CASE REPORT

Toe walking after three: how serious could it be?

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SUMMARY

A 4-year-old girl with no significant medical or family history presented with toe walking, leg pain, unsteady gait and frequent falls for 2 months. Examination revealed upper motor neuron signs in the lower extremities. Laboratory tests were normal including creatinephosphokinase and lactate dehydrogenase. Brain and lumbar spine MRI were normal. MRI cervical and thoracic spine showed a large intradural and extradural mass arising from the right C7 nerve root, widening of the neural canal with evidence of cord compression. She underwent C6–C7 laminectomy with excision of the tumour. Pathology revealed spindle cell tumour with extensive expression of S100 protein and CD56, with Ki-67 proliferation index of 1%–2% consistent with benign schwannoma. She made an excellent recovery following surgery and physiotherapy sessions. Review of literature shows rare reported case of schwannoma prior to the third decade of life.

BACKGROUND

The prevalence of toe walking is between 2% and 12% in children.¹ Idiopathic toe walking is a diagnosis of exclusion when it persists after the age of 3 years.^{1,2} It is important to appropriately evaluate toe walking, as it has been associated with neurological deficits including cerebral palsy, global developmental delay and muscular dystrophy. One study

found a relationship between idiopathic toe walking and higher rates of admission to the neonatal intensive care unit and perinatal complications.¹

We describe an interesting case of toe walking in a 4-year-old, which was initially thought to be of no clinical significance. She was later diagnosed to have a schwannoma of the cervical spine, a rare condition in this age group with an even more unusual tumour location.

CASE PRESENTATION

A 4-year-old girl (ex—preterm 32 week twin) presented with right lower extremity pain and unsteady gait for over 2 months. She was noted to be a toe walker since 12 months of age. Her developmental milestones were within normal limits. For the previous 2 months, she had difficulty standing and parents noted frequent falls. Her parents denied weakness of the upper limbs, lack of coordination, urinary or bowel incontinence. There was no history of trauma or recent infections.

During the initial neurological evaluation, she had normal strength in upper and lower extremities, tight hamstrings and Achilles' tendons. Upper and lower extremity reflexes were normal except for bilateral Babinski signs. There were no dysmorphic features, sacral dimples; tufts of hair or scoliosis but two café au lait spots were noted on her back. Brain and lumbar spine MRI were normal.

At 2-week follow-up, she was crawling because of progressive lower extremity weakness. She had sustained clonus in both ankles and +3/4 right patellar reflex. MRI of cervical and thoracic spine was requested.

INVESTIGATIONS

Initial laboratory studies revealed normal basic metabolic panel, liver function tests, complete blood counts, creatine kinase (CK) (164 U/L), aldolase (61 U/L), ammonia (42.8 µg/dL) and lactate dehydrogenase (LDH) (263 U/L).

Brain and lumbar spine MRI were normal. Electromyography (EMG) demonstrated normal motor and sensory nerve conduction but globally reduced motor unit potential activation in both lower extremities.

MRI of the cervical and thoracic spine demonstrated a large intradural and extradural mass arising from the right C7 nerve root associated with widening of right C7 neural canal that displaced the edematous spinal cord, to the left. The tumour was 5 cm in transverse diameter with the extraspinal portion measuring 2.2 cm×2.3 cm (figure 1).

Pathology revealed spindle cell tumour with extensive expression of S100 protein and CD56,

Figure 1 MRI fluid attenuation inversion recovery of cervical spine with sagittal image showing a large intradural (arrow) and extradural mass arising from the C7 region displacing and severely compressing the spinal cord.

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Figure 2 Pathological examination of spindle cell neoplasm with a mostly fascicular architecture with extensive expression of S100 protein and CD56. The cells were not mitotically active with the Ki-67 proliferation index of 1%–2% in most of the sample, focally up to 5%, consistent with benign schwannoma (WHO grade I).

with Ki-67 proliferation index of 1%–2% consistent with benign schwannoma (figure 2).

DIFFERENTIAL DIAGNOSIS

Lower extremity weakness in children can be attributed to an array of causes. However, when it occurs in the context of toe walking, the differential diagnosis narrows.

The differentials for toe walking include neurological impairments such as cerebral palsy (CP), which given her history of prematurity and twin gestation certainly raises the suspicion. However, the regression to crawling and progression to limb weakness is not characteristic of CP, idiopathic toe walking or tight heel cords.² Toe walking and lower extremity weakness can also be seen in Duchene's muscular dystrophy; however, it is rare for females to phenotypically express this X-linked inherited disease.^{3,4} Hereditary spastic paraplegia is another collection of genetic conditions with variable severities and modes of inheritance.⁵ People with uncomplicated types can have progressive lower extremity weakness, spasticity and urgency incontinence.⁵ This diagnosis is rare and should be considered only after ruling out common aetiologies. Other less common causes of progressive weakness are myopathies, such as sarcoglycanopathies and other congenital muscular dystrophies.^{6,7} However, the normal CK and aldolase with the presence of upper motor neuron signs are not typical for any of these conditions.

In any case of sudden inability to walk, infectious aetiologies and cord compression should be evaluated. The presence of normal inflammatory markers and the absence of constitutional symptoms or systemic signs including that of urinary urgency made infection or tethered cord less likely.⁸ Hence, spinal cord mass remained an active differential diagnosis. A number of possible benign spinal masses are developmental tumours (dermoid, epidermoid, teratoma), intraspinal lipomas, neurofibromas or schwannomas.⁹ Some potential malignant paediatric spinal tumours to consider are neuroblastomas, ependymomas or astrocytomas.⁹

The upper motor signs in this patient raised suspicion for central nervous system pathology rather than a peripheral nerve disease.^{10,11} The timing of symptoms, progressive change in gait,

toe walking and tight hamstrings suggested more of a chronic or subacute aetiology. This child lacked the stigmata of neural tube defects. However, overt pathology such as tethered cord can remain concealed until linear growth increases and more tension is placed on the spinal cord. Back pain, numbness, scoliosis and overlying skin changes are commonly seen in patients with tethered cord, but some may present with only gait abnormalities. In children without a history of spinal surgery, the aetiology of tethered cord is often congenital except in cases where a mass entraps the spinal cord.¹²

Spastic or scissor gait rather than weakness from spastic paralysis is a more obvious manifestation of upper motor neuron injury, as seen in individuals with CP. Toe walking may be a subtle finding of hypertonicity of the gastrocnemius and soleus muscles.¹³

An intraspinal mass remained high in the differential, which is why further imaging was pursued. In retrospect, the patient's history of toe walking was possibly an early upper motor sign in response to her growing schwannoma.

TREATMENT

She was admitted to the hospital for neurosurgical intervention after pretreatment with dexamethasone. C6–C7 laminectomy with excision of extradural, intradural extramedullary tumour was performed. Pathological examination revealed a spindle cell neoplasm with a mostly fascicular architecture, with extensive expression of S100 protein and CD56. The cells were not mitotically active and the Ki-67 proliferation index was 1%–2% in most of the sample, focally up to 5%, consistent with a benign schwannoma.

OUTCOME AND FOLLOW-UP

She made an excellent recovery and began walking after surgery and a few physiotherapy sessions.

Follow-up at 3 and 6 months reveals normal neurological examination.

DISCUSSION

The most intriguing features of the patient's presentation were her upper motor neuron signs with progression to lower extremity weakness. These are long tract myelopathic findings, which are common in intradural extramedullary tumours.^{14,15}

The presence of abnormalities on neurological examination warrants a thorough evaluation. The persistence of toe walking beyond the age of 2–3 years, when the heel-toe pattern is typically established, can represent a neurological deficit.¹ In autism and idiopathic toe walking, a sensory processing dysfunction was previously proposed as a cause of toe walking.^{13,16} Differences in EMG findings during resisted knee extension and quadriceps activation with an extended knee (quad set) can help distinguish cerebral palsy from idiopathic toe walking.^{8,13,15,17} Both demonstrate premature recruitment of the gastrocnemius during swing phase but the duration of coactivation of the gastrocnemius with the quadriceps is longer during a resisted knee extension and quad set in cerebral palsy compared with idiopathic toe walking and controls.^{14,17}

It is prudent to evaluate for the presence of supraspinal and spinal lesions in the workup of upper motor neuron signs.¹⁸ The clinical picture of upper motor neuron syndrome is more dependent on the site of the lesion. In cerebral lesions, spasticity tends to be mild and often involves the extensors.¹⁸ Spinal cord lesions often have severe spasticity in the flexor muscles and clonus. Of the principle descending motor tracts in the spinal cord involved

in the production of spasticity, a lesion in the inhibitory dorsal reticulospinal tract (DRT) is critical in production of spasticity and extensor plantar response.¹⁷ Furthermore, involvement of the lateral corticospinal tracts results in spastic paralysis, ipsilateral hypertonia and hyper-reflexia. Given the size of her schwannoma and lateral intrusion on the spinal cord, it is probable that her neurological findings were signs of a DRT lesion. The localisation of a cervical schwannoma was critical in the diagnosis of this previously healthy girl.

Schwannomas are slow-growing benign tumours that originate from Schwann cells.^{19–21} They are common intradural tumours that typically present in the third decade of life and later. Our patient's age at diagnosis is young for the typical demographic of patients with schwannomas. To the best of our knowledge, she is one of the youngest presentations of a schwannoma. Early age presentations of multiple schwannomas or neurofibromas should raise concern for a genetic syndrome such as schwannomatosis or neurofibromatosis.^{16–20} Close monitoring for recurrence or for development of additional neoplasms is indicated in these cases.

Toe walking can be a stage in the early development of gross motor skills. There is also growing awareness of its association with some neuropsychiatric, musculoskeletal or neuromuscular disorders. Few clinicians may be aware that toe walking can be a subtle neurological finding in the absence of weakness, behavioural issues or developmental delays. By describing the muscle mechanics of toe walking instead of characterising it as a gait pattern, practitioners may be better attuned to the potential diagnoses that should be excluded prior to labelling toe walking in a child as idiopathic.

Patient's perspective

'Thank God! She can walk again, we were so scared!!' (mother of the child).

Learning points

- Idiopathic toe walking is a diagnosis of exclusion when it persists beyond the age of 3 years.
- Toe walking can occur secondary to a central nervous system disorder, myopathy or peripheral neuropathy.
- Toe walking in the presence of red flag signs such as hyper-reflexia, spasticity, weakness and progressive symptoms warrants a thorough evaluation for supraspinal and spinal aetiology.
- Toe walking is a manifestation of both benign structural conditions such as tight heel cords or more limiting and complex central nervous system pathologies, but can be distinguished with a thorough musculoskeletal and neurological examination.

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