TITLE: Sotos syndrome: a look at cerebral gigantism. Case report.

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ABSTRACT

Antecedents: Soto's syndrome also known as Brain gigantism. It is one of prevalent overgrowth syndromes. Macrocephaly and tall stature is a frequent characteristic of these childs, it is characterized by a distinctive facial appearance (broad and prominent forehead with a dolichocephalic, poor frontotemporal hair, among others); learning disability and overgrowth. For the diagnosis, molecular genetic analysis is performed for the patient's NSD1 gene. Treatment is in pursuit of promoting neurological development. Clinical case: we present the case of a preschool child who in the infant stage showed increased head circumference and poor progress in neurodevelopment, dolichocephaly, bulging forehead, narrow fissures, hypoplastic spine, nostrils pointing upward, full palate, pavilions rotated posteriorly, back with significant lumbar kyphosis, a molecular study was sent, which identifies a heterozygous variant, missense c.5165G> C; p.Cys1722Ser in the NSD1 gene. The patient receives multidisciplinary support with progress in neurodevelopment. **Conclusion:** in spite of its worldwide distribution, Sotos syndrome may not be reported. In addition to its characteristic clinical picture, molecular genetic tests are also highly recommended to reach a diagnosis.

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