What is the frequency of Sotos syndrome in children with global developmental delay?



Summary

Sotos syndrome: Sotos syndrome is characterized by unusually large head size, unusual facial features, and a markedly advanced bone age.

Macrocephaly: A diagnosis given when a child's head circumference is above the 98th percentile of children of the same age and sex.

The main aim of this study was to try to find out how often Sotos syndrome occurs in children with global developmental delay. Sotos syndrome is characterized by unusually large head size (called macrocephaly), unusual facial features, and a markedly advanced bone age. It has traditionally been under-recognized. The study found that among a large group of children with global developmental delay, just under one in ten (approximately 7%) had macrocephaly. Of those with macrocephaly, a further one in five (approximately 17%) were diagnosed with Sotos syndrome through a simple x-ray test for bone-age, which is considered a substantial percentage.

What families should know

In patients with global developmental delay accompanied by macrocephaly, Sotos syndrome is not uncommon.

What practitioners should know

Children with macrocephaly and global developmental delay should have a bone age determined in order to determine if the child has Sotos syndrome.

Reference

Srour, M., Mazer, B., & Shevell, M.I. (2006). Diagnosing Sotos Syndrome in the setting of global developmental delay and macrocephaly. *Journal of Child Neurology*, 21, 287-290.