CASE REPORT

CLEIDOCRANIAL DYSOSTOSIS- A CASE REPORT

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Abstract

Objective: First report of cleidocranial dysostosis (CCD), a rare genetic disorder, in Ghana.
Case report and intervention: The condition presented in a 13 year old boy with most of the classical features. He had cranial features in the form of open fontanelles, underdeveloped paranasal air sinuses and hypertelorism. Thoracic features were severely underdeveloped clavicles which allowed him to appose both shoulders in the midline. He had an open bite, several unerupted permanent teeth and several retained teeth most of which were discoloured which was why he was seen at the Dental department. After restoration of some of these teeth the patient was very satisfied.
Conclusion: Though the definitive treatment in these patients can be sophisticated major craniofacial surgery, early restoration of malformed teeth can help ease patient's anxiety and improve quality of life.

Key Words: Cleidocranial dysostosis, Ghana, genetic, disorder, jaws, dentition

Introduction

Cleidocranial dysostosis (CCD) is a rare congenital disorder of bone and dentition[1, 2, 3]. Also called cleidocranial dysplasia, mutational dysostosis and Marie-Sainton disease[4], the condition is mainly characterized by clavicular aplasia or deficient formation of the clavicles, delayed and imperfect ossification of the cranium, moderately short stature, and a variety of other skeletal abnormalities[5] The principal oral manifestations are a delayed exfoliation of primary teeth, delayed or multiple impactions of the permanent dentition, and multiple impacted supernumerary teeth[6]

It may be inherited as an autosomal dominant pattern or occur as de novo mutation of the affected gene. The gene has been mapped on the short arm of chromosome 6p21, core binding factor a-1 (CBFA1). This disorder can be caused by a mutation in the transcription factor CBFA1 (RUNX2). The CBFA1 gene controls differentiation of precursor cells into osteoblasts and is, therefore, essential for both membranous and endochondral bone formation. This may be related to delayed ossification of the skull, teeth, pelvis, and clavicles[7] Diagnosis of CCD is usually based on clinical and radiographic findings and affected individuals may live a normal life[4,5,8].

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Cases have been published in South Africa[9] and Nigeria[10] but to the best of our knowledge, this is the first reported case of the condition in Ghana

Case Report

A 13 year-old male presented to the Dental Surgery clinic, 37 Military Hospital, Accra, with a complaint of discoloured and carious teeth in both upper and lower jaws which had been present since childhood. He requested improvement in his appearance and the colour of his teeth.

His only relevant medical history was the surgical removal of a sixth finger next to the right little finger which was carried out under local anaesthetic without complications a "few" years previously. He also said he could approximate his shoulders to meet in the midline

He had never visited a dental surgeon in the past. There was no social or family history of note. The patient's stature was small for his age but otherwise looked generally well. Initial examination focused on the head and neck.

The site of the anterior fontanelle was soft. He had hypertelorism and flaring of alae nasi.
The skeletal jaw relationship was class III with a protrusive mandible and an anterior open bite.

Almost all his deciduous teeth were retained. The only permanent teeth present were the first permanent molars. There was discolouration of several teeth in both jaws. Some of the teeth had caries. There was a notch in the midline of the palate which otherwise looked flat. On further examination he could demonstrate the almost meeting in the midline of his two shoulders with ease. He also had pes planus. Fig 1

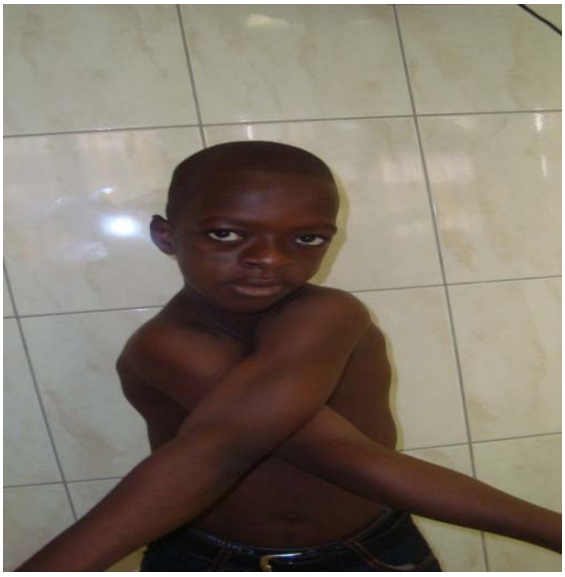

Figure 1: Shows the apposition of the shoulders close to the mid-line.

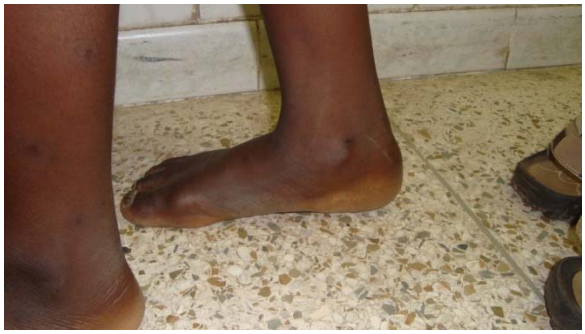

Figure 2: Presence of pes planus in the patient

Plain radiographs of the patient's jaws (Orthopantomogram [OPG] and lateral skull views) and posterior- anterior radiographs of the chest were requested on account of the missing teeth, the skeletal class III appearance and the ability to bring his shoulders close to the midline with ease.

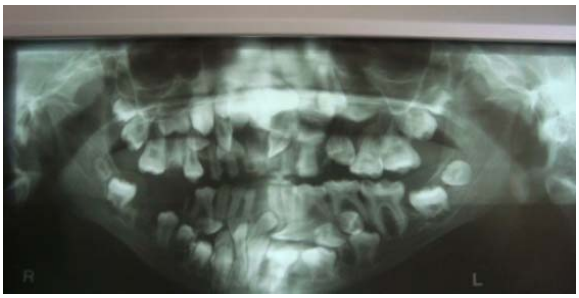

Figure 3: Orthopantomogram view of the jaws showing several unerupted teeth.

The OPG and lateral skull views showed several teeth impacted and unerupted in both jaws. Some of these were supernumeraries. (Fig. 3, 4).

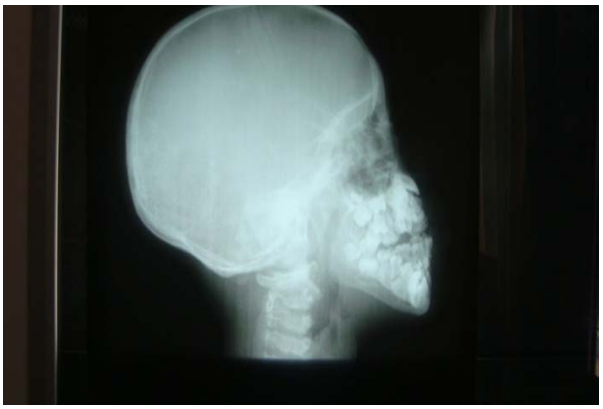

Figure 4: Lateral skull radiograph showing open skull sutures, large fontanelles, and underdeveloped paranasal sinuses.

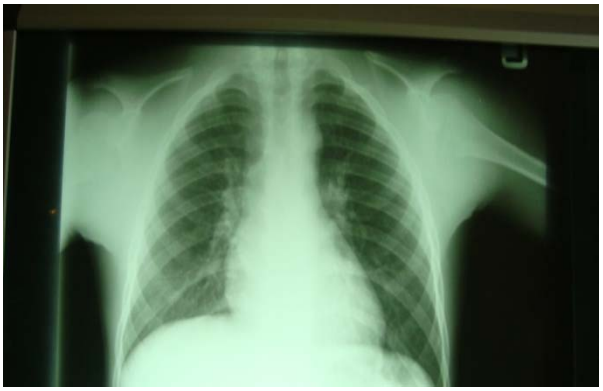

Figure 5: Chest radiograph (PA View) of the patient showing thinning and hypoplasia of the clavicles and bell shaped rib-cage.

The lateral skull view also confirmed the skeletal III jaw relationship. The chest radiograph showed clavicular hypoplasia and bell shaped rib-cage (Figure 5). These radiographic and clinical features confirmed our diagnosis of CCD.

Management: The patient and the parents were reassured. Dietary advice and oral hygiene instructions were reinforced. Composite restorations of his carious teeth as well as veneers for the discoloured teeth were carried out fig 6. Badly damaged primary teeth were extracted and where there was a high chance for eruption of the permanent teeth, the corresponding primary teeth were extracted to make way for their permanent counterparts. Future orthodontic repositioning of some of the teeth and artificial replacement of missing teeth as well as orthognathic surgery was discussed and this will be considered at a future date.

The patient was also referred to the ENT, Orthopedics and medical departments for further investigations. These further examinations and evaluations did not reveal any new clinical features.

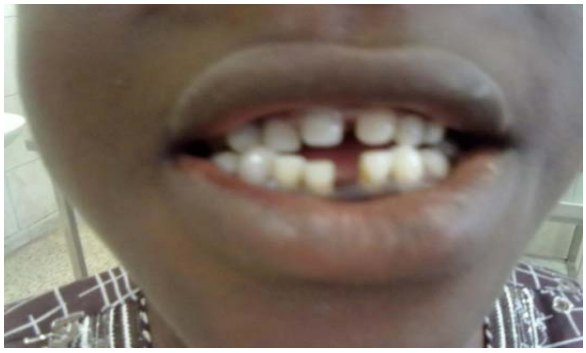

Figure 6: Appearance of dentition after composite restorations.

Discussion: The descriptive term cleidocranialdysostosis was first used by Pierre Marie and Paul Sainton in 1898[11, 12]. The condition was originally thought to involve bones of intramembranous origin only, namely the bones of the skull, clavicles and other flat bones, hence the name cleidocranial. Hesse was first to describe in detail the association of the defects of dentition and jaws[13]

The prevalence of CCD is one per million of the population. It is inherited as autosomal dominant trait with complete penetrance and variable expressivity[8]. It is most likely underdiagnosed because of the relative lack of medical complications in comparison with other skeletal dysplasias. It may be discovered at any age, but the cranial deficiencies may be noticed at birth. There is no gender predilection. The defect often appears in several successive generations[14, 15, 16]

The most characteristic and pathognomonic feature of this disorder is hypoplasia or aplasia of the clavicles, which results in hypermobility of the shoulders allowing the patients to approximate the shoulders in the midline. Muscle attachments to the clavicles may also be dysplastic, leading to distortion of the neck. Defects of the cervical and lumbar vertebrae are included in the clinical findings. Absence of the pubic symphysis and hypoplasia of the pelvis is common in females. Postural defects and spinal curvature are common. Frequently, genuavalga and pes planus are found in children younger than 5 years of age. In our case our patient had pes planus. They are of moderately short stature[14, 15, 16]

Delayed ossification of the cranial sutures and fontanelles occurs, and may remain open throughout life. The skull is usually large and broad.

The face appears small in relation to the cranium with hypoplastic maxillary, lacrimal, nasal, and zygomatic bones. The paranasal sinuses may be underdeveloped. The bones of the middle part of the face are also less well developed than the cranial bones. Defects in the skull appear to be always symmetrical. The frontal, parietal, and occipital bones are prominent. The maxillary sinuses may be small or missing, and the maxilla is underdeveloped, causing a Class III skeletal relationship and a maxillary retrusion (a relative mandibular prognathism). The palate may be abnormally high, and, occasionally, a cleft palate has been reported. Ocular hypertelorism and mild exophthalmus are seen[16].

In the hands and the feet, various abnormalities have been found, the most constant and curious being the presence of epiphyses at both ends of the metacarpals and metatarsals, particularly of the second and fifth, and an abnormally long second metacarpal. The intermediate phalanges may be small. Association with mental retardation has been shown, but most patients apparently possess normal intelligence. These individuals have no significant physical handicap[16].
The eruption of primary teeth is normal or sometimes delayed, but the exfoliation of primary teeth is always delayed and may be due to the failure of most permanent teeth to erupt[17]. The presence of supernumerary teeth has been hypothesized to result from incomplete or delayed resorption of the dental lamina. Dental manifestations include delayed eruption or failure of eruption of the primary dentition. Delayed tooth development has been reported in association with malocclusion and supernumerary teeth[15]. The significant variability in clinical expression of this syndrome reflects a degree of phenotypic polymorphism[16]

Confirmation of the diagnosis was based on the examination of the cranium, the face, the intra-oral features (mainly the dentition), clavicles, including shoulder mobility.

Our patient had most of the classic features.
Our patient had an open anterior fontanelle, hypertelorism, flared alae nasi, a relative mandibular prognathism, an anterior open bite, several carious and discoloured teeth, shoulders that nearly met in the mid-line, pes planus and a notch in the hard palate.

Radiographically, a PA chest radiograph confirmed his clavicles were nearly missing, and an OPG showed he had several supernumerary teeth, several impacted and unerupted teeth, a maxillary antrum nearly filled with unerupted teeth and skeletal mandibular prognathism or maxillary hypoplasia.

The patient's treatment is restricted in our environment. Quality of life is such as the facilities we have in our health service will allow. There was a huge smile after the initial treatment. There are plans to involve other specialists to provide an integrated team service, made up of a pedodontist, an orthodontist, and an oral and maxillofacial surgeon, however we could not confirm this with the parents. No doubt aesthetic improvement brought a smile to their face and to some extent improves their quality of life.

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