Diabetes Mellitus and Steinert's Myotonic Dystrophy

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Abstract:

Background:

Steinert's disease, or Steinert's myotonic dystrophy, is a muscular disorder characterized by myotonia and multi-organ involvement associating varying degrees of muscle weakness, cardiac dysfunction, cataract, endocrine involvement (hypogonadism, carbohydrate intolerance), baldness.

Patients & methods:

We report a case associating diabetes mellitus and Steinert's disease with the aim of highlighting the characteristics of this type of diabetes.

Case report:

Male patient, aged 47, known diabetic for 23 years, initially treated with metformin, then Insulin and metformin. Diabetes was discovered in a context of overweight without acid ketoacid decompensation. He underwent surgery for bilateral cataract and ptosis with good evolution. Given the notion of congenital bilateral ptosis associated with baldness since the age of 30, bilateral cataract and signs of erectile dysfunction with primary infertility, the diagnosis of Steinert's myotonic dystrophy was made at the age of 38. The patient was referred to us for management of his diabetes. Clinical examination revealed a conscious patient with asthenia, overweight with a BMI of 26 kg/m², a non-palpable thyroid and a peripheral neurogenic syndrome on neurological examination. On paraclinical examinations: HbA1c was 8%, ophthalmological examination showed no sign of diabetic retinopathy, 24-hour microalbuminuria was normal. Lipid profile showed hypercholesterolemia. electrocardiogram was normal. Hormonal profile showed normal thyroid function, with TSH at 1.32 mIU/l. Testosteronemia at 4.96 mg/l, FSH at 4.5 iu/l and LH at 6.7 iu/l, and normal prolactinemia. Management was based on therapeutic education, optimization of antidiabetic treatment and initiation of atorvastatin. The evolution was favorable.

Discussion & Conclusion:

Steinert's myotonic dystrophy is a rare autosomal dominant neuromuscular disorder with incomplete penetrance and variable expressivity. It's caused by an abnormality in the DMPK gene, which codes for a protein called "myotonin" involved in several physiological functions, explaining the diversity of disorders encountered, including diabetes.

Clinical severity, and hence prognosis, varies from the severe form in newborns to the late-onset form in adults. Diabetes affects 6% of Steinert's disease patients. Its pathogenesis incriminates insulin resistance via a pre-receptor mechanism, which could be due to a deficiency in muscle protein kinase. Management ideally includes multidisciplinary follow-up. Disease progression is generally slow, but rapid deterioration can sometimes be encountered. Life expectancy is reduced by the increased mortality associated with pulmonary and cardiac complications.