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Dominant Sotos's syndrome

Sir,

Since the first report of a syndrome of cerebral gigantism in childhood by Sotos *et al.*¹ many similar cases have been described. Sotos *et al.*² reviewed 105 other cases as well as 10 of their own. The condition was generally sporadic, but some reports indicated a genetic basis. Until now, there have been two reports of 4 families³⁻⁴ in which a dominant mode of inheritance was shown, considered most likely to be autosomal dominant, although dominant X-linked inheritance was not excluded. We report here the fifth example of dominant Sotos's syndrome.

We recently examined a 4-year-old girl because of speech delay and large body size—her height was over 97th centile, weight on 90th centile, and head circumference on 97th centile. Examination showed other signs of cerebral gigantism—such as a high arched palate, a high bossed forehead, hypertelorism, a flat nasal bridge, and prognathism. She had large hands and her shoes were size 3, the size worn by an average 10-year-old child. Bone age was advanced at 5.75 years. Banded karyotype was normal. She had slight mental retardation.

The mother was also examined as she, too, was large. Her height, weight, and head circumference were all above the 90th centiles, she had large hands and feet, pronounced prognathism, a very high palate, coarse facial features, and a deep hoarse voice. She had a full scale IQ of 85. Banded karyotype was normal. There was some enlargement of the lateral ventricles shown by computerised tomography. We consider that she has Sotos's syndrome. There was no evidence of gigantism in any male relative.

For both mother and daughter, the differential diagnosis was gigantism due to general genetic endowment, or gigantism due to Sotos's syndrome. Apart from the gigantism, both of them have many of the clinical signs of Sotos's syndrome. The diagnosis is more obvious in a child than an adult. This point is exemplified by the fact that until now the syndrome in adults has been detected only through the investigation of their children,⁴ so that the pediatrician plays an important role in detecting adult cases.

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