CASE SERIES

DELAYED ERUPTION IN CLEIDOCRANIAL DYSPLASIA

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Cleidocranial dysplasia is a rare autosomal dominant disorder of bones mainly affecting bones formed by intra-membranous and endochondral ossification. It presents clinically as brachycephalic skull, frontal bossing, depressed nasal bridge, hypertelorism, delayed closure of fontanelles, delayed eruption of permanent teeth, missing clavicles and wide pelvis. It runs in families; sporadic cases are rarer. Just over than 1000 cases have been reported. We are presenting here 2 cases of cleidocranial dysplasia in siblings.

Keywords: Cleidocranial Dysplasia; Supernumerary Tooth; Pseudarthrosis of Clavicle; Congenital

Citation: Ahmed S, Shaheen MN, Hameed F. Delayed eruption in Cleidocranial Dysplasia. J Ayub Med Coll Abbottabad 2020;32(3):406–8.

INTRODUCTION

Cleidocranial dysplasia is a rare disorder of bones. It is an autosomal dominant condition running in families. Previously considered dystosis, it is now named as dysplasia since it is a generalized disorder.[1] The CBFA1 (core binding protein factor subunit Alpha 1) gene responsible for this disease. it is mapped to 6p21 chromosome.[2] Its characteristics features are presented in table-1:[1]

Table-1: Key features of Cleidocranial dysplasia[3]

• Frontal bossing
• Open/delayed closure of fontanelles.
• Hypoplastic maxilla.
• Delayed/impacted permanent teeth.
• Supernumerary teeth.[3]
• Scoliosis.
• Short stature
• Depressed nasal bridge
• Partial/absence of clavicles
• Dentigerous cyst
• Thinner roots.
• Sparse/absent cementum on permanent and deciduous dentition

Activation of CBFA 1 gene during postnatal growth leads to this disease. Hearing problems and Musculature underdevelopment[1] is also evident in these patients. Reduced paranasal sinuses along with dysfunction of auditory tube leads to frequent bouts of inflammation and infection of middle ear and sinuses.[4,5] Diagnosis is on basis of clinical and radiographic features.[5]

Management includes protection from blunt trauma to the head.[5] Vitamin D and calcium supplements are prescribed for the generalized decreased in bone density. Orthodontic surgery for hypoplastic maxilla. Orthodontic treatment for eruption of permanent teeth, if partly effective then prosthetic rehabilitation of the patient.[2] Treatment usually patient tailored.

CASE-1:

An 11-year-old boy reported to Out-patient department of Oral & Maxillofacial Surgery with complaint of impacted permanent teeth. Patient had history of dental check-up from different hospitals for the same problem. He was under treatment for short height with vitamin D and calcium supplements. On examination the patient appeared short stature and pale. Frontal bossing was prominent with open anterior fontanelles. Nasal bridge and maxilla were depressed with missing deciduous maxillary and mandibular Central and lateral incisors 51, 52, 61, 62, 72, 81 and 82. Oral hygiene was poor with calculus around right mandibular anterior teeth. Decayed mandibular right posterior tooth. Patient was asked to bring both shoulders together showing abnormal mobility of the shoulders showing absence of clavicles. [Figure-1] Patient was advised posteroanterior view face and OPG and bone profile tests. Posteroanterior view showed open anterior fontanelle and metopic suture. OPG showed impacted permanent teeth. [Figure-2] Albumin and calcium levels were deranged. Patient was referred to a paediatrician for further evaluation.

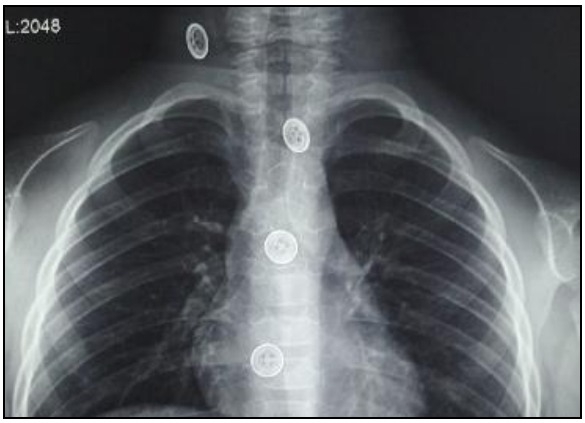

Figure-1: Chest x-ray showing missing clavicles

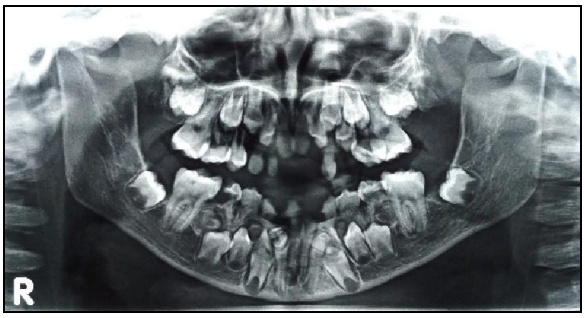

Figure-2: Orthopantamotomogram showing impacted permanent teeth with only upper and lower first molars erupted. Parallel anterior and posterior border of mandible

CASE-2:

A 9-year-old female patient, sister of above boy came for impacted permanent teeth. She had same characteristics features:

- Delayed closure of anterior fontanelle.
- Frontal bossing.
- Impacted permanent with only erupted lower left central incisor and maxillary and mandibular all four first molars.
- Short stature.
- Missing clavicles.

She was also diagnosed to have cleidocranial dysplasia advised OPG and bone profile. Chest X-ray showing missing clavicles and slight scoliosis of spine. OPG showed unerupted permanent teeth and retained deciduous teeth with hypoplastic sinuses and ascending ramus being narrower. Albumin and calcium levels were also deranged. She was also referred to paediatrician for further evaluation and treatment.

DISCUSSION

Cleidocranial dysplasia is diagnosed clinically on basis of delayed eruption of permanent teeth supernumerary teeth, with wide open/delayed closure of fontanelles, missing or hypoplastic clavicles and wide pelvis. It is 70% due to deletion in RUNX2 gene.[1] Till today three cases[7–9] were reported in Pakistan with just above 1000[1] worldwide. 70% of the cases are inherited due to consanguineous marriages only few are sporadic ones.[6] Usually the intellect of the patient is unimpaired.[6] Supernumerary teeth add to the crowding of the dentition. Delayed union of mandibular symphysis and sloping of shoulders is also present.[3] Case reported by Dr. Nazir showed a case of CCD with neurofibromatosis type 1, which is was not reported in literature previously. Our findings were similar to Dr. Qiam and Dr. Baig's reports documenting short stature, delayed closure of fontanelles, delayed eruption along with cysts, frontal bossing and missing clavicles. Differential diagnosis includes Congenital Pseudarthrosis, Pyknodystosis, Mandibulofacial dysplasia, Yunis Varun syndrome, Gardner syndrome and Kenney Caffey syndrome [Table-2].[10]

Table-2: Syndromes similar to Cleidocranial dysplasia[3]

Disease	Symptoms
1. Congenital pseudoarthrosis	Unilateral missing clavicles
2. Pyknodystosis	Osteosclerosis Delayed closure of suture/ fontanelle Hypoplasia of clavicle Mal aligned teeth Anodontia and delayed eruption
3. Mandibuloacral dysplasia	Hypoplastic clavicle Wide sutures Multiple wormian bone Acro osteolysis with loss of bone from distal phalanges.
4. Yunis Varun syndrome	Lethal condition Growth deficiency Failure to thrive Wide clavarial sutures Enlarged fontanelles Agenesis/hypoplasia of thumb and big toes
5. Gardner syndrome	Supernumerary teeth
6. Kenney Caffey syndrome	Increased bone density Impaired bone growth Decreased mineralization

Genetic counselling is one of the important factors as this disease is autosomal dominant with 50% chances of inheritance.[6] In both cases delayed eruption and anterior open fontanelles along with missing clavicles were the diagnostic points. Dental treatment usually is around the following goals:

1. Restoration of deciduous as permanent fail to erupt.
2. Ortho treatment to direct eruption of permanent if deciduous Exfoliates.
3. Orthognathic surgery to compensate maxillary hypoplasia
4. Extraction of supernumerary teeth as well as enucleate associated cysts.
5. Prosthetic rehab/implant.

In the current cases patients were advised surgical exposure and orthodontic traction but then both failed to turn up for follow up.

Acknowledgement: All authors have viewed and agreed to submission

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Submitted: February 23, 2019	*Revised: October 27, 2019*	*Accepted: January 12, 2020*

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