening blood samples for hexosaminidase activity
- the disease can be detected by biochemical analysis of cultured amniocytes

2. Sickle cell anemia

- manifests itself practically exclusively in blacks
- the molecular basis is abnormal hemoglobin – HbS
- this is a severe hemolytic disease with a characteristic tendency of red blood cells to take an obviously abnormal form in an environment with reduced partial pressure of oxygen
- clinical picture: anemia, icterus and "crisis", vascular obstruction and painful infarcts of various organs (bones, spleen, lungs)

3. Thalassemia

- heterogeneous group of disorders of HbA chain formation
- caused by damaged genes for hemoglobin, possibly. deletions

Screening α-fetoprotein in maternal serum (MSAFP)

- possibility of detection of split defects, event. fetal Down syndrome warning

Links

Related Articles

- Indication of chromosomal examination
- Inherited disorders of amino acid metabolism
- Newborn Screening

Source

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