Congenital medullary tubular stenosis

A case of Caffey-Kenny syndrome

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We report a case of congenital medullary stenosis of the tubular bones, or Caffey-Kenny syndrome. The patient was referred to our department with retarded psychomotor development and contractures.

Congenital medullary stenosis of the tubular bones, or Caffey-Kenny syndrome, was originally described by Kenny and Linarelli (1966). It is characterized by proportionate dwarfism, transient hypocalcemic convulsions, normal intellectual and motor development. Radiographically, the medullary cavities of the tubular bones are reduced, with normal or increased cortical thickness and possible absence of the diploic space of the calvaria in addition to delayed closure of the anterior fontanelle (Caffey 1967, Frech and McAllister 1968, Kenny and Linarelli 1966, Sarria et al. 1980).

Our case had clinical characteristics different from earlier reports.

Case report

The patient, a girl, was born during the 37th week and weighed 2,100 grams. She had an Apgar score of 8/9, neonatal jaundice, bilateral congenital cataracts, and osseous abnormalities. She also had repeated generalized seizures that responded to antiepileptic medication. She was hospitalized several times for gastroenteritis, urinary tract infection, and pneumonia. The psychomotor development was retarded; she first sat up at 3 years of age, stood at 4.5 years, began to walk at 5, and started to speak at aged 6 years.

Both the maternal grandparents and the parents were consanguineous. A maternal uncle was mentally retarded. The patient's mother and brother had radiographic evidence of medullary tubular stenosis, although no clinical signs or symptoms of the disorder were found.

The girl was referred to us at aged 6 years after several episodes of seizures. She had severe motor and intellectual impairment along with generalized hypotonia, platyrrhiny, hypoplastic mandible, cleft palate, crossbite, pseudohypertelorism, mongoloid palpebral fissures, mild bilateral epicanthus, exophthalmos, and cataracts. The hands and feet were in hyperextension with rigid interphlangeal articulations. She also presented a mild kyphosis, flatfeet, valgus knees, and a waddling gait. Moderate contractures of the elbows, hips, and knees were noted.

Laboratory data: calcium 2.2 mmol/L, phosphorus 1.6 mmol/L, alk. phosphatase 3.2 μmol.s^{-1}/L, LDH 9.0 μmol.s^{-1}/L, CPK 1.8 μmol.s^{-1}/L, aldolase 74.9 nmol.s^{-1}/L. The patient also had hypogammaglobulinemia. All the other laboratory values were normal.

The electroencephalogram showed bilateral slow waves particularly over the frontal areas. A computed axial tomography revealed an area of low density in the left frontal region and lateral ventricular asymmetry. Radiography was compatible with medullary tubular stenosis, and bone maturation was retarded 2–3 years (Figure 1). A bone marrow biopsy revealed increased cortical thickness with increased calcification, narrowing of the medullary cavity with reduced osseous matrix, and widening of the trabeculae.

Discussion

Medullary stenosis of tubular bones, or Caffey-Kenney syndrome, is a constitutional bone disease with autosomal dominant inheritance, as in

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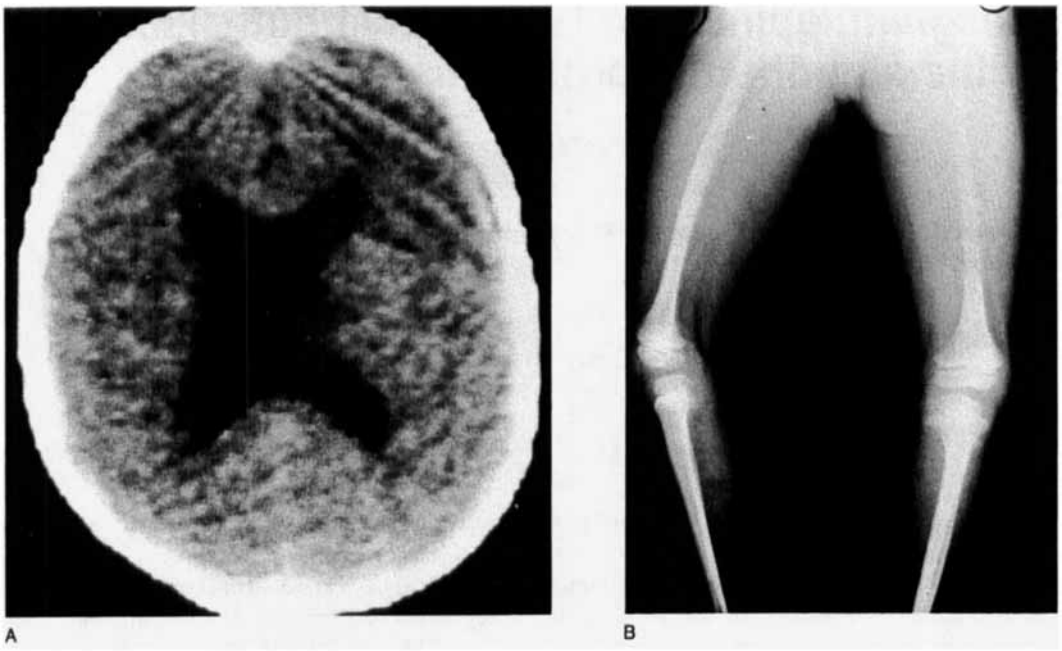

Figure 1. A 6-year-old girl with congenital medullary stenosis of the tubular bones; Caffey-Kenny syndrome.
A. CT. Ventricular asymmetry.
B. Increased cortical thickness and narrowing of medullary cavity.

our case (Beighton et al. 1984). In contrast, Sarria et al. (1980), however, proposed heterogeneous genetic transmission, with both dominant and recessive autosomal varieties.

During the patient's hospital stay at 7 years of age, she presented generalized convulsions not associated with hypocalcemia. The cases reported by Kenny and Linarelli (1966) and by Frech and McAllister (1968) had hypocalcemic seizures that did not respond to phenobarbital, but were effectively treated with calcium. Our patient's EEG results, the effectiveness of clonacepan, and the absence of seizures while on treatment with phenobarbital (Luminal®) made us believe that we were facing a coincidental case of epilepsy, hypotonia, delayed psychomotor development, and contractures secondary to a cerebral lesion.

The unique features in our case are the delayed psychomotor development and contractures manifested in a patient with Caffey-Kenny syndrome. Even though Epstein et al. (1968) referred to 2 reported cases of tubular stenosis with kyphosis and contractures, these did not have many of the major clinical signs of the Caffey-Kenny syndrome. Our patient not only had the typical clinical, laboratory, and radiographic signs, but also demonstrated alterations not previously observed in the classical descriptions.

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