# **Physical Medicine and Rehabilitation Research**

# **Research Article**



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# Prevalence of associated diagnoses in children who present with toe walking

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#### Abstract

Purpose: The purpose of this study was to assess the relative prevalence of underlying diagnoses among children who presented to an orthopedic clinic with a primary complaint of toe walking.

Methods: This was a retrospective cross sectional descriptive study carried out at a children's orthopedic clinic. Out of 638 patients who presented from 2004 to 2013 with a primary gait abnormality, 262 were documented as walking on their toes. Inclusion criteria were set as children who came to the clinic with a primary complaint of toe walking and were between 2 and 18 years of age. Underlying diagnoses such as cerebral palsy, autism spectrum disorder, Charcot-Marie-Tooth, or excluding a cause and listed as idiopathic were recorded and the proportion was calculated for each.

**Results:** Diagnoses of idiopathic toe walking, cerebral palsy, autism spectrum disorder and concomitant diagnoses with autism spectrum disorder accounted for 70.2%, 13.4%, 7.6% and 1.9% respectively – culminating in 93.1% of the associated diagnoses.

Conclusions: Understanding the relative likelihood of underlying diagnoses for children who present with toe walking has the potential to guide their treatment and increase access to appropriate services.

#### Introduction

Toe walking is used to describe a toe-toe, toe-heel, or early heel rise gait pattern persisting after two years of age [1]. Diagnoses associated with toe walking include cerebral palsy, myopathies, neuropathies, spinal dysraphism, and autism spectrum disorder (ASD) [2]. When other diagnoses are excluded as the cause of toe walking, children are given a diagnosis of idiopathic toe walking [3]. In addition, toe walking has been associated with developmental delay, motor delay and language delay [4]. When a child presents with a concern of toe walking, the probability that it is associated with cerebral palsy, ASD, other diagnoses, or is unknown (idiopathic) has not been reported. The purpose of this study is to describe which diagnoses are most commonly associated with toe walking in children who present with toe walking as the primary complaint to a children's orthopedic hospital.

#### Materials and methods

Local Institutional Review Board approval was obtained for this retrospective cross sectional descriptive study. The hospital database at a children's orthopedic hospital was queried for all patients from 2004 (implementation of electronic medical records) to 2013 and coded for gait abnormality as the primary concern (n=638). Coding was not specific enough to identify toe walking as the primary concern so these 638 charts were reviewed further to determine patients that should be included in the study. Those children with documented toe walking as a primary concern during examination or as a final diagnosis were included, resulting in a sample size of n = 262. The age range of participants was 2 to 18 years of age. Patients who presented with toe walking as a minor concern among multiple primary gait abnormalities were excluded.

Underlying diagnoses (e.g. cerebral palsy, ASD, etc.) were recorded for the 262 participants identified as children who toe walk. The diagnosis of ASD was based on physician and/or caretaker report, or recorded characteristics of children based on interactions during examinations that were noted by providers as consistent with ASD when there was a suspected autism spectrum disorder.

For each diagnosis, the proportion of the total of 262 participants was calculated. The 95% confidence intervals of each proportion were also calculated using binomial probabilities with Wilson's score interval method [5] using R statistics software (Hmisc package) [6,7]. The binomial probabilities confidence intervals provided improved estimates with smaller samples sizes (such as the diagnoses associated with toe walking).

#### Results

The disorders associated with toe walking, along with percentage and confidence intervals are shown in Table 1. Diagnoses of idiopathic toe walking, cerebral palsy, ASD and concomitant diagnoses with ASD accounted for 93.1% of the patients seen for toe walking. The percentage of patients with ASD (either alone or concomitant with another

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Diagnosis	Number of Subjects	Percentage	95% Confidence Interval
Idiopathic Toe Walking	184	70.2	64.4-75.4
СР	35	13.4	9.8-18.0
ASD	20	7.6	5.0-11.5
ASD and CP/FAS	5	1.9	0.8-4.4
Other	18	6.9	4.4-10.6
CMT	3		
Dopa responsive dystonia	2		
FAS	2		
Cerebral folate deficiency	1		
Clubfeet	1		
Congenital myopathy	1		
CMTC syndrome	1		
Down syndrome	1		
Incomplete spinal cord injury	1		
Myotonic dystrophy	1		
Neurofibromatosis	1		
Pierre-Robin syndrome	1		
Schizencephaly	1		
Williams syndrome	1		

 
 Table 1. The Prevalence of Disorders Associated with Toe Walking Along with Percentage and Confidence Interval (for Binomial Probabilities using Wilson score interval [5]).

 $\label{eq:asymptotic} ASD-autism spectrum disorder; CMT-Charcot-Marie-Tooth; CMTC-cutismarmoratatelangiectaticacongenita; CP-cerebral palsy; FAS-fetal alcohol syndrome$ 

diagnosis) was 9.5%. Fourteen diagnoses were grouped together in a single category and accounted for the remaining 6.9% of patients seen for toe walking.

The mean age of the participants at the time of first clinic visit was 7.1 years (range 2.1 – 18.1 years). Over 90% of children across all diagnostic groups initially presented at the clinic between 4 and 12 years old. Approximately 50% of the children with cerebral palsy, idiopathic toe walking and "other" diagnoses presented between 4 and 8 years of age. Children with ASD presented initially in nearly equal percentages (20%) for 2-year groupings between 2 and 12 years.

The number of male participants was 139 (53.1%) and the number of female participants was 123 (46.9%). Compared to the group of participants as a whole, there was a marked difference in ratio of sex within the ASD group of 19 male to 6 female participants (3.2:1 ratio).

## Discussion

This study established the relative prevalence of diagnoses among children who presented to a children's orthopedic hospital between 2004 and 2013 with a primary complaint of toe walking. The results indicated that if a child presents with a primary complaint of toe walking, they had a 93.1% probability of falling into 1 of 3 diagnostic categories: idiopathic, cerebral palsy, or ASD.

Previous research reported findings similar to the current study regarding diagnosis percentages for children who toe walk. A population based study determined that 4.9% of children had previously walked on their toes and stopped, or were still actively walking on their toes at 5.5 years, n = 70 out of a sample size of 1436 [8]. Of the 70 children who toe walk, 90% were considered to have idiopathic (63/70) and 10% neuropsychiatric (7/70) causes [8]. Neuropsychiatric diagnoses included autism spectrum disorder, unspecified developmental delay and attention-deficit/hyperactivity disorder. Children with motor disorders such as cerebral palsy and spina bifda were excluded from the sample of 70 identified children who walked on their toes, and

therefore there was no ability to compare to the current study. In the current study, the equivalent comparison was to consider only children who walked on their toes diagnosed with idiopathic toe walking and ASD. The current study found the prevalence of ASD was 10.2% when considering only children with idiopathic toe walking and those with ASD. Therefore, the current study and a previously reported study [8] found the prevalence of ASD was 10% for children that toe walk when considering only children with idiopathic toe walking and those with ASD. Further, the percentage of children who toe walk that were male (53% current study, 60% Engström) and female (47% current study, 40% Engström) were similar between the two studies. The ratio of male to female for children with a diagnosis of ASD in the current study (3.2:1) was broadly consistent with expectations reported for the ASD population as a whole (4.5:1) [9].

A limitation of this study was that it was conducted at a tertiary pediatric orthopedic center. Therefore, diagnosis of ASD relied on parental report or observation by an orthopedist, rather than a report in all cases by the diagnosing provider. Another limitation of the study could be that referral to our center was predicated on ability to provide orthopedic treatment. It was possible that those patients with severe ASD might not seek orthopedic treatment for toe walking (thus under-representing this group), as toe walking can be seen as a secondary problem by parents as they pursue other medical concerns [10]. However, it was likely that our pediatric orthopedic center provides services for a high percentage of children who toe walk in our catchment area, meeting the goal of determining underlying diagnosis associated with toe walking.

Among children presenting with toe walking, the likelihoods of various underlying diagnoses such as cerebral palsy, autism spectrum disorder, or idiopathic (diagnosis of exclusion) are not well understood. This study has presented estimates of these likelihoods for children presenting to an orthopedic center with a primary complaint of toe walking, thus enhancing the understanding of the basic question. Understanding the likelihoods of possible underlying diagnoses for children who toe walk beyond two years of age may help identify such diagnoses and guide treatment, thus improving overall access to needed services for this population.

## **Competing interests**

Mark L. McMulkin, Andi B. Gordon, Sarah Genung, Michael Huck, Paul M. Caskey, and Glen O. Baird declare that they have no competing interests.

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