Scheithauer-Marie-Sainton Syndrome

Kundoor Vinay Kumar Reddy[1], Krishna Priya Pasupuleti[2], Anurag Ellanki[3], Kotya Naik Maloth[4,*], Kesidi Sunitha[5]

[1]Professor & Head, [2,3]PG Student, [4,5]Assistant Professor, Dept. of Oral Medicine & Radiology, Mamata Dental College & Hospital, Telangana

***Corresponding Author:**
Email: dr.kotyanaik.maloth@gmail.com

Abstract

Cleidocranial dysplasia (CCD) is an autosomal dominant developmental anomaly with craniofacial, skeletal and dental malformations. The gene causing CCD has been mapped to chromosome 6p21 within a region containing CBFA1, a member of the runt family of transcription factors. It is characterised by failure of membranous ossification resulting in absence or pseudoarthrosis of clavicle, open fontenelles, impacted and supernumerary teeth. The short stature, long and narrow neck with marked droopeding shoulders. CCD patients exhibit prognathic mandibular appearance due to maxillary hypoplasia and also exhibit relatively short cranial base. Here with describing a case of Scheithauer-Marie-Sainton syndrome, in a 30 year old male patient.

Key words: Brachycephalic, Hypertelorism, Hypoplastic, Supernumerary teeth.

Introduction

Cleidocranial dysplasia (CCD) is a rare autosomal dominant developmental disorder which effects entire skeletal system and teeth. First described by Marie and Sainton in 1897, who termed the condition cleidocranial dysostosis.[1] CCD is a rare disease with a prevalence of less than 1 per million.[2] Till now over 1000 cases have been published in the medical literature.[3] Marie-Sainton's disease and Scheithauer-Marie-Sainton syndrome are the other names for cleidocranial dysplasia.[4]

Case Report

A 30 year old male patient reported to the department of Oral Medicine & Radiology with the complaint of dislodged upper fixed prosthesis. His medical and family history was non-contributory. On general examination the patient had normal I.Q, short stature, with thin built, scoliosis and a long neck. Brachycephalic head with frontal bossing and depressed mid-face profile with hypertelorism were noticed. He had a drooping right shoulder and not able to bring his shoulders towards midline which confirmed incomplete clavicle formation; and he was able to easily perform this procedure. [Fig. 1] Intra oral examination showed high arched palate, prolonged retention of deciduous teeth and missing permanent teeth. Orthopantramogram (OPG) [Fig. 2] revealed abnormally retained primary teeth with multiple permanent and supernumerary impacted teeth. PA-skull and lateral cephalogram [Fig. 3 and 4] revealed open sutures, calvarial thickening and characteristically sunken sagittal suture giving the skull a flat appearance and absence of frontal sinuses. Lateral Cephalogram showed hypoplastic maxilla and nasal bone with prognathic mandible. PNS view revealed absence of frontal sinus and hypoplastic maxillary sinus. Chest radiograph [Fig. 5] revealed absence of right clavicle and hypoplastic left clavicle. The hand wrist radiograph [Fig. 6] revealed tapering of the terminal phalanges and short intermediate phalanges. Based on the clinical and radiographical findings diagnosed as Cleidocranial dysplasia.

Fig. 1: Extra oral view showing hypertelorism, hypermobility of shoulders

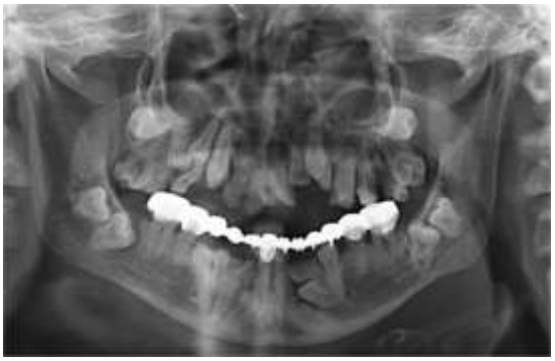

Fig. 2: OPG showing multiple impacted permanent & supernumerary teeth

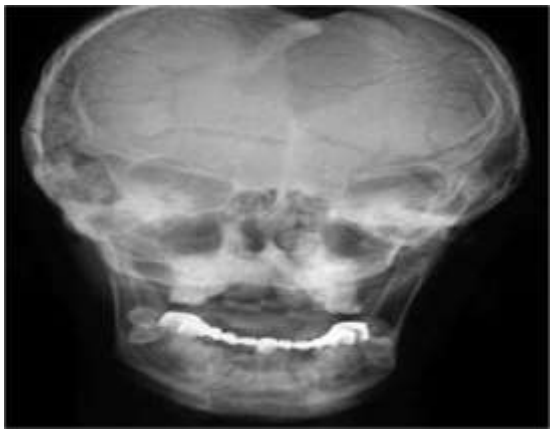

Fig. 3: PA skull view showing open sutures, hypoplastic paranasal sinuses & absence of frontal sinus

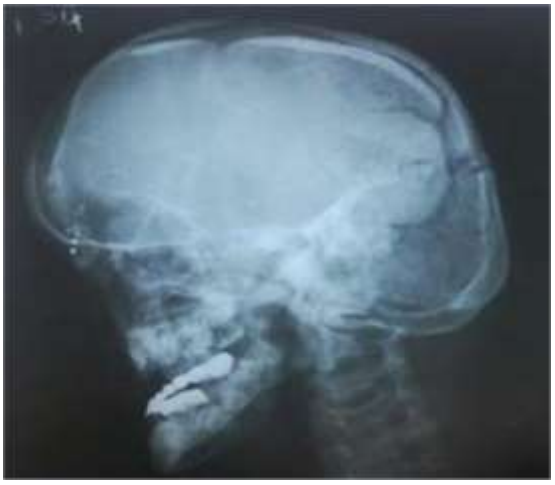

Fig. 4: Lateral cephalogram showing obtuse angle of mandible and open fontanelle

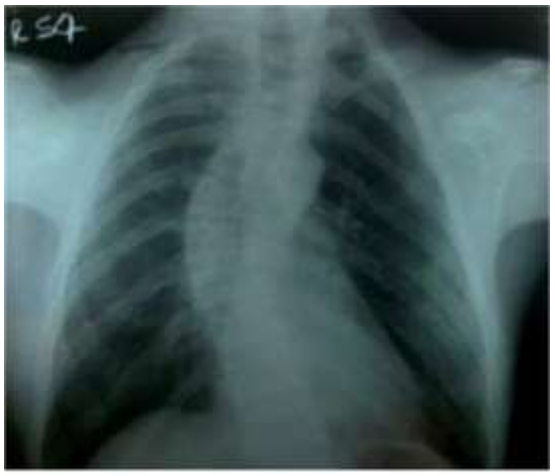

Fig. 5: Chest radiograph showing absence of right clavicle & hypoplastic left clavicle

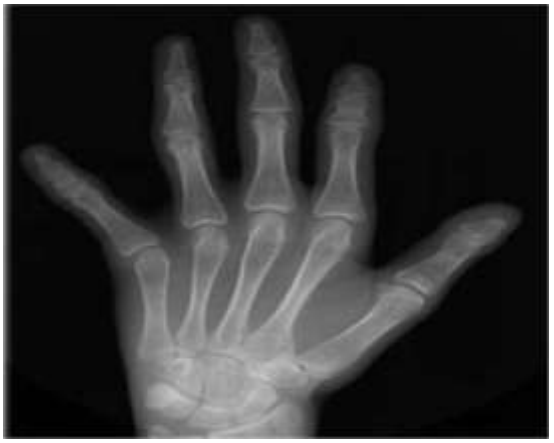

Fig. 6: Hand wrist radiograph showing tapering of distal phalanges & short intermediate phalanges

Discussion

CCD initially was thought to involve only bones of membranous origin.[5] Recent clinical investigations have shown that CCD is a generalised skeletal dysplasia affecting not only the clavicles and the skull but the entire skeleton. CCD is thus considered a dysplasia rather than a dysostosis.[6] The midline osseous structures are primarily involved, though there might be generalized involvement.[7] The aetiology is not completely known but it was thought that the disorder was due to mutation in CBFA1 (core binding factor activity gene defect on the short arm of chromosome 6p21 was considered.[5] In 1997 short arm of chromosome 6 was mapped with RUNX2 gene and was considered as aetiological factor of CCD.[2] It is now recognized that the condition affects the entire bony skeleton and extra-skeletal structures, such as skeletal muscles.[8] Late closure of fontanelle results in frontal bossing. The disease is characterized by persistently open or delayed closure of sutures, hypoplastic or aplastic clavicles and short stature.[3] The skull deformities observed in this case were all consistent with cleidocranial dysplasia. Delayed or disturbed eruption of permanent teeth accompanied by multiple supernumerary teeth is often presented consistent with our case. A study of teeth from patients with cleidocranial dysplasia revealed a paucity or complete absence of cellular cementum on both erupted and unerupted teeth.[1] The clavicles are the first bone to ossify and are commonly affected, being either hypoplastic or aplastic causing unusual mobility of the shoulders. When there is unilateral absence, it is usually in the right clavicle, which was consistent with our present case.[5] In the present case the right clavicle was absent and the left was hypoplastic. It is of clinical significance to the dentist due to involvement of facial bones, altered eruption patterns and presence of multiple supernumerary teeth.[9] Hands show poorly developed terminal phalanges which gives a tapered appearance to the digit which was seen in wrist radiograph.[6] They exhibit small and bell shaped

thoracic cage with short ribs which was also evident.[3] patient usually exhibit a short stature with CCD.[8] Vertebral abnormalities include scoliosis, vertebral synostosis, lordosis, and kyphosis,.[7] Our patient had scoliosis. Less common findings include shortened or absent nasal bones, reduced or absent paranasal sinuses.[9] In the present case hypolplastic paranasal sinuses and absence of frontal sinuses is seen. Life expectancy is normal.[10]

Conclusion

Since the disorder is rare, medical consultation is often associated with "one patient-one-doctor" phenomenon.[3] There is no treatment for cleidocranial dysplasia, although care of the oral condition is important. The retained deciduous teeth should be restored if they become carious.[5] An interesting fact is that most of the features described in this present paper involve the cranio-maxillo-mandibular complex. Therefore, it is possible to diagnose CCD based on panoramic radiograghs, calvarial radiographs, lateral cephalometric radiograghs and hypermobility of shoulders.

References

1. Pereira CM, Alves VF, Gasparetto PF, Souza RCN, Botelho TL. Cleidocranial dysplasia associated with the dentigerous cyst: review of the literature and report of the clinical case. Journal of Health Science Institute. 2010;28(2):137-9.
2. Verma P, Verma KG and Gupta SD. Cleidocranial dysplasia: a dilemma in diagnosis? Archives of Orofacial Sciences 2010;5(2):61-4.
3. Golan, Baumert U, Hrala BP and Mubig D. Dentomaxillofacial variability of cleidocranial dysplasia: clinic-radiological presentation and systematic review Dentomaxillofac Radiol 2003;32:1-10.
4. Kuruvila VE, Bilahari N, Attokkaran G, Kumari B. Scheuthauer-Marie-Sainton syndrome. Contemp Clin Dent. 2012;3(3):338-40.
5. Suresh SS. A family with cleidocranial dysplasia and crossed ectopic kidney in one child. Acta Orthopaedica Belgica 2009;75(4):521-7.
6. Mundlos S. Cleidocranial dysplasia: clinical and molecular Genetics. J Med Genet 1999;36:177–82.
7. Sharma A, Yadav R and Ahlawat K. Cleidocranial Dysplasia. J Indian paediatrics 1995;32:588-92.
8. Salem G. Non-familial Cleidocranial Dysplasia (Dysostosis): A Case Report. Saudi Dental Journal 1990;2(3):112-5.
9. Jafferson et al. Cleidocranial dysplasia: importance of radiographic images in diagnosis of condition. J Oral Science 2006;48:161-6.
10. SK Verma, Jain P and Sharma NC. Scheuthauer-Marie-Sainton syndrome - a rare entity imaging findings. J Radiol Imaging 2004;14:175-6.