Cancer Prone Disease Section
Mini Review

Sotos syndrome (SOS)
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Identity
Alias: Cerebral gigantism
Inheritance: Generally sporadic, a few inherited cases. The familial case reported in 2003 was proved to have NSD1 mutation.

Clinics
Phenotype and clinics
Excessive growth, advanced bone age, typical facial gestalt, developmental delay.
In infancy growth is rapid, but settles down above the >97th centile in early childhood.
The adult height remains close to normal.
Large hands and feet.
Characteristic facial gestalt: macrocephaly (>97th centile), frontal bossing, prognathism, hypertelorism, and antimonogolid slant of the palpebral fissures.
Occasional hypotonia and delay in motor and language development.
Cardiac, urogenital, musculoskeletal, and ophthalmologic anomalies are observed.

Neoplastic risk
Relatively high. Neoplasms in SoS are found with a frequency of 2.2-3.9%.

Cytogenetics
Inborn conditions
Routine chromosome analysis usually shows normal karyotype. Chromosomal abnormality concerning 5q35 were reported. In addition, several chromosomal translocations other than 5q have been published.
References


Douglas J, Hanks S, Temple IK, Davies S, Murray A, Upadhyaya M, Tomkins S, Hughes HE, Cole TR, Rahman N. NSD1 mutations are the major cause of Sotos syndrome and occur in some cases of Weaver syndrome but are rare in other overgrowth phenotypes. Am J Hum Genet. 2003 Jan;72(1):132-43


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