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SOTOS SYNDROME PRESENTING WITH EPILEPSY*

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A fourteen and a half year-old boy of tall stature presenting with epilepsy was diagnosed as having Sotos syndrome (cerebral gigantism) at the Hacettepe University Children's Hospital. Since EEG abnormalities are known manifestations in this syndrome, a case presenting with epilepsy is very rare. We believe that a presumed cerebral defect could create convulsions and would thus be the cause of cerebral gigantism whose etiology is still unclear. Thus patients with this syndrome should be followed up for the risk of epilepsy. *Key words: Sotos syndrome, epilepsy.*

Sotos syndrome (cerebral gigantism) was first described as a syndrome by Sotos and co-workers[1] in 1964. A hypothalamic defect has been suggested as a cause, but this has not been demonstrated functionally or at necropsy. Thus, the etiology of the disease remains unknown[1-4]. In 1983, Ashcraft et al[5] described an unidentified growth factor in the serum of patients with this syndrome. This factor also stimulated the erythroid precursor cells of a Laron-type dwarf whose cells were unresponsive to growth hormone. But the authors claimed that although this factor could be responsible for the altered somatic growth observed in this condition, the physiologic role of this growth factor in normal man as well as its pathogenic role in subjects with the syndrome remains to be established. In this paper, we report a case of Sotos syndrome presenting with epilepsy and review the literature.

Case Report

A fourteen and a half year old boy of tall stature was diagnosed as having epilepsy and was referred to the Hacettepe University Children's Hospital for further investigation. His past history revealed that he had graduated from a private school for mentally retarded children and was unable to continue his education.

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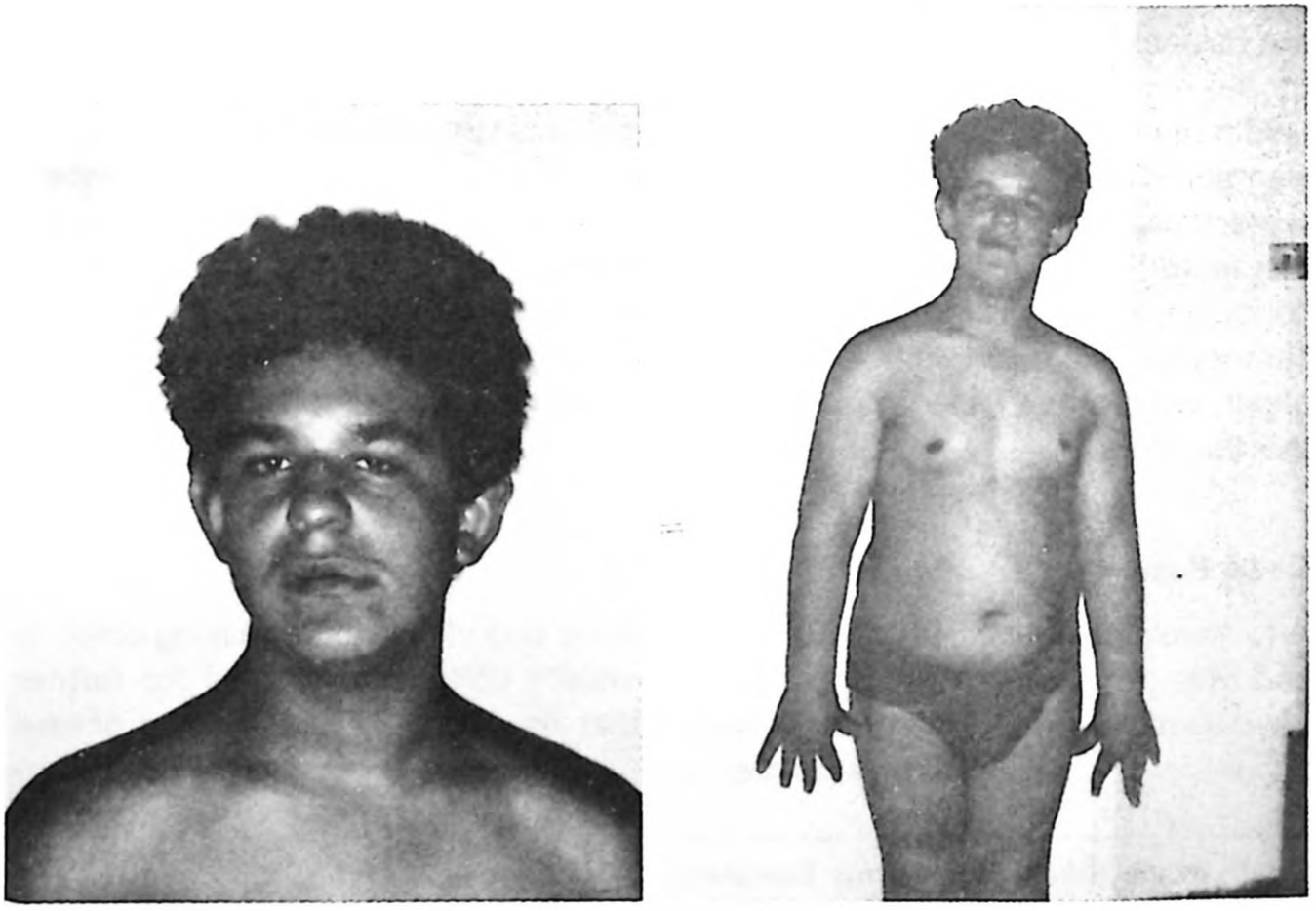
He had been having generalized convulsions for five years, and the diagnosis of epilepsy with abnormal EEG findings was established. Diphenylhydantoin therapy was initated in a daily dose of 5 mg/kg, and he responded well to treatment. Two years ago his enlarged hands and feet were noticed by his family.

Physical examination revealed a boy whose weight was 95 kg (above 95th percentile) and whose height was 177 cm (above 90th percentile). He had an arm span of 179 cm, an upper/lower ratio of 0.9 and a head circumference of 63 cm. He had a large head, prominent jaw and forehead, large hands and feet (Figs. 1,2), and wore a size 45 shoe. His sexual development was Tanner Stage IV. He was mentally retarded, had an awkward gait and his skeletal age was 15 years. Radiographs (Figs. 3-5) showed a large skull, no signs of increased intracranial pressure, and the sella turcia and clinoid processes were of normal contour. Radiographs of the hands showed them to be large and there was slight tufting of the terminal phalanges[5]. Lateral foot radiographs showed thickening of the soft tissue from the calcaneus. Computerized axial tomography demonstrated no enlarged ventricles.

Repeated fasting growth hormone levels were 0.50, 1.49, and 0.87 ng/ml (normal, 0-10 ng/ml). The oral glucose tolerance test yielded normal results.



Figs. 1, 2: General appearance of the patient; note the large head, prominent jaw and forehead, and large hands.

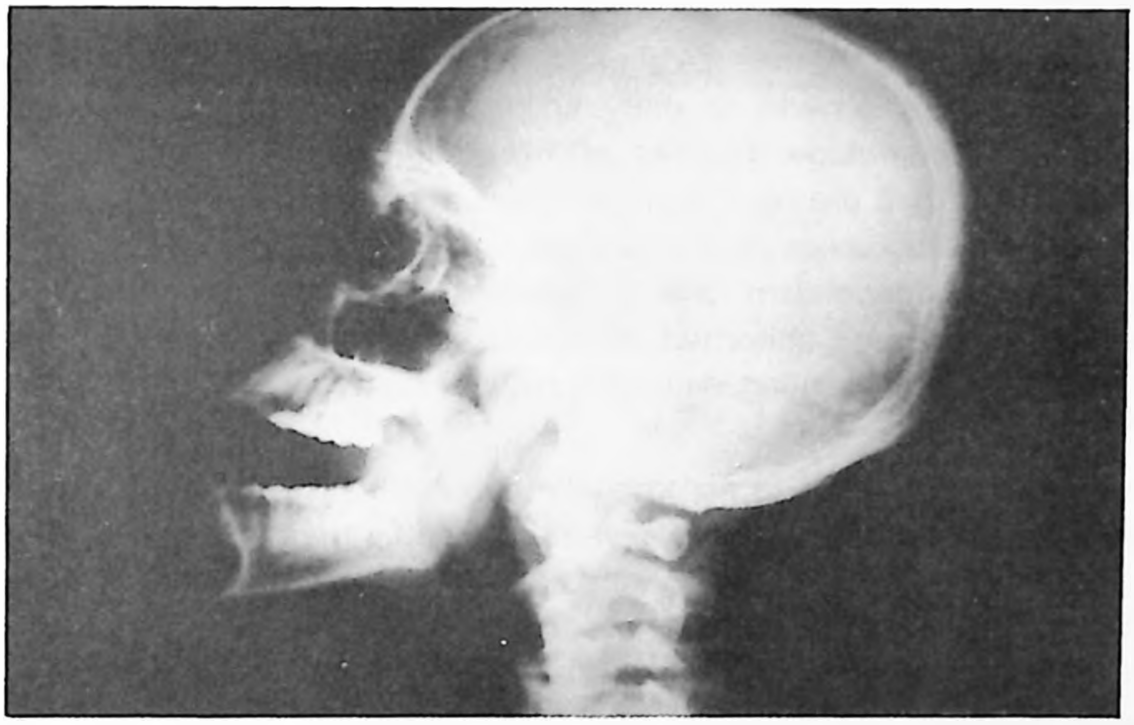


Fig. 3: Skull radiogram of the patient (lateral view); note the absence of signs of increased intracranial pressure and the normal appearance of sella turcica and clinoid processes.

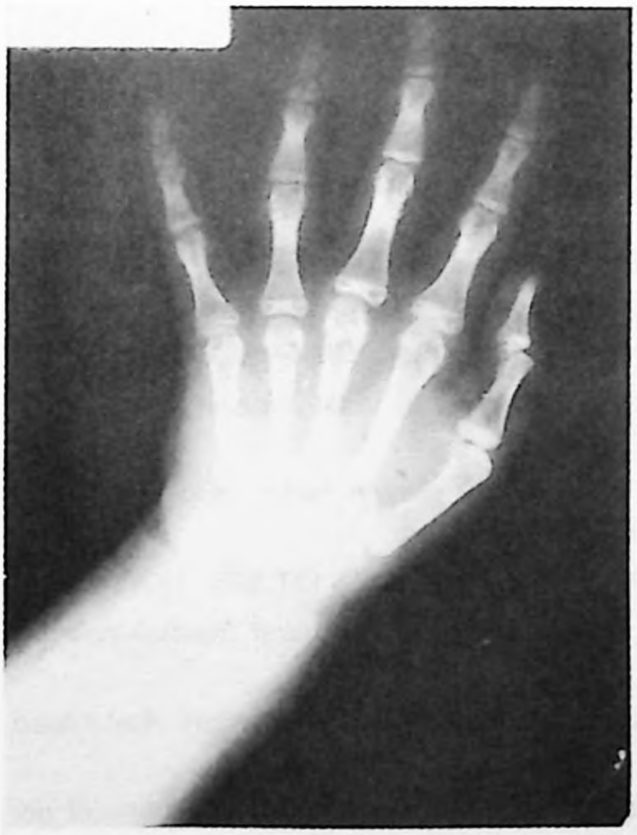


Fig. 4: Wrist X-ray demonstrating bone age of 15 years; note enlarged hand.

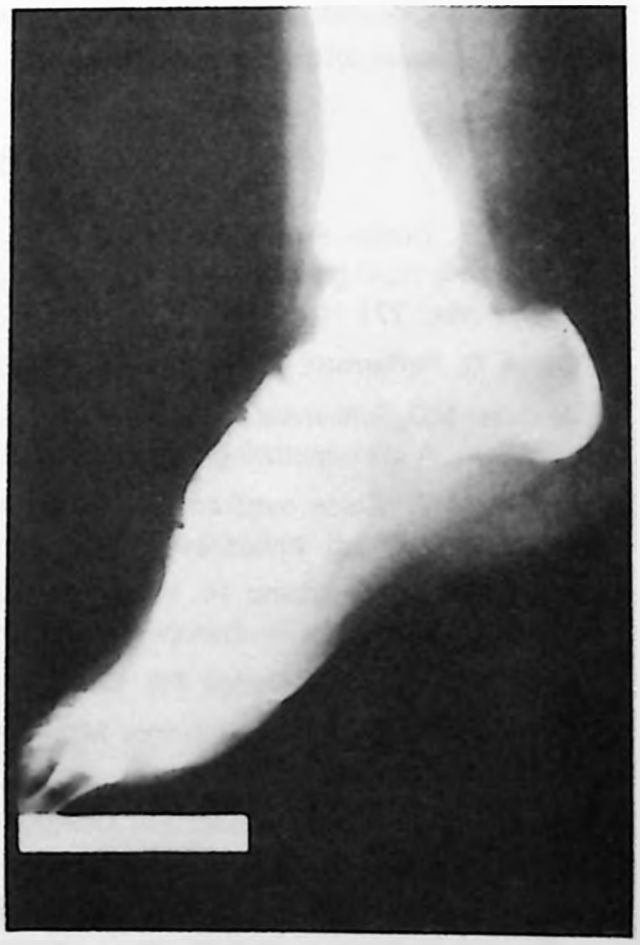


Fig. 5: X-ray of foot shows enlargement.

Discussion

According to Sotos et al[1], the typical pattern of cerebral gigantism is characterized by excessive rapid growth in early childhood, acromegalic features, and a non-progressive neurologic disorder with mental retardation. Mental retardation and an awkward gait are mostly present in patients with Sotos syndrome[1,4,6]. Several reports emphasize endocrinological abnormalities including thyrotoxicosis, primary hypothyroidism and a fourteen percent incidence of glucose intolerance[7-9]. Although abnormal electroencephalograms are common, to our knowledge a case presenting with epilepsy as the initial symptom is extremely rare[14].

Our case presented definite acromegalic characteristics, mental retardation an awkward gait, and epilepsy. There were no signs of intracranial pressure but since EEG abnormalities are known manifestations in this syndrome, a presumed cerebral defect in our patient could create convulsions[1-4]. His generalized convulsions responded well to 5 mg/kg diphenylhydantoin therapy. Although advanced osseous maturation is said to accompany cerebral gigantism, our patient did not show any osseous maturation[1-4].

Patients with Sotos syndrome may be at high risk for developing neoplasia, hepatic carcinoma and Wilms' tumor[10,11]. Therefore, at follow-ups, patients should be evaluated for these risk factors including epilepsy.

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