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The Genetic Causes of Toe Walking in Children

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Abstract

This article describes how, methodologically, the genes which can be linked to idiopathic toe walking have been identified. Additionally, the article gives an overview of the relevant genes which have been identified and classifies them according to the Types of Toe Walking scheme by Pomarino. It explains, why this new research gives reason to the claim that idiopathic toe walking does not in fact exist.

Keywords: Idiopathic Toe Walking, Genetics; Genes AIFM1; EGR2; PMP22

Introduction

The research presented in this article can for the first time identify the medical causes of Idiopathic Toe Walking (ITW). The term Toe Walking (TW) describes a gait anomaly, which causes patients to walk on their forefoot, rather than applying pressure to the entire sole of the foot. The roll over motion which is present in normal subjects cannot always be found in toe walkers. The causes for this can be of a neurological or Orthopaedic nature. Therefore, the patient will be examined for any neurological or Orthopaedic problems which might cause him/ her to walk on their toes.

If no medical cause for toe walking can be found in children over the age of two, who continue to walk on their forefoot for more than 50% of the time, the diagnosis of idiopathic or habitual toe walking is secured [1]. This diagnosis is only agreed on, if all alternative reason for the abnormal gait pattern can be excluded [2]. The existing literature describes toe walking as having a possible hereditary cause, and further research into possible genetic causes is recommended in order to further extent the existing research into the condition [3].

The results of the analysis of possible genetic causes for idiopathic toe walking have led to the identification of a number of genes, which can be linked to patients who are predominantly walking on their forefoot.

A significant statement which can be made on the basis of the research at hand is that idiopathic toe walking as a condition with no medical cause does not in fact exist.

The genes which the research has linked to toe walking can be classified according to the Types of Toe Walking scheme by Pomarino.

It defines toe walkers of Type I as having a late onset of the gait anomaly and normal reflexes up until the age of 14. Patients display a hypotrophy of the calf muscle, a pes cavus (high-arched foot) and delayed language development. Additionally, some children suffer from a tremor of the upper extremities. This cluster of symptoms has now been linked to changes in the area of the genes PMP22, EGR2 & AIFM [4,5].

In contrast, toe walkers of Type II display early onset toe walking and a hypertrophy of the calf muscle. Patients with Type II toe walking have at least one other family member who displays symptoms in 90% of all cases [6]. Furthermore, the children have motoric difficulties and, in a few cases, suffer from limited mobility in their upper ankle. The cluster of symptoms for Type II toe walking has been observed in patients with changes to the genes MORC2, DHTKD1, GDAP1, KIF1B, FGD4, SBF2, SH3TC2, NAGLU, NEFL & PRX [5].

Whole Exome Sequencing and the Identification of Genes which can be Linked to ITW

The method of whole exome sequencing is used to genetically test the 1-2% of the human genome that accommodates approximately 85% of all mutations that cause illnesses [5,7]. This small section is known as the exome and can be sequenced using genetic tests 1. Even gene variants which are known to cause rare genetic disorders can be found during the sequencing process [5,8]. Mutations on the genes can lead to the wrong proteins being produced or halt the production of individual proteins altogether [8,9].

Geneticists using this method are able to identify the cause of unspecific and complex symptoms [5]. Even if doctors have spent years trying to secure a diagnosis for a patient the method can aid in establishing the cause of the symptoms at hand [9].