A case of cleidocranial dysostosis

W W S D Mendis¹, P Hewavitharana², S I Samarasekera³


(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 dentition¹². It is mainly inherited as autosomal dominant¹. One third are fresh mutations¹.

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 (50⁰ centile), height 90 cm (<3⁰ centile), weight 16 kg (between 10⁰ and 50⁰ 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 normal¹.

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.

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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. It affects more of the skeleton than the name would imply. It can be suspected clinically even at birth and confirmed by radiological investigations.

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.

These patients have normal intelligence quotient (IQ) and normal life span. 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.

References


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