



RESEARCH

Children presenting with toe walking: when should concern arise?

Parmak ucunda yürüme ile başvuran çocuklar: ne zaman endişe duyulmalı?

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Abstract

Purpose: This study aimed to evaluate children who present with tiptoe walking (TW) and to identify potential indicators of underlying medical conditions.

Materials and Methods: Out of the 248 patients who visited the Pediatric Neurology Outpatient Clinic for gait disturbances, 90 individuals aged 1-17 years were identified as exhibiting TW. After excluding those with systemic neurological diseases and pervasive developmental disorders (PDD), the study ultimately included 47 patients.

Results: Among the participants, 19 were female (40.5%) and 28 were male (59.5%). The mean age of the patients was 4.9 years (SD ± 3.53). When evaluating the etiology of TW, 30 patients (63.8%) were found to have idiopathic tiptoe walking (ITW), 8 (17%) had cerebral palsy (CP), 6 (12.7%) had Achilles tendon shortness (ATS), 2 (4.2%) had hereditary spastic paraplegia (HSP), and 1 (2.1%) had syringomyelia. Imaging was not performed for 17 patients (36.1%), while cranial magnetic resonance imaging (MRI) was conducted for 13 (27.6%), spinal MRI for 2 (4.2%), and both cranial and spinal MRI for 15 (31.9%). Pathology was detected in 5 of the patients who underwent imaging (10.6%): 4 (8.5%) showed hypoxic-ischemic processes, and 1 (2.1%) showed syringomyelia.

Conclusion: In cases where patients exhibit normal neurological examinations and neuromotor development, TW is often identified as idiopathic. However, for patients presenting with risk factors in their personal or family history, delays in neuromotor milestones, or abnormal neurological findings, a diagnosis should be pursued at an earlier stage.

Keywords: Idiopathic toe walking, cerebral palsy, children

Öz

Amaç: Bu çalışmanın amacı parmak ucunda yürüme (PUY) ile başvuran çocukları değerlendirmek ve altta yatan tıbbi durumların potansiyel göstergelerini belirlemektir.

Gereç ve Yöntem: Çocuk Nöroloji Polikliniği'ne yürüme bozukluğu nedeniyle başvuran 248 hastadan 1-17 yaş arası 90'ında parmak ucunda yürüme tespit edildi. Sistemik nörolojik hastalıkları ve yaygın gelişimsel bozuklukları (YGB) olanlar dışlandıktan sonra, çalışmaya 47 hasta dahil edildi.

Bulgular: Hastaların 19'u kız (%40,5) ve 28'i erkekti (%59,5). Hastaların ortalama yaşı 4,9 yıldır (SD ± 3,53). PUY etiyojisi değerlendirildiğinde, 30 hastada (%63,8) idiyopatik parmak ucunda yürüme (IPUY), 8 hastada (%17) serebral palsi (SP), 6 hastada (%12,7) Aşil tendon kısalığı (ATK), 2 hastada (%4,2) herediter spastik parapleji (HSP) ve 1 hastada (%2,1) siringomiyeli saptandı. Hastaların 17'sine (%36,1) görüntüleme yapılmazken, 13'üne (%27,6) kraniyal manyetik rezonans görüntüleme (MRG), 2'sine (%4,2) spinal MRG ve 15'ine (%31,9) hem kraniyal hem de spinal MRG yapıldı. Görüntüleme yapılan hastaların 5'inde (%10,6) patoloji tespit edilmiş olup bunlar; 4'ünde (%8,5) hipoksik-iskemik süreçler ve 1'inde (%2,1) siringomiyeli olarak sıralandı.

Sonuç: Hastaların nörolojik muayenelerinin ve nöromotor gelişimlerinin normal olduğu durumlarda, TW sıklıkla idiyopatik olarak tanımlanır. Bununla birlikte, kişisel veya aile öyküsünde risk faktörleri, nöromotor kilometre taşlarında gecikmeler veya anormal nörolojik bulgular ile başvuran hastalar için tanı daha erken bir aşamada takip edilmelidir.

Anahtar kelimeler: İdiyopatik parmak ucunda yürüme, serebral palsi, çocuk

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INTRODUCTION

Toe walking (TW) is a common condition encountered in pediatric and pediatric neurology outpatient clinics. The term describes the pattern whereby children walk on their toes beyond the age at which they would normally develop a normal gait pattern. Although many children with TW have no other identifiable abnormality, clinicians should have a high index of suspicion for possible underlying pathologies. Common conditions associated with TW include cerebral palsy (CP), muscular dystrophies, and neuropathies¹. Other neurological disorders of childhood, such as autism spectrum disorder (ASD) and global developmental delay (GDD), may also present with TW²⁻⁴. Various focal pathologies including traumatic injuries or tumours are predominantly associated with unilateral disease⁵.

Idiopathic toe walking (ITW) is a term used when all identifiable medical conditions are ruled out; generally, the reason for this walking pattern's initiation and continuation remains unclear^{5,6}. ITW may be associated with Achilles tendon shortness (ATS), which may limit ankle mobility; however, most children do not have evidence of such contracture³. ITW is generally considered a diagnosis of exclusion and usually carries no long-term functional consequences. Prevalence of ITW is variable with literature quoting the rates from 7% to 24% in the childhood population, although recently a large-scale study quoted prevalence of approximately 5%⁷. Family history is positive in 10% to 85% of cases, with an autosomal dominant pattern of inheritance^{4,8}. ITW is more common in boys than in girls^{8,9}.

Despite its frequency, there are relatively few studies on the clinical approach to TW. Existing studies primarily focus on specific conditions associated with TW, such as CP and ASD, but there is a notable gap in comprehensive assessments of TW covering a wide range of potential etiologies. In addition, there is limited guidance on distinguishing ITW from other causes of TW, which is crucial for appropriate clinical management.

Previous studies on TW and ITW have predominantly been conducted by the orthopedics, child psychiatry, and physical medicine and rehabilitation departments. Orthopedics and physical therapy tend to address the issue from a mechanical and functional perspective, whereas child psychiatry focuses on the relationship with ASD. However, we

did not find any study in the literature examining the approach to TW by a pediatric neurology department.

Our hypothesis is that if a child's neurological development and neurological examination (apart from TW) are normal, there is usually no significant underlying disease, and TW tends to resolve over time. In contrast, in children with delayed neuromotor milestones and/or pathological findings in the neurological examination, along with TW, TW should be taken more seriously and early investigations must be carried out to exclude possible pathological causes.

The clinical impact of ITW can range from benign, self-limited conditions to more serious underlying disorders. It is crucial to differentiate ITW from other potential etiologies through comprehensive evaluation, including detailed history, physical examination, and appropriate diagnostic tests. This study aimed to evaluate patients presenting to the child neurology outpatient clinic with TW complaints and identify possible clues for underlying pathologies.

MATERIALS AND METHODS

Study design and sample

In this retrospective study, we initially screened 248 patients who presented with gait disturbances at the Pediatric Neurology Outpatient Clinic of Ankara Etlik City Hospital, a tertiary care hospital, between February 2023 and February 2024. These patients included individuals with various abnormal gait complaints, such as delayed walking, inability to walk, ataxic gait, and foot drop. From this initial group, 90 individuals aged between 1 and 17 years were identified as having TW.

We excluded those patients who already had one of the diagnosis of CP, HSP, GDD, or any neurodegenerative diseases to focus particularly on TW complaints. Patients with incomplete records, not followed up, or still in the process of diagnosis without a definitive diagnosis were also excluded. This resulted in a final sample size of 47 participants who presented solely with TW complaints.

The power analysis was designed to achieve an 80% power ($\beta = 0.20$) and a 5% significance level ($\alpha = 0.05$). Based on these parameters, a minimum sample size of 45 patients was required. Ultimately, 47 participants were included in the study, ensuring

adequate power for detecting differences in the prevalence of TW and its associated conditions.

All procedures regarding data extraction and analysis were conducted according to relevant ethical guidelines and standards for clinical research adopted by the hospital. Ethical approval for this study was taken from Ankara Etlik City Hospital Ethics Committee on March 6, 2024, with the number AEŞH-BADEK-2024-184.

Data collection

Demographic information, such as age and gender, was recorded from patient files as documented at the time of their initial presentation. The duration of TW was primarily reported by parents or caregivers during clinical visits and documented in the medical records. In cases where this information was unclear, clinicians reviewed previous medical records and consulted with parents for confirmation. Personal and family history, including prematurity, intensive care hospitalization, seizures, cerebral hemorrhage, and family history of neurological disease, was collected from medical records and corroborated with parental reports during clinical evaluations.

Neurological examination and diagnostic workup

Neurological examination findings, such as deep tendon reflexes (DTRs), gait patterns, and other abnormalities, were assessed and documented by the examining pediatric neurologists.

Neuroimaging results from cranial MRI, spinal MRI, and combined cranial and spinal MRI were reviewed and recorded based on radiology reports in the patient files. Additionally, results from tests such as electromyography (EMG) and genetic testing for patients with suspected HSP were obtained from the respective diagnostic reports in the medical records.

Diagnostic criteria and outcome measures

The methodology used for the diagnosis of various underlying pathologies associated with TW included a combination of clinical evaluations, neuroimaging studies, and diagnostic tests. CP was diagnosed based on clinical findings such as spasticity, abnormal reflexes, and delayed motor milestones, supported by cranial MRI findings of hypoxic-ischemic lesions. Muscular dystrophies and neuropathies were ruled out through clinical examination, family history, and

specific tests such as creatine kinase levels, EMG, and genetic testing when necessary. HSP was diagnosed based on clinical symptoms, EMG findings, and confirmed through genetic testing.

Final diagnoses included ITW, CP, ATS, HSP, and syringomyelia. It also documented the treatment and follow-up results such as physical therapy, surgical interventions, and the use of botulinum toxin.

Statistical analysis

Statistical analyses were done with the SPSS 20.0 package program. Associations of categorical variables such as sex versus neurological abnormalities were tested using the chi-square test. For continuous variables, such as age, the Spearman correlation coefficient was used to assess the relationship between variables. The significance level was set at 0.05, with a p-value of less than 0.05 considered indicative of a statistically significant relationship. Additionally, descriptive statistics, including means and standard deviations, were calculated for demographic variables and clinical findings.

RESULTS

Of the patients studied, 19 were female (40.5%) and 28 were male (59.5%) and there was no statistically significant difference between genders ($p > 0.05$). The mean age of the patients was 4.9 years ($SD \pm 3.53$). Detailed gait evaluation revealed that 38 patients (80.8%) had walked on their toes since they started walking.

Identifiable pathologies are given in Table 1. Idiopathic disease was observed in 30 patients (63.8%), cerebral palsy in 8 patients (17%), ATS in 6 patients (12.7%), hereditary spastic paraplegia in 2 patients (4.2%), and syringomyelia in 1 patient (2.1%). All patients diagnosed with CP had findings of previous hypoxia on cranial MRI. All patients diagnosed with ATS were evaluated and diagnosed by the orthopaedics and/or physical therapy and rehabilitation department. One of the two patients with HSP was diagnosed based on EMG findings, while the other patient was diagnosed genetically.

Consideration of past medical and family history revealed that 8 patients had a history of prematurity, all of whom were diagnosed with cerebral palsy (CP). Among those diagnosed with ITW, 6 patients were found to use a baby walker (BW). Unfortunately, a

family history of ITW was not questioned in any of the files we retrospectively analysed. But none of the patients reported a family history of neurological diseases.

All patients exhibited tiptoe walking during their first neurological examination. However, additional neurological abnormalities were found in 11 (23,4%) patients. DTRs were hyperactive in 5 patients (10,6%), 3 patients (6,3%) displayed a scissoring gait

pattern with hyperactive DTRs, 2 patients (4,2%) had a stepping gait with hyperactive DTRs, and 1 patient (2,1%) showed increased joint laxity. Of the 5 patients with hyperactive DTRs, 4 (80%) were diagnosed with CP, and all 3 patients (100%) with a scissoring gait and hyperactive DTRs were diagnosed with CP. Two patients (100%) with a stepping gait and hyperactive DTRs were diagnosed with hereditary spastic paraplegia (HSP) (Table 2).

Table 1. Etiologies of patients with toe-walking

Etiology	Frequency
Idiopathic toe-walking	30 (63.8%)
Cerebral palsy	8 (17%)
Achilles tendon shortness	6 (12.7%)
Hereditary spastic paraplegia	2 (4.2%)
Syringomyelia	1 (2.1%)
Total	47 (100%)

Table 2. Neurological abnormalities and final diagnosis

Patient Characteristics	Number of Patients	Diagnosis
Hyperactive DTRs	5	4 diagnosed with CP
Scissoring Gait + Hyperactive DTRs	3	All diagnosed with CP
Stepping Gait + Hyperactive DTRs	2	Diagnosed with HSP
Increased Joint Laxity	1	-
Total	11	

:DTRs:Deep Tendon Reflexes, CP: Cerebral Palsy, HSP: Hereditary Spastic Paraplegia

Imaging was not performed for 17 patients (36.1%) as there were no positive examination findings, and their mental and motor development was normal except for TW. Cranial MRI was conducted on 13 patients (27.6%), spinal MRI on 2 patients (4.2%), and both cranial and spinal MRI on 15 patients (31.9%). Positive pathological MRI findings were observed in 5 patients (10.6%), with 4 (8.5%) displaying hypoxic-ischemic processes and 1 (2.1%) exhibiting syringomyelia. EMG was carried out on 2 patients; one was normal, while the other was consistent with HSP. The patient with normal EMG and genetically diagnosed HSP was a 5-year-old boy who had been TW for two years. On neurological examination, in addition to TW, he had a stepping gait and hyperactive DTRs. The patient with normal EMG and genetically diagnosed HSP was a 5-year-old boy who had been TW for two years. On neurological examination, in addition to TW, he had a stepping gait and hyperactive DTRs. In the family history, it was discovered that his parents were consanguineous, and his older brother was being followed up with a diagnosis of HSP. Therefore, the patient was referred to the Pediatric Genetics

department, where he was found to have a heterozygous c.1632_1641del p.(Tyr544Ter) stop-gain variant in the SPAST gene (NM_014946.4). This variant is classified as a possible pathogen. The patient was thus diagnosed with autosomal dominant spastic paraplegia 4 (182601). Validation from the patient and segregation in family members is planned by the Pediatric Genetics department.

In terms of treatment, physiotherapy and rehabilitation were prescribed for 32 patients. Among these, 2 patients underwent Achilles tendon lengthening surgery due to ATS, and one patient was treated with botulinum toxin. The remaining 15 patients were not undergoing any treatment. These patients were evaluated together with the physiotherapy and rehabilitation department, and regular follow-up was recommended.

DISCUSSION

TW is one of the most common complaints in pediatric and pediatric neurology outpatient clinics. Families often worry about how long toe walking will

persist and whether it indicates a serious underlying condition. This also creates uncertainty for physicians regarding which patients require detailed investigations and which can be monitored through regular follow-up visits. In this study, we found that ITW was the most common cause of TW among the patients, accounting for 63.8% of cases. This finding aligns with existing literature, which suggests that ITW is a prevalent condition in pediatric patients presenting with toe walking. Our study contributes to the current understanding by highlighting the necessity of differentiating ITW from other potential underlying conditions through comprehensive evaluations. The presence of ITW as the predominant diagnosis underscores the importance of considering this condition in the differential diagnosis when children present with TW, especially in the absence of neurological abnormalities.

Our study also identified other underlying conditions such as CP, ATS, HSP, and syringomyelia in 36.2% of the patients. These findings emphasize the critical need for a thorough diagnostic workup, including neuroimaging and genetic testing, in patients with abnormal neurological findings or risk factors in their medical history. The detection of hypoxic-ischemic processes on MRI in patients with CP and the use of EMG and genetic testing for diagnosing HSP highlight the role of advanced diagnostic tools in identifying specific etiologies of TW.

TW is quite common as part of normal neurological development. In toddlers, some of these may resolve in a short time, while others may persist until the age of 3 years or even longer. According to previous studies, TW is estimated to affect 7%-24% of the childhood population. However, a more recent large-scale population study reported a prevalence of approximately 5%¹⁰. Family history is positive in 10-85% of cases, with autosomal dominant inheritance^{4,8}. No gender difference has been reported for TW, whereas ITW is more common in boys than in girls^{8,9}.

The most common cause of TW identified in our study was ITW, observed in 30 patients (63.8%). ITW is a broad term used for conditions where tiptoe walking is not linked to any medical condition. Before confirming the diagnosis, it's essential to exclude potential differential etiologies, including CP, peripheral neuropathy, and myopathy^{5,10}.

Given that ITW is a diagnosis of exclusion, a comprehensive history is crucial to thoroughly

explore potential predisposing factors for CP and other neurological conditions. Additional investigations are warranted if there's a history of prematurity, delays in neuromotor development, a family history of neurological disorders, or abnormal findings in neurological examinations. MRI and EMG are important tests in this regard and may be particularly beneficial for these assessments^{7,10}. In our study, MRI was performed on 30 patients, and 2 patients underwent EMG.

The underlying diseases other than ITW were CP, ATS, HSP, and syringomyelia. CP is the most common motor disability in childhood, occurring at a rate of 1 in every 1000 births across Europe. The risk factors for CP cover a broad timeline, including the period before and around conception, throughout pregnancy, the perinatal phase, and up to 2 years of age. Factors known to have the potential for leading to CP encompass genetic variations, congenital anomalies, preterm birth, kernicterus, intrauterine growth restriction, infections, hypoxic-ischemic events, cerebrovascular insults during pregnancy and infancy, along with accidental or non-accidental brain injuries. These factors can converge in various ways to injure motor pathways leading to CP¹¹. Spastic CP, the predominant variant of the condition, is characterized by symptoms including increased muscle tone, weakness of the affected extremities, and atypical walking patterns¹². In our study, 8 patients (17%) were diagnosed with CP and all of these patients had a history of premature birth and hyperactive DTRs.

Engström and colleagues studied a large population of healthy children and divided TW into two distinct groups. The predominant group displayed TW without any ankle contracture, indicating that tiptoeing in these cases is likely a temporary phenomenon. Remarkably, 79% of these children naturally ceased tiptoeing by the age of 10 without developing secondary ankle contractures. The researchers recommend identifying this phenomenon as ITW. Conversely, the second, the small group of younger children, exhibited early contracture of the triceps surae muscle and showed favorable outcomes with early intervention. For these cases, the team advocates for maintaining the precise diagnosis of congenital short tendo calcaneus, commonly referred to as ATS³. In our study, 6 patients exhibited ATS. Out of these, only one had a history of premature birth; however, this patient's DTRs and both cranial MRI and spinal MRI were normal. The other 5

patients had no noteworthy personal or family history that might have explained their medical condition.

HSP, characterized by spasticity and weakness in the lower extremities, can be inherited in several genetic patterns, including autosomal dominant, autosomal recessive, X-linked, or maternal (mitochondrial) traits. When symptoms appear in early childhood and have a non-progressive course, they resemble spastic diplegic CP¹³. In our study 2 patients (4.2%) were diagnosed as HSP. They both had hyperactive DTRs and lower extremity weakness. One of the two patients diagnosed with HSP did not have a history of consanguinity, while the other did.

Not all children with ASD engage in TW. The underlying mechanisms of this behavior are not fully understood, and its occurrence varies in intensity. Two main theories have been proposed: TW may result from problems with sensory integration and may be a residual behavior from a past tonic labyrinth reflex when lying on the patient's back. Symptoms of sensory integration disorder are not seen in every child with autism, and when they do appear, they manifest with varying levels of intensity. Either an overreaction or underreaction to sensory stimuli or an unusual focus on sensory elements of the surroundings is now recognized as part of the diagnostic criteria for ASD¹⁴. In our study, none of the patients were diagnosed with ASD. The reason may be that they are firstly evaluated in general pediatric outpatient clinics and are directed to the child mental health department before coming to our clinic.

In our study, all patients diagnosed with CP and HSP had abnormal neurological examinations. Among the patients diagnosed with ITW, only one had detectable DTRs, while the neurological examination of the rest showed no abnormalities. Thus, patients with normal neurological examinations can be followed up relatively more safely. On the other hand, those with pathological findings on neurological examination need to be evaluated in more detail.

The management of ITW varies, with options ranging from orthotics and physical therapy to botulinum toxin injections, serial casting, and surgical procedures to lengthen the muscles or tendons. Non-surgical methods, while safe from operative risks, show limited success and have uncertainty regarding the durability of achieving a normal walking pattern. In contrast, surgical intervention, particularly Achilles tendon lengthening, has been consistently effective

without reported serious complications, offering significant improvements or complete resolution in toe-walking cases⁶. In our study, physiotherapy and rehabilitation were prescribed for 32 patients. Among these, 2 patients underwent Achilles tendon lengthening surgery by the orthopedics and traumatology department due to ATS, and one patient was treated with botulinum toxin. The remaining 15 patients were not receiving any treatment.

Generally, by age 10, 79% of children with TW will naturally transition to a normal walking pattern without any intervention and don't show any signs of ankle contractures. Neurodevelopmental comorbidities are frequently observed in children who persist in TW³. Since our study was a retrospective study conducted over a short period of time, we do not have any information about how long patients diagnosed with ITW continued to tiptoeing

In a recent review evaluating the etiology of ITW, BW was not listed among the causes¹⁵. However, in some studies directly analyzing the possible relationship between BW and ITW, it has been reported that BW may be a risk factor for ITW. In a study by Mete et al.¹⁶ involving 193 infants aged 8-84 months, gait disorders and toe-tip walking were found to be higher in children who used a BW for at least 30 minutes a day for more than one month. Krirova et al.¹⁷ included a total of 749 infants in their study on 3 retrospective cohorts, and 363 of them used BW. The use of BWs was identified as a factor contributing to the formation of the toe-walking pattern and as a possible causal factor of ITW. Similarly, BW use was present as a predisposing factor in 6 of the patients diagnosed with ITW in our study. These results shed light on the need for more studies on the possible cause-and-effect relationship between ITW and BW use and the importance of stating that BW should not be used in routine pediatric follow-up visits.

For children with ITW, a detailed history and physical examination should be the first steps. This includes assessing the family history of similar gait patterns, neuromotor development milestones, and any signs of neurological abnormalities. When the physical examination and history do not reveal any concerns, ITW can be considered, but it is vital to monitor these patients regularly to detect any evolving conditions.

In cases where neurological abnormalities are present or there are risk factors such as prematurity,

developmental delays, or a family history of neurological diseases, further investigations are warranted. Cranial and spinal MRI, as well as EMG, can provide essential insights and help in differentiating conditions like CP or HSP from ITW. Our study demonstrated that patients with CP often had a history of hypoxia detectable on MRI, while HSP could be confirmed through genetic testing or EMG findings.

A multidisciplinary approach involving pediatric neurology, orthopedics, and physical therapy is recommended for managing these patients. Early intervention, particularly for those with ATS, through physiotherapy, and in some cases, surgical procedures, can prevent long-term complications and improve gait patterns. Regular follow-up is crucial to reassess the diagnosis and adjust the treatment plan as necessary.

Our study has some limitations, including that it was conducted in a single center and over a short period of one year, the lack of standardization in which patients would be imaged due to the retrospective nature of the study, and the small number of patients. If our study had been designed to cover a longer time interval, perhaps diagnostic changes could have been observed. For example, we might have seen that patients diagnosed with CP evolved into different diagnoses, such as HSP, over time. Additionally, we could have obtained data on how long patients diagnosed with ITW walked on tiptoe.

This study highlights the importance of a comprehensive diagnostic approach to children presenting with TW, particularly in distinguishing idiopathic cases from those associated with underlying neurological conditions. While our findings underscore the prevalence of ITW as the most common cause, the identification of cases linked to CP, HSP, and other conditions emphasizes the need for careful evaluation.

In conclusion, early diagnosis is advisable for those with risk factors in their personal or family history, neuromotor development delay, or unusual neurological exam results. Also, persisting TW after the age of 2 warrants further investigation for underlying neuromuscular conditions. Such persistence may indicate the early signs of neuromuscular disorders, necessitating evaluations for conditions like CP or muscular dystrophy.

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