**Case Report**

**Cleidocranial Dysplasia – A Rare Autosomal Dominant Syndrome**

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**Abstract**
Cleidocranial dysplasia is a rare disease which can occur either spontaneously or by a dominant autosomal inheritance, with no predilection of genre or ethnic group. The odontologist is, most of the times, the first professional who patients look for to solve the problem, since there is a delay in the eruption and/or an absence of permanent teeth. Many others odontological problems are also present in this syndrome, such as, retained teeth, absence of deciduous teeth resorption. The premature diagnosis allows a proper orientation for the treatment, offering a better life quality for the patient.1.

**Keywords:**
Cleidocranial dysplasia; Dysostosis; Dentofacial deviation.
**Introduction**

The cleidocranial dysplasia, also known as Marie and Sainton Disease, Scheuthauer Marie-Sainton Syndrome and Mutational dysostosis (Shafer *et al.*, 1979) is a rare disease which can occur either spontaneously or by a dominant autosomal inheritance, with no predilection of genre or ethnic group. The first case of clavicular defects was reported by Martin in 1765. Another case with both clavicles and the skull affected was reported in 1871 by Scheuthauer. In 1897 Marie and Sainton coined the descriptive term cleidocranial dysostosis. The condition was originally thought to involve bones of intramembranous origin only, namely the bones of the skull, clavicles and flat bones, hence the name cleidocranial. Hesse was first to describe in detail the defects of dentition and jaws associated with cleidocranial dysostosis.

It is characterized by a generalized skeletal dysplasia. The absence of the clavicles, which occurs in 10% of cases or the presence of hypoplastic clavicles allow the patient the movement of the shoulders up to the medial plan of the body without any discomfort.

They are slightly dwarfed and slender and have long necks and drooped shoulders. In general, CCD patients are of a normal intelligence quotient and health. They have persistent fontanels of the cranium or late closure of the same. The parietal bones, frontal and occipital are prominent, the paranasal sinus underdeveloped and many other cranial abnormalities might be present.

Regarding the manifestations of odontological interest it is important to mention that these patients show an arcate, narrow and deep palate. The maxilla can be underdeveloped and shorter than normal in relation to the mandible, resulting as a consequence, in a pseudo mandibular prognathism. The zygomatic and lacrimal bones can also present themselves underdeveloped. Another important finding is the presence of supranumerary teeth, impacted and ectopic, presenting crown and root anomalies. The dental eruption is retarded, and an absence of root resorption in the deciduous teeth, hypodontia and dentigerous cysts can be observed.

**Case Report**

A 8 years old male patient came to the department of oral and maxillofacial surgery in Guru Nanak Dev Dental College Sunam with the chief complaint of prolonged retention of deciduous teeth.

In the extra-oral physical examination we could notice short height, Large head, Slight ocular hypertelorism, Broad base of nose with depressed nasal bridge, prominent frontal and parietal bones, Frontal and occipital bossing determining an increase in the cranial perimeter with Flat appearance of skull. When asked to
move the shoulders, he was capable of bringing closer the humeral heads, which characterized the hypermobility of the shoulders.

**Discussion**

Cleidocranial dysplasia is very rare in occurrence, incidence being 1:1000000. This pathology was first described in 1765, whereas only in 1898 Mane & Sainton had described cases of the disease and associated them with patterns of inheritance. Later, Bauer apud Kallialla (1962) suggested the genetic mutation as an etiological factor of the disease. In 1946 Lasker apud Forlan (1962) had concluded that it was a genetic disease with an autosomal dominant inheritance, and in some cases, external interferences in the fetal period could cause this mutation that is transferred to the progeny.

Other names of disease are Marie & Sainton’s Disease, Scheuthouer-Marie Sainton Disease & Mutational Disorder. Disease is of unknown etiology, often not always hereditary. It is an autosomal-dominant heritable skeletal disease caused by heterozygous mutations in the osteoblast-specific transcription factor RUNX2 gene, transmitted by either sex with Male & Female same predicition. It is characterized by a generalized skeletal dysplasia. Clinical features include short stature, membranous bones are affected – skull, clavicles, Skull is large and open fontanels/ Delayed closure. Sutures remain open. Frontal /parietal/occipital bones are prominent with (frontal and parietal bossing. Wormian bones are commonly present.

The average height of adult man with Cleidocranial dysplasia is 156.6 to 168.8 cm and for a women is 144.6 to 148.5 cm. Midfacial skeleton is involved with hypoplastic lacrimal and zygomatic bones. There is also hypoplasia of nasal bone & paranasal sinuses. The nose usually is broad based with depressed nasal bridge. Head is brachycephalic i.e cephalic index is increased. It is wide & short with increased transverse diameter of skull. There is complete absence of clavicles (10%) to partial or even simple thinning of one/both clavicles. This allows the patient the movement of the humeral heads up to the medial plan of the body without any discomfort. Cervical or thoracic vertebral defects, supernumerary ribs, thoracic and lumbor scoliosis, kyphosis or lordosis, pelvic bony abnormalities and anomalies of phalangeal, tarsal, metatarsal, carpal and metacarpal bones are all systemic findings.

**Oral Manifestations**

Maxilla is underdeveloped smaller and mandible is large in 70% cases. In the región of the symphysis of the mandible an incomplete fusión is commom to be observed. Dentigerous cysts can be
observed. In radiographs increase in the osseous density of some regions as a result of an abnormal remodeling of the bone can be seen. High and narrow arched palate can be observed with increased prevalence of Cleft palate\textsuperscript{1,3}.

The dental eruption is retarded, and an absence of root resorption in the deciduous teeth, hypodontia and dentigerous cysts can be observed. There is prolonged retention of deciduous teeth with delayed eruption of succedaneous teeth. Patient gives premature aged appearance.

The roots of teeth are characteristically short and thin. Sometime dilacerations of roots can be seen. Numerous unerupted supernumerary teeth can be appreciated in radiographs, most prevalent in mandibular premolar & incisor areas. There is absence/paucity of secondary/cellular cementum on roots of permanent teeth. This may be related to failure of eruption (Rushton). Smith confirmed absence of secondary/cellular cementum on both permanent/deciduous teeth with increased thickening of primary acellular cementum\textsuperscript{1,7}.

Pycnodysostosis / Maroteau Lamy Syndrome is syndrome with most of features of Cleidocranial dysplasia along with dwarfism, partial agenesis of terminal phalanges of Hands & Feet and Dense fragile bones.

**Treatment**

The treatment for the alteration in the dentoalveolar complex, in the patient of the cleidocranial dysplasia, involves the teeth restoration of the deciduous teeth when they present cavities, because their extraction does not necessarily induces the eruption of the permanent teeth (Shafer et al). It also involves the orthognathic surgery, to fix the maxillary hypoplasia, and the surgery in the impacted teeth in association with the ortodontical and/or prosthetic therapy. The implantology can be carried out recently as another therapeutic option in the oral rehabilitation of these patients. The combined orthodontic and oral surgical procedures can be carried out for uncovering teeth and orthodontic repositioning.

**Summary**

This case discusses several characteristic anomalies in a patient with cleidocranial dysplasia. Orthodontic treatment is usually indicated to direct the eruption of the malposed and often impacted teeth. Extraction of some of the supernumerary teeth may be needed. Timing of the intervention is critical, and many surgeries may be required. Patients with cleidocranial dysplasia require a team approach involving an orthodontist, pedodontist and an oral surgeon with good cooperation and communication from the patient.
Figure 1: Unusual mobility of shoulders

Figure 2: Panoramic image shows retained deciduous teeth

Figure 3: Increased circumference of skull and open fontanels

Figure 4: Patients father also showed unusual mobility of shoulders

Figure 5: Patients father panoramic image shows retained deciduous teeth and impacted permanent teeth


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