Friedreich's ataxia

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This is the most common **hereditary ataxia**. The basis of this autosomal recessively inherited disease is the intronic expansion of GAA repeats of the X25 gene on chromosome 9. The gene mutation causes impaired **frataxin** function and subsequent mitochondrial oxidative stress. It belongs to the **neurodegenerative** diseases.

Incidence

1-2/100,000 inhabitants

Clinical picture

The disease manifests most often at the end of the second half of the first decade of life. Rarely, patients from other age groups have been described.

The initial symptom is **paleocerebellar symptomatology**, often interpreted by the environment only as clumsiness, which worsens after closing the eyes or in the dark. The first symptoms may be asymmetric. The coordination of the movements of the upper limbs gradually deteriorates, **tremor**, choreiform dyskinesias of the limbs or mimic muscles are observed, possibly. shaking his head. Finding of **nystagmus** and rapidly worsening **dysarthria**. Other cranial nerves can also be affected in the sense of external or internal ophthalmoplegia, various degrees of **hearing and vision disorders**.

Higher nerve functions are usually intact during the orientation examination, special tests in some cases show cognitive dysfunctions. Tendon-muscle reflexes of the limbs usually go out in the early stages of the disease, deep sensation can be maintained for a long time in younger patients. Babinsky's pyramid irritation phenomenon is well equipped. In Friedreich's ataxia with an **adult** onset, reflexes can be preserved and pyramidal phenomena of irritation are **missing**! Tactile and algic sensation is usually preserved, alteration can occur only in the late stages of the disease, disorders of autonomic innervation of the surface layers of the skin have also been described.

Bone deformities include **pes cavus**, **hammer-like position of the fingers**, deformities of the hands and flexion contractures of the joints, less often pedes plani. Common finding of **kyphoscoliosis**. Orthopedic surgery is not beneficial, because the body may not be able to compensate for a sudden change in the center of gravity of the body and the cerebellum.

Other manifestations

Hypertrophic cardiomyopathy with ECG changes in 90% of patients with juvenile form. Diabetes mellitus most often in the third decade and is difficult to compensate with insulin. Conditions of hypothermia, intermittent vomiting and ventilation disorders have also been described.

Diagnostics

The results of ancillary examination methods are often pathological but **non-specific**. MRI examination shows **atrophy of the upper part of the cervical spinal cord** in the early stage of the disease, the cerebellum atrophies later.

Therapy

Intensive **rehabilitation**, polyvitaminosis therapy, nootropics, cognitives. Monitoring in cardiology, endocrinology and orthopedics. Studies are ongoing with the coenzyme Q10 analogue idebenone, which, according to preliminary results, slows disease progression and has a symptomatic effect.

External links

- https://www.neurologiepropraxi.cz/pdfs/neu/2007/05/04.pdf
- https://www.upjs.sk/public/media/7402/2013-03-07-LEKARSKE%20LISTY%20-%20Hereditarne%20a%20ziskan%C3%A9%20spinocerebellarne%20ataxie%20-%20Skovranek.pdf

Reference

AMBLER, Zdeněk. *Základy neurologie.* - vydání. Galén, 2006. 351 s. ISBN 9788072624331.