A Case of Childhood Onset Dystonia Due to KMT2B Gene Mutation

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Introduction

- Dystonia is a hyperkinetic movement disorder characterized by sustained or intermittent muscle contractions that cause abnormal, often repetitive movements and postures affecting the extremities, trunk, neck, and face
- Childhood-onset dystonia can be acquired or genetic. It may occur as an isolated movement disorder, or it may occur together with other movement disorders, neurological or systemic symptoms
- Dystonia 28 is an autosomal dominant inherited disorder characterized by the onset of progressive dystonia in the first decade of life
- The disease is seen due to mutations in the lysine methyltransferase 2B (KMT2B) gene and is inherited in an autosomal dominant
- This text was aimed to emphasize the KMT2B gene mutation

Case

- A nine-year-old male patient presented with inward walking in his left foot
- The patient, who could walk and run until the age of eight, had complaints of inward walking in the left foot, inability to run, and pain in the left foot for the last year. It was learned that similar gait and pain were also present in the father who passed away
- In the patient's neurological examination, bilateral lower extremities' deep tendon reflexes were hyperactive. Spasticity was present in the bilateral lower extremities, especially on the left. The patient had a dystonic gait and had pain with walking
- The patient's cranial and spinal magnetic resonance imaging findings were normal
- In the whole-exome sequence analysis. c.3646_3653del heterogeneous pathogenic mutation was detected in the KMT2B gene. This variant has not been reported before, but evaluations in databases have classified the variant as likely pathogenic. This mutation, which was confirmed by Sanger sequence analysis, was not detected in the mother, and samples could not be taken from the father due to death
- The patient was diagnosed with Dystonia 28
- The patient was initially started on levodopa therapy for the treatment of dystonia. however, the patient's treatment was discontinued because he did not benefit from levodopa treatment (400 mg/day)
- Botox was injected into the bilateral lower extremity muscles of the patient and his walking improved significantly after the injection
- Physical therapy is continued regularly

Conclusion

- Dystonia should definitely be evaluated in the examination of patients who apply with the complaints of gait disturbance and spasticity that started in the late childhood
- If there are similar complaints in family members, hereditary dystonia should be considered in the differential diagnosis
- Next-generation sequencing analyzes, especially whole exome and whole genome sequencing, are very important in diagnosing these diseases