Dandy-Walker syndrome

Dandy-Walker syndrome is a rare **disease**, affecting the development of the **brain**. It manifests with a triad of symptoms

1. complete or partial **agenesis of vermis cerebellum**
2. cystic **dilatation** of the fourth ventricle of brain
3. **enlargement** of the posterior cranial fossa[1]

Enlargement of the posterior cranial fossa may be associated with membranous atrasia of the apertures of the fourth ventricle, leading to pathological **accumulation** of cerebrospinal fluid in the ventricles.

These abnormalities most often result in problems with movement, coordination but also with intellect. Psychiatric illness may also occur[2].

Affected individulas may survive up to their second decade of life.

Malformations

In most cases, individuals with Dandy-Walker syndrome develop symptoms of **abnormal brain development** during the first year of life. Clinically, it is manifested in most children by the accumulation of cerebrospinal fluid in the brain - hydrocephalus, which can cause macrocephaly.

Mental disability is often present[3], although some individuals also have a normal intellect. Children often have **delayed development**, especially in the field of motor skills (crawling, walking, coordination of movements). Muscle stiffness and paralysis - spastic paraplegia - may occur. Older children experience symptoms of increased intracranial pressure such as irritability, vomiting, convulsions and nystagmys. Less frequently other cerebral malformations are present such as agenesis of the corpus callosum connecting the right and left hemispheres, occipital encephalocele or abnormal clefts in the brain - schizencephaly and gyrification disorders. These brain defects are associated with more or less severe signs and symptoms. Dandy-Walker syndrome can also include **heart defects, malformations of the urogenital tract, limbs and face**. A milder form of the disease is the Dandy – Walker variant, which includes hypoplasia of the cerebellar vermis without agenesis, mild or no enlargement of the posterior fossa and fourth ventricle, without hydrocephalus.

Genetics

Mutations in certain genes occur in individuals affected by Dandy-Walker syndrome, but these mutations account for only a small number of all known cases. Dandy-Walker syndrome is also **associated with chromosomal abnormalities on most chromosomes.'' *It is most common in people with trisomy 18, but can also occur with trisomy 13,* *21* or **9**.** Dandy-Walker syndrome has also been reported in fetuses with *triploidy*, a fatal condition in which individuals have an extra complete set of chromosomes in each cell. Dandy-Walker syndrome can also be **caused by mutations of specific genes** (FOXC1 contains instructions for the preparation of a protein that regulates the activity of other genes, ZIC1 and ZIC4 act as transcriptional activators and participate in CNS organogenesis). Brain malformations associated with Dandy-Walker syndrome can occur also in isolation, and may not be associated with a genetic disease, with the cause being often unknown.

Environmental influence

Dandy-Walker syndrome can be caused by environmental factors that affect the fetus before birth. For example exposure of the fetus to rubella, toxoplasmosis or due to substances that causes damage to the fetus, (teratogens). A more frequent occurrence of fetuses with Dandy-Walker syndrome also occurs in the fetuses of mothers who suffer from diabetes.

Diagnosis and incidences

Prenatal diagnosis is possible by ultrasound examination and performing amniocentesis to examine the karyotype of the fetus. It is estimated that **Dandy-Walker syndrome occurs in 1:25000 to 1:30000 newborns**. The occurrence of cases of Dandy-Walker syndrome is rare and does not have a clear pattern of inheritance. Only immediate relatives *of people with Dandy-Walker syndrome have an increased risk* of developing the disease compared to the general population.

Therapy

Therapy consists in removing the obstruction, and hydrocephalus is treated by introducing a shunt.

Links

Related articles

- CNS malformations
- Congenital defects of the nervous system

External links

- http://www.priznaky-projevy.cz/geneticke-nemoci/449-dandy-walker-syndrom-dandy-walkerova-malformace-priznaky-projevy-symptomy

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