Journal of Orthopaedics 15 (2018) 685–689

Contents lists available at ScienceDirect

Journal of Orthopaedics

journal homepage: www.elsevier.com/locate/jor

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 exercise[10]. 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 point[16].

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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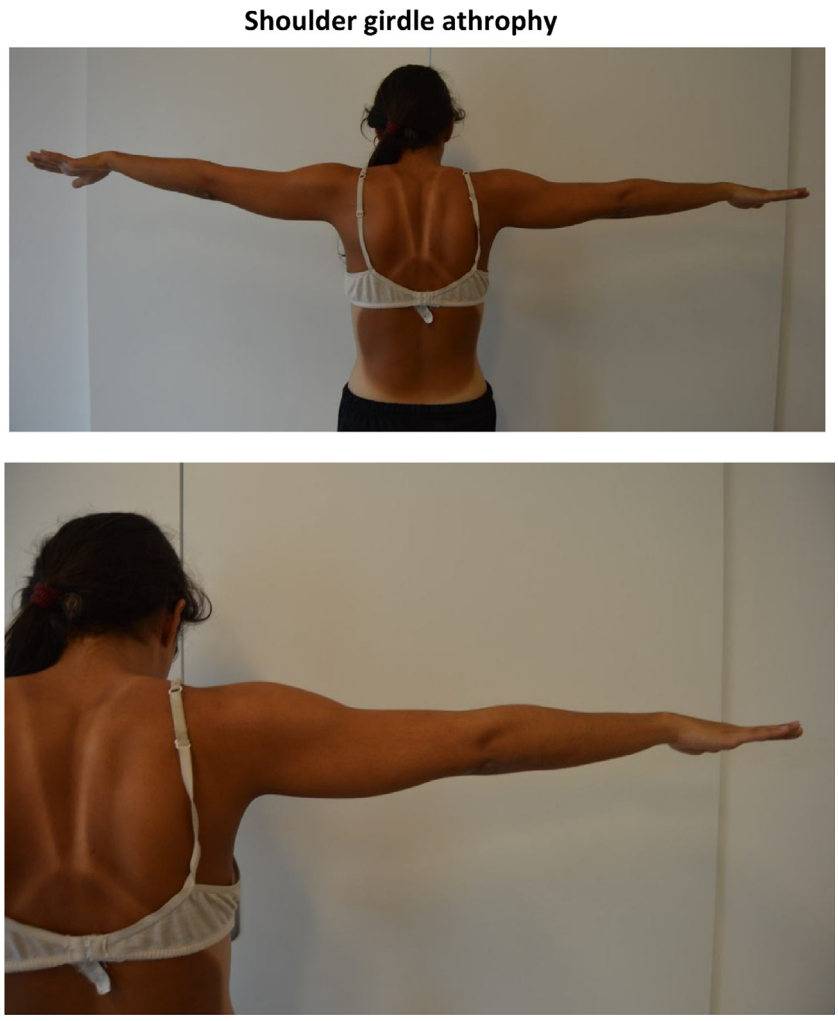
https://doi.org/10.1016/j.jor.2018.05.024
Received 18 April 2018; Accepted 6 May 2018
Available online 08 May 2018



Figs. 1 and 2. Show the shoulder girdle atrophy on the 18 years old young woman. Lateral portion of the triceps brachii muscles and the long head of the biceps brachii muscles show signs of hypotrophy.

intensity of the pain increased with physical exertion.

In the upper extremity the deltoid muscles are apparently normal, but the lateral portion of the triceps brachii muscles and the long head of the biceps brachii muscles show signs of hypotrophy; and there is a general atrophy of the shoulder girdle (Figs. 1–3). Additionally, they presented myoclonus affecting the hands.

In the lower extremity, there is a hypertrophy of the gastrocnemius muscle while the belly of the muscle is more proximal (Fig. 4); the forefoot is wider (Figs. 5, 6), and the ankle's range of motion (ROM) is decreased compared to children with a plantigrade walking pattern.

The "spin" and "walking after spinning" tests were positive for toe walking in both subjects; in the "heel walking test" the young woman was able to heel walk with several compensations (trunk flexion and knee hyperextension). The boy was not able to perform this test. (The tests were taken from "*Idiopathic Toe walking, tests and family predisposition" (1)).*

The 18-year-old woman reported tiredness after activities such as running or walking; however, she actively swims 3–4 h per day, attempting to increase her stamina and speed. She does not report any problems during the training in the water; however, when the training includes weight lifting, she complains about muscular aches and she uses lighter weights and reduces repetitions.

The 18-year-old subject was diagnosed using clinical characteristics of McArdle disease. By recognizing the fatigue, muscle cramping, the atrophy on the shoulder girdle and in particular the second wind phenomena it was possible to diagnose McArdle in the 18-year-old woman

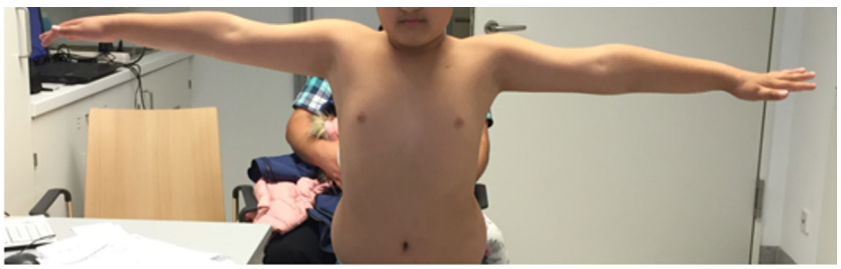


Fig. 3. Shows the shoulder girdle atrophy on the 10 years old boy. Lateral portion of the triceps brachii muscles and the long head of the biceps brachii muscles show signs of hypotrophy.