Friedreich ataxia: The Search for a Treatment

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Friedreich ataxia is a progressive neurological disease affecting 1 in 50,000 people. The major symptom of the disease is gait and limb ataxia, lack of voluntary coordination of muscle movements. Cardiac complications also occur in a majority of patients and are the leading cause of death. Presently, there are no existing therapeutic treatments to slow or halt the disease progression, instead the treatment is directed to managing the symptoms. There are several therapeutic treatments in research and clinical trials and hopefully soon there will be a drug on the market.

What is the cause of Friedriech ataxia?

The disease Friedreich ataxia involves a slow progression of ataxia, usually detected at the age of 10 – 15, and the disease rarely breaks out after 25 years of age. The underlying cause of the disease is a mutation in a gene called X25 which is located on chromosome 9. The mutation causes a deficiency of the protein frataxin. This protein plays a crucial role in the regulation of iron in humans. Frataxin is primarily active in mitochondria, the part of the cell that is responsible for metabolism, where it affects a variety of processes related to iron metabolism. The lack of frataxin causes iron to accumulate in the mitochondria, which leads to the formation of toxic substances which in turn leads to cell damage and death. The function of the protein is still under discussion. Therefore, it is difficult to say exactly how frataxin deficiency contributes to the pathogenesis of Friedreich ataxia.

Treatment of Friedriech ataxia

At present, treatment for Friedreich ataxia is aimed at symptom relief and frequent health checks to be able to detect secondary diseases such as diabetes, so treatment can be started early in the disease process. The complex and varied symptomatology of Friedreich ataxia patients leads to the need for a broad interdisciplinary approach in the management of the patient group and its treatment. The treatment is based on extending patient independence in daily life and the maintaining a good quality of life. Over the course of development, a lot of studies have been conducted to find disease-modifying substances, but so far there are no drugs that stop the progression of the disease. That is why today's treatment focuses on managing the symptoms and the problems they give rise to.

Although there is no drug available on the market for Friedreich ataxia at present, there are several treatments under investigation. Either the treatment focuses on correcting the problem caused by the frataxin deficiency or eliminating the mutation that has occurred and recreate a healthy gene. Determining the exact function of frataxin could be an aid in the development of medicines for the disease. The search for an effective treatment for Friedreich ataxia continues and the future will show what the solution will be, either if the most effective treatment will be based on the increase of frataxin levels by upregulation of the gene or gene therapy. Or if the future treatment will target the metabolic consequences of the disease.

Further reading

If you are interested to know more about this subject, you can read the following article Åkerström J. 2017. Vad är den bakomliggande mekanismen till Friedreich ataxia och kan vi utveckla en mer effektivare behandling? Independent Project in Biology, Uppsala University.