Nigerian Journal of Paediatrics 2002; 29 (4): 122-4

Camptomelic Dysplasia in One of Twins -A Case Report

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Summary

Lagunju IA, Omokhodion SI, Brown BJ, Adeniyi OF. Camptomelic Dysplasia in One of Twins – A Case Report *Nigerian Journal of Paediatrics* **2002; 29:122.** Camptomelic dysplasia, a very rare and lethal form of skeletal dysplasia is reported in one of a set of twins. He presented on the second day of life with short, deformed lower limbs and respiratory distress from birth and died on the fifth day of life. This report highlights the features of camptomelic dysplasia and draws attention to its occurrence in a Nigerian child. With advances in neonatal life support, there is need to proffer solutions to the problems that presently make the prognosis of camptomelic dysplasia, poor.

Introduction

CAMPTOMELIC dysplasia is one of the numerous genetic skeletal dysplasias.[1] It is characterized by dwarfism, osseous and cartilage anomalies including prenatal bowing of the lower extremities, with pretibial skin dimpling, peculiar facies, respiratory distress and neonatal death.[2] The affected bones are long, slender and usually bent at their mid-points. Cutaneous dimples may overlie the points of maximal curvature.[3] Most patients with this condition die from respiratory insufficiency during the neonatal period.[4,5] The incidence of camptomelic dysplasia is unknown but it is reported to be very rare.[4,5] This report describes a case of camptomelic dysplasia in one of a set of twins, admitted into the children's emergency ward of the University College Hospital, Ibadan. To the best of our knowledge, it is the first such case to be reported in a Nigerian child.

Case Report

AK, a male neonate and second of a set of twins, was first seen on the second day of life, in February 2002 at the University College Hospital, Ibadan. He presented with breathlessness, weak cry, and deformity of both lower limbs. He was a product of twin pregnancy and was delivered to a para^{1+0} woman at a local government health centre in Ibadan. Pregnancy was uneventful and his mother who took routine haematinics during pregnancy, received antenatal care at the local government health centre. There was no history of the use of other drugs, herbal medications or exposure to irradiation during pregnancy. Labour was spontaneous, lasted about eight hours and both babies were delivered by spontaneous vaginal delivery, vertex presentation. AK, the second twin, was delivered five minutes after the first. The Apgar scores were not taken at the place of delivery. He was however, noticed to be breathless from birth, had a weak cry and was unable to suck. He was also observed to have deformity of both lower limbs.

Examination revealed a dysmorphic male neonate in severe respiratory distress, with flaring alae nasi, intercostal and subcostal recession. He had a flattened facial appearance with a prominent forehead, micrognathia, low set ears and bilateral pre-auricular skin tags (Fig. 1). There was a skin dimple over the anterior aspect of both tibiae, with shortening and bowing of the lower limbs, and bilateral talipes equinovarus deformity. He weighed 2.5kg, his length and occipito-frontal circumference were 36.5cm and 36cm, respectively. He had a respiratory rate of 72 breaths per minute and was grunting; the breath sounds were however, vesicular and there were no added sounds. His pulses were regular and of normal volume.

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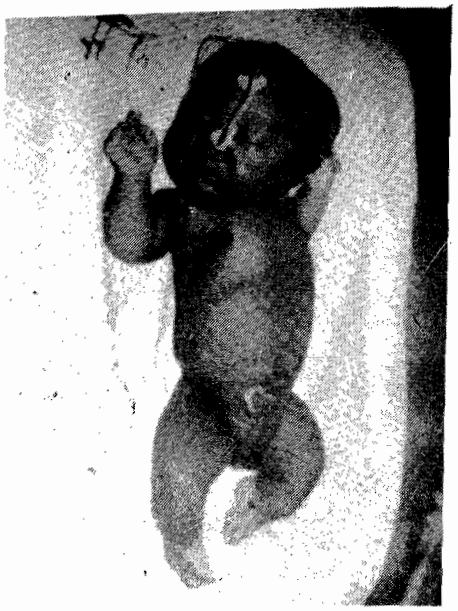

Fig. 1. Camptomelic dysplasia. Note the large head, flat face, low-set ears, short and bowed lower limbs, with dimples at the maximum point of bowing.

Fig. 2. Radiograph of the lower limbs showing the poorly developed slender, angulated femurs and tibiae, with hypoplasia of the fibulae.

His heart rate was 136 beats/min and he had a grade 3/6 systolic murmur, which was maximal at the left lower sternal edge. Abdominal examination did not reveal any abnormality. The external genitalia was ambiguous. Radiograph of the lower limbs showed slender femurs and tibiae, with bowing and angulation

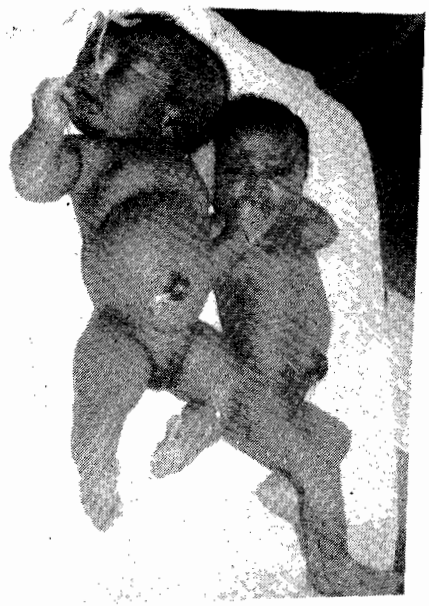

Fig. 3. AK and the second twin. Note the patient's very short limbs and the normal second twin.

of these bones, and hypoplasia of the iliac bones (Fig. 2). He was placed on intravenous fluids and intranasal oxygen. A full sepsis screen was done and no evidence of sepsis was found. His condition however remained poor with severe respiratory distress, until he died on the fifth day of life. The second twin was found to be a normal male infant (Fig. 3).

Discussion

Camptomelic dysplasia is a very rare condition.[4] One of the earliest descriptions of this condition was by Bound *et al*[6] in 1952. Bain and Barrett[7] also reported a case of congenital bowing of the long bones associated with skin dimpling in 1959. The disease became more broadly recognized as camptomelic dysplasia in the 1970s when Spranger *et al* [8] utilized the term "camptomelique" meaning "bent limb" to epitomize the disorder. Camptomelic dysplasia is associated with short-limbed dwarfism affecting selectively, the lower limbs with anterior bending of the femur and tibia over which there are pretibial skin dimples.[1,4,5] The calvarium is large with disproportionately small facies. The ears are low set and there is micrognathia. [1,4,5] Other abnormalities that have been described in these patients include cardiac defects (patent ductus arteriosus), hydronephrosis, renal medullary cysts, cleft palate, tracheomalacia and mental retardation with various brain abnormalities. The typical radiographic features of this condition include long, slender, bent tibiae and

femurs, hypoplastic fibulae, scapulae and iliac bones; others include 11 pairs of narrow, wavy ribs with a small thoracic cage and slender clavicles. There is retardation of bone maturation and there may be associated kyphoscoliosis or scoliosis.[1,3-5] Our patient exhibited most of the typical features of the disease as enumerated above. He had a cardiac murmur but the exact nature of the cardiac defect could not be ascertained because of the poor clinical state.

The exact cause of camptomelic dysplasia is unknown[4] but it is widely believed to be genetic with autosomal recessive inheritance.[1,5] Females are more commonly affected than males,[3-5] but cases of intersex have also been reported.[3] AK had an ambiguous genitalia but the genetic sex could not be ascertained by karyotyping before death. The majority of patients die during the neonatal period from respiratory insufficiency presumably due to a small thoracic cage, narrow larynx and hypoplasia of the tracheal rings. With advancements in neonatal life support facilities, Coscia *et al*[9] have reported increasing survival beyond the neonatal period among these patients but some lingering difficulties remain due to feeding problems, failure to thrive, spinal deformities requiring orthopaedic management and severe mental retardation.

Since pre-natal diagnosis is possible, the almost uniformly poor prognosis that presently marks this condition might tempt some to advocate termination of pregnancy on detection. Such a line of action would be controversial, considering the raging battle between pro-life and pro-abortionists. In any case, with further advancements in knowledge and understanding of this condition, it may not be too long before most of these problems are overcome, enabling longer life for these rather unfortunate individuals. Questions regarding the risk of recurrence of the disease in subsequent siblings are also bound to arise from the parents of affected children. These questions require appropriate answers and underscore the need to study the children more closely. The present report therefore draws attention not only to the occurrence of this condition in Nigeria but also to the need to provide solutions to the problems that presently make the prognosis of camptomelic dysplasia, poor.

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