

Clinical Research

Idiopathic Toe Walking

Family Predisposition and Gender Distribution

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Abstract: *Current literature is inconsistent concerning the causes and the frequency of idiopathic toe walking (ITW). Available studies vary widely in their results. The aim of this study is to supply gender-related data particularly regarding the genetic influence on toe walking. Methods. The ITW patterns of 836 children were recorded and analyzed during a period of 4 years. Questionnaires and clinical measurements were evaluated along with clinical tests, assessing the occurrence and severity of toe walking. Information about the incidence of toe-walkers in the family was recorded. Results. Of the 836 toe-walkers, 64% were boys and 42% had a positive family history (PF-TW). About 60% of the PF-TW children had fathers with a positive toe-walking pattern. PF-TW children were on average half a year younger than children with a negative family predisposition (NF-TW). Conclusions. This study shows that a genetic component might be factor in toe walking. PF-TW children were more severely affected in all performed clinical tests than NF-TW children.*

Levels of Evidence: Prognostic,
Level IV

Keywords: toe walking; idiopathic; family history; genetic; heredity; forefoot; walking

Toe walking is a common gait pattern that is found in various pediatric pathologies such as muscular dystrophy or cerebral palsy.¹ Idiopathic toe walking is diagnosed if this gait abnormality is present and a medical condition associated with toe walking can be excluded.¹

Toe-walkers start showing their typical gait as they learn to walk. Nevertheless they are able to support their entire foot on the ground on request or when concentrating on walking.^{2,3} ITW was first mentioned in 1967 by Hall et al.⁴ They described it as caused by a congenitally short Achilles tendon; recent studies apply the term *idiopathic toe walking* (ITW) to children who walk on their toes in the absence of any medical condition.⁴⁻⁶ The etiology of ITW is unknown. Several authors have

described ITW as the persistence of an early gait pattern in which the children ambulate on their toes.⁵ One etiological factor suggested is hereditary transmission. In 1973, Levine⁷ first described a case study with 5 family members all of whom had a congenitally short Achilles tendon.

Katz and Mubarak⁸ in 1984 reported 7 patients who all had a positive family history and suggested a “genetic” Achilles tendon contracture as a possible cause.

“Toe walking is a common gait pattern that is found in various pediatric pathologies such as muscular dystrophy or cerebral palsy.”

They also found that ITW predominates in boys with a family history of toe walking and hypothesized an inheritance pattern with autosomal dominance.⁸ In the past years, several studies have noted hereditary factors as a reason for toe walking with ranges from 30% to 71%.^{6,9-12} In these studies, however, only

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Table 1.

Age Distribution of Toe-Walkers and Normal Walkers.

Age (Years)	Toe-Walkers (TW)			Normal Walkers (NW)		
	Total	Males	Females	Total	Males	Females
N	836	537	299	55	30	25
Mean	6.2	6.2	5.9	6.2	6.5	5.9
Standard error of mean	0.09	0.12	0.16	0.41	0.52	0.64
Standard deviation	2.7	2.7	2.8	3.0	2.9	3.2
Minimum	2.0	2.0	2.0	2.0	2.0	2.0
Maximum	13.9	13.9	13.8	13.6	13.6	13.4
Homogeneity test: $P(TW\text{-total, NW-total}) = .706$ (Kolmogorov-Smirnov)						

a small number of cases were evaluated. Furthermore, clinical characteristics like reduced range of motion of the upper ankle joint are still under discussion and several studies have mentioned a severe decrease of range of motion for ITW.^{11,13} This is often the only clinical characteristic that is used to describe toe walking.

During gait analysis in children with ITW, electromyography shows that there are children that develop a toe-toe gait pattern.⁵ There is an early "onset of gastrocnemius activity at the end of the swing phase, abnormal activity of the tibialis anterior, overlapping activity of gastrocnemius and tibialis anterior."⁵

The primary purpose of this study is to show a possible inheritance pattern of toe walking in a large observational group of children with ITW. Another objective of this study is to present statistically significant differences between toe walking with a positive family history (PF-TW) and toe walking with a negative family history (NF-TW).

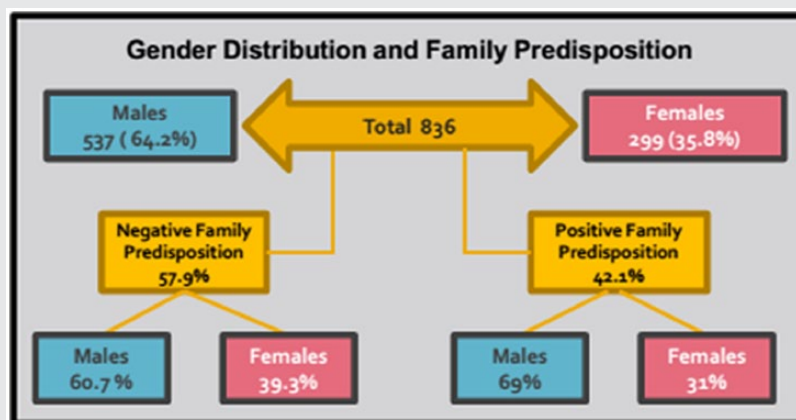
Methods

Procedure and Participants

Between June, 2006 and June, 2010, 923 children between 24 months and 13 years of age were referred to our center

Figure 1.

Gender distribution and family predisposition in percentages.



for treatment of toe walking. All the children were diagnosed with idiopathic toe walking, they had been referred exclusively by local pediatricians. All children had been permanently walking on their toes for at least 6 months. The referring pediatricians had not found any signs or evidence of underlying neurological or orthopedic conditions.

Twenty-four children were excluded from the study because of the late onset of their toe walking and 12 children were excluded because the

toe-walking pattern had been present for less than 6 months. Questionnaire 1 shows the standardized history (see the appendix).

The first examination and the follow-up examinations were performed by 5 different investigators who were trained and supervised by the first author. All measurements were taken in accordance with a standardized examination guide. Retests with healthy volunteers and toe walking were carried out and indicated an interrater and retest reliability of more than 80%.

Table 2.

Gender Distribution and Family History.

	Statistics of Gender Distribution and Family History				$\chi^2(P)$	WM(P) ^a
(1)	N 836 TW					
	Mean 6.25 ± 0.09 (1.5) ^b					
	Males 537 (64.2 ± 3.2) ^c , females 299 (35.8 ± 3.2)				<.001	
	Mean: 6.42 ± 0.12 (1.9), 5.93 ± 0.16 (2.7)					.028
	Negative Family History (NF)		Positive Family History (PF)			
(2)	NF-TW 484 (57.9 ± 3.3)		PF-TW 352 (42.1 ± 3.3)		<.001	
	Mean: 6.45 ± 0.12 (2.9)		5.96 ± 0.15 (2.4)			.045
	Males	Females	Males	Females		
	295 (60.7 ± 4.4)	189 (39.3±4.4)	242 (69.0±4.8)	110 (31.0±4.8)	.016	
	Mean: 6.67±0.16 (2.3)	6.11±0.20 (3.3)	6.14±0.18 (2.9)	5.58±0.25 (4.3)		.034
						.221
(3)	N 348, PF-TW					
	Males 240 (69.0 ± 4.8), females 108 (31.0 ± 4.8)				<.001	
	PF-TW With Paternal Relatives		PF-TW With Maternal Relatives			
	207 (59.5 ± 5.2)		141 (40.5 ± 5.2)		<.001	
	Mean: 6.01 ± 0.20 (3.3)		Mean 5.95 ± 0.23 (3.9)			.666
	Males	Females	Males	Females		
	147 (71.0 ± 6.2)	60 (29.0 ± 6.2)	93 (66.0 ± 7.8)	48 (34.0 ± 7.8)	1.0346	

^aMann-Whitney *U* test and Kolmogorov-Smirnov test.^bYears ± Standard Error of Mean (%)^cNo. (% ± 95% CI).

TW, toe walker; NF-TW, Toe Walkers with a Negative Family Predisposition; PF-TW, Toe Walkers with a Positive Family Predisposition.

Examination

Parent's Questionnaire. For this study, the results of 3 predefined questions from the standardized history form were evaluated (see questionnaire 1 in the appendix). A total of 75 participants were excluded from the study because of conflicting answers in the pretest questionnaire and the follow-up test questionnaire or because these had not been completed properly.

Clinical Examination. Five clinical tests were used in order to find out if there were clinical differences between children with a positive family

predisposition and children with a negative family predisposition, tests taken from Idiopathic Toe Walking, tests and family predisposition¹⁸ (see clinical examination in the appendix).

- Spin test performance
- Walking test performance
- Heel walking test performance
- Range of motion (ROM) of the ankle joint in dorsiflexion
- Angle of the lumbar lordosis

Statistical Analysis. For statistical analysis, SPSS18 was used: chi-square and/or Fisher-Yates test, $\chi^2(P)$,

Kolmogorov-Smirnov test, KS(*P*), Wilcoxon-Mann-Whitney Test, WM(*P*).

Results

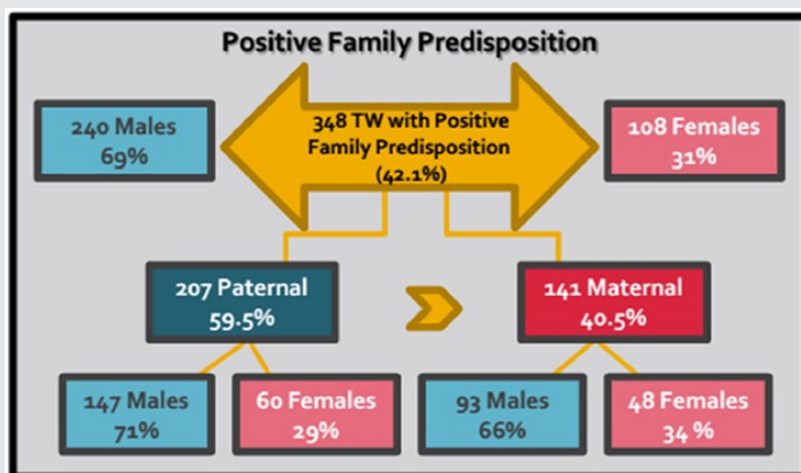
Gender Distribution and Family History

The age distribution is presented in Table 1. Figure 1 and Table 2 show the results of gender distribution and family history.

Gender Distribution. The average age of the 836 toe-walkers was 6.25 years. There were 537 (64.2%) boys and 299

Figure 2.

Positive family predisposition results Data in percentages.



(35.8%) girls. The average age of the boys was 6.42 years, and of the girls was 5.93 years. Boys were significantly more often affected by toe walking than girls (difference 28.4%) and they were significantly older (on average 0.49 years) than girls at the onset of treatment for toe walking.

Positive or Negative Family

Predisposition. A total of 484 (57.9%) of the 836 TW children had a negative family history (NF-TW) and 352 (42.1%) of them had a positive family history (PF-TW). The difference between the number of PF-TW and NF-TW children (15.8%) was significant. The PF-TW children were significantly younger than NF-TW children (on average 0.49 years).

Overall, 295 (60.7%) of the NF-TW children and 242 (69.0%) of the PF-TW children were boys, 189 (39.3%) of the NF-TW and 110 (31.0%) of the PF-TW were girls. The number of male PF-TW was significantly higher (difference 8.3%) than the number of male NF-TW. The number of male PF-TW was significantly greater (difference 38.0%) than the number of female PF-TW.

In the NF-TW group, on average, the 295 boys were aged 6.67 years and the 189 girls were aged 6.11 years. In the

NF-TW group, boys were significantly older than girls (on average 0.56 years, $P = .034$). In the PF-TW group, the average age of the 242 boys was 6.14 years, and of the 110 girls was 5.58 years. In this group, the boys were on average 0.48 ($P = .221$) years older than the girls; this difference, however, was not significant.

A Family Inheritance Pattern. When asked "Which of your family members is a TW or was one as a child?" 348 of the 352 PF-TW answered clearly with "father," "mother," "relatives of father," or "relatives of mother." Two boys and 2 girls were excluded because they only had 1 sibling and had no maternal or paternal relatives who were toe-walkers.

Out of the PF-TW group of 240 (69.0%) were boys and 108 (31.0%) were girls, this difference is significant (difference 38.0%).

In the PF-TW group, 207 out of 348 (59.5%) had paternal relatives and 141 (40.5%) maternal relatives who were also toe-walkers. The number of "TW with paternal relatives" was significantly greater than the number of "TW with maternal relatives" (difference 19.0%). The average ages of the "TW with paternal relatives" and the "TW with

maternal relatives," however, did not significantly differ.

In the PF-TW group, 147 (71.0%) with paternal relatives were male and 60 (29.0%) were female, while 93 (66.0%) with maternal relatives were male and 48 (34.0%) were female.

The number of "TW with paternal relatives" was greater than the number of "TW with maternal relatives" (difference 5%). This difference, however, was not significant. All average ages of boys and girls with paternal and maternal relatives were not significant and are not displayed in Table 2. Figure 2 shows the positive family predisposition results.

Discussion

The etiology of toe walking is a topic that has often been debated in recent decades. In 1967, Hall et al⁴ first suggested a positive family history and reported on 2 families that had more than 1 child affected by toe walking. In 1973, Levine⁷ published a study on a family with 5 members who toe-walked. In 1984, Katz and Mubarak⁸ mentioned a positive family history in 7 of the study samples and found an autosomal dominant pattern of inheritance with variable expression for toe walking. In their review on ITW, Sala et al⁵ reported 3 more studies from Stricker and Angulo,⁶ Kalen et al,⁹ and Shulman et al,¹⁰ which investigated the family history factor. Eiff et al¹⁴ reported a range of between 30% and 71% for this factor in several studies. All the study samples in the previous reports had, however, been quite small.

This is the first study that presents the findings of family history for a large observation group differentiated by gender distribution. The findings in this study point out statistical evidence that ITW possibly underlies hereditary transmission. A total of 352 (42.1%) of these children had family members who were either toe-walkers or used to toe-walk as children; 484 (57.9%) of them did not have a direct family association; however, a possible hereditary factor cannot be excluded (Table 2).

Table 3.

Parents' Questionnaire "First Occurrence of Toe Walking".

Question	PF-TW, n (% ± 95% CI); N = 352	NF-TW, n (% ± 95% CI); N = 484	$\chi^2(P)$
Onset of toe walking?			
Since onset of walking	245 (69.6 ± 4.8)	295 (61.0 ± 4.3)	.010
Months after onset of walking	51 (14.5 ± 3.7)	90 (18.6 ± 3.5)	.134
Years after onset of walking	50 (14.2 ± 3.6)	89 (18.4 ± 3.5)	.111
Only recently	6 (1.7 ± 1.4)	10 (2.1 ± 1.3)	.802

Furthermore, boys were significantly more often affected than girls in both the PF-TW and the NF-TW groups. The results (Table 2 and Figure 1) show that 537 (64.2%) of all 836 TW were male and 299 (35.8%) were female. 242 (69.0%) of the 352 PF-TW group were male and 110 (31.0%) female. 295 (60.7%) of NF-TW group were male and 189 (39.3%) were female. The difference of 8.3% between male PF-TW and male NF-TW was significant ($P = .016$).

According to these results, we can conclude that boys have a significantly higher predisposition to be affected by TW than girls (PF-TW and NF-TW). The proportion of boys was significantly higher in the PF-TW group than in the NF-TW group.

There were 537 TW participants with no family history. Perhaps there is not just an autosomal dominant factor, but is also another unknown factor that causes the TW condition.

The PF-TW had been investigated to find out whether gender-specific differences exist and whether genetic factors are involved (Table 2, lower part). It was found that fathers passed on toe walking to 207 (59.5%) of their children ($P < .001$); while mothers passed on toe walking to only 141 (40.5%) of their children ($P < .001$). Furthermore, it was also found that there were 5% more affected boys with a positive paternal predisposition than boys with a positive maternal predisposition; however, these differences were not significant ($P =$

.346). We cannot prove that fathers pass on toe walking significantly more often to their sons than to their daughters, although a tendency may be assumed.

In addition to family history, several characteristics of idiopathic toe walking are described in the current literature. Range of motion of the ankle joint, electromyography studies, and kinematic profiles of gait analysis were evaluated to describe and differentiate this condition.^{5,6,9,14-17}

According to the information provided by the parent's questionnaire (1.3: When did your child start toe walking?), 245 (69.6%) of PF-TW had been toe walking "since onset of toe walking", the result for NF-TW was 295 (61.0%) (Table 3). This finding supports the assumption that toe walking appears significantly more often right from the onset of walking if there is involvement of the genetic factor ($P = .010$).

Conclusion

The assumption that a hereditary transmission pattern exists for ITW has been made in several studies. This is the first study that reports on a large number of children with this condition including their family history of toe walking. The findings suggest that ITW possibly has a strong genetic factor, and also that boys are significantly more often affected than girls. However, there certainly are other nongenetic causes associated to this idiopathic condition.

Appendix

Questionnaire 1

Please answer the following questions

- 1.1 "Are there other TW in the family?" with predefined answers: yes____ no____
- 1.2 If response to 1.1 is "yes": "Which relatives?" with predefined answers:
 - Father ____
 - Mother____
 - Brother____
 - Sister____
 - Relatives of father____
 - Relatives of mother____
- 1.3 "When did your child start toe walking?" with predefined answers: Since onset of walking____
 - Months after onset of walking____
 - Years after onset of walking____
 - Only recently____
 - n/a____

Clinical Examination 1

Spin Test Performance. The child was exposed to a challenging situation by spinning fast on the same place. A maximum of 10 spins should be performed and the number of the spin at which the child began toe walking was noted (1-10, 99 = no findings). The test is positive when the child stands on the forefoot after the third spin.

Walking Test Performance. Right after the spin test the child was requested to walk 10 steps in a straight line. The step in which the child started toe walking

was noted (1-10, 99 = no findings). The test is considered positive when the tiptoe walking pattern occurs during the first 10 steps.

Heel Walking Test Performance. The child was requested to heel walk. Heel walking test is considered possible, if the child manages to perform at least 4 steps on its heels. The test is considered negative when the child is not capable to heel walk. Evasive movements such as flexion and/or external rotation in the hip were allowed. Predefined input: [yes, no].

Range of Motion for the Ankle Joint in Dorsiflexion. Measurements of passive ankle dorsiflexion at initial examination were made two times by using a goniometer. First time, in supine position (SP) with the knee joint extended, and the second time in prone position (PP) with the knee bent at 90°. Maximum ankle dorsiflexion was measured with the heel in neutral valgus and rounded up to the nearest 5° interval.

Angle of the Lumbar Lordosis. The goniometer was placed at the point of the greatest convexity at the lumbar spine when the child was in upright and neutral zero position. Lumbar lordosis

angle was measured and rounded up to the nearest 5° interval. **FA5**

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