A case of cleidocranial dysostosis

W W S D Mendis[1], **P Hewavitharana**[2], **S I Samarasekera**[3]

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(Key words: Cleidocranial dysostosis)

Cleidocranial dysostosis is characterized by varying degree of hypoplasia of membranous bone and to a lesser extent of endochondral bone and dentition[1,2]. It is mainly inherited as autosomal dominant[1]. One third are fresh mutations[1].

Case report

A four and half year old girl was admitted to General Hospital, Kalutara for investigation of short stature. Her head circumference was 51 cm (50th centile), height 90 cm (<3rd centile), weight 16 kg (between 10th and 50th centiles). She had a brachycephalic head with open anterior fontanelle. Both clavicles were absent so that she could move her shoulders across the chest. There was no chest deformity. Her dentition was norma[1].

X-ray skull (figures 1 and 2) showed defective mineralization of the skull bones and widely open fontanelles.There was frontal and parietal bossing with multiple wormian bones. X-ray chest (figure 3) revealed absent clavicles, narrow thorax and short ribs which were obliquely directed downward. Physical features and x-ray findings confirmed the diagnosis of cleidocranial dysostosis.

[1]*Consultant Paediatrician,* [2]*Consultant Radiologist,* [3]*Registrar in Paediatrics, General Hospital, Kalutara.*

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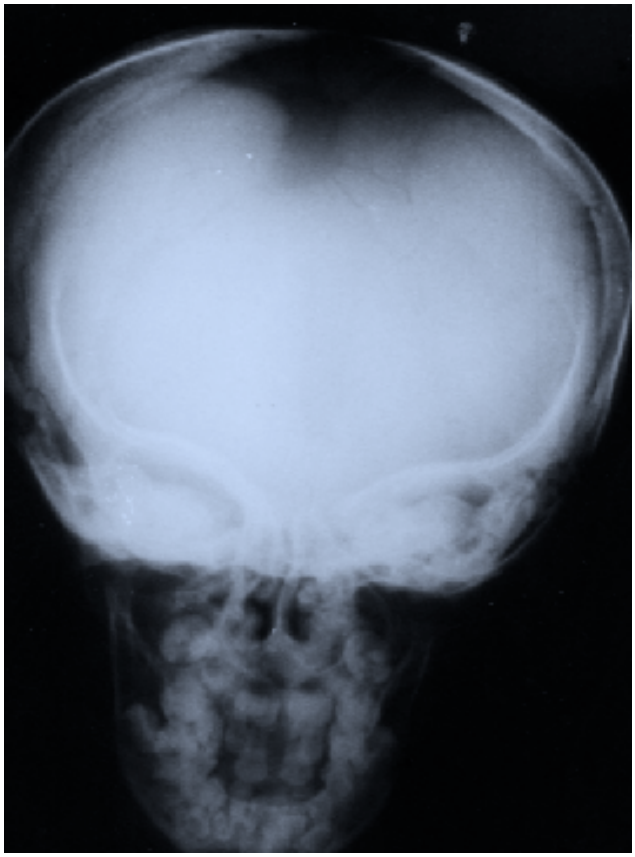

Figure 1. *Skull Xray PA*

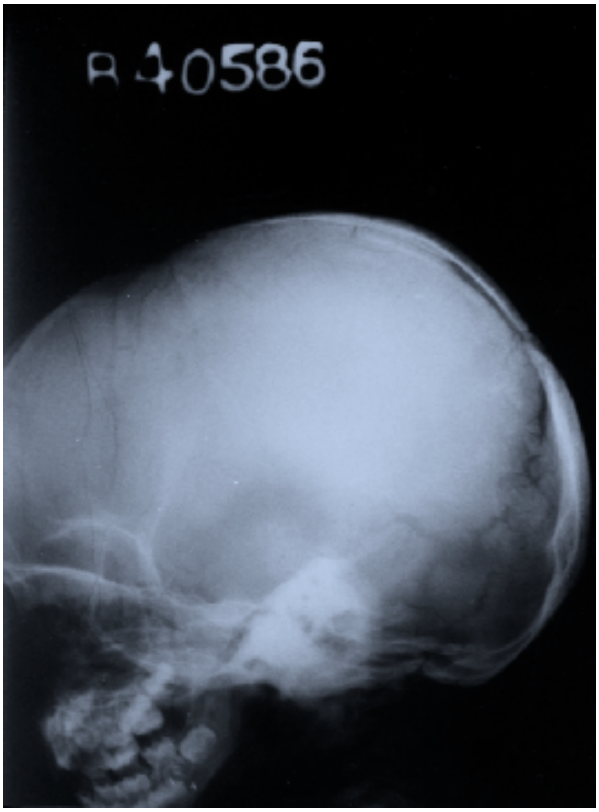

Figure 2. *Skull Xray lateral*

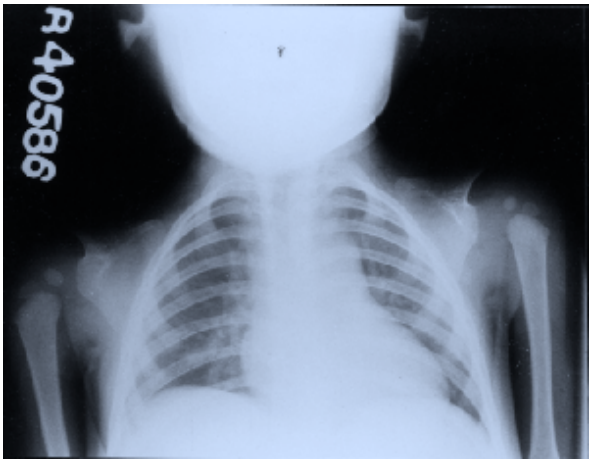

Figure 3. *Chest Xray PA*

Discussion

Cleidocranial dysostosis covers a wide range of phenotype variability and deformities are so unobtrusive that the sufferer may not be aware of having the condition[2]. It affects more of the skeleton than the name would imply[3]. It can be suspected clinically even at birth and confirmed by radiological investigations[3].

Frequently seen abnormalities are short stature, brachycephaly with frontal, parietal, occipital bossing; late closure of fontanelles and mineralization of sutures; incompletely developed accessory sinuses; wormian bones; midfacial hypoplasia with high arched palate; hypoplastic clavicles with small thorax due to short and oblique ribs; asymmetric length of fingers with tapering of distal phalanges; narrow pelvis with wide symphysis pubis; broad femoral head with coxa vara[1,2,3].

These patients have normal intelligence quotient (IQ) and normal life span[1]. Dental problems, conductive deafness, respiratory distress in early infancy, cephalo-pelvic disproportion necessitating caesarean section are the problems encountered. Although no treatment is available for underlying disorder, multidisciplinary approach for above problems and genetic counselling are of prime importance[3].

References

1. Jones K L, editor. Smith's recognizable patterns of human malformation. 5th ed. Philadelphia; W B Saunders, 1997.

2. Sutton D, editor. Textbook of Radiology and Imaging, 5th ed, Edinburgh; Churchill Livingstone, 1993, 14-5.

3. http://www.icondata.com/health/pedbase/files/CLEIDOCR.HTM

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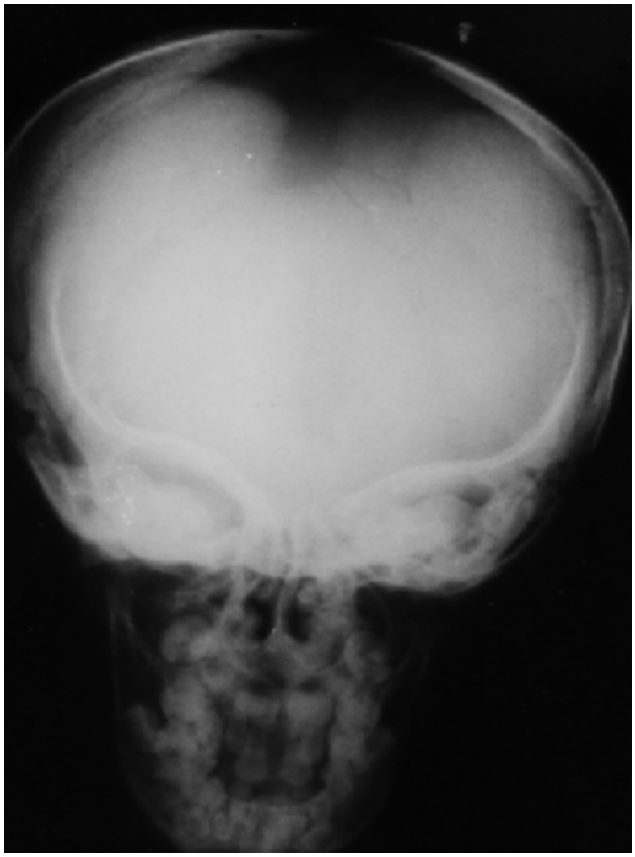

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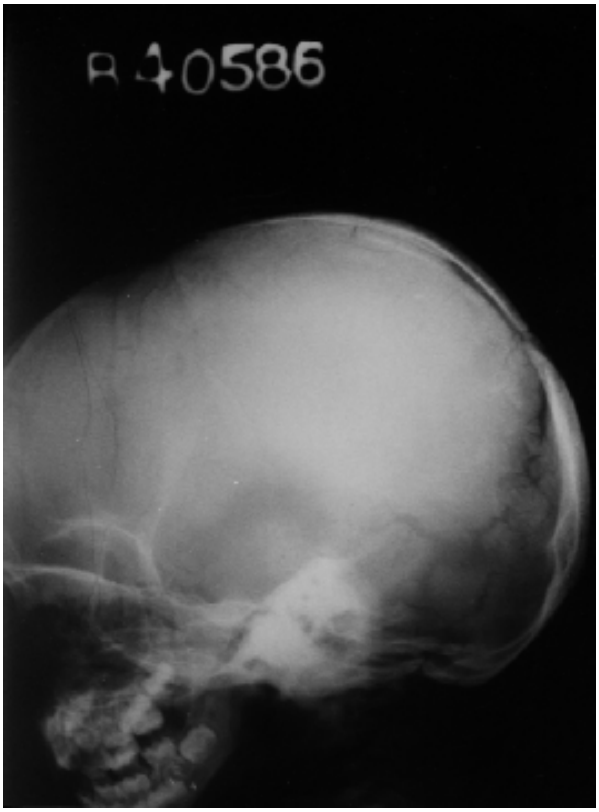

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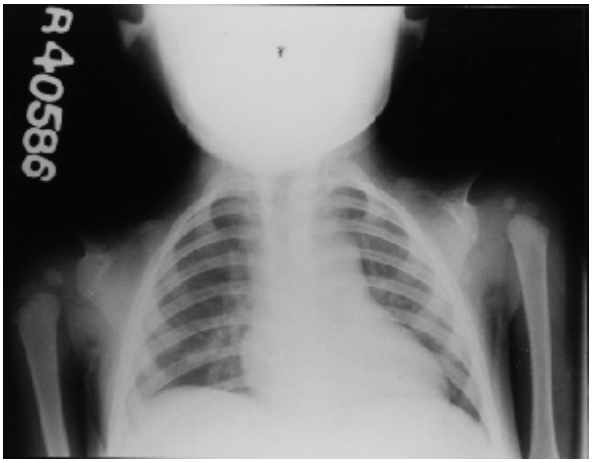

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