Duane Retraction Syndrome

Duann's retraction syndrome (DS) is characterized as a disease manifested by strabismus with non-progressive horizontal ophthalmoplegia (inability to move the eyes) primarily affecting the nucleus and fiber of the abducens nerve (VI) and its neuromuscular connection with the rectus lateralis muscle. After birth, the child has a limited range of movement of the bulb in abduction and adduction, during which the bulb retracts into the orbit and at the same time the eye slit narrows. Strabismus tends to be very noticeable, however, most sufferers learn to use a compensatory head position and thus prevent the loss of binocular vision and the development of diplopia. DS is most often isolated to one eye. The patient more often has amblyopia.

Etiology

DS arises from insufficient development of motor neurons in the nucleus of the abducens nerve or in its course and neuromuscular connection with the rectus lateralis muscle. Originally, the origin was assumed to be muscle fibrosis, however, recent research and autopsies have shown the absence of motor neurons and, at the same time, the compensatory involvement of nerve fibers from the oculomotor nerve. The EMG recording then shows a simultaneous contraction of both the rectus lateralis muscle and the rectus medialis muscle , which causes retraction of the bulb.

Pathogenesis

The majority of DS sufferers have a defect in the CHN1 gene, which is autosomally inherited with incomplete penetrance. Each child of the proband conceived with a healthy individual will have a 50% chance of inheriting the pathological combination of alleles and thus of developing the disease. Isolated familial cases can also arise from a de novo gene defect. The products of the CHN1 gene are the regulatory molecules α1-chimaerin and α2-chimaerin, which play an important role in the early development of the nervous system. In general, they help regulate the formation of a complex of signaling molecular cascades during the development of axons and dendrites of a neuron. α2-chimaerin is characteristic for the development of the nerves of the head and neck, especially the abducens nerve (VI).

Diagnosis

Diagnosis is based on the clinical picture. At the same time, a sequential genetic test of the proband's family for the CHN1 gene can be performed. DS should always be considered when:

- congenital defect in the movement of the eyeballs (abduction and adduction)
- retraction of the bulb during adduction
- narrowing of the orbital fissure during adduction.

Therapy

Amblyopia is treated with glasses or contact lenses. The leading eye is often covered. A specialized surgical procedure is also available abroad, which improves the position of the head and alleviates deviations in eye movements, but does not solve the cause.

Links

External links

https://emedicine.medscape.com/article/1198559-treatment

https://www.youtube.com/watch?v=QMpB08S-G74

Source

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