Psychology and Psychiatry: Open access

Further preliminary results of enzyme replacement therapy for late-onset Pompe disease

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Abstract

Pompe disease is a rare hereditary disease related to storage diseases: due to a genetically determined deficiency of the enzyme alglucosidase alpha, pathological glycogen deposition is noted in all tissues and organs of the patient, which leads to various symptoms. The most common manifestations of late onset are slowly progressing waist-limb myodystrophy and respiratory failure. Other manifestations may be diverse [1]. In the routine practice of a doctor the Pompe disease is so rare that every experience has a high value [2].We present a clinical case of a late onset Pompe disease with an assessment of the dynamics of clinical symptoms before the start of enzyme replacement therapy and at an early stage of its use.

A 34-year-old woman complained of slowly progressing muscle weakness from the age of 25. Upon examination, the previous diagnosis, the patient visually did not have obvious signs of muscle hypotrophy against the background of excess body weight. The pterygoid scapula became noticeable only when the anterior limbs were raised anteriorly, and gluteal muscle hypotrophy was detected by palpation. Nevertheless, the patient showed not too obvious weakness of the muscles of the pelvic girdle, getting up from squats was significantly difficult, and elements of an overwhelming gait were noted. Hereditary myopathy was suspected. A study of the activity of alglucosidase alpha and the subsequent identification of mutations in the GAA gene confirmed the diagnosis of late onset Pompe disease.

It is known that enzyme replacement therapy is more effective against cardiomyopathy, to a lesser extent - striated muscle myopathy[3]. The effectiveness of enzyme replacement therapy in Pompe disease is recommended to be judged in 2 years from it's beginning[4]. In the clinical case we present, for a short time — only 4 months — of enzyme replacement therapy with myozyme, there was a slight encouraging regression of muscle weakness of the hip flexors, and an improvement in the results of the 6-minute walk test. In the next 4 months there is further efficiency of enzyme replacement therapy. Among the observation features, it is necessary to highlight an improvement in the motor function of the intestine. It is known that glycogen in Pompe disease is deposited everywhere, including the walls of the intestine, what can lead to impaired intestinal motility[5]. Getting rid of constipation and a decrease in muscle pain were the first signs of improvement during therapy. Laboratory results also showed a steady improvement from the beginning of the enzyme replacement therapy.

Biography

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Citation: Liubov P. Smertina, Further preliminary results of enzyme replacement therapy for late-onset Pompe disease, June 08-09, 2020, PP. 06