

Cleido-Cranial Dysplasia: A Case Report

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Abstract

Cleido-Cranial Dysplasia is an autosomal dominant condition with generalised dysplasia of bone and teeth. It is characterized by short stature, typical facial features and skeletal anomalies affecting skull and clavicle.

CASE REPORT

An 11 year old girl child presented with short stature and abnormal head shape. On examination height was 127 cm and weight was 22.1 kg (both < 3rd centile). She was only child born to non-consanguineous parents by normal vaginal delivery. Her development was normal. Head circumference was 50cm. She had brachycephaly, frontal bossing, hypertelorism and slopping of shoulders. She had hypermobility of shoulders and she was able to approximate her shoulders adjust to each other (Fig.1). Her skull x-ray showed asymmetry of skull vault with wormian bones. She also had defective dentition.

Figure 1

Figure 1: Clinical photograph of child with cleido-cranial dysplasia showing brachycephaly, frontal bossing and slopping of shoulders. She was able to approximate her shoulders adjust to each other



Her chest x-ray revealed small clavicles with hypoplasia of the lateral third (Fig.2). She was diagnosed to have cleido

cranial dysplasia based on the clinical and radiological findings.

Figure 2

Figure 2: Chest x-ray revealing small clavicles with hypoplasia of the lateral third.



DISCUSSION

Cleido cranial dysplasia was first described in a patient with congenital absence of clavicle in 1765 by Martin.¹ But it was well described by Marie and Sainton in 1898.² Only few cases were reported from India.³ The main pathogenesis is the defective midline ossification which results in patent anterior fontanelle, metopic suture, wormian bones, cleft palate, hypoplastic or absence of clavicle.⁴

The most characteristic and pathogenic skeletal feature is the absent or hypoplastic clavicles which give rise to hypermobility of shoulders with tendency to approximate shoulders anteriorly. This disorder also has delayed

meneralisation of pubic bone, wide symphysis pubis, narrow pelvis, spondylosis, short stature, syringomelia or spina bifida

occulta, kyphosis, scoliosis, lordosis, hypoplastic maxillary, lacrimal and zygomatic bones and vertebral synostosis.⁵ Incomplete development of accessory sinuses and mastoid air cells, small sphenoid bones, calvarial thickening and hypertelorism can also be seen. The primary dentition appears late and secondary dentition is also delayed with malalignment of teeth and supernumerary teeth. It is a relatively benign condition with normal intelligence and normal life expectancy but complications during child birth can occur.⁵ It is not associated with any other diseases.

A gene for this disorder osteoblast specific transcription factor 2 (Osf2)/Core binding factor activity 1 (Cbfa1) has been mapped to chromosome 6p21.⁶

This case is presented to show the significance of thorough clinical examination of children with short stature including palpation of clavicles.

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References

1. Martin S. Sur Undepacement naturel de la clavicle. J Med Chir Pharmacol 1765; 23: 456
2. Marie P, Sainton P. Sur La dysostose cleidocranienne hereditaire. Rev Neurol 1898; 6: 835
3. Gargaya S, Sharma S, Pal LS, Bahl L, Puri DS, Sarin NK. Cleido cranial dysostosis in five generations of a family with some atypical roentgenologic features. Indian Pediatr. 1979; 16 (12):1145-50.
4. Jensen B. Somatic development in cleido-cranial dysostosis. Am J Med Genet 1990; 35: 69-74
5. Nebgen D, Wood RS, Shapiro RD. Management of a mandibular fracture in a patient with cleido-cranial dysplasia. J Oral Maxillofac Surg 1991; 49: 405-409
6. Shapiro IM. Discovery:Osf2/Cbfa 1, a master gene of bone formation. Clin Orthod Res 1999; 2: 42-46

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