PYCNODYSOSTOSIS : REPORT OF TWO CASES

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SUMMARY

Two familial cases of pycnodysostosis are presented. The clinical and radiological features of cases in a boy with traumatic intracerebral hematoma and a girl with anemia are presented with a brief review of the literature.

Key Words : Anemia, Intracerebral hematoma, Pycnodysostosis

INTRODUCTION

The term pycnodysostosis was coined by Maroteaux and Lamy in 1962 (1). The typical findings of this syndrome have been described as follows; short stature, separated cranial sutures and open fontanels, loss of mandibular angle, partial aplasia of terminal phalanges of the fingers and toes, generalized condensation of bone shadow, bone fragility, and autosomal recessive inheritance (1,2). It is considered by many to be a variant of osteopetrosis (3).

The purpose of this paper is to report two familial cases of pycnodysostosis observed by the authors.

CASE REPORTS

A boy aged 8 years was admitted to our clinic with left hemiparesis complaint, after minor blunt head injury. He was the second child in a family of four children (two boys and two girls). The parents were first step cousins (from step grandfather). Both parents, one brother, and one sister were normal. Second sister was on treatment for anemia. There was no previous pycnodysostosis history in the family.

Upon examination, it was observed that the boy was of markedly short stature, and his weight was that of a five - year old. Head showed markedly frontal bossing, open fontanels and sutures, low hairline, receding chin, prominent eyes.

Skeletal survey : Skull examination revealed open anterior fontanel, separated sutures, presence of wormian bone, aplastic frontal sinuses, hypoplastic other paranasal sinuses, increased craniofacial ratio, hypoplastic maxillary bones, prominent occipital and frontal bones, absence of mastoid aeration, hypoplastic mandible, and flattened mandibular angle (Figs. 1a, 1b).

Extremities : Narrowed medullary canal in long tubular bones, increased bone density, bilaterally coxa valga deformity, relatively short and widened middle phalanges, slightly acroosteolysis at terminal phalanges, were observed.

Pelvis and spine : Diffuse increased bone density, anterior notching at lumbar vertebras (Fig. 2).

Cranial computed tomography showed right intracerebral hematoma, widely open anterior and posterior fontanels (Fig.3).

The patient was followed-up with anti-edema therapy for intracerebral hematoma and was discharged with full recovery.

In the search of consanguinity, pycnodysostosis was found in one of the sisters (Case 2). She was five years old, but the appearance and the measurements were according to that of a three - year - old. The typical findings of pycnodysostosis, as the first case were present. Anemia and hepatosplenomegaly were also present.

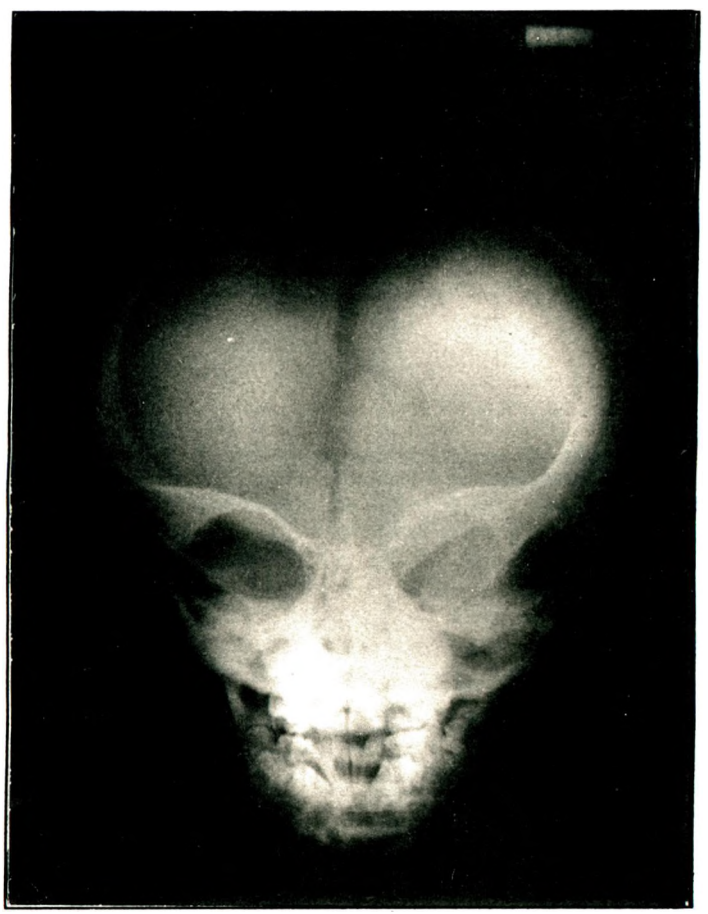
DISCUSSION

Pycnodysostosis is a disease of autosomal recessive inheritance, the abnormal chromosome being a member of the G 22 pair (4,5). Consanguinity of parents has been reported in about 25 % of cases. Both sexes are equally affected (5). Only 125 cases have been reported in the literature (6).

Pycnodysostosis is to be differentiated from osteopetrosis, cleido-cranial dysostosis, and acroosteolysis syndromes (5,7,8). The unique combination of the radiographic findings in pycnodysostosis makes the diagnosis (2,6,8). Our findings are in accordance with literature.

In the recent relevant literature, traumatic linear fracture, epidural hematoma, hydrocephalus, visceromegaly and anemia caused by extramedullary hematopoesis, complications of upper airway obstruction as part of pycnodysostosis, rarely seen bone fractures, eye and teeth anomalies are reported beside of the typical findings of pycnodysostosis (3,9-13).

a



b

Fig 1 (a - b) X - ray film of the skull shows separated cranial sutures, open fontanels, wormian bone, hypoplastic facial bones and mandible, loss of mandibular angle and absence of frontal sinuses.

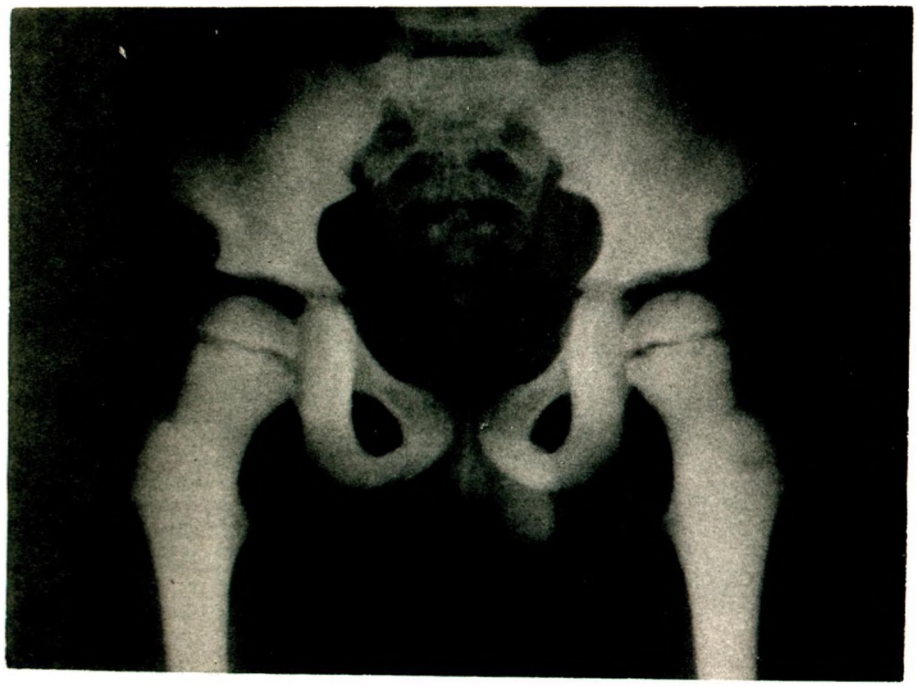


Fig 2. Roentgenogram of the pelvis and femora showing generalized increased density of bones and coxa valga deformity. Note the markedly narrowed medullary canals.

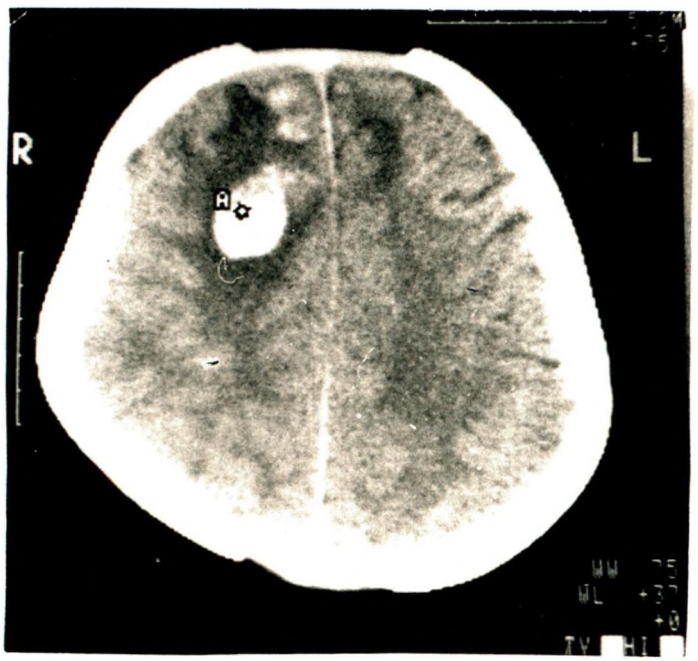


Fig 3. Cranial computed tomography shows intra-cerebral hematoma in the right frontal lobe. Note that the anterior and posterior fontanels are open.

Complications, such as atypical fractures due to bone fragility, are well known in pycnodysostosis. However, there was minor blund injury resulted with frontal intracerebral hematoma in the present case.

Therefore, we suggest that open fontanels and sutures in pycnodysostosis may be providing better protection of the head.

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