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Craniodiaphyseal Dysplasia : A Case Report[1]

Gye Yeon Lim, M.D., Jeong Mi Park, M.D., Jae Mun Lee, M.D., Choon-Yul Kim, M.D., Kyung Sub Shinn, M.D.

Craniodiaphyseal dysplasia is a rare hereditary bone dysplasia characterized by craniofacial hyperostosis and diaphyseal dysplatic changes. We reviewed the clinical, radiologic and laboratory features of one such case.

Index Words : Bones, dysplasia
Hyperostosis

Craniodiaphyseal dysplasia, as described by Gorlin (1), is a very severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. Craniodiaphyseal dysplasia remains a rare disorder, with fewer than 25 case reports published (2). Progressive bony encroachment upon cranial foramina leads to severe neurologic impairment in childhood.

Using plain radiographs, we encountered a case of craniodiaphyseal dysplasia.

CASE REPORT

A 17 month-old girl, the first-born of the family, was admitted for the evaluation of recurrent otitis media and left facial nerve palsy. No significant birth or family history was uncovered.

The pregnancy and delivery were unremarkable with no abnormalites noted at birth. Early physical and mental development were normal. On physical examination, a flattening of the nasal bridge, mild hypertelorism and conjunctival injection were seen. Except for left facial nerve palsy, no other cranial or skeletal abnormality was detectable.

Radiologic studies, however, revealed moderate to severe sclerosis and hyperostosis involving the cranium and facial bones (Fig. 1a). There was obliteration of the sinuses and mastoids (Fig. 1b) ; the ribs and clavicles were thickened and sclerotic (Fig. 2) ; the long bones were generally cylindrical in appearance because of diaphyseal, endosteal, cortical thickening, and showed a lack of modelling (Fig. 3). The short tubular bones of the hand lacked normal tubulation and also showed some diaphyseal thickening (Fig. 3).

Laboratory studies revealed mild elevation in the level of serum alkaline phosphatase, calcium and phosphate levels were normal. The patient was kept under continous surveillance in the out-patient's department and was treated symptomatically.

DISCUSSION

Gorlin et al (1) suggested in 1969 that the term "craniodiaphyseal dysplasia"(CDD) should be used to refer to a group of diseases within a spectrum of craniotubular dysplasia rather than to a single discrete disease entity. They defined it is "a very severe bone disorder characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones"(1).

Typically, patients with CDD may present in early infancy with facial abnormalities. Bony overgrowth results in paranasal bossing and apparent hypertelorism (2). Affected infants attract to medical attention, however, because of respiratory difficulty owing to nasal obstruction before the characteristic facial appearance has developed. Recurrent dacryocystitis by progressive stenosis of the nasolacrimal duct and recurrent otitis media may occur (3). The infant in our case came to our attention because of recurrent otitis media and conjuctivitis in association with bony overgrowth. Bony encroachment upon cranial foramina causing cranial nerve compression (especially Ⅱ and Ⅷ) leads to progressive visual and auditory impairment with ultimate blindness and deafness (2, 4, 5). In our case, facial nerve paralysis was caused by marked bony overgrowth over temporal bone.

[1]Department of Radiology, Catholic University Medical College
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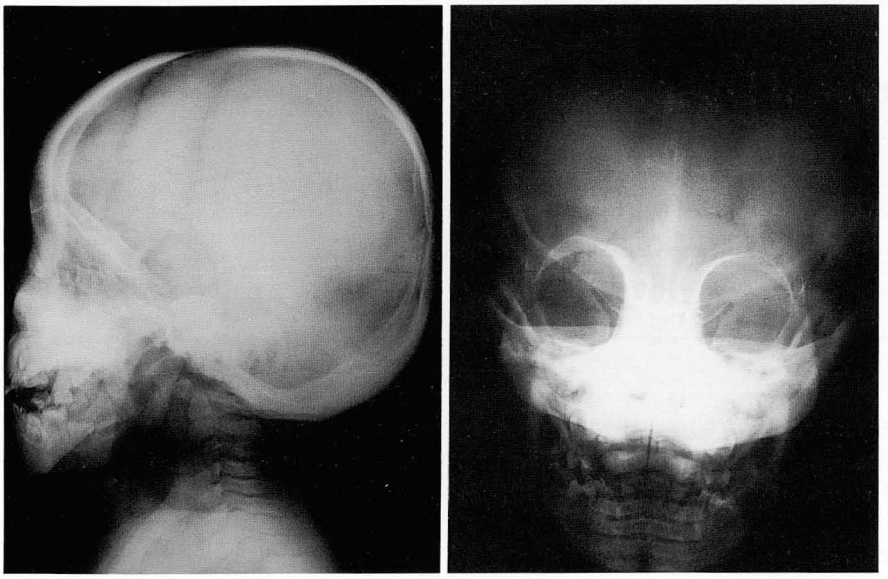
Address reprint requests to : Gye Yeon Lim, M.D., Department of Diagnostic Radiology, St. Mary's Hospital # 62, Youido-dong, Yongdungpo-gu, Seoul, 150-101 Korea. Tel. 82-2-789-1277



a b

Fig. 1. Radiographs of the skull show hyperostosis of the calvarium, with marked sclerosis of facial bones and skull base (a). The paranasal sinuses and mastoids are obliterated (b).

The underlying cause of this dysplasia is unknown. A review of the metabolic profiles of CDD cases reveals that the only consistent biochemical abnormality is a marked rise in serum alkaline phosphatase levels, suggesting that a major abnormality is grossly excessive osteoblastic activity (6). Calcium and phosphate levels are normal, however, as in our case.

Radiologically, the whole skull including facial bones and mandible shows severe sclerosis and hyperostosis with nasal obstruction and obliteration of the sinuses. The appearance of long bones varies in severity and distribution, but the long bones are generally cylindrical in appearance because of diaphyseal, endosteal, cortical thickening, and a lack of modelling.

There is usually moderate thickening and sclerosis of the ribs, clavicles and pelvis. Sclerosis of the spine has been found in some cases, but no discernable abnormality was demonstrated in our case.

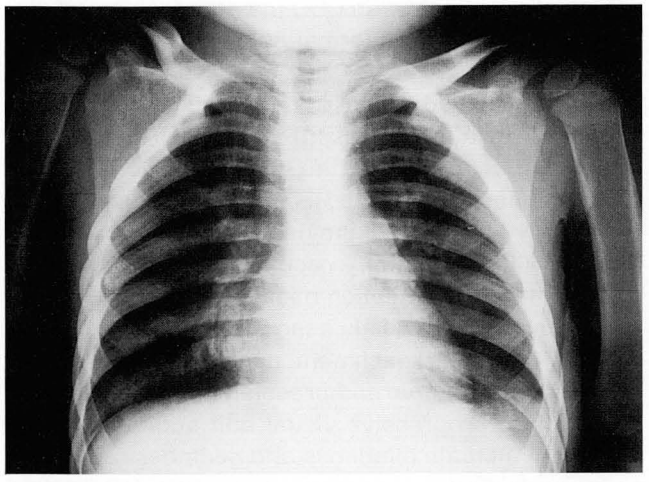


Fig. 2. Radiograph of chest shows marked sclerotic thickening of ribs and clavicles.

CDD must be distinguished from Engelmann's disease (diaphyseal dysplasia). Compared with the former, the latter shows a milder degree of craniofacial involvement and milder changes occurring in long bones (7).

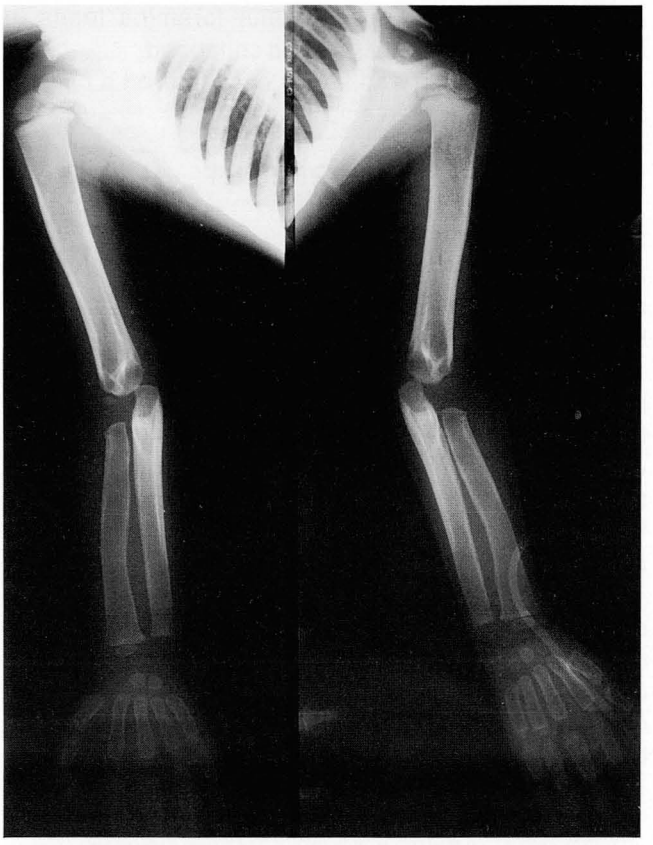


Fig. 3. Radiographs of right (a) and left (b) upper extrimities show expanded diaphyses of long bones with a straight cylindrical shape. Both hands demonstrate short tubular bones with broad diaphyseal expansion.

Cases of craniometaphyseal dysplasia (CMD) resemble those of CDD in that craniofacial changes are similar, but radiologic examination of the long bones distinguishes the two conditions. In CMD, metaphyseal widening and cortical thinning gives rise to a club-shaped configuration of long bone, whereas in CDD, diaphyseal changes appear as a cylindrical configuration(3, 7).

Surgical decompression of cranial foramina is possible, but there is risk of brainstem compression and the benefits of such a procedure are only short-term, as bony overgrowth inevitably recurs (3). The partial responsiveness of CDD to synthetic calcitonin therapy has been reported (8).

Several severely-affected children have died between the ages of 7 and 16 years (9).

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두개-골간 이형성증 : 1예보고[1]

[1]가톨릭대학교 의과대학 방사선과학교실

임 계 연 · 박 정 미 · 이 재 문 · 김 춘 열 · 신 경 섭

두개-골간 이형성증(craniodiaphyseal dysplasia)은 두개안면부의 골화과잉증(hyperostosis)과 골간 이형성증을 특징으로 하는 매우 드문 유전성 질환이다. 최근 반복적인 중이염과 안면신경마비 증상으로 내원한 여아에서 두개-골간 이형성증 1예를 경험하였기에 이의 특징적인 방사선학적 소견 및 임상적 특징을 문헌고찰과 함께 보고하는 바이다.

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