

# HEREDITARY CLEIDO — CRANIAL DYSOSTOSIS

## WITH A NOTE ON THE ANOMALY OF CEMENTUM

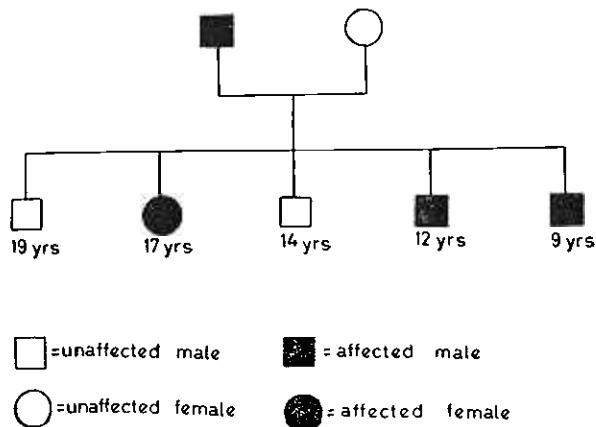
By Lee Khee Wee and V. K. Pillay

(The Departments of Dentistry and Orthopaedic Surgery, University of Singapore)

This curious syndrome of developmental anomalies affecting the skeletal structures and associated with characteristic dental abnormalities has been recorded in Singapore (Paul, 1957). Its occurrence in a family however has not been known in this country, and it is felt that the following cases, which involved a father and three children out of a family of five, are worth recording.



Fig. 1. Radiograph of shoulder region of Case I  
Note: Absent outer ends of clavicles and somewhat shallow glenoids.



### CASE REPORTS

#### Case 1

A male Chinese, aged 41 years, presented himself at the Dental Clinic, General Hospital, Singapore, requesting dental treatment. On examination, the patient was short and stocky, with a height of 4 ft. 11½ inches. His skull was brachycephalic, with prominent frontal bossing and a vertical depression over the front of the forehead. His maxillae were hypoplastic and there was relative prognathism of his mandible. He could approximate his shoulders in front and stated that three of his five children were afflicted in the same manner. His wife and his parents were normal. There was no history of epilepsy in the family and the degree of intelligence of all members was normal.

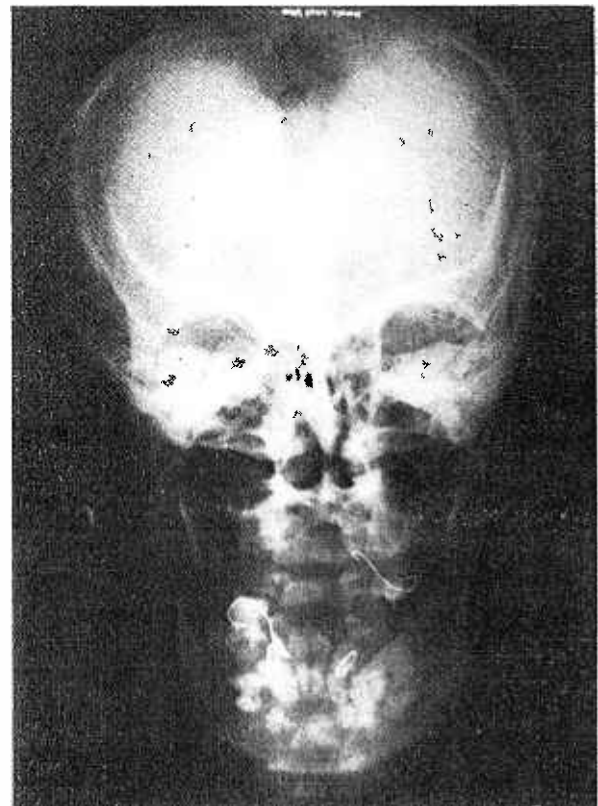


Fig. 2. Radiograph of skull of Case I  
Note: Flattened vertex, bossing of eminences, wormian bones, patent anterior fontanelle and buried teeth.

Radiographs showed that there was bilateral absence of the outer thirds of the clavicles (Fig. 1). There was no widening of the symphysis pubis and the necks of the femora were short and broad. The anterior fontanelle was patent and many wormian bones were present in the skull (Fig. 2). The following teeth were present and fully erupted.

				45678
6	321		1	E

Two mandibular premolar teeth, one on each side, were partially erupted. Bony prominences palpable on both sides of the mandible, and on the anterior part of the right maxilla indicated the possible presence of buried

teeth. Dental radiographs confirmed the presence of the following unerupted teeth.

			321
ss54			234s5

The space of the dental follicle around 3J was widened and suggested a cystic change (Fig. 3).



Fig. 3. Full mouth dental radiographs of Case I. Note the presence of the many unerupted teeth and the widened space of the dental follicle around 3J.

### Case II

*Daughter:* 17 years. Refused examination at the General Hospital, Singapore. The father, and the almoner who visited the home, assured us that the clinical appearance of the daughter was similar to that of the father, and there was little doubt that she was similarly afflicted.

### Case III

*Son:* 12 years old. Height 3 ft. 11½ inches. He possessed a less marked but still characteristic facial appearance. The radiographic appearances of the skull and clavicles were similar to those of the father. In addition, there was delayed ossification of the pubic bones, the femoral necks were short but there was no coxa vara (Fig. 4) and spina bifida was present in the cervico-dorsal region (Fig. 5).



Fig. 4. Radiograph of pelvis and hips of Case III. Note: Delayed ossification in pubis, short necks of femora but no coxa vara.

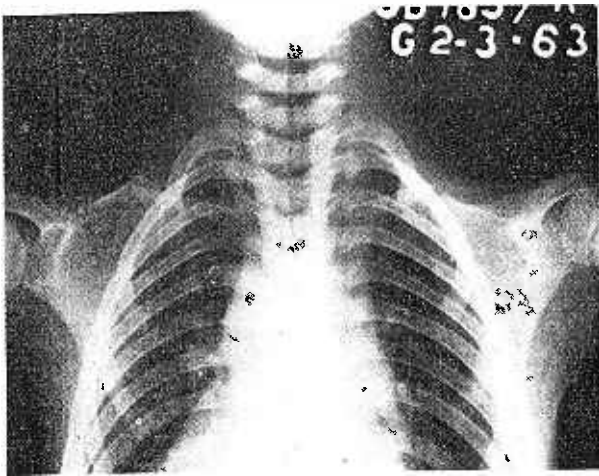


Fig. 5. Radiograph of shoulder region of Case III. Note: Absent outer ends resembling father's clavicles, somewhat shallow glenoids, spina bifida in cervico dorsal region.

The following teeth were present clinically:—

6EDC	1	BCDE6
6E	CBA	ABC 6

[6 was just beginning to erupt.

Radiographs showed the presence of the following unerupted teeth.

87	5432	12345	78
87	5s4321	1234s5	78

The degree of dental development was comparable to that of a normal child of about 7 years of age.

*Case IV*

*Son:* 9 years old. Height 3 ft. 5 inches. The clinical features of the abnormality were more obvious in this boy than in his brother. The radiographic features resembled those of the brother (Fig. 6) and in addition accessory ossific centres were present in the metacarpals and the second metacarpals were longer than the others.

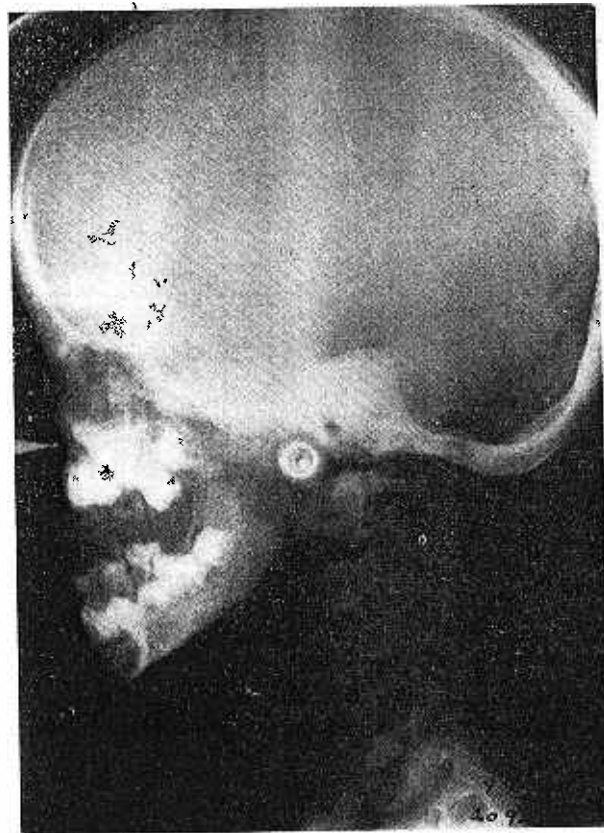


Fig. 6. True lateral radiograph of skull of Case IV. Note: The hypoplastic maxillae and normal mandible.

The following teeth were present clinically:—

EDC1	CDE
EDC	CDE

1] was just beginning to erupt.

Radiographs showed the following unerupted teeth.

765432	1234567
7654321	1234567

The degree of dental development was comparable with that of a normal child of about 6 years of age.

THE ANOMALY OF CEMENTUM

Rushton (1956) examined 9 permanent teeth from 5 cases of cleido-cranial dysostosis and found that there was extreme deficiency of cellular cementum in 8 of them. He suggested that the deficiency of cellular cementum was an additional hitherto undescribed dental defect characteristic of the dysostosis. Of the 9 specimens examined, only one tooth was fully erupted and one partially erupted, the remainder buried.

He further suggested that it would be interesting to know when further material became available, whether the defect was as common in the erupted, as it appeared to be with buried teeth. Only one report published since Rushton's paper (Alderson, 1960) makes reference to this anomaly of cementum, and Alderson found three partially erupted teeth to possess this anomaly. A number of teeth were extracted from the father (Case I) for prosthetic reasons; ground and decalcified sections (stained with picrothionin) were prepared from them and examined for the presence of this anomaly.

The observations of the present study, which together with those of Rushton and Alderson are summarised in Table I, are as follows:—

- (1) All the buried and partially erupted teeth were covered with acellular cementum (Fig. 7) except for small areas near apices in some teeth where cementocytes were indentifiable.



Fig. 7. Apex of root of partially erupted [4 covered with acellular cementum. (Picrothionin X 78).

- (2) Of the two erupted teeth, both maxillary molars, there was a complete lack of cellular cementum in the roots of

one of them while cellular cementum was present in the other.

- (3) Considerable variation in cementum thickness was observed and the tendency for cellular cementum to fill in hollows and grooves when it was found, as was noted by Rushton, was seen in one specimen (Figs. 8 & 9).

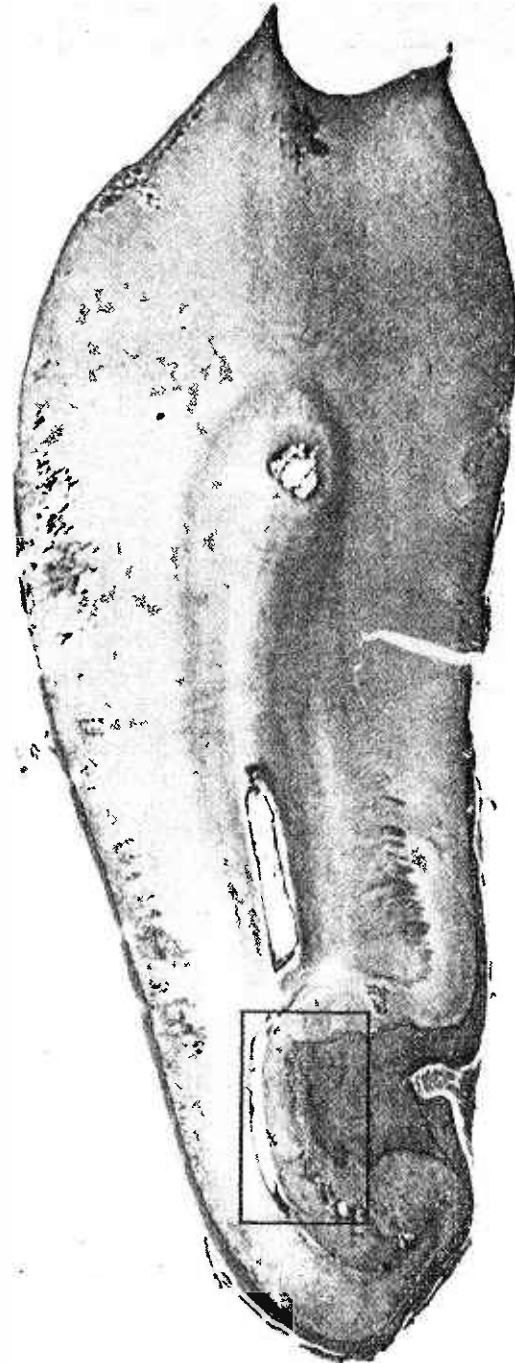


Fig. 8. Decalcified section of partially erupted [4] showing concavity near apex of twisted root. (Picrothionin X 12).

TABLE I. SHOWING THE PRESENCE OF THE ANOMALY OF CEMENTUM IN SPECIMENS OF TEETH FROM PATIENTS WITH CLEIDO-CRANIAL DYSOSTOSIS EXAMINED BY RUSHTON (1956), ALDERSON (1960), AND THE PRESENT AUTHORS (1963).

Tooth	Age & Sex of Patient	Eruption Status	Cementum
<i>Rushton's Specimens</i>			
1. Lower Incisor	42 ♂	Erupted	Acellular
2. Lower Incisor		Buried	Acellular
3. Lower Incisor		Buried	Acellular
4. Upper Lateral Incisor	50 ♀	Buried	Acellular
5. Upper Premolar	29 ♂	Buried	Cellular
6. Lower Incisor	36 ♂	Buried	Acellular except small patch in concavity of root.
7. Upper Canine	21 ♀	Buried tip of crown showing	Acellular except for minute patch in hollow near middle of apex.
8. Upper Premolar		Buried	Acellular
9. Upper Canine	41 ♀	Buried	Acellular (? Partial dysostosis).
<i>Alderson's Specimens</i>			
[2	45 ♀	Partially erupted	Thin layer of ecellular cementum. Apex resorbed.
[3	52 ♀	Partially erupted	Acellular. No resorption.
[3		Partially erupted	Uniform layer of acellular cementum.
<i>The Present Case</i>			
<i>Case 1</i>			
1]	41 ♂	Buried	Acellular
2]		Buried	Acellular
3]		Buried	Acellular except for small area near apex.
[6		Erupted	Cellular in all three roots.
[7 (4 rooted)		Erupted	Acellular in all four roots.
[4		Partially erupted	Chiefly acellular a few cementocytes seen near apex.
4]		Partially erupted	Acellular except for small patch in concavity near apex of twisted root.

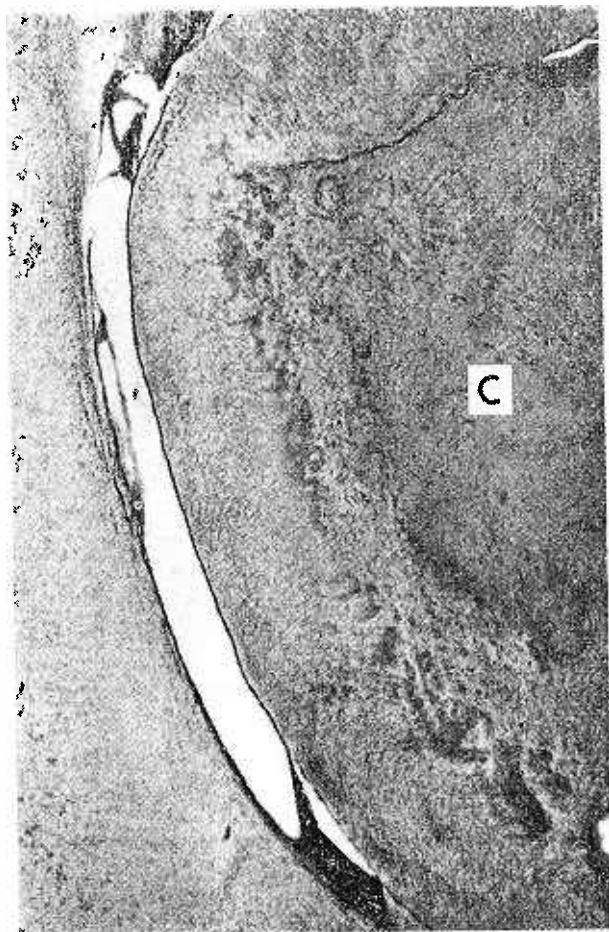


Fig. 9. Higher magnification of blocked area in Fig. 8. Showing cellular cementum (c) filling in concavity (Picrothionin X 78).

## DISCUSSION

The hereditary nature of cleido-cranial dysostosis was first fully documented by Marie and Sinton in 1898 and they listed four signs as pre-requisites of the condition:—

1. Clavicular aplasia, either unilateral or bilateral.
2. Exaggerated development of transverse diameter of the cranium and delayed ossification of the fontanelles.
3. Defects in the teeth.
4. Hereditary transmission.

It was thought that the defects were confined to membrane bones, but cartilaginous bones are affected as well. Indeed, a wide range of variations of skeletal and dental defects exists.

Of the variation of clavicular defects, in the majority of cases it is the outer thirds that are absent. The next common was presence of two fragments for each clavicle. Presence of the outer end is rarer still, and complete

absence very rare indeed. Cases have been reported with unilateral defects and also presence of skull changes without clavicular change. In the three cases examined (Cases I, III and IV) the defect in the clavicles is similar — bilateral absence of the outer third. It would appear that the abnormal gene interferes with growth of this part of the clavicle.

The pelvis may show delayed appearance of the pubis and a wide symphysis pubis. The neck of the femur usually shows a coxa vara. No coxa vara was seen in the present series of cases though the femoral necks are short and broad. The vertebrae may exhibit spina bifida which is usually in the cervico-dorsal or lumbar regions. In the two boys in this series, spina bifida is present in the cervico-dorsal region. The hands and feet show presence of epiphysis at both ends of the metacarpals and metatarsals, the second metacarpals are usually disproportionately long and rarely the lateral condyles of the humeri are hypoplastic. Case IV shows these extra ossific centres and the long second metacarpal, but in the three cases studied no changes were seen in the elbow.

The nervous system seems to be involved often. Paul's case had epilepsy, though the present cases are free from this condition. Cysts of the frontal lobes, haemorrhagic lesions of the brain, failure of development of some basal nuclei and deafness have been reported. The degree of intelligence however is normal.

There is universal agreement that the condition arises from the transmission of a dominant (completely penetrant) gene. New cases are accounted for by mutation occurring in the gametes of one of the parents of the affected individual. Here a mutation must have occurred in the gametes of one of the parents of Case I. The condition is not disabling and the person afflicted can lead a normal and useful life.

The defects of the facial skeleton and the dentition are clinically even more significant than defects of other bones. The maxillae are hypoplastic, though the mandible is normal, which results in relative prognathism and a prenatal occlusion. The exfoliation of the deciduous dentition is delayed and many permanent teeth remain buried. Supernumerary teeth are common, and they too fail to erupt; there is a tendency for dentigerous cyst formation. The shape and structure of the teeth are often abnormal, the crowns are usually conical and hypoplastic, and the roots bent and twisted. Except for the anomaly of

cementum these dental defects are well known (Rushton, 1937).

Failure of the permanent teeth to erupt gives rise to several problems. Attempts have been made to expose the crowns surgically to hasten their eruption so that some sort of occlusion is achieved but it is difficult to evaluate this procedure as the number of cases reported is small (Kjellgren, 1952). Because in the adult, removal of the buried teeth entails the excision of a thick layer of overlying bone, it is the accepted practice to remove these teeth when the individual is fairly young. At this age the surgical removal is a relatively simple procedure (Rushton 1947). In elderly patients only unerupted teeth which are superficially placed and those which are partially erupted need be removed, leaving those which are deeply buried (Burkett, 1961). This procedure was adopted in the case of the father. The further dental development of the two boys is being kept under observation, and it is intended to surgically expose the crowns of the buried teeth at the appropriate time.

The study of the roots of the extracted teeth bears out Rushton's observations that the teeth in patients afflicted with this anomaly are peculiarly deficient in cellular cementum. Unfortunately, the number of erupted teeth examined is too small, and no conclusion could be drawn from the evidence presented.

### SUMMARY

1. Four new cases of cleido-cranial dysostosis in a Chinese family are recorded.

2. The deficiency of cellular cementum in the roots of teeth as an additional dental defect characteristic of the dysostosis has been further borne out by examination of material obtained from one of these cases.

### APOLOGY

It has not been possible to include clinical photographs of the cases as consent for their publication could not be obtained.

### ACKNOWLEDGEMENTS

We would like to thank Professor F. A. C. Oehlers for his many helpful suggestions in the preparation of this paper and to the technical staff of the two departments for their help in the production of the illustrations.

### REFERENCES

- Alderson, C.G.P. 1960: Hereditary Cleido-Cranial Dysostosis. *Brit. dent. J.* 108, 157.
- Burkett, L.W. 1961: *Oral Medicine*. 4th edition p.306. Philadelphia: J.B. Lippincott Co.
- Kjellgren, B. 1952: Bite Conditions and Treatment in cases of Cleido-cranial Dysostosis. *Int. dent. J.* 3, 83.
- Marie, P., et Sainton, P. 1898: Sur la dysostose cléidocranienne héréditaire. *Rev. Neurol.* 6, 835.
- Paul, F.M. 1957: Cranio-cleido Dysostosis. *Malaya, University Alumni Ass. Proceedings.* 10, 91.
- Rushton, M.A. 1937: The Failure of Eruption in Cleido-cranial Dysostosis. *Brit. dent. J.* 63, 641.
- Rushton, M.A. 1947: Cases of failures of tooth eruption in 2 families. *Dent. Gaz.* 13, 504.
- Rushton, M.A. 1956: An anomaly of Cementum in Cleido-cranial Dysostosis. *Brit. dent. J.* 100, 81.