

PYCNODYSTOSIS — REPORT OF A CASE.

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SYNOPSIS

A case of pycnodysostosis is reported. This appears to be the first case reported in Malaysia. The characteristic clinical and radiological features are described. The differentiating diagnostic features of pycnodysostosis, osteopetrosis, cleidocranial dysostosis and osteogenesis imperfecta are tabulated.

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INTRODUCTION

Pycnodysostosis was initially described as a variant of osteopetrosis (marble bones disease, Aibers-Schonberg disease) and cleidocranial dysostosis. In 1962, Maroteux and Lamy (1, 2) coined the term and gave it a distinct clinical entity with characteristic clinical and radiological features. The term pycnodysostosis is derived from the Latin words; 'pycnos' meaning 'dense', 'dys' meaning 'defective' and 'ostosis' meaning 'bone'.

Elmore (3) reviewed 33 cases in 1967. Since then, occasional cases have been reported in the medical literature.

This appears to be the first case reported in Malaysia.

CASE REPORT

A three and a half-year-old Chinese boy was referred to the Paediatric Unit of the Alor Setar General Hospital, Kedah by a general practitioner. The significant findings noted by the doctor were separated cranial sutures, widely open anterior and posterior fontanelles and frontal bossing. His diagnosis was hydrocephalus.

The patient was seen by a doctor in the Paediatric Unit and he confirmed the general practitioner's clinical findings. He thought the patient had hypothyroidism and he admitted the patient for further management.

The patient was born in the Alor Setar General Hospital. It was a normal full-term delivery and he weighed 3200g at birth. The maternal history was unevenful. The neonatal history was unremarkable. He was immunised against tuberculosis, diphtheria, tetanus, whooping cough and poliomyelitis. His developmental milestones were normal. His parents denied any history of recent trauma to the child. There was no past history of sustaining

fractures after minor trauma. He did not have frequent respiratory tract infections. There was no history of a tendency to vomit or to regurgitate after feeds in the past. He has never been hospitalised before. He is the youngest of 4 siblings. The other siblings and the parents are normal and are of normal stature. His parents did not give a history of consanguinity.

On admission, he weighed 12.2kg (25th percentile*). His standing height was 88cm (10th percentile*) and his head circumference was 48.5cm (50th percentile*).

He had a relatively narrow face with a small chin. His eyes had a slight antimongolian slant. His sclerae were blue in colour. The eyes were not proptosed. The nose and the ears were normal (Figs. 1a and 1b).

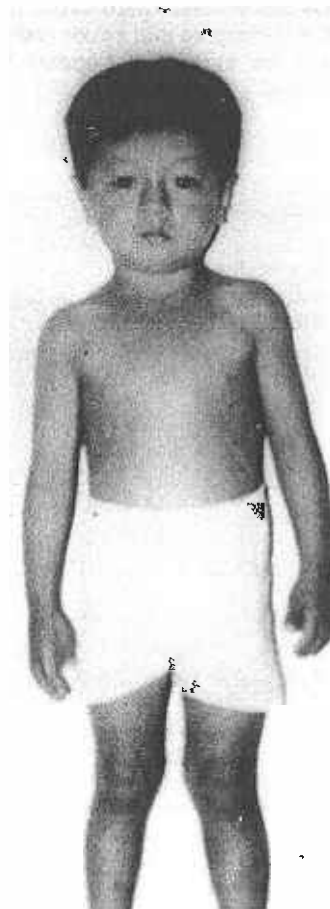


Fig. 1a

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Fig. 1b

He had frontal and bilateral parietal bossing. The sagittal, coronal and lambdoid sutures were separated and the anterior and posterior fontanelles were widely open.

Examination of the mouth revealed a narrow, high-arched, grooved palate. The teeth were hypoplastic and had a yellowish stain. There were multiple dental caries.

His fingers and toes were short. There was cutaneous wrinkling of the distal parts of the fingers and toes. The nails were dystrophic and spoon-shaped (Fig. 2).

There was no significant hepatosplenomegaly or lymphadenopathy.

The cardiovascular, respiratory and the central nervous systems were normal.

He had normal intelligence.

The results of laboratory investigations revealed a haemoglobin level of 11.0g pr 100ml, total white blood cell count 8,800 per mm^3 (with a differential count of 17% polymorphs, 80% lymphocytes and 3% eosinophils) and a platelet count of 400,000 per mm^3 . The bleeding and clotting times were normal. The serum calcium, serum inorganic phosphate and serum alkaline phosphatase were normal.

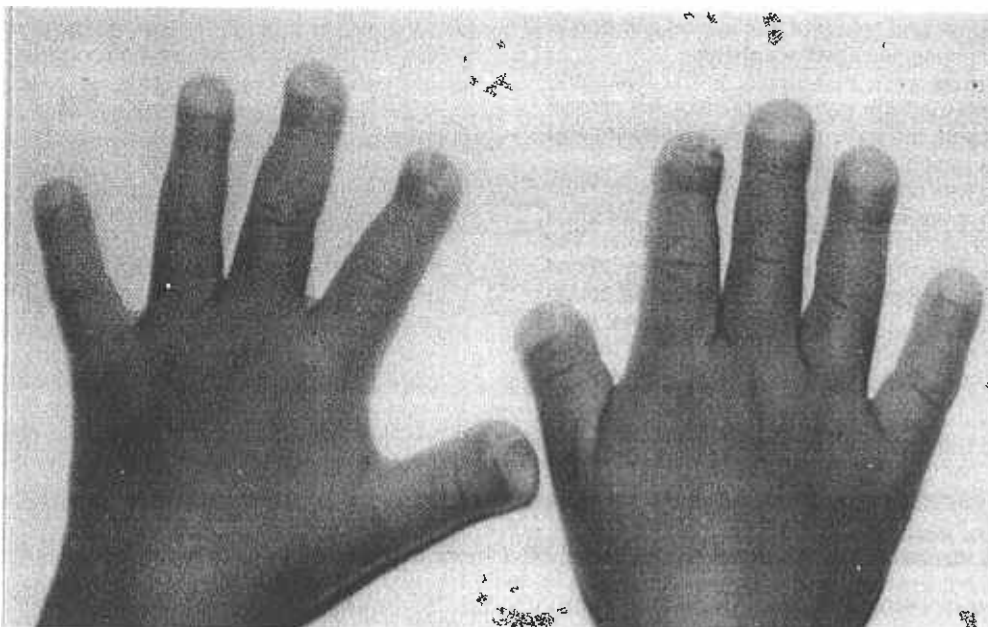


Fig. 2

The radiological findings were significant. The skull x-ray films showed widely separated cranial sutures and widely open anterior and posterior fontanelles. The facial bones and the paranasal sinuses were underdeveloped. The angle of the mandible was straightened (Fig. 3a and Fig. 3b). There was generalised increase in bone density. The medullary cavity of the long bones was narrowed but not obliterated. The acromial ends of the clavicles were hypoplastic. Apart from generalized osteosclerosis, x-ray of the pelvis was unremarkable (Figs. 4a and 4b). X-ray

films of the lower limbs showed transverse healed fractures of the midshafts of both tibias (Fig. 5). There was mild splaying of the distal part of the radius and the femur (Fig. 5 and Fig. 6). There was atrophy of the terminal phalanges of the fingers and the toes (Fig. 6). The bone age corresponded to his chronological age.

He was diagnosed as a case of pycnodysostosis on the basis of characteristic clinical and radiological findings.

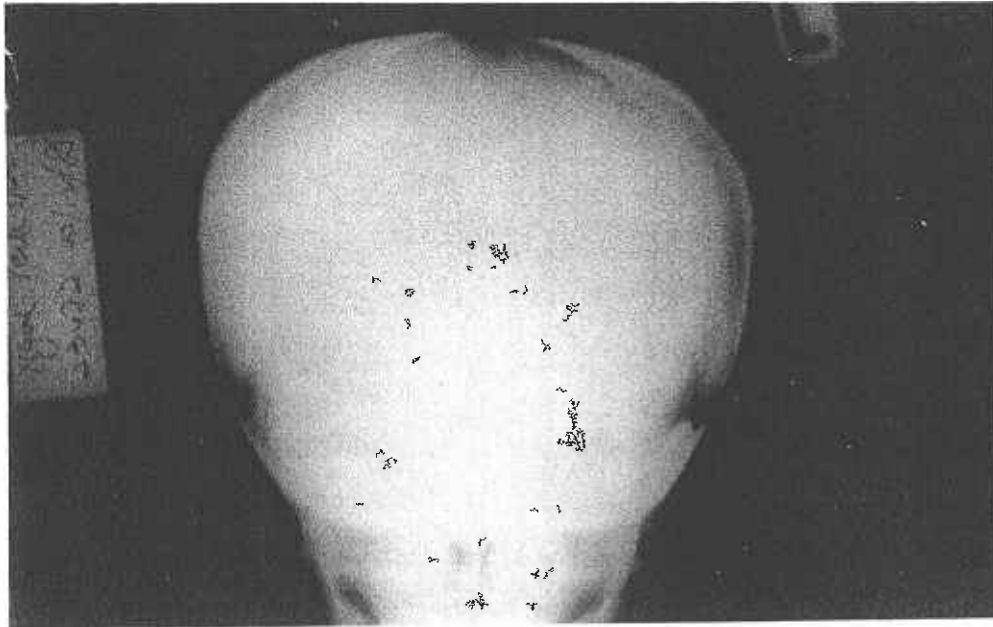


Fig. 3a

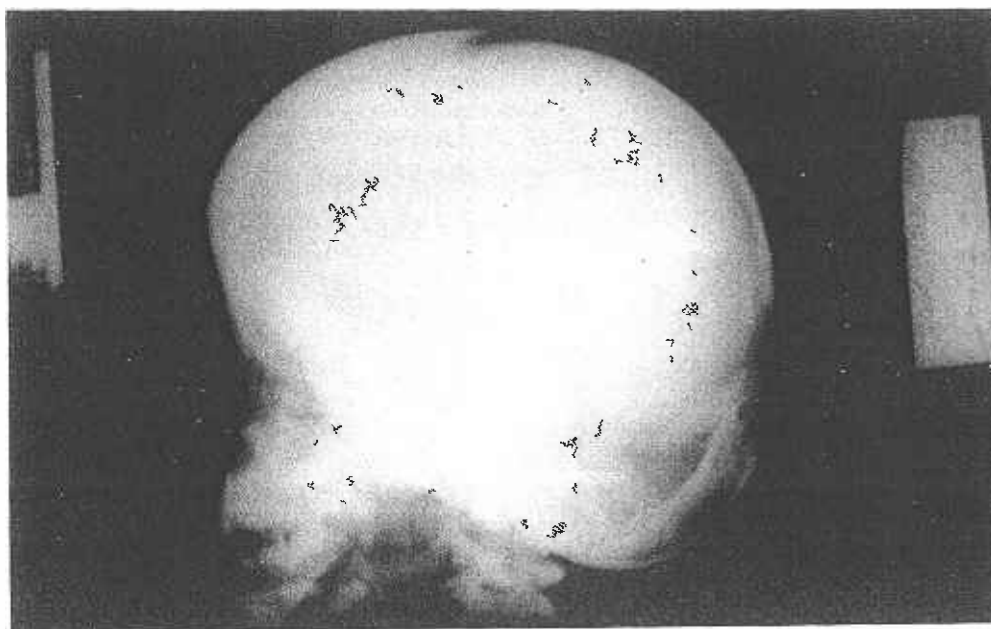


Fig. 3b

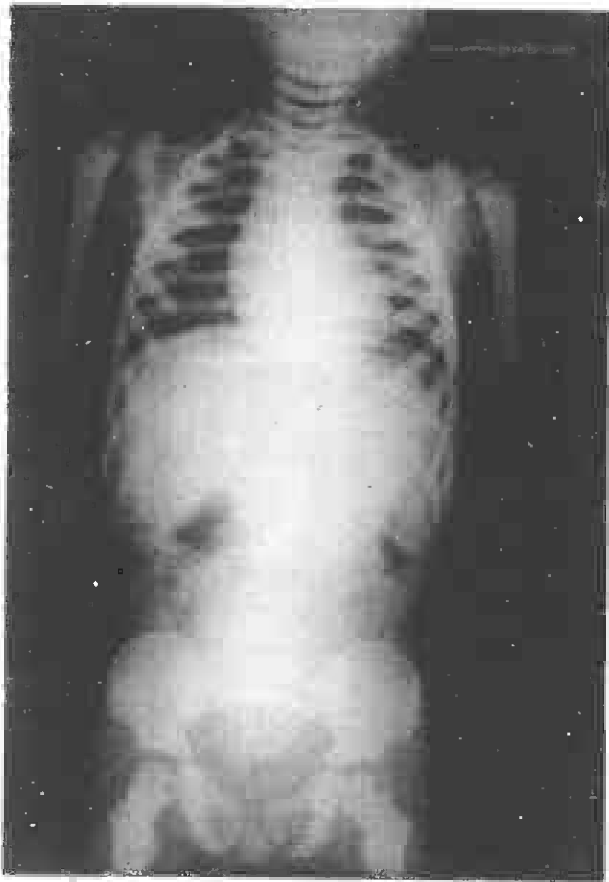


Fig. 4a

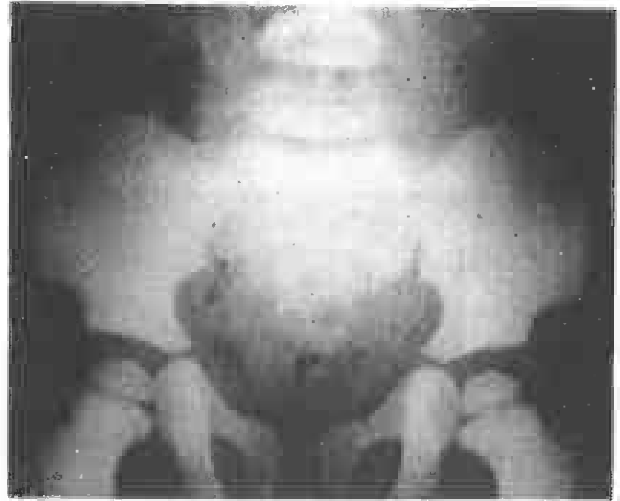


Fig. 4b



Fig. 5



Fig. 6

DISCUSSION

Awareness of the condition is necessary before one can make a diagnosis of pycnodysostosis. The main differential diagnosis are osteopetrosis, cleidocranial dysostosis and osteogenesis imperfecta. The differentiating diagnostic features are listed in Table 1.

This patient showed most of the characteristic features of pycnodysostosis. The majority of patients present with a history of sustaining fractures after trivial trauma. However, this patient did not present in this manner although radiological findings showed evidence of fractures in the past. Fractures are most often found in the lower limbs but clavicles and mandible are often affected too.

Other dental abnormalities which have been described are a double row of teeth and partial anodontia.

The nose is often beaked, a feature which is not seen in this case. Blue sclera is a rare finding in pycnodysostosis.

Nielsen(4) described 6 cases of pycnodysostosis in 1974. His patients had one or more of the following; pronounced snoring, stridulous respiration, frequent respiratory tract infections and a tendency to vomit or regurgitate during feeding. He attributed these to a long soft palate which almost reaches the root of the tongue in his cases. Previously, there was only one case who was reported to have frequent respiratory tract infections.(5) Other features have not been reported previously and this patient did not have any of these features.

Radiologically there is nonpneumatization or underdevelopment of the paranasal sinuses. The acromial ends

of the clavicles are often hypoplastic. The pelvis may have shallow acetabular angles. Bilateral coxa valga of the hips may be present. A lateral x-ray of the spine often show a notch described as a 'fish-tail deformity' on the anterior border of the vertebral bodies.

Reports in the medical literature have frequently cited this disorder as genetically determined with an autosomal recessive mode of inheritance. Sporadic cases have also been reported.

Affected patients are often of normal intelligence.

Very few histopathological or metabolic studies have been done. Metabolic studies done on one patient showed reduced bone formation and reduced bone resorption rates(6). Histopathological studies done on another patient showed the defect in bone is primarily one of disorganization of bone structure at the level of lamellar bone formation and the osteon(7). The basic defect in pycnodysostosis is still obscure.

The main medical problem is a tendency to fractures. Otherwise, the prognosis is good.

*Based on growth charts of Singapore children by H.B. Wong, C.Y. Tye & K.M. Quek and H.B. Wong and J.S.H. Tay.

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Table 1
DIFFERENTIATING DIAGNOSTIC FEATURES OF PYCNODYSTOSIS, OSTEOPETROSIS, OSTEogenesis IMPERFECTA AND CLEIDOCRANIAL DYSOSTOSIS.

Disease entity	Pycnodysostosis	Osteopetrosis	Osteogenesis Imperfecta	Cleidocranial
Genetic	autosomal recessive	autosomal dominant (mild type) autosomal recessive (severe type)	autosomal dominant (mild type) autosomal recessive (severe type)	dominant
Stature	short	usually normal	short	normal
Open fontanelles and cranial sutures	+	-	+	+
Frontal bossing	+	-	+	+
Blue sclerae	+	-	+	-
	(Occasional)			
Compression of cranial nerves	-	+	-	-
Tendency to fractures	+	+	+	-
Generalized osteosclerosis	+	+	-	-
Wormian bones (parietal)	+	-	+	+
Hypoplasia of facial bones	+	-	-	+
Paranasal sinuses underdeveloped/non-pneumatized	+	+	-	-
Angle of mandible straightened or obtuse	+	-	-	-
Hypoplasia/aplasia of clavicles	+	-	-	+
Atrophy of terminal phalanges	+	-	-	-
Obliteration of medullary cavity of long bones	-	+	-	-
Pelvis	coxa valga	coxa vara	coxa valga	dysplastic pubic rami
Anemia	-	+	-	-
Hepatosplenomegaly	-	+	-	-
Prognosis	fair	bad	bad	good

Note: + present
 - absent

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