A family study of phosphorylase deficiency in muscle

Abstract

The first description of a myopathy due to a defect of muscle glycolysis and characterized by muscular weakness and pain on exertion was made by McArdle in 1951 (1). He described a 30-year-old man with a lifelong history of pain, stiffness, and weakness of muscles occurring with exertion. The symptoms lasted minutes to hours and disappeared with rest but became more severe if exercise was continued after the onset of difficulty. McArdle showed that this patient did not produce lactic acid during ischemic exercise of the forearm. He concluded that the patient had a defect of muscle glycolysis and noted

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Myophosphorylase deficiency (McArdle's syndrome) is an uncommon condition characterized by exercise intolerance, muscle cramps, and myoglobinuria. This study was supported by a grant from the Prinses Beatrix Fonds.