Case Report

Early onset primary torsion dystonia with DYT1 positive in an 8-Year-old child: a case report

Naher N[a], Begum N[b], Bhuiyan SJ[c]

ABSTRACT

Apart from tremor, primary clinical symptom of primary torsion dystonia (PTD) is dystonia. No history or evidence of neuronal degeneration or acquired origin are present. Out of the seven different loci reported for PTD, only two genes have been identified.

This case is being presented with a diagnosis of PTD. The patient, an 8-year-old boy from non-consanguineous parents, had problem in walking for one year, mostly in his left leg and had trouble using his left hand to pick up objects. He had dystonia in the left upper and lower limbs, had intact sensory function with intact cranial nerve function. All his laboratory tests were normal including brain imaging; nevertheless, genetic testing revealed DYT1 was positive.

Key words: *Primary torsion dystonia, tremor, DYT1 positive.*

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INTRODUCTION

Movement disorders like dystonia are characterized by persistent or sporadic muscular spasms that result in atypical postures and/or motions.[1] The etiology, age at symptom onset and distribution of afflicted body regions are used to classify dystonia.[2]

In DYT1 variety of dystonia, the inheritance is autosomal dominant with reduced penetrance.[3] For the majority of instances of typical DYT1 dystonia, the TOR1A gene's exon 5 has a heterozygous 3-bp deletion.[1] People with isolated dystonia who do not exhibit any other abnormalities on neurologic examination (apart from tremor), normal routine neuroimaging and no known past cause of acquired dystonia (e.g. exposure to neuroleptic medications, cerebral trauma, infarct, infection), should be suspected of having it.[4]

The most common and severe type of primary dystonia is early-onset dystonia brought on by the DYT1 mutation. Of patients with pediatric onset dystonia (POD), it affects 16%–53% of non-Jewish groups and 80%–90% of Ashkenazi Jewish communities.[2] DYT1 dystonia usually first appears in childhood or adolescence. The initial symptom to appear is usually dystonia involving one limb, which can present as writing dystonia or walking dystonia with foot inversion or eversion. Patients with arm dystonia may present later in life with an average age of 15 year and are less likely to experience generalized dystonia.[5]

Here a case is being presented with early onset dystonia who has DYT1 positive.

CASE REPORT

An 8-year-old Bangladeshi male child presented with difficulty in walking started from left leg along with difficulties in picking object with left arm for 1 year. He had no history of fever, altered level of consciousness, seizure, any offending drugs or any head trauma.

The patient was born at term and was the only child of non-consanguineous parents with appropriate birth

Author information

a. Nurun Naher, Consultant, Paediatric ICU, Evercare Hospital, Dhaka, Bangladesh.

b. Noorjahan Begum, Senior Specialist, Paediatric ICU, Evercare Hospital,Dhaka.

c. Sarwar Jahan Bhuiyan, Senior Consultant, Department of Paediatrics and Neonatology, Evercare Hospital, Dhaka.

Address of correspondence: Nurun Naher, Consultant, Paediatric ICU, Evercare Hospital, Dhaka, Bangladesh. Email: nahernurun004@gmail.com

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weight along with normal perinatal and developmental history. There was no family history of similar disease or any other neurological disease.

His examination findings revealed, higher psychic function was normal with intact sensory function and cranial nerves. There was increased tone on left side of body with intermittent dystonic movement compared to right. No Kayser-Fleischer ring was detected on eye examination. Other systemic examination including skin survey revealed no abnormalities.

His Complete blood count, Liver function test, Thyroid function test, serum ceruloplasmin and urinary copper all came out negative. Brain imaging (MRI and MRA) also revealed normal findings. Genetic test for DYT1 came out positive. He responded well with anti-dystonic medications trihexyphenidyl.

DISCUSSION

The majority of cases of generalized primary dystonia with childhood onset are caused by DYT1 dystonia, a severe form of primary torsion dystonia that manifests in individuals under 26 years of age.[6] This case study serves as a reminder to patients with dystonia to look into DYT1 gene deletion as a differential diagnosis. In the Asian region, very few cases involving Chinese people have been documented. Three cases of primary torsional dystonia with DYT1 gene mutation were reported by Lan YW et al[7], these cases also showed limb dystonia, which is similar to our case.

It is important to work toward obtaining an early and precise genetic diagnosis since it will reduce the need for pointless investigations and enable the application of suitable genetic counseling and early medical or surgical interventions.[6] The goal of treatment is to reduce symptoms. Usually, oral drugs are tried initially.[4] Trihexyphenidyl is the preferred oral medication and it alleviate the symptoms of dystonia by reducing the excitability of neuromuscular junctions. The only oral drug that has been shown to be successful in double-blind, placebo-controlled trials is trihexyphenidyl[8], which has an efficacy rate of 71% that steadily declines with time. Other oral drugs that have been used to treat dystonia with varying degrees of success include benzodiazepines (commonly used drugs include diazepam and clonazepam), antidepressants, levodopa, antiepileptics, baclofen.[4,9] For patients with more widespread dystonia, where certain muscle groups cause incapacitating symptoms, botulinum toxin injections may also be used in conjunction with oral drugs.[9] Our presented case was also improved with trihexyphenidyl. Recent uncontrolled trials on a small patient group revealed that deep brain stimulation improved patients with primary dystonia brought on by DYT1 gene mutation by 90 to 100 percent.[6]

Conclusion

Patients with early-onset dystonia may exhibit comparable typical clinical characteristics. Healthcare professionals, particularly those who work with patients who are dystonic, should be aware of this issue because early genetic diagnosis would enable early, appropriate therapy and genetic counseling.

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Consent: Taken from parents for this publication.

Conflicts of interest: Nothing to declare.

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