Toe Walking as the Initial Symptom of a Spinocerebellar Ataxia 13 in a Patient Presenting with a Mutation in the KCNC3 Gene

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Abstract

This article at hand described a 4-year-old child patient who initially presented with the symptoms of toe walking. As part of the diagnostic process, the patient was genetically tested to find the cause of the gait anomaly. The genetic test found a mutation in the KCNC3 gene. The variant c.1268G>A; p.Arg423. His was found in a heterozygotic state. This variant is frequently described as a cause for spinocerebellar ataxia type 13 (SCA13) in the literature. Apart from toe walking as the most pronounced symptom, the patient displayed an instable gait with frequent falls and delayed speech development. The genetic test to determine the cause of the gait anomaly successfully diagnosed the patient with a previously undiscovered SCA13 and subsequently enabled the recommendation of personalized further treatment.

Keywords

► SCA 13
► spinocerebellar ataxia 13
► toe walking
► idiopathic toe walking
► habitual toe walking
► genetic testing toe walking
► pes cavus

Introduction

The term “toe walking in children” describes a characteristic gait anomaly, where patients walk on their forefoot for most of the time. Toe walking can have different neurological and orthopaedic causes and is often associated with further illnesses.1 This makes both the professional treatment of patients and a thorough diagnostic process very important. If the anamnesis finds no alternative clinical cause of the gait anomaly, the patient’s gait is often described as “idiopathic toe walking”. This terminology is misleading, as recent publications suggest that previously undiscovered genetic mutations lead to the symptoms of toe walking when there is no apparent alternative clinical cause.5

For the first time, this article described the case of a mutation in the KCNC3 gene in relation to toe walking. The full clinical report and the results of the genetic test are described in the subsequent paragraphs.

Case Description

The male patient was sent to our specialist practice for gait anomalies at the age of 4 years, following a recommendation by their pediatrician. The patient had previously been treated by a physiotherapist as part of an integrated service at their kindergarten. The anamnesis found that the patient was spontaneously born in the 40th week of pregnancy following an infection with B-streptococcus. The birth
weight was 3680 g and the patient’s height at the time of
birth was 49 cm. The patient developed pneumonia in in-
fancy and was treated with antibiotics and mask-assisted
breathing in hospital; following treatment the patient was
released in generally good health. The patient took his first
independent steps when he was 11 months old. From gait
onset, the patient walked with an unstable gait and fell
frequently; after walking for 30 minutes the patient was
extremely exhausted. No signs of muscle weakness or epi-
lepsy were found. The toe walking intensified substantially
as the patient got older. There were no known chronic or
neurological health conditions or gait anomalies in the
family. The patient’s parents are not consanguine.

The neurological-motorscopic report, following an exami-
nation at the age of 4 years, found an atactic gait pattern, with
bilateral toe walking, as well as a broadened forefoot, bilat-
eral drop foot, and pes cavus. The patient reported no pain
resulting from the abnormal gait. Furthermore, the patient
exhibited a hyper lordosis of the lumbar spine with an angle
of 40 degrees. The mobility of the upper ankle joint for dorsal
extension/plantar flexion was 5–0–50 degrees with a
straight knee and 10–0–50 with the patient’s knee bent. The
examination also found a straight hip and spine position.
The proprioceptive reflexes were successfully triggered bi-
laterally, reflex zones were not broadened, and the Babinski
sign was negative. No disruption was found in the cranial
nerve region and there was no dysarthria or disruption of the
ocular movement. An magnetic resonance imaging found a
pronounced narrowing of the cerebellar Gyri and of the
responding marked Sulci as signs of a cerebellar atrophy.
No further structural abnormalities were found.

Genetic testing found a mutation in the KCNC3 gene of the
heterozygotic variant c.1268G > A; p.Arg423His. This genetic
change was found to be the likely cause of the present SCA13-
ataxia. This variant has been linked to a significant loss of
activity in the tension-dependent potassium channel in
previous studies. SCA13-ataxia is hereditary via the autoso-
mal dominant channel and displays a characteristically slow
progression with a very varied age of onset ranging from
infancy to late adulthood. The clinical variant was not
found in either of the patient’s parents.

**Therapy**

The therapeutic treatment process included a consultation
with a pediatric neurologist. The treatment of the gait
anomaly was conducted using lower leg night splints and
personalized pyramid insoles to achieve an improved gait
and reduce the intensity of toe walking in the patient.
Following treatment, the parents reported that the patient
would rarely walk on the forefoot and would display toe
walking predominantly when going barefoot. The patient can
walk unassisted at the age of 5 years but struggles with a
coordinative weakness and frequent falls. The maximum
walking distance is significantly reduced and the patient
experiences frequent trembling because of the underlying
disease. The use of a wheelchair was recommended to help
manage this and avoid the need to carry the patient on longer
walks. The use of a therapy bike is also being considered. It is
recommended that the night splints continue to be worn and
cupped insoles have been prescribed to be worn during the
day. Additionally, it was recommended that physiotherapeu-
tic treatment should be continued.

**Conclusion**

This article provided the first-hand description of a patient,
who displays toe walking as a main symptom of a SCA13-
ataxia as a result of a mutation in the KCNC3 gene. Conse-
quentially, the patient exhibits toe walking with a genetic
cause. The diagnosis was secured following genetic testing,
where the identification of the ataxia was relevant for
subsequent treatment and contributed to the correct treat-
ment choice for the patient.

**Ethical Approval and Consent to Participate**

Ethical approval was obtained by the Research Team, and
the patients’ parents gave informed consent to participate
in consultations relating to the data collected for this
article.

**Availability of Data and Material**

The data cited in this article is available from the patients’
files and can be reviewed for professional review purposes
pending the consent of the patients’ parents as their legal
guardians.

**Authors’ Contributions**

D.P. is the owner of the practice for toe walking, a
specialist for the gait anomaly, and the head of the
research team for the genetic causes of toe walking.
Data from his examination of the patient was included
in this article. J.R.T. works for D.P. as a research assistant
and combined the data with background research to
produce this case study. A.T. is an expert for the ortho-
pediatric causes of toe walking and has examined this
patient for the case study. K.R. is a neurologist with
experience in treating and examining patients with
SCA13; and examined the patient to verify that his initial
symptom was toe walking and that other known indica-
tions of the condition were not as pronounced. J.S. works
as a neuropediatrician and examined the patient for this
case study.

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None.

**Conflict of Interest**

D.P. is the owner of a practice specializing in the treatment
gait anomalies and has developed a treatment method
for toe walking. Rest authors declare no conflict of
interest.

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kindly agreeing to the publication of this case study.
References