MACROSOMIA IN CONJUNCTION WITH DEVELOPMENTAL DELAYS LEADS TO DIAGNOSIS OF GENETIC ABNORMALITY

Guy Hamilton, DO , Moon Suh, DO , Diane Kudes, MD
Department of Family Medicine, Suburban Family Medicine

Suburban Community Hospital
Extraordinary People. Extraordinary Care.

Background

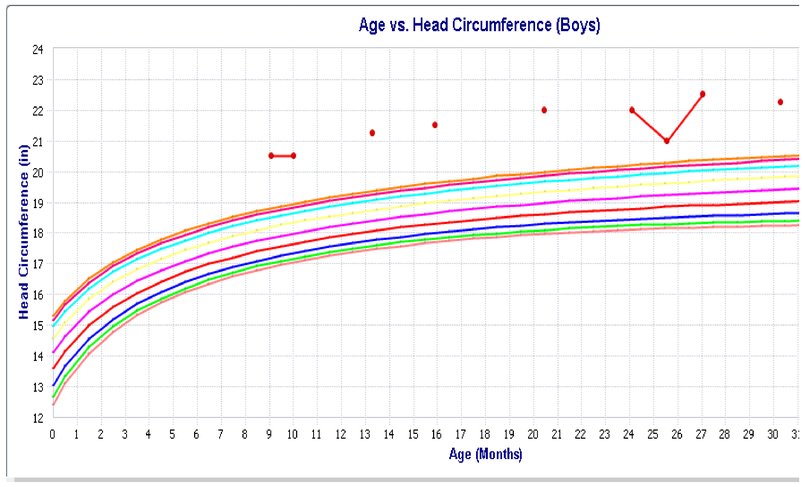
A child's growth is an essential part of a patient's well-child care. At times, a patient will fall under abnormal percentages of height, weight, or head circumference. The rate of change should be noted as it may be even more vital to the diagnosis of a crucial condition.

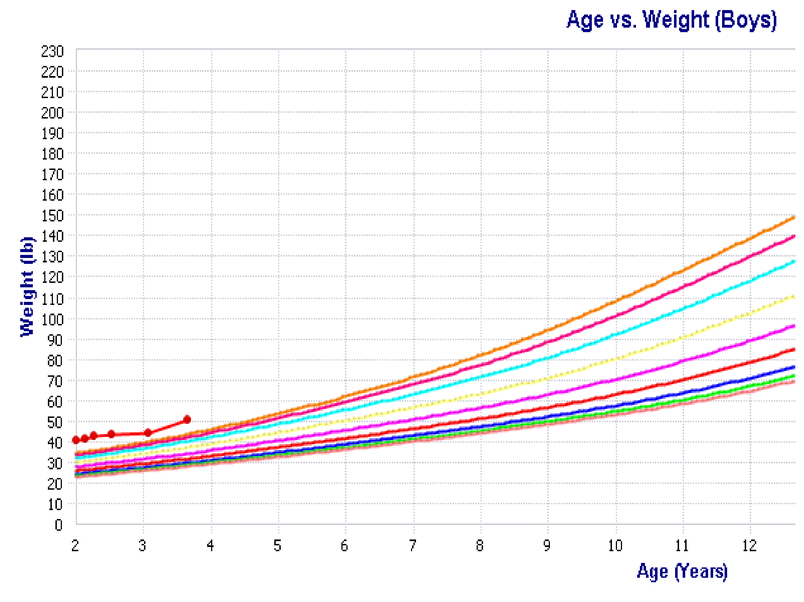
Case Report

A 6 month male presented to clinic for his well child check. The patient was born at forty weeks and birth history only significant for macrosomia. His head circumference, height, and weight were all greater than the 95th percentile. Physical exam was significant for hypotonia. The family denied a history of vomiting or lethargy. Patient had developmental delays of not rolling from prone to supine, or sitting independently. Due to worsening macrocephaly a CT was obtained. The results of the CT lead to a medical work up for child abuse including a negative skeletal survey and ophthalmologic exams. Neurosurgery evaluated the patient and did not feel intervention was needed. The patient was then evaluated by Genetics and formally diagnosed with Sotos syndrome

Labs/Imaging

Head CT revealed bilateral chronic subdural hematomas, mild enlargement of the extra-axial CSF spaces and prominent ventricles in supratentorial spaces.





Discussion

- Sotos syndrome is a congenital overgrowth syndrome associated with intellectual disability. Sotos syndrome is a well-known anomaly syndrome characterized by overgrowth, characteristic facial gestalt, and developmental delay. Haploinsufficiency of the NSD1 gene has been revealed as one of the major genetic causes..

Conclusion

- When patients in a primary care setting present with developmental delay, and abnormal growth, a wide differential should be considered. Accurate diagnosis leads to proper care and appropriate referrals, including genetic testing. Sotos syndrome should be considered when pediatric patients present with developmental delay, hypotonia, and macrocephaly

References

1. Legler, J. D., & Rose, L. C. (1998). Assessment of Abnormal Growth Curves. American Family Physician, 153–158.
Retrieved from https://www.aafp.org/afp/1998/0701/p153.html#ref-list-1
2. Sotos syndrome - Genetics Home Reference - NIH. (2020, March 31). Retrieved April 11, 2020, from https://ghr.nlm.nih.gov/condition/sotos-syndrome#
3. Baujat, G., Cormier-Daire, V. Sotos syndrome. Orphanet J Rare Dis 2, 36 (2007). https://doi.org/10.1186/1750-1172-2-36