عنوان مقاله:

Headache: A Presentation of Pompe Disease; A Case Report

محل انتشار:

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خلاصه مقاله:

Pompe disease, also termed glycogen storage disease type II or acid maltase deficiency, caused by deficient activity of acid alpha-glucosidase (GAA), the glycogen degrading lysosomal enzyme. As a result, massive lysosomal glycogen deposits in the numerous organs including the muscles. In Pompe disease weakness of truncal muscles is a prominent presentation which results in respiratory failure as a main clinical presentation in the early stages of the disease. Even sleep may be affected by nocturnal respiratory disturbances. Specific treatment with enzyme replacement (human recombinant GAA) is available. Here we present a case of progressive muscular weakness which had been misdiagnosed with limb girdle muscular dystrophy. A history of severe morning headaches led authors to think about sleep apnea, which was confirmed by polysomnography and therefore provided a clue for appropriate diagnosis of Pompe disease. As a conclusion, Morning headaches and sleep insufficiency in any stage of a progressive muscular disorder can lead us to think about respiratory muscle involvement, which is more prominent in Pompe disease.

کلمات کلیدی:

Pompe Disease; Sleep Apnea, Obstructive; Lysosomal Storage Diseases; Headache