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**Case Report**

# Scheuthauer-Marie-Sainton syndrome: a case report

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## ABSTRACT

The term Scheuthauer-Marie-Sainton syndrome is also known as cleidocranial dysplasia or cleidocranial dystosis and is derived from ancient greek words cleido (collar bone), knanion (head) and dysplasia (abnormal formation). It is an uncommon but well known genetic skeletal condition and an autosomal dominant malformation affecting bones and teeth. The most common skeletal and dental abnormalities in affected individuals are hypoplastic/ aplastic clavicles, open frontanelles, short stature, retention of primary teeth, delayed eruption of permanent teeth, supernumerary teeth and multiple impacted teeth. Affected person have a characteristic facial appearance with a bulky forehead, hypertelorism and midfacial hypoplasia. General health is usually good and the intellect is unimpaired. This article describes clinical and radiographic features of Scheuthauer-Marie-Sainton syndrome in a 29 years old male patient.

**Keywords:** Cleidocranial dysplasia, Hypoplastic/aplastic of clavicles, Open frontanelles

## INTRODUCTION

Cleidocranal dysplasia (CCD) is a highly polymorphous autosomal dominant skeletal disorder with a wide variety of expressivities, mainly affecting bones undergoing intramembranous ossification. It is characterized by retarded cranial ossification, patent sutures and fontanelles, supernumerary teeth, short stature and a variety of other skeletal abnormalities.[1] It is a rare disorder with a prevalence of less than 1 per million. The disease gene, which has been mapped to chromosome 6p21 within a region containing core binding factor activity 1 (CBFA1), a member of the runt family of transcription factors controls differentiation of precursor cells into osteoblasts and is essential for both membranous and endochondral bone formation.

The different clinical manifestations reflect the basic mechanisms of skeletal development, patterning, bone and cartilage formation, growth and homeostasis. Cleidocranial dysplasia presents with skeletal defects of several bones, the most striking of which are the partial or complete absence of clavicles, late closure of the fontanels, presence of open skull sutures and multiple wormian bones.[2] Late closure of fontanels is also a feature of Basal cell nevus syndrome and Crouzon syndrome, but together with other characteristic features, cleidocranial dysplasia can be easily differentially diagnosed. The base of the skull is dysplastic and its growth is reduced, leading to an increase in the width of the skull, resulting in brachycephaly and hypertelorism. Delayed closure of anterior fontanel and metopic sutures result in frontal bossing.

Thoracic cage is small and bell shaped with short ribs. Typically, clavicles are underdeveloped to varying degrees and in approximately 10 percent of cases, are completely absent. This allows excessive mobility of the shoulder girdle. Other bones may also be affected including long bones, the vertebral column, the pelvis and the bones of hands and feet.[3] Cooper et al recorded the natural history of 90 probands and 56 first- and second-

degree relatives, findings highlight the clinical variability of this condition within affected members of the same family who harbor the same pathogenic variant. Roberts et al reviewed their experience with more than 100 affected individuals in South Africa. Males and females are affected equally.[4]

Characteristically, patients with cleidocranial dysplasia, show prolonged retention of deciduous dentition and delayed eruption of permanent teeth. Adults with cleidocranial dysplasia have mixed dentition in their oral cavities.[5] In addition, patients with this condition, frequently show a large number of unerupted supernumerary teeth, often mimicking a premolar.[6] As many as 63 unerupted supernumerary teeth have been documented in one patient. Maxilla is also underdeveloped along with ill-formed paranasal sinuses. This condition is of clinical significance to every dentist due to the involvement of the facial bones, altered eruption patterns and multiple supernumerary teeth.[7]

## CASE REPORT

A 29 years male patient reported to the Department of Oral Medicine and Radiology, Karpaga Vinaya Institute of Dental sciences, with a chief complaint of the pain in his upper front tooth region for the past 4 days. Past medical history reveals no history of diabetes mellitus, hypertension, tuberculosis, ischaemic heart disease, bronchial asthma, and bleeding disorders. No relevant drug history. No adverse habit history. Past dental history reveals patient underwent uneventful extraction before 2 weeks. Family history reveals Patient is unmarried and his elder brother has a similar deformity and all other family members are apparently normal. On General physical examination, the patient was well built with short stature. He is conscious, cooperative, and well oriented to time, place, and person. On extraoral examination, facial asymmetry, short stature, short fingers of upper limb, hypertelorism and absence of clavicles on both right and left side.

On intraoral examination, spacing was seen in upper anterior tooth region, retained deciduous in relation to 74,75,83, generalised gingival recession.

### *Differential diagnosis*

Differential diagnosis was Crane-Heise syndrome, Mandibuloacral dysplasia, Pycnodysostosis, Yunis-Varon syndrome, CDAGS syndrome, Hypophosphatasia, Congenital hypothyroidism (disturbed thyroid metabolism).

### *Investigations*

Orthopantomogram (OPG) reveals presence of paramolar in relation to 17,27, mesioangular impaction was 13,38, horizontal impaction was 23,48, vertical impaction was 35,34,33,43, presence of odontome distal to 48, narrow ramus on both sides.

### *Chest X-ray*

Chest X-ray reveals absence of clavicle on both right and left side.

### *Paranasal sinus view*

Paranasal sinus view reveals presence of open fontanelles and open suture bones with many wormian bones.

### *Final diagnosis*

Final diagnosis revelas alveolar osteitis in relation 23 region, Scheuthauer-Marie-Sainton syndrome, partially edentuluous in relation to 23, 45 regions, retained deciduous in relation to 74,75,83, chronic generalized periodontitis.

### *Treatment plan*

Treatment plan reveals saline irrigation, oral prophylaxis, multidisciplinary approach (extraction of retained deciduous teeth-74,75,83, extraction of impacted-48, 38, orthodontic treatment for impacted 13,23,35,34,33,43).

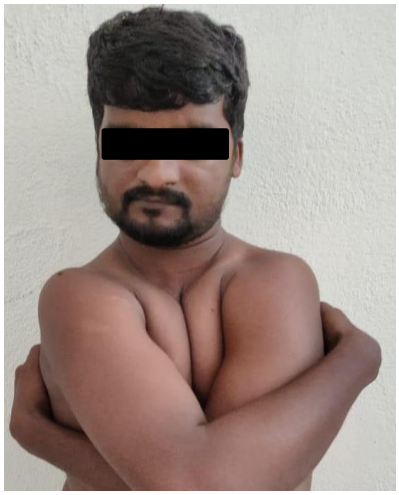

**Figure 1: Extra oral image.**

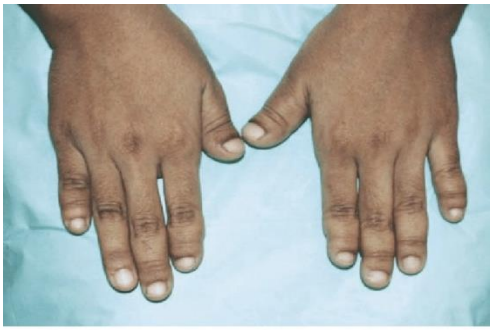

**Figure 2: Short fingers.**

**Figure 3: Intraoral image.**

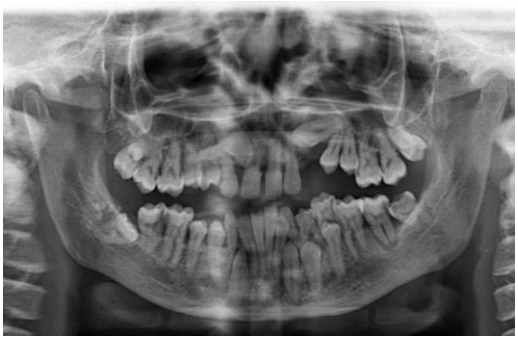

**Figure 4: OPG.**

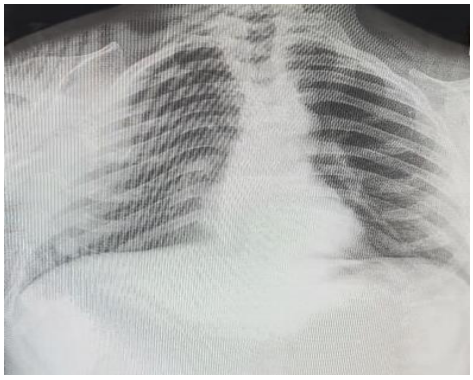

**Figure 5: Chest X-ray.**

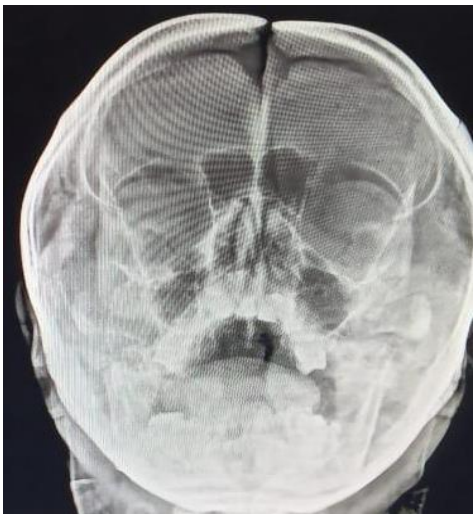

**Figure 6: Paranasal sinus view.**

## DISCUSSION

Cleidocranial dysplasia (CCD) spectrum disorder is a skeletal dysplasia representing a clinical continuum ranging from classic CCD (triad of delayed closure of the cranial sutures, hypoplastic or aplastic clavicles, and dental abnormalities) to mild CCD to isolated dental anomalies without the skeletal features. Most individuals come to diagnosis because they have classic features. CCD spectrum disorder affects most prominently those bones derived from intramembranous ossification, such as the cranium and the clavicles, although bones formed through endochondral ossification can also be affected.[8]

### *Classic CCD*

The most important clinical findings in people with classical CCD are listed under suggestive findings and include: Abnormally enlarged and wide fontanelles at birth can remain open for life, clavicle hypoplasia leads to narrow, sloping shoulders that may be antagonized in the midline and abnormal dentition.

Cleidocranial dysplasia (CCD) spectrum disorder should be suspected in individuals with the following clinical and radiographic findings.[9]

### *Clinical findings*

A fontanelle that is abnormally wide open at birth may remain open for life. A wide and open metopic suture leads to separation of the frontal bones by the Metopic sulcus. The forehead is broad and flat and the skull is brachycephalic.[10]

Frontal and parietal bossing and mid-face retrusion. Narrow, sloping shoulders that can be opposed at the midline due to clavicular hypoplasia or aplasia. Abnormal dentition including delayed eruption of secondary dentition, failure to shed the primary teeth, variable numbers of supernumerary teeth along with dental crowding, and malocclusion. Hand abnormalities including brachydactyly, tapering fingers, and short, broad thumbs. Short stature (typically moderate). Normal intellect in individuals with classic CCD spectrum disorder.[11]

### *Radiographic findings*

*Cranium*

Wide-open sutures, patent fontanelles, presence of wormian bones (small sutural bones). Delayed ossification of the skull. Poor or absent pneumatization of the paranasal, frontal and mastoid sinuses. Impacted, crowded and supernumerary teeth.

*Thorax*

Conical rib cage with narrow upper rib cage diameter. Typically, bilateral (but not necessarily symmetric)

clavicular abnormalities ranging from complete absence to hypoplastic or discontinuous clavicles. The lateral portions are more affected than the medial aspects of the clavicles. Hypoplastic scapulae

*Pelvis*

Delayed ossification of the pubic bone with wide pubic symphysis. Hypoplasia of the iliac wings. Widening of the sacroiliac joints. Elongated femoral head with short femoral neck and elongated epiphyses ("chef-hat" appearance). Coxa vara.

*Hands*

Pseudoepiphyses of the metacarpal and metatarsal bones, which may result in a characteristic lengthening of the second metacarpal. Hypoplastic distal phalanges. Deformed and short middle phalanges of the third, fourth and fifth digits with cone-shaped epiphyses.[12]

*Others*

Osteopenia/osteoporosis with evidence of decreased bone mineral density by DXA, some affected individuals sustain multiple fractures.

Further medical problems identified in individuals with CCD spectrum disorder include the following:

***Height***

Individuals with CCD spectrum disorder are often shorter than their unaffected sibs: males are on average six inches shorter than their unaffected brothers and have an average height of 165 cm (±8 cm). Females are on average three inches shorter than their unaffected sisters and have an average height of 156 cm (±10 cm).[16]

***Skeletal/orthopedic problems***

Affected individuals are more likely to have other bone-related problems: pes planus (flat feet) in 57%, genu valgum (knock-knee deformity) in 28%, scoliosis in 18%, osteoporosis, found in 8/14 (57.1%) affected individuals and osteopenia, identified in 3/14 (21.4%) individuals with CCD spectrum disorder, other less common orthopedic problems include joint dislocation at the shoulder and elbow.[16]

***Dental complications***

Up to 94% of individuals with CCD spectrum disorders have dental findings such as supernumerary teeth (often not losing deciduous teeth) and failure to erupt permanent teeth.[16] The most consistent dental findings in individuals with CCD spectral defects were the presence of second permanent molars with primary dentition (80%), wide spacing of lower incisors, excess tooth germ (70%) and parallel predominance.[16] Individuals with CCD spectrum disorders are more likely to have underbites and cysts in the gums, which usually form around extra teeth.[16]

***ENT complications***

Recurrent sinus infections and other upper respiratory complications are observed significantly more frequently in individuals with CCD spectrum disorders than in the general population. If symptoms suggest upper airway obstruction, a sleep evaluation is needed and surgery may be needed. Conductive hearing loss occurs in 39% of affected individuals. Individuals with CCD spectrum disorders of all ages are more likely to have recurrent ear infections.

***Endocrinology***

Individuals with CCD spectrum disorders may have low levels of IGF-1. Low vitamin D has also been reported and is not consistently associated with osteoporosis.[16] Rarely, individuals with CCD spectrum disorders have low levels of alkaline phosphatase.[16]

***Development***

Intelligence is typically normal. Children under the age of 5 may exhibit slight motor delays, especially with respect to gross motor skills. This delay may be related to orthopedic complications such as flat feet and valgus knees. No significant difference is seen in children of primary school age.

***Prevalence***

CCD spectrum disorder is present at a frequency of one in 1,000,000 individuals worldwide. It affects all ethnic groups found the frequency to be 0.12 per 10,000 individuals in the Utah (USA) population, suggesting that the frequency may be higher than previously recognized.[13,14,16]

On comparing with the present case, patient has short stature, short and broad thumbs, narrowing of shoulders with absence of clavicles, abnormal dentition and open and wide fontanelles with numerous wormain bones.

***Differential diagnosis***

*Crane-Heise syndrome*

Crane-Heise syndrome reveals similar features such large head, poorly mineralized calvarium, cleft lip and palate, low-set, dysplastic ears, hypoplastic clavicles and scapulae, hypoplasic/absent phalanges, absence of cervical vertebrae, genital hypoplasia and distinguishing feature such as multiple joint contractures, severe vertebral and limb anomalies with or absence of cervical vertebrae

*Mandibuloacral dysplasia*

Mandibuloacral dysplasia reveals similar features such as short stature, delayed closure of cranial sutures, mandibular hypoplasia and dysplastic clavicles, scalp hair sparse by 3rd decade, progressively stiff joints, acroosteodysplasia of fingers and toes with delayed ossification of carpal bones, micrognathia, early tooth loss, atrophic skin with or without subcutaneous fat and distinguishing feature such as acroosteolysis, hyperpigmentation, lipodystrophy, alopecia

*Pycnodysostosis*

Pycnodysostosis reveals similar features such as short stature, osteopetrosis with increase in bone fragility and short terminal phalanges, failure of closure of cranial sutures with persistence of an open fontanelle, increase in radio-opacity of all bones due to increase in density of trabecular bone but not cortices and distinguishing feature such as osteopetrosis, acrosteolysis,

*Yunis Varon syndrome*

Yunis Varon syndrome reveals similar features such as prenatal growth deficiency, wide-open fontanelles and sutures, unusual mineralization of the skull and hypoplastic clavicles, hypoplastic or absent thumbs and great toes and distinguishing feature such as absence/hypoplasia of thumbs, halluces and distal phalanges, gracile bones, brain malformations

*CDAGS syndrome*

CDAGS syndrome reveals similar features such as craniosynostosis, delayed closure of fontanelles, cranial defects and clavicular hypoplasia, anal and genitourinary malformations, skin eruption and distinguishing feature such as craniosynostosis, anal anomalies, skin lesions (porokeratosis)

Hypophosphatasia

Hypophosphatasia reveals similar features such as generalized defect of mineralization with delayed ossification of multiple skeletal elements, children with infantile form may present with very poorly mineralized cranium, widened cranial sutures short ribs and narrow thorax, very low alkaline phosphatase activity in serum and tissues and distinguishing feature such as clavicles least affected, no supernumerary teeth, premature deciduous tooth loss, rachitic skeletal changes, nephrocalcinosis, hypercalcemia.

***Genetic counselling***

Genetic counseling is the process of providing individuals and families with information about the nature, mode of inheritance, and effects of genetic disorders to help them make informed medical and personal decisions. It deals with clarifying the genetic status of the family using genetic risk assessment, family history and genetic testing. It is not intended to address all personal, cultural, or ethical issues that may arise, or to replace the advice of a geneticist.[13-15]

## CONCLUSION

Cleidocranial dysplasia is a rare condition. However, its clinical and radiological manifestations have been characterized. Early diagnosis allows proper focus on treatment and improves quality of life. A holistic approach considers all aspects, including major pathological and psychological aspects. Dental management of CCD requires an interdisciplinary approach, and indeed requires special skills of paediatric dentists to treat such cases.

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