P-753 - SOTOS SYNDROME - CASE STUDY

A.P.Martins[1], R.Coelho[1,2], R.Grangeia[1]

[1]Psychiatry and Mental Health, Hospital São João, [2]Psychiatry and Mental Health, Faculdade de Medicina da Universidade do Porto, Porto, Portugal

Sotos syndrome, also known as cerebral gigantism, was first described by Juan Sotos in 1964 in five patients with prenatal and postnatal overgrowth, characteristic facial appearance, advanced bone age and developmental delay. Some patients have serious clinical manifestations, such as congenital heart defects, dysplastic kidneys, severe mental retardation and leukemia.
A wide spectrum of emotional, cognitive, and behavioral difficulties occur in patients with Sotos syndrome, who have a high risk of developing attention deficit disorder, hyperactivity disorder, and temper tantrums. Other cases have been described in which affected children have feeding and sleeping problems, phobias, and irritability. Many Sotos syndrome children exhibit adaptive problems in social functioning, and some present autistic features.
In this study authors present a 30-year-old woman with Sotos syndrome and behavioural difficulties. The patient had Sotos syndrome diagnosed at childhood, with typical facial gestalt, macrocephaly, advanced bone age, scoliosis, kidney abnormalities, pulmonary chronic disease and migraine.
At the age of 26 she developed irritability with marked impulsivity, episodes of agressivity and later of self-harm (skin-picking).
She was treated with atypical anti-psychotics with positive clinical response.
Clinical and laboratory examinations should be performed to prevent and manage any unusual medical aspect of the syndrome. Facial gestalt and macrocephaly, rather than advanced bone age, are the strongest indications for clinical diagnosis.

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