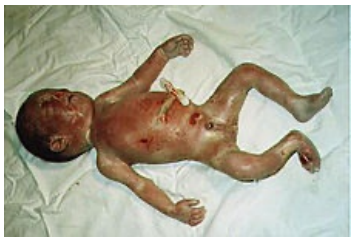
Wolf-Hirschhorn syndrome

Wolf-Hirschhorn syndrome is a structural chromosome aberration, specifically a **deletion of a subtelomeric section on the arm of the 4th chromosome**. The region responsible for Wolff-Hirschhorn syndrome is located at 4p16.3. Deletion can be of varying extent. A deletion smaller than 3.5 Mbp results in a milder form of this syndrome, also called Pitt-Rogers-Danks syndrome. The typical phenotypic features of the syndrome were first described in 1961 by Cooper and Hirschhorn, and cytogenetic examination revealed a deletion at 4p. In 1965, an article was published in *Humangenetik* by the authors Wolf and Hirschhorn, who brought the syndrome to the wider awareness of geneticists.



Wolf-Hirschhorn syndrome Clinical picture of microcephaly, hypertelorism, *carp mouth*, cleft lip, postpartum hypotonia, defects of the heart, kidneys, skeletal system, hypospadias Cause of chromosomal aberration on the 4th chromosome Prenatal diagnostics, genetic testing Investigation in the Czech Republic CG21 Worldwide incidence 1:50,000 Prognosis by phenotype; 20-40 years

Symptoms

Patients are severely psychomotor retarded. Seizures (epilepsy) are common. The most typical phenotypic manifestations are visible in the face. These include microcephaly, a wide beak-like nose, hypertelorism, a special shape of the mouth, the so-called **carp mouth**, protruding glabella, epicanthus, cleft lip and/or palate. Growth retardation is also evident, already in the prenatal period. Hypotonia is noticeable after delivery. Common organ malformations include defects of the heart, kidneys, skeletal system. In boys, the opening of the Urethra at the base of the penis (hypospadia) and non-descension of the testicle (cryptorchis) are common.

Genetics

Approximately 87% of deletions occur *de novo* and more often on the paternal chromosome. Up to 15% of deletions are caused by a balanced translocation of the maternal chromosome. The incidence of individuals with Wolf-Hirschhorn syndrome is 1:50,000 births. For unknown reasons, the syndrome occurs twice as often in women as in men.

Treatment and care

There is no comprehensive treatment for Wolf-Hirschhorn syndrome. It depends on the specific needs of individual patients. Some sufferers have more serious organ defects requiring surgical treatment. Seizure periods must be treated with antiepileptic drugs containing, for example, valproic acid. Nutritional problems, low weight, and poor growth often require high-calorie artificial nutrition administered through a percutaneous endoscopic gastrostomy (PEG). Physiotherapy, ergotherapy and speech therapy will help improve the mental state of patients and are therefore welcome.

Prognosis

The prognosis depends on the nature and number of congenital defects. Babies with Wolf-Hirschhorn syndrome may be stillborn or may die shortly after birth. Approximately 35% of patients with this syndrome will die within 2 years of age. Individuals that survive to adulthood have more difficulty walking or do not walk at all. Some are able to speak in short sentences. Life expectancy has not been studied further, patients live between 20-40 years.

Links

External links

- Wolfhirschhorn.org (http://wolfhirschhorn.org/about-wolf-hirschhorn-syndrome/)

References

- KOČÁREK, Eduard – PÁNEK, Martin. *Klinická cytogenetika I : úvod do klinické cytogenetiky.* 2. edition. Prague : Karolinum, 2010. ISBN 978-80-246-1880-7.