



McArdle's disease: A differential diagnosis of idiopathic toe walking

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ABSTRACT

Idiopathic toe walking (ITW) is a pathological gait pattern in which children walk on their tip toes with no orthopedic or neurological reason. Physiological characteristics of the gastrocnemius muscles, the Achilles tendon, and the foot of toe walkers differ from subjects with a plantigrade walking pattern. McArdle's disease is characterized by the inability to break down muscle glycogen. It is an autosomal-recessive condition, characterized by low exercise tolerance, muscular atrophy at the shoulder girdle, episodes of myoglobinuria after vigorous physical activities and the occurrence of the second wind phenomenon. The aim of this review is to present the case studies of two subjects who were originally diagnosed as idiopathic toe walkers, but were then found to have McArdle's disease. This review will describe some physical characteristics that distinguish McArdle's disease from Idiopathic toe walkers.

1. Introduction

Idiopathic Toe Walking (ITW) is defined as a walking pattern in which the weight bearing occurs on the forefoot. It is diagnosed in the absence of any neurological or orthopedic condition if the toe walking persists after 2 years of age^{1,2}. The etiology of toe walking is still unknown; however, literature suggests a positive family predisposition in about 40% of the cases^{1,3–5}, a congenital short Achilles tendon^{6,7} or a sensory processing dysfunction (SPD)^{2,8,9}.

McArdle's disease is a genetic Type V Glycogen storage disease (located on the chromosome 11). As a result of this deficiency, patients with McArdle disease experience muscle cramps, muscle injury, and myoglobinuria induced by vigorous exercise¹⁰. One of the main characteristics of this medical condition is the second wind phenomenon^{10,11}.

The second wind phenomenon is characterized by a period of time when pain decreases and exercise efficiency increases after a period of muscle pain. Before the appearance of the second wind, patients experience tiredness and an increase of the heart rate. The second wind phenomenon occurs after 6–8 min of exercise; typically the patients with McArdle have already stopped exercising or have reduced their pace at this point¹⁶.

This review presents two cases of patients who exhibit a toe walking pattern with no signs of neurological or orthopedic conditions. The two subjects were diagnosed as idiopathic toe walkers. Later findings and clinical examination suggest McArdle's disease. The main goal of this review is to point out some clinical characteristics which children affected by ITW and children with McArdle's disease can have in common and also how they differ. It is very likely that some of the children diagnosed with ITW actually have McArdle's syndrome. Describing these clinical characteristics will help health care practitioners find the correct diagnosis in these cases.

2. Review of case studies

We are reporting the cases of a 10-year-old boy and an 18-year-old woman. In both cases the subjects were diagnosed as idiopathic toe walkers, based on the fact that they walked on their toes, their gait pattern was bilateral and symmetric, and there were no signs of a neurological or orthopedic condition.

During the physical examination, they both reported to have been toe walkers since the onset of walking with a normal neurological and motoric development. Both of them reported having recurring pain in different body parts with difficulties localizing a specific area. The

The 10-year-old boy was tested using next-generation sequencing which was positive for the glycogen storage disease type V or McArdle's disease. The genetic finding was as follows: PYGM, c. 2128_2130delTTC; p.Phe710del (het.), NM_005609.2, rs527236147;

PYGM, c. 1620 + 1G > A; p.? (het.), NM_005609.2;

Of note, c. 1620 + 1G > A has not been described so far. Both mutation are most likely in compound heterozygous state and can be considered pathogenic.

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