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CORRESPONDENCE

Cervical Dystonia—A Rare Presentation of Spinocerebellar Ataxia Type 35

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To the Editor: A 14-y-old developmentally normal boy presented with progressive torticollis to the left side for 6 mo and positional lumbar scoliosis for 3 mo. Examination revealed cervical dystonia, hyperkinetic dysarthria, and intentional hand tremors. He was born to third-degree consanguineous parents. His evaluation for Wilson disease was negative. The neuroimaging was normal. There was no satisfactory improvement with trial of levodopa, baclofen, trihexyphenidyl, and clonazepam. At this juncture, we considered congenital forms of dystonia and performed a targeted genetic analysis which revealed a heterozygous missense variation in exon 9 of the *TGM6* gene (chr20:g.2403628G>A) that resulted in the amino acid substitution of asparagine for aspartic acid at codon 381 (p.Asp381Asn;ENST00000202625.7) confirming the diagnosis of spinocerebellar ataxia (SCA) type 35. We administered intramuscular botulinum toxin injection (300 IU) around the neck for his cervical dystonia. He had significant improvement in torticollis as observed at a one-month follow-up. Subsequently, three-monthly injections of botulinum toxin were planned.

SCA is characterized by progressive ataxia with autosomal dominant inheritance [1]. SCA type 3 is the commonest among 40 types [2]. The first case of SCA type 35 was reported in the year 2010 with a mutation in the transglutaminase 6 (*TGM6*) gene [3]. Dysarthria and gait disturbances are the commonest presenting symptoms. Spasmodic torticollis, cerebellar dysarthria, intentional tremor, dysmetria, pyramidal and extrapyramidal signs, and dystonia are less common. Thirty cases of SCA type 35 have been reported till now with the age of onset between 20 and 54 y [4]. Our index case is the youngest of SCA type 35 cases reported till now. He had torticollis, hyperkinetic dysarthria, and intentional hand tremors at initial presentation.

We conclude that cervical dystonia can be a rare presenting feature of SCA type 35 and it responds well to botulinum injections. SCA type 35 should be considered a differential diagnosis in young children presenting with cervical dystonia.

Declarations

Informed Consent Written informed consent was obtained from the parents for publication of the child's clinical details.

Conflict of Interest None.

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