Gait & Posture 68 (2019) 300–304

Contents lists available at ScienceDirect

Gait & Posture

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

Full length article

Idiopathic toe walking—A follow-up survey of gait analysis assessment

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ARTICLE INFO

Keywords:
Idiopathic toe walking
Kinematics
Electromyography
Gait analysis

ABSTRACT

Background: Toe-walking is a normal variant in children up to 3 years of age but beyond this a diagnosis of idiopathic toe-walking (ITW) must be considered. ITW is an umbrella term that covers all cases of toe-walking without any diagnosed underlying medical condition and before assigning these diagnosis potential differential diagnoses such as cerebral palsy, peripheral neuropathy, spinal dysraphism and myopathy must be ruled out. Gait laboratory assessment (GLA) is thought to be useful in the evaluation of ITW, and kinematic, kinetic and electromyography features associated with ITW have been described. However, the longer term robustness of a diagnosis based on GLA has not been investigated. The primary aim of this study was to examine if a diagnosis of ITW based on GLA features persisted.

Methods: All patients referred to a national gait laboratory service over a ten year period with queried ITW were sent a postal survey to establish if a diagnosis of ITW which had been offered following GLA persisted over time. The gait and clinical parameters differentiating those reported as typical ITW and not-typical-ITW following GLA were examined in the survey respondents.

Results: Of 102 referrals to the laboratory with queried ITW, a response rate of 40.2% (n = 41) was achieved. Of the respondents, 78% (n = 32) were found to be typical of ITW following GLA and this diagnosis persisted in the entire group at an average of 7 years post GLA. The other nine subjects were reported as not typical of ITW following GLA and 44.4% (n = 4) received a subsequent differential diagnosis. The clinical examination and gait analysis features differentiating these groups were consistent with previous literature.

Conclusion: GLA appears to be a useful objective tool in the assessment of ITW and a diagnosis based on described features persists in the long-term.

1. Introduction

Toe-walking has been described as the inability to heel strike during the initial contact of gait and the absence of full foot contact during stance phase [1]. Toe-walking is regarded as a normal variant in children up to 3 years of age [2] but beyond this a diagnosis of idiopathic toe-walking (ITW) must be considered. Hall et al. [3] first described a group of 20 neurologically normal patients who presented with tightness of the tendo-achilles and a tendency to walk on their toes. Subsequent reports documented many cases of ITW without static contracture and so the classic presentation of ITW is now described as one who is otherwise neurologically normal, possesses normal muscle strength and selective control, and demonstrates a preference for walking on the balls of the feet [4]. The term idiopathic toe walking has become an umbrella term that covers all cases of toe-walking without any underlying medical condition [5] and so before assigning a diagnosis of ITW, potential differential diagnoses such as cerebral palsy (CP), peripheral neuropathy, spinal dysraphism and myopathy must be ruled out [6]. ITW had been estimated to occur in 7%–24% of the childhood population [7] but a more recent large scale population study documented a prevalence of approximately 5% [5]. The variation in reported prevalence rates of ITW probably reflects the inherent difficulty in establishing a diagnosis of exclusion and the reduced prevalence in the more recent study may reflect increased use of further diagnostic testing. For example, Haynes et al. [6] found that a significant portion of those thought to be ITW were subsequently found to have another diagnosis following genetic testing.

The kinematic, kinetic and surface electromyography (EMG) features associated with ITW have been described [2,4,8] and have been suggested as a potential means of diagnosing ITW and in particular ruling out common differential diagnoses, particularly CP. Kelly et al. [2] reported normal sagittal plane knee kinematics in ITW which was

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https://doi.org/10.1016/j.gaitpost.2018.12.011
Received 3 September 2018; Received in revised form 30 November 2018; Accepted 5 December 2018

not seen in those with mild bilateral CP. The same authors described the sagittal plane ankle kinematic associated with ITW as initial dorsi-flexion in swing phase followed by rapid plantar-flexion to pre-position the foot for toe-contact. Westberry et al. [4] confirm a normal knee kinematic in those with ITW and report that while there were abnormalities in the ankle kinematics and kinetics, ITW is associated with the ability to normalise these deviations. EMG patterns during gait in ITW vary and it has been reported that EMG did not provide sufficient data to diagnose ITW [4]. However, the use of surface EMG during resisted knee extension has been shown to help differentiate ITW from mild bilateral CP and on resisted knee extension those with CP demonstrate co-contraction of the gastrocnemius which is less likely to be seen in ITW [9].

Our gait laboratory is a national referral centre for gait difficulties and we assess an increasing number of children with a queried diagnosis of ITW. In attempting to establish a diagnosis of ITW or out rule a differential diagnosis, the above GA features are combined with our standardised clinical examination and if the overall Gait Laboratory Assessment (GLA) data is felt to best match the features of ITW, this diagnosis is offered. If the data is not typical of ITW, this is communicated to the referrer along with suggestions for further investigation.

In a previous retrospective study we reviewed 102 patients referred to the gait laboratory with a presumed or queried diagnosis of ITW over a ten year period [8]. We compared the GLA (gait and clinical data) of those felt to be typical-ITW versus those who were thought to be not typical-ITW. Consistent with previous studies, we found that the not-typical-ITW group had increased knee flexion at initial contact and asymmetry in the sagittal plane ankle kinematics which warranted further investigation [2,4,10]. On clinical examination, we found that those not typical of ITW were more likely to have an up-going Babinski response and spasticity in the gastrocnemius suggestive of a pathological diagnosis [6]. However, while these findings confirmed that the clinical and gait characteristics of those diagnosed as ITW based on GLA were consistent with previous literature, we were unable to report if this diagnosis persisted or if any differential diagnosis was subsequently established.

Our previous study [8] noted that a large portion of the referrals to the gait laboratory came from medical consultants (orthopaedic, paediatric or neurology) suggesting that the cohort of potential ITW referred were those who had already been screened by the primary physician. A concerning birth history, delay in initial ambulation and late onset of toe-walking have all been associated with a potential pathological diagnosis beyond ITW [6,10] and it may be that a gait analysis is sought as confirmation of a diagnosis of ITW or further investigation in those who do not fit this history. This highlights both the potential importance of gait analysis in the assessment of ITW and the need examine the robustness of such a diagnosis based on GA.

The primary aims of this study were to-

1 Survey all patients referred to a gait laboratory with a queried diagnosis of ITW over a 10 year period to determine if a diagnosis of ITW offered following gait laboratory assessment persists.
2 Examine birth and walking history in those referred to the gait laboratory to determine if this information may aid in diagnosis.
3 Record treatment history and any on-going concerns relating to gait.

2. Methods

Ethical approval for this study was obtained from the local ethics committee.

A retrospective review of our gait laboratory database identified 102 patients who were referred with queried ITW between May 2002 and May 2013 and the clinical examination and gait analysis characteristics differentiating typical-ITW from not-typical-ITW in this population have previously been reported [8].

Using the available contact details, this same group were invited to participate in a retrospective postal questionnaire.

The gait analysis and clinical examination variables relevant to ITW were re-analysed for the survey respondents in this study. The clinical variables analysed were gastrocnemius range on a fast and slow stretch, hamstring range and, Babinski response. The gait variables examined were ankle ground-contact position, maximum ankle dorsi-flexion in stance and swing and, maximum knee extension in stance. The degree of asymmetry in maximum ankle dorsi-flexion between left and right lower limbs was also assessed. In addition, the results of a resisted knee extension test (RKE) [9,11] with surface EMG were reviewed in those for whom this data was available. A positive test was one in which co-activation of the gastrocnemius was evident on visual inspection of the EMG signal while the quadriceps were extending the knee against resistance.

Significant differences were examined using a Mann-Whitney *U* test apart from Babinski response and RKE which were tested using a chi-squared test. Significance level was set at $p < 0.05$.

The follow-up questionnaire focussed on 3 areas-

1 Diagnosis and follow on investigation following gait analysis
2 Birth History and History of Crawling/Walking
3 Treatment and persistence of ITW

Following initial dispatch of the questionnaire, attempts were then made to maximise the response rate by telephone follow-up 2 weeks after initial dispatch (week 2) to remind parents to fill in the questionnaire, follow-up postal dispatch of the questionnaire one week later (week 3) and a final repeat phone call one week after that (week 4).

The questions were as worded below and the results of each question are described narratively.

3. Results

Of the 102 questionnaires sent out a response rate of 40.2% (n = 41) was achieved. The characteristics of the over-all study population and the responders were similar as shown in Table 1.

The referrals to the gait laboratory for the population as a whole came from –orthopaedic consultants 43%, paediatricians 21%, GPs 14%, physiotherapists 14% and neurologists 9%. Of the 41 respondents the referrals came from orthopaedic consultants 37.5%, paediatricians 28.1%, GPs 18.8%, physiotherapists 9.4% and neurologists 6.3%.

The questionnaire was completed by the child's parent in the majority of cases (n = 38/41). The remaining 3 questionnaires were completed by the child's grandmother, foster parent and one 17 year old participant completed the questionnaire themselves.

Of those who responded, 78% (n = 32/41) were felt to be typical-ITW based on the gait assessment and 22% (n = 9/41) were reported as not-typical.

The characteristics of these two groups are outlined in Table 2 below and, the gait analysis and clinical examination characteristics differentiating typical-ITW and not-typical ITW in the 41 survey respondents are summarised in Table 3.

The questionnaire responses are summarised after each question below.

Has your child been given any other diagnosis apart from idiopathic or habitual toe-walking?

Table 1
Characteristics of the complete study population versus responders.

	Population (n = 102)	Responders (n = 41)
Age at gait analysis (years)	7.6 ± 3.2	7.8 ± 3.5
Age at questionnaire (years)	14.0 ± 4.0	14.6 ± 4.0
Follow-up time (years)	5.9 ± 3.4	6.3 ± 3.6
%Male:Female	73%:27%	73%:27%
%Typical-ITW:Not-typical ITW	79%:21%	78%:22%

Table 2
Characteristics of the typical-ITW and not-typical ITW questionnaire responders.

	Typical ITW (n = 32)	Not Typical ITW (n = 9)
Age at gait analysis (years)	7.9 ± 3.1	7.2 ± 4.7
Age at questionnaire (years)	15.1 ± 3.8	12.8 ± 4.2
Follow-up time (years)	7.2 ± 3.5	5.1 ± 3.7
%male: female	78% : 22%	56% : 44%

Table 3
Clinical examination and gait analysis parameters in typical-ITW and not-typical ITW.

	Typical ITW	Not Typical ITW	p-value
Clinical Examination Measures			
Gastrocnemius Range-Slow (°)	95.0 (10)	91.5 (5)	0.10
Gastrocnemius Range-Fast (°)	95.0 (8)	90.0 (6)	0.01
Hamstring Range (°)	40.0 (15)	37.5 (18)	0.88
Positive Babinski Response	3/32 (9.4%)	3/9 (33.3%)	0.07
Gait Analysis Measures			
Ankle Ground Contact[a] (°)	−9.3 (16.5)	−13.5 (13.3)	0.16
Knee Ground Contact (°)	7.0 (6.6)	10.0 (16.8)	< 0.01
Maximal Ankle Dorsi-Flexion in Stance[a] (°)	6.8 (5.9)	6.4 (10.6)	0.92
Maximal Knee Extension in Stance (°)	6.0 (6.5)	4.7 (13.1)	0.93
Degree of Asymmetry in Ankle Dorsi-Flexion (°)	2.0 (2.7)	6.8 (3.4)	< 0.01
Maximal Ankle Dorsi-Flexion in Swing[a] (°)	−3.0 (12.0)	−8.5 (12.0)	0.04
Positive Knee Extension EMG Test	30.8% (8/26)	71.4% (5/7)	0.05

Data reported are median (interquartile range) apart from Positive Babinski Response and Positive Knee Extension EMG Test.
[a] Positive values = dorsi-flexion, negative values = plantar-flexion.

None of the 32 reported as typical-ITW following GLA had received a subsequent diagnosis at the time of questionnaire.

Of the not-typical-ITW group, 44.4% (n = 4/9) subsequently received an alternative diagnosis (spastic paraparesis = 2, cerebral palsy = 1, peripheral neuropathy = 1).

Please list any investigations carried out (such as X-rays, MRI Scans), and the result if known.

37.5% (n = 12/32) of the typical-ITW group had further investigation. Of the 6 who had MRI, 3 reported a 'normal' result and 3 did not know the result. Of the 7 who had x-ray, 2 reported 'normal' result and 5 did not know the result.

88.9% (n = 8/9) of the not-typical-ITW group had subsequent investigation. (MRI = 7, x-ray = 4, nerve conduction = 2, blood test = 1). Of the 7 who had MRI, 4 reported 'normal', 1 reported 'mild CP' and 2 did not know the result. Of the 4 who had x-ray, 2 did not know the result, 1 reported 'normal' and 1 reported 'bilateral knee fractures'. The participant who had a blood test was reported 'clear' and the individual who had nerve conduction studies was subsequently given a diagnosis of peripheral neuropathy.

Was this patient born early or late and if yes, how many weeks early/late?

In the typical-ITW group, one respondent did not supply this information. 41.9% (n = 13/31) self-reported early birth (mean 6.0 ± 4.9 weeks early) and 32.3% (n = 10/31) late birth (mean 1.5 ± 0.7 weeks late).

In the not-typical-ITW group, 11.1% (n = 1/9) reported early birth (2.5 weeks early) and 44.4% (n = 4/9) late birth (mean 1.4 ± 0.5 weeks late).

Were there any problems during the pregnancy and if yes, please describe?

Among those who responded, the rate of self-reported problems was 27.6% (n = 8/29) in the typical-ITW group and 33.3% (n = 3/9) in the not-typical-ITW group.

Were there any problems during the delivery and the birth of the patient and if yes, please give a brief outline of any problems?

The rate of self-reported problems in those who responded was 25.8% (n = 8/31) in the typical-ITW group and 77.8% (n = 7/9) in the not-typical-ITW group.

At what age did the patient begin to- a/crawl; b/take first steps?

For those who were able to supply this information, time to crawl and time to first steps are summarised in Table 4 below.

Three of the typical ITW group and one not-typical-ITW reportedly never crawled.

Did the child walk on toes from the onset of walking?

All but one patient in the typical ITW group were able to supply this information. 74.2% (n = 23/31) of respondents reported toe-walking from the outset in the typical-ITW group and 88.9% (n = 8/9) in the not-typical-ITW group.

Please list any treatments the patient has had related to his/her walking and if treatment resolved the toe-walking.

81.3% (n = 26/32) typical-ITW reported receiving treatment. Seventeen had physiotherapy with reported resolution of toe-walking in 3 of those. 5 had casting +/- botulinum toxin with resolution in 1 of those cases. Six had surgical intervention with 4 of those reporting resolution of toe-walking.

Do you currently have any concerns regarding the patient's walking or movement and if so, what are your current concerns?

Of the 32 typical-ITW, 53.1% (n = 17/32) had current concerns. The concerns were- toe walking (n = 12/17), muscle tightness (n = 2/17), cramps (n = 1/17), flat feet (n = 1/17) and clumsiness (n = 1/17).

88.9% of the not-typical-ITW group (8/9) had on-going concerns which were listed as- in-toeing gait (n = 2/8), walking balance (n = 1/8), toe walking (n = 3/8), speed and mobility (n = 1/8) and muscle tightness (n = 1/8).

Table 4
Time to crawl and first steps for typical-ITW and not-typical ITW responders.

	Typical ITW	Not Typical ITW
Mean age to crawl (months)	8.5 ± 0.7 (n = 23[a])	7.5 ± 0.7 (n = 7[a])
Mean age of first steps (month)	15 ± 1.4 (n = 30[a])	16.0 ± 2.8 (n = 9[a])

[a] Nine typical ITW and 2 Not-Typical-ITW either never crawled or did not supply this information; 2 typical ITW did not supply age of first steps.

4. Discussion

The aim of this study was to establish if a diagnosis of ITW offered following GLA persists on long-term follow-up. Based on parental questionnaires we found that, at an average of 7 years following GLA, all those diagnosed as ITW still carried this diagnosis and none had subsequently received a differential diagnosis.

The GLA features differentiating typical and not-typical ITW were similar to those we previously described in a larger population of 102 [8] and are consistent with the existing literature [2,4,6,10]. On clinical examination, those in the not-typical-ITW group had reduced gastrocnemius range on a fast stretch indicating potential spasticity and while the proportion of the not-typical-ITW with a significant Babinski response was higher (33.3% versus 9.4%), this was not statistically significant. During gait, the not-typical-ITW group had increased knee flexion at initial contact, reduced ankle dorsi-flexion in swing and, increased asymmetry in the sagittal plane ankle kinematics. The RKE using EMG was positive (co-activation of the gastrocnemius) in 30.8% of the typical-ITW group and 71.4% of the not-typical-ITW group. This was just short of statistical significance (p = 0.05). The original description of the RKE [9] also reported that a proportion (25%; 2/8) of

the ITW population had a positive resisted knee extension test and suggested that a diagnosis of mild CP should be considered based on this EMG test result. However, our clinical experience is that getting younger patients to isolate the required knee extension against resistance can sometimes be difficult which may result in a 'false positive'. Therefore, while we feel the RKE is a useful additional piece of information to add to the assessment we would caution against any diagnosis using this test in isolation.

Our findings suggest that GLA can be useful in assigning a persisting clinical diagnosis of ITW but, perhaps more importantly, GLA identified those who did not appear typical of ITW and therefore warranted further investigation. While only four of the nine not-typical-ITW group had received another diagnosis five years following GLA, it is probably appropriate that gait laboratories err on the side of caution in suggesting further investigation in this group rather than wrongly assign the more benign diagnosis of ITW.

The persistence of a diagnosis of exclusion such as ITW will partly depend on what further testing is carried out and a recent study found that many of those thought to be ITW were subsequently found to have an underlying pathologic diagnosis on further testing which included MRI, EMG, nerve conduction studies and genetic testing [6]. Of those reported typical-ITW in this study, only 37.5% had further investigation (x-ray or MRI) with no significant findings reported by the parents. None of this group had follow-up nerve conduction studies, diagnostic EMG or genetic testing. It might be that the gait analysis findings served as re-assurance for both referring clinician and parents regarding the diagnosis and so further tests were less likely to be requested unless there was a change in the clinical presentation. A repeat gait analysis might be an appropriate follow-up if concerns persist and based on any changes or deterioration in the gait pattern, further testing might then be considered.

Haynes et al. [6] included autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) among the alternative diagnoses to ITW. While it has previously been reported that those with ITW display more neuropsychiatric problems and increased prevalence of neurodevelopmental diagnoses than age matched children [5,12], these have traditionally been considered as co-diagnoses rather than differential diagnoses. Engstrom et al. [5] report that, similar to those without a neurodevelopmental disorder, ITW was a transient condition for the majority of those with a neurodevelopmental disorder which appears to support the argument that these are independent co-diagnoses. However, it has been suggested that a sensory processing dysfunction can cause toe walking but this theory is still under investigation and further studies are suggested [13]. Therefore, in ruling out primarily orthopaedic or neurological reasons for toe-walking, we have considered any neuropsychiatric or neurodevelopmental issues as co-diagnoses and while it is likely that some of the typical-ITW population in this study have such co-diagnoses we did not specifically examine this.

Overall, basic questions on pregnancy, birth and walking history found no major differences between the groups and time to crawling and walking was within normal limits in both groups [6]. It is somewhat notable that the majority of the not-typical-ITW group (77.8%) self-reported difficulties during delivery and birth but analysis is limited by the small number in this group (n = 9). Overall, these findings suggest that those with significant findings in their history are more readily assigned differential diagnoses and therefore, are not referred for GLA. This is consistent with the finding that the vast majority (78%) were found to be typical- ITW based on the GLA.

The reported interventions were in-line with standard treatment for ITW and appear to have been primarily aimed at improving ankle dorsiflexion [14,15] and the outcomes of treatments appear to be consistent with the literature and orthopaedic surgery demonstrated the most sustainable effect [14] A higher portion of those in the not-typical-ITW group had on-going concerns compared to typical-ITW (88.9% versus 53.1%) and the majority of concerns in the typical-ITW group related to persistent toe-walking. This is possibly suggestive of stability in clinical presentation in the typical ITW group consistent with a persisting clinical diagnosis of ITW. In contrast, most of those with on-going concerns in the not-typical-ITW group were related to issues other than toe-walking (in-toeing, tightness, speed and mobility, and walking balance).

There are a number of limitations to be considered when interpreting the results of this study. As with all questionnaire surveys the robustness of the findings depends on the response rate. Our response rate of 40.2% was reasonable and the characteristics of the respondents were very similar to the population as a whole in terms of age at GLA, follow-up time, gender and percentage diagnosed as ITW. Our findings are based on parental questionnaires rather than formal follow-up or review of medical files. This was primarily because, as a national gait analysis laboratory, referrals come from nationwide and so access to medical files would not always be possible. This issue is highlighted by the finding that a number of parents reported that they did not know the result of follow-up investigations. However, we feel that, while the results of radiological tests may not have been known or recalled by parents, they would have been informed of any subsequent change in diagnosis and so the primary finding that an ITW diagnosis persisted in this group is valid. As previously highlighted, a diagnosis of exclusion such as ITW will in part depend on what further diagnostic tests were carried out. The typical-ITW group in this study had less follow-up testing than the not-typical-ITW group which in turn may have been influenced by the diagnosis offered following GLA. To attempt to definitively diagnose ITW in this group would potentially involve undertaking follow-up MRI, nerve conduction studies, diagnostic EMG and genetic testing on the study population. A more achievable, and potentially valuable, future study might be to carry-out a long-term follow-up GLA to ascertain if the features typical of ITW either normalised or continued to match described ITW features as would be expected.

The results of this study suggest that GLA can be useful in the assessment of ITW and parental surveys show that a diagnosis of ITW offered following GLA persisted over time. We therefore recommend GLA as a relatively non-invasive and inexpensive objective tool to be used as part of an assessment of ITW.

Conflict of interest statement

The authors have no relevant conflicts of interest to declare.

Acknowledgements

Khalid Munir received funding from the RCSI Undergraduate Research Summer School Student Fund, RCSI Alumni, The Charitable Infirmary Charitable Trust and the Association of Physicians of Great Britain & Ireland.

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