

Dental Management Of Patient With Scheuthauer-Marie-Sainton Syndrome

Sandhyarani B¹, Dayanand Huddar², Sachin Gunda¹, Shridevi Tamagond¹.

¹(Department of Pedodontics, Bharati Vidyapeeth Dental College and Hospital/ Bharati Vidyapeeth deemed University, Sangli, India).

²(Department of Prosthodontics, Bharati Vidyapeeth Dental College and Hospital/ Bharati Vidyapeeth deemed University, Sangli, India)

Abstract: Scheuthauer –Marie –Sainton Syndrome is a rare autosomal dominant skeletal dysplasia which is commonly known as cleidocranial dysplasia. General manifestations of scheuthauer-marie-sainton syndrome includes open or delayed closure of fontanel, hypoplastic or aplastic clavicles, short stature and tooth abnormalities. Dental manifestation includes retention of multiple deciduous teeth, impaction or delayed eruption of permanent teeth, presence of supernumerary teeth. Here is a case report of 13 year old female child suffering from scheuthauer-marie-sainton syndrome and a chief complaint of missing upper front teeth which was treated by operculectomy.

Keywords: Cleidocranial dysplasia, Operculectomy, Scheuthauer- Marie- Sainton Syndrome.

I. Introduction

Scheuthauer- Marie –Sainton Syndrome, commonly known as cleidocranial dysplasia is a rare congenital defect of autosomal dominance inheritance which was first described by Marie and Sainton in 1898.¹ It is a well defined clinical phenotype arising from deregulation of intramembranous and endochondral ossification due to mutation in Cbfa1.² It has been suggested that 70% of patients with cleidocranial dysplasia have a point mutation involving Runx2 and 13% have a deletion. In patients whose mutations are not found by traditional sequencing, the deletion/duplication assay, either Reverse transcription - quantitative real time polymerase chain reaction (RT-qPCR) or Multiplex Ligation-dependent Probe Amplification (MLPA), needs to be done.² Occurance in males and females are equal.³

Characteristic features of cleidocranial dysplasia are absence or reduced size of one or both clavicles, brachycephaly with parietal and frontal bossing, hypoplastic maxilla and zygoma, and relative mandibular prognathism. Other skeletal abnormalities include delayed closure of the fontanels, pubic symphysis and coxa vara, as well as anatomical changes in the vertebrae and phalanges.⁴

Oral manifestations include short lower facial height in older individuals, acute gonial angle, anterior inclination of the mandible, and mandibular prognathism, however young individuals have normal jaw proportions and mandibular morphology. These differences are attributed to pronounced horizontal mandibular growth resulting from impaired vertical maxillary growth and eruption of permanent teeth. The dental manifestations include delayed exfoliation of primary teeth, delayed eruption or noneruption of permanent teeth, supernumerary teeth, retention cysts, and enamel hypoplasia.⁵

Hence here is a case of 13 year old female child suffering from cleidocranial dysplasia. Her main concern was bad appearance because of missing upper front teeth. As the incisal bulge was present but was delay in eruption, operculectomy was performed for fast eruption of tooth.

II. Case Reports

A 13 year old female child reported to the department of pedodontics with the chief complaint of missing upper front teeth. There was a family history of consanguineous marriage but with no specific medical history. All the siblings were found to have no abnormalities. On extra oral examination, there was increased intraorbital distance, appearance of saddle nose i.e., bridge of nose was wide and flat, underdeveloped maxilla and slight prognathic mandible were present (figure 1). Her shoulders were drooping and she could approximate her shoulders anteriorly. On intraoral examination, upper front teeth i.e., tooth number 11 and 21 were missing. Teeth present were 16, 15, 14, 53, 52, 62, 63, 24, 25, 26, 36, 75, 74, 73, 32, 31, 41, 42, 84, 85, 46. Generalized spacing was present with no carious tooth except for little stains and plaque (Figure 2). As approximation of shoulders was a classic sign of cleidocranial dysplasia further investigations were done. PA view radiograph revealed open anterior fontanelle, orthopantomograph revealed multiple impacted teeth, chest x ray revealed hypoplastic

clavicles thus ensuring the diagnosis as cleidocranial dysplasia or also called as Scheuthauer –Marie –Sainton Syndrome (figure 3,4). As the chief complaint of patient was missing front teeth, operculectomy was performed to expose the incisal edges of 11 and 21 (figure 5). The patient is under continuous observations for eruption of teeth.

III. Figures And Tables



Figure 1



Figure 2

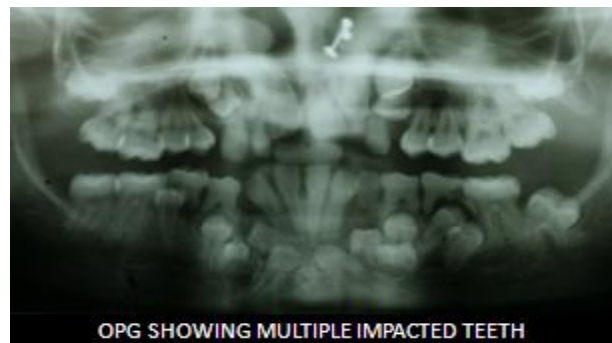


Figure 3

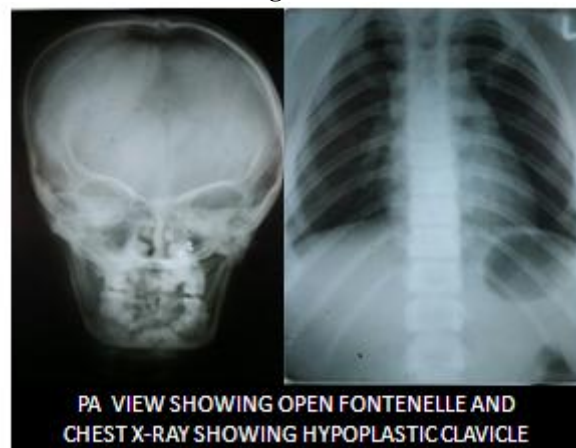


Figure 4



Figure 5

IV. Discussion

Scheuthauer- Marie –Sainton Syndrome, also called as cleidocranial dysplasia, cleidocranial dysostosis or mutational dysostosis is a rare congenital disorder which occurs due to haploinsufficiency caused by mutations in the CBFA1 gene (also called Runx2), located on the short arm of chromosome 6, which encodes transcription factor required for osteoblast differentiation. It results in delayed ossification of midline structures of the body, particularly membranous bone.⁶ CBFA1 gene is vital for differentiation of stem cells into osteoblasts, so any defect in this gene will cause defects in membranous and endochondral bone formation.⁷ Clinical features of this syndrome has a wide range of variability. There can be introcession on the forehead and occiput⁸ bossing (bulging) of the forehead, open skull sutures, large fontanelles, hypertelorism, delayed ossification of bones forming symphysis pubis producing a widened symphysis coxa vara can occur limiting abduction and causing trendelenburg gait, short medial fifth phalanges, sometimes causing short and wide fingers, vertebral abnormalities, on rare occasions, brachial plexus irritation can occur, scoliosis, spina bifida and syringomyelia have also been described.^{9,10}

Dental problems include hyperdontia of both primary and permanent teeth which may lead to crowding, impaction of teeth and malocclusion. There can be midfacial hypoplasia which can worsen the condition. Articulation and mastication may be compromised, and the cosmetic appearance of the dentition may be unsightly. Excess teeth may be normal or misshapen and situated in front, behind, or within the normal upper and lower rows of teeth. The supernumerary teeth may be arranged uniformly as a double row or placed chaotically on the jaws.¹¹

Treatment plan varies according to the expression of the dental anomalies where in orthodontic and surgical corrections may be required. Prosthetic appliances can be required for missing teeth which includes placement of removable or fixed dentures or implant supported dentures depending upon jaw growth and age of the patient.^{12,13} In the present case the main concern of the patient was the missing upper front teeth and the clinical bulge was seen with it. Thus operculectomy was performed which is exposure of incisal edges of teeth for fast eruption of teeth. Continuous monitoring of teeth is must for further remaining treatment.

V. Conclusion

Dentist has a very important role in diagnosis and management of Scheuthauer- Marie –Sainton Syndrome and dental problems are of more concern for the patient as esthetic point of view. Thus treatment plan depends on chronological and dental ages of patient which may better the psychology and quality of life.

References

- [1]. Marie P, Sainton P: Sur la dysostose cleido-cranienne hereditaire. Rev Neurol 1898, 6:835-838.
- [2]. Nagarathna C, Bethur S S, Mathew S, Krishnamurthy N H, Yumkham R. Cleidocranial dysplasia presenting with retained deciduous teeth in a 15 year old girl; a case report. Journal of Medical Case Reports 2012, 6:25.
- [3]. S Rizvi, H Raihan, T Rizvi. Cleidocranial dysplasia –A case report. Biomedical Research 2006; 17 (2): 129-132.
- [4]. W. Kim Seow, J Hertzberg. Dental development and molar root length in children with cleidocranial dysplasia. Pediatric Dentistry- 17:2,1 995.
- [5]. TKN Park, K Vargervik, S Oberoi. Orthodontic and surgical management of cleidocranial dysplasia. Korean J Orthod. 2013 Oct; 43(5): 248–260.
- [6]. Turek's Orthopaedics: Principles And Their Application. Lippincott Williams & Wilkins. 2005. pp. 251–252.
- [7]. Saraswathivilasam S. Suresh, A Family With Cleidocranial Dysplasia And Crossed Ectopic Kidney In One Child, Acta Orthop. Belg. 2009, Vol. 75; 4:521-527.
- [8]. S Wang, S Zhang, Y Wang, Y Chen, Li Zhou. Cleidocranial dysplasia syndrome: clinical characteristics and mutation study of a Chinese family. Int J Clin Exp Med 2013;6(10):900-907
- [9]. Vanderwerf, Sally (1998). Elsevier's medical terminology for the practicing nurse. Elsevier. p. 65.
- [10]. Menkes, John (2006). Child Neurology, 7e. Lippincott Williams & Wilkins. p. 307.
- [11]. Tina R, Lawrence S, Peter B. Cleidocranial dysplasia: A review of the dental, historical, and practical implications with an overview of the South African experience. Oral surgery oral medicine oral pathology and oral radiology. Volume 115, Issue 1, January 2013, Pages 46–55.
- [12]. R.K. Hall, A.L. Hyland. Combined surgical and orthodontic management of the oral abnormalities in children with cleidocranial dysplasia. International journal of oral surgery 1978, vol 7(4); 267-273.
- [13]. J Daskalogiannakis, LPiedade, T C. Lindholm, G K.B. Sándor, R P. Carmichael. Cleidocranial Dysplasia: 2 Generations of Management. J Can Dent Assoc 2006; 72(4):337–42.