Neural tube defects

Neural tube clefts occur early in fetal development. During the 3rd to 4th week, the basis of the spinal cord and brain develop. Neural tube formation is complete by approximately **day 28 of pregnancy** (usually before pregnancy is detected). defects occur If the neural tube is not closed along its entire length. The severity of the damage depends on the location and surrounding tissues. No association with cleft defects of the face or abdominal wall was proven .

Division

clefts in the region of the brain

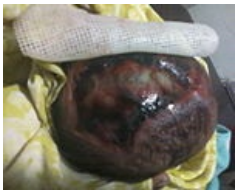
- **Acrania** – absence of a skull cap with protrusion of brain structures.
- **Anencephaly** – missing skull cap and brain tissue at the level of the orbits, often caused by the toxic effect of amniotic fluid on exposed brain tissue.

clefts with protrusion of intracranial structures (cephalocele)

- **Meningocele** – an isolated protrusion of the meninges, more often with open herniations of the spine.
- **(Meningo)encephalocele** – a protrusion of the brain tissue together with the brain cases.

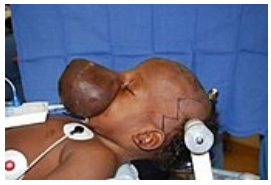
Spina bifida

- ***Spina bifida aperta*** – an open split of the spinal vertebrae together with a protrusion of the spinal cord and spinal tissue
- ***Spina bifida occulta*** – a closed split of the spinal vertebrae without a herniation.



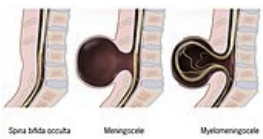
acrania

encephalocele



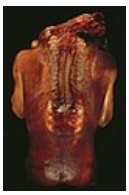
Encephalocele in the facial area

Meningocele in an adult



Types of spina bifida

Spina bifida aperta



Extensive spina bifida with anencephaly

Etiology

The occurrence is multifactorial (genetic predisposition and environmental influence). But in 90-95% of cases, this defect is described in newborns, in whose family no one suffered from this defect.

The risk of developing is significantly increased with a **positive family history** , certain diseases during pregnancy (diabetes , epilepsy treated with antiepileptic drugs), with c**hromosomal defects and genetic syndromes** , with malnutrition with **folic acid deficiency**

Diagnostics

Basic screening during pregnancy is an early and repeated **two-dimensional ultrasound examination**. To clarify the diagnosis, three-dimensional ultrasound and magnetic resonance are performed. The typical signs on the head (*lemon sign* = head shaped like a lemon) **and in the brain** (*banana sign* = erebellum shaped like a banana) alert us, it is more recognizable than the defect of the spine itself. Closed spine defects are discovered mostly by chance due to unusual skin pigmentation.

Open cleft neural tube defects are often associated with congenital brain defects.

Invasive diagnostics

Cleft neural tube is a frequent symptom of chromosomal aberrations – **trisomy of the 18th chromosome** (Edwards syndrome) and trisomy of the 13th chromosome (Patau syndrome). If there is a significant suspicion of these pathologies, we can indicate an invasive diagnostic method for taking material and determining the karyotype of the fetus. Today, it is possible to use fetal DNA diagnostics from the mother's peripheral blood (more expensive, but less risky).

Prevention and treatment

In prophylaxis, the dispensary of the pregnant woman and the determination of the degree of pregnancy risk are applied. Women with a known diagnosis of diabetes are advised to have a planned pregnancy to minimize the risk of developing developmental defects. **Education** about the effect of teratogenic substances and careful consideration of pharmacotherapy during pregnancy are also important . In the first trimester, we recommend an **increased intake of folic acid** (protective effect against VVV).

Treatment of the defect itself is carried out by a surgical intervention usually only postnatally (shortly after birth). During the first year of life, a so-called **shunt** is introduced into the cerebral ventricles. In the event of a risk of toxic effects of amniotic fluid on the brain tissue, we can indicate prenatal closure of the neural tube (**fetal surgery**). Of course, such operations are associated with a high risk of premature birth (fetal immaturity) or further injury to the fetus.

Links

Related Articles

- cleft defects
- the third week of embryo development

References

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