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Light and Electron Microscopy in a Case of Myophosphorylase Deficiency (Glycogenosis V or McArdle Disease)

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We are presenting the light and electron microscopic findings in a 27 year-old female with McArdle disease who presented with exercise induced myalgia and cramps.

Myophosphorylase deficiency is a rare skeletal muscle disorder that produces weakness, pain and cramping in skeletal muscles upon exertion (exercise intolerance), often with temporary paralysis. This is sometimes associated with myoglobinuria due to muscle fiber breakdown [1].

The disease results from a genetic abnormality that produces an incomplete expression of myophophorylase activity. Specifically, the gene responsible for human myophosphorylase deficiency is found on chromosome 11. Inheritance is autosomal recessive, with a male preponderance [1].

In fact, several disease-associated mutations may occur in the myophosphorylase gene, but in Caucasians a nonsense mutation at codon 49 accounts for three fourths of the cases. [2]. The disease usually expresses itself before age twenty but may occur later in life [3]. A fatal infantile form also exists [4].

As a result of myophophorylase deficiency, glycogen cannot be broken down as a needed energy source to fire muscle fibers. Symptoms occur upon exercise when myophophorylase is reduced to 30%-50% [5]. Glycogenosis V is the specific type for mature skeletal muscle [1].

In the present patient, the most important microscopic findings consisted of lack of myophosphorylase staining in the sarcolemma of myofibers using light microscopic histochemical techniques (See Fig. 1) and the accumulation of rich subsarcolemmal and intracyotplasmic deposits of glycogen on electron microscopy (See Fig. 2). Additionally, a few fibers were atrophic, some displayed central nucleation and scattered ones showed subsarcolemmal vacuoles. The intensity of the PAS stain was diffusely increased, consistent with the glycogen deposits. These features are classic for myophosphorylase deficiency (Glycogenosis V or McArdle disease).

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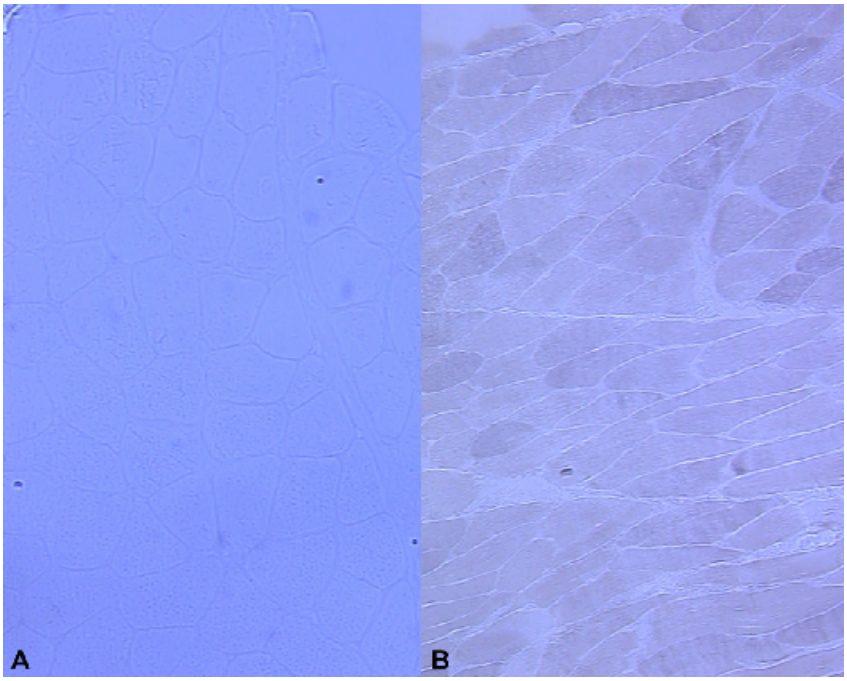

FIG. 1. Light microscopy showing phosphorylase activity in muscle fibers A=Case, phophorylase absent; B=Control, phosphorylase present

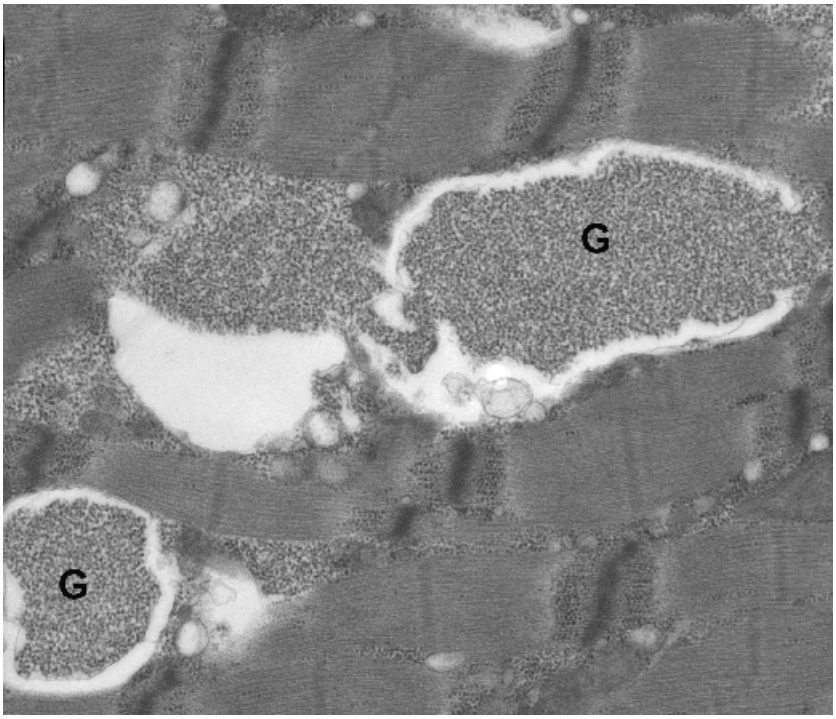

FIG. 2. Electron Micrograph showing glycogen accumulation (G) in muscle

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