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# Chromosome 16p13.3 Duplication Syndrome: A Case Report

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**Received:** October 30, 2024; **Published:** November 25, 2024

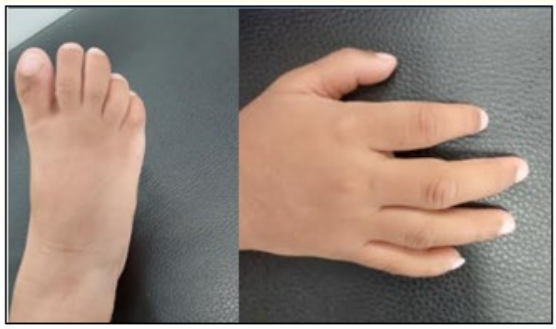

***Figure 1:*** *Short fingers of our case.*

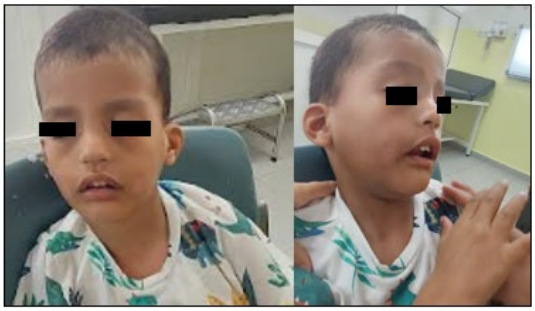

***Figure 2:*** *Facial features.*

We report the case of a 4-year-old child, born at term via vaginal delivery, from non-consanguineous parents, with no history of neonatal distress or epilepsy in the family. The child exhibited psychomotor delay: head control was achieved at 6 months, sitting at 12 months, and walking at 2 years, along with mental retardation and delayed language acquisition. The illness began at the age of 8

months with the onset of generalized convulsive seizures characterized by eye rolling in a context of fever, and was treated with sodium valproate. The progression was marked by a decrease in the frequency of seizures. Clinical examination revealed facial dysmorphism: long face, micrognathia, low-set and protruding ears, bulbous nose, with short fingers and toes. The EEG showed abundant interictal epileptic anomalies in the form of generalized slow-wave spikes in prolonged bursts, compatible with epileptic encephalopathy. Brain MRI spectroscopy was normal, and the ophthalmologic examination was unremarkable. The karyotype was normal (46: XY) and was further complemented by exome sequencing, which revealed a 16p13.3 duplication. The patient is currently on sodium valproate at a dose of 30 mg/kg/day, along with speech therapy.

16p13.3 duplication syndrome is a rare genetic disorder characterized by the duplication of a small region of chromosome 16, including the CREBBP gene. Discovered in 2010, this syndrome remains poorly studied, with only 26 cases reported in the medical literature. Affected individuals present with specific developmental features that manifest consistently, regardless of the size of the duplication, including those as small as 240 kilobases. These features include mild to moderate intellectual disability, facial dysmorphism, limb anomalies, as well as occasional malformations of the eyes, palate, genitalia, and heart. Although the CREBBP gene is also implicated in Rubinstein-Taybi syndrome, which results from a deletion of this same gene, the clinical manifestations of the two syndromes should not be confused. It is estimated that 16p13.3 duplication syndrome affects approximately 1 in 97,000 to 146,000 newborns.

Exome sequencing is a powerful tool for studying 16p13.3 duplication syndrome. It allows for the identification and characterization of the genetic variations responsible, correlating genotypes with phenotypes, and guiding diagnostic and treatment strategies. By combining technological advances in genetics with an integrated clinical approach, it is possible to improve the management of patients affected by this rare and complex condition [1-5].

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**Volume 7 Issue 12 December 2024**