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# Cerebral gigantism with hydronephrosis: a case report

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A case of cerebral gigantism with hydronephrosis in a 20-month-old boy is described. Hydronephrosis is believed to be an additional association of the syndrome not hitherto reported.

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Since the first report of cerebral gigantism (Sotos Syndrome) more than 100 cases displaying similar features have been described. Additional findings have also been reported in association with this syndrome (Milunsky et al. 1967, Appenzeller & Snyder 1969, Evans 1971, Mikulowski et al. 1972). To these we add hydronephrosis.

## Case Report

A 20-month-old boy was admitted to King Khalid University Hospital with a history of rapid growth for one year. His mother noticed that he looked much bigger than other children of his age. He was born normally after full-term normal pregnancy. His birth weight and length were not recorded but the mother noticed that he looked bigger than her previous children. On the third day of his life he was admitted to hospital with cyanosis when he cried and jaundice. The parents were told that one of his kidneys was abnormal but no specific defect was given.

On examination, his milestones were delayed. He had a large body size. His anthropometric measurements showed a height of 97.5 cm (above the 90th percentile), weight of 19.4 kg (above the 90th percentile) and head circumference of 55.5 cm (97th percentile). He had a dolicocephalic head, prominent forehead, hypertelorism, high-arched palate, large hands and feet, and dorsolumbar scoliosis. He was mentally retarded. Otherwise he was normal. Neither the kidneys nor the bladder were palpable.

His haemogram, blood chemistry, renal and liver functions, and endocrine profile were normal. His bone age was 38 months. Computerized scan of the brain showed mild dilation of the ventricular system. Intravenous pyelography (Fig. 1) and ultrasound demonstrated hydronephrosis in the left kidney but the micturating cystourethrogram was normal.

## Discussion

The syndrome of cerebral gigantism was first described by Sotos et al. in 1964. Jacken & Vander Schueren-Lodoweycky (1972) in a review of 60 cases, stated that the most common clinical features were gigantism

**Fig. 1.** I. V. P. demonstrating hydronephrosis.

(100%), prominent forehead (96%), high-arched palate (96%), hypertelorism (91%), dolicocephaly (84%), developmental retardation (83%), large hands and feet (83%), advanced bone age (74%) and lack of fine motor control (67%). Our patient conforms closely to the above criteria. Certain additional findings have also been occasionally reported. Among these are cataract (Weber & Hirch 1978), juvenile macular degeneration (Ferrier 1980), syndactyly (Mikulowski et al. 1972), kyphoscoliosis (Milunsky et al. 1967), peripheral dysotosis (Evans 1971), and functional megacolon and autonomic failure with persistent fever (Appenzeller & Snyder 1969). Sugerman et al. (1977) reported a patient with cerebral gigantism who developed a hepatoma. Our patient has hydronephrosis on the left side in addition to cerebral gigantism, an association which has not been described before. It was first detected when the patient was only a few days old suggesting that it is most probably of congenital origin. The patient never had any problems with his urinary tract at any time. The pathogenesis of this disorder is obscure. Various modes of inheritance have been described (Gemelli et al. 1974, Zanana et al. 1977). The investigations we have done in our patient as well as those in all previous studies could not detect an underlying cause which therefore still remains unknown.

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