Friedreich Ataxia Diabetes

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Introduction:

Friedreich's ataxia (FA) is the most common hereditary ataxia, causing neurodegeneration, which leads to neurological function decline at various stages, it may also be associated with a major cardiovascular, ophtalmologic, auditory and metabolic desorders suchas diabetes mellitus due to both insulin deficiency and resistance.

Objective:

This study aims to outline the different physiopathological and therapeutic aspects of diabetes in friedreich's ataxia through a clinical case.

Observation:

24 years old patient, without specific medical assessment, except maternal diabetes, under treatment, and without any similar familial case. The patient reported insidious and progressive installation of walking and balance disorders worsening significantly over the past year. All evolving in a context asthenia, weight loss with polyuropolydipsic syndrome. Physical examination finds a cerebellous syndrome with a positive Romberg test, impairment of vibrational sensitivity and amyotrophy of the lower limbs. The biological assessment reported an elevation of HbA1c and fasting blood glucose assessment. The Electroneuromyography approved a pure sensory neuropathy with sensitive axonal involvement , compatible with Friedreich ataxia. Genetic examination is underway. The patient was treated with metformin and basal bolus insulin therapy with a spectacular evolution and a normalization of blood glucose testing.

Conclusion:

Friedreich ataxia is suspected in individuals with a combination of family history, clinical neurologic findings, musculoskeletal features, optic atrophy, hypertrophic non-obstructive cardiomyopathy, endocrinologic disorders such as glucose intolerance, diabetes mellitus. The confirmation is established by detection of biallelic pathogenic variants in FXN. Our patient had a number of arguments in favor of this disorder, apart from heredity, although the genetic study is still in progress.

The physiopathology includes an oxidative stress by iron accumulation in mitochondria leading to the celle apoptosis and increasing the cardio-vascular and metabolic risk.

The management of this specific diabetes mellitus is intricated including a combination of insulin-sensitizers and insulin therapy, while anti-oxidant agents remain to delay the disease's progression.

Keywords:

Diabetes mellitus Friedreich ataxia, insulin therapy, insulin-sensitizers