Case Report

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Dentistry Section

Unilateral Ankylosis of Temporomandibular Joint (TMJ) with Contralateral Condylar Aplasia and Related Orthopedic Deformity – Syndromic or Nonsyndromic?

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ABSTRACT

Condylar aplasia which means "failure of development" is a rare condition and can be unilateral or bilateral. Mandibular condylar Aplasia without any association with syndrome is extremely rare. Temporomandibular joint (TMJ) ankylosis results from trauma, infection and inadequate surgical treatment of the condylar area. Congenital cases are very rare. We report case of congenital unilateral aplasia of left mandibular condyle with ankylosis of right condyle, with an associated orthopedic deformity in a nine-year-old male patient, which may be a part of some unreported syndrome that has not been mentioned so far in literature. As per our best knowledge, no other case including such clinical features has been reported.

Keywords: Developmental disturbances, Gene mapping, Retrognathia

CASE REPORT

A 9-year-old presented, with a chief complaint of inability to open mouth. On careful history taking the patient's mother revealed the childbirth to be a normal delivery. On further questioning, the mother revealed that the child suffered from jaundice within seven days of birth, and developed a papule at the same time on the left lower limb, which was surgically removed (the patient's family was from poor social-economic status and thus his mother was not able to give detailed report of any pediatric consultation or surgical history and also they had not preserved any past medical records). At the age of six months, it was noticed that the patient had restrictions in mouth opening. There was no history of trauma or middle ear infection. Also, walking difficulty was detected, at around one year of age. The child slept in a semi-supine position and still had loud snoring and wheezing. The food intake was limited to liquids or semi-liquids. The patient's parents had no history of consanguinity and his siblings were normal; no blood incompatibility was found. There was no family history of any craniofacial abnormalities.

CLINICAL EXAMINATION

The clinical findings included low weight (13 kg) and short stature (96 cm) for his age with noisy breathing. The patient had a bird-like facial profile with double chin and retrognathia of the mandible and short neck [Table/Fig-1a,b]. There was no deviation of mandible. On palpation left condyle was not detected and had a Class II with division 1 malocclusion and collapsed bite. The inter-incisal opening was about 1 mm. Lateral and forward mandibular movements were relatively difficult [Table/Fig-1c]. The patient also had limb deformity that was depicted by walking difficulty [Table/Fig-1d].

INVESTIGTAIONS

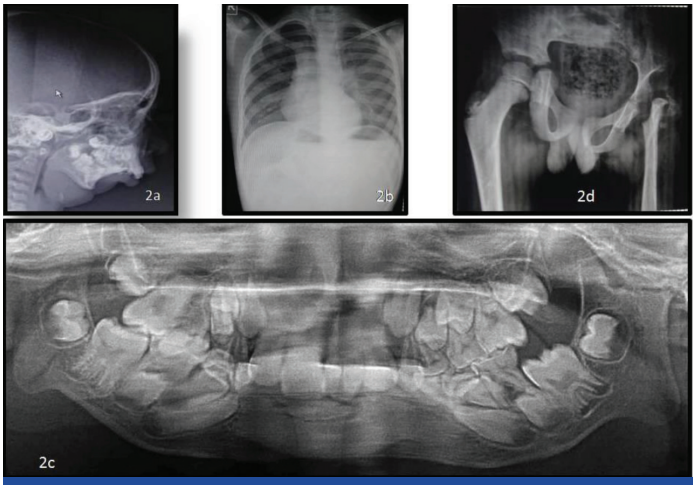
Blood Examination

Patient's blood profile showed that he was anemic, with hemoglobin being 5 gm% and his blood group was B negative. Rheumatoid factor test was done to rule out rheumatoid arthritis and was negative.

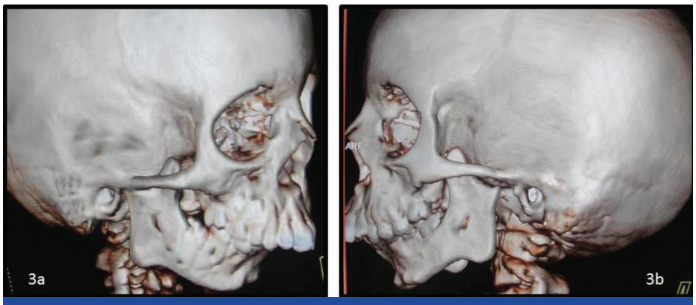
Radiological Examination

Lateral Cephalogram, orthopantomogram and 3D CT were done to study the facial skeleton. The lateral cephalogram revealed that facial skeleton appeared small in comparison with the neurocranium and exhibited mandibular retrusion. Tooth buds of erupting molar teeth were also seen [Table/Fig-2a].

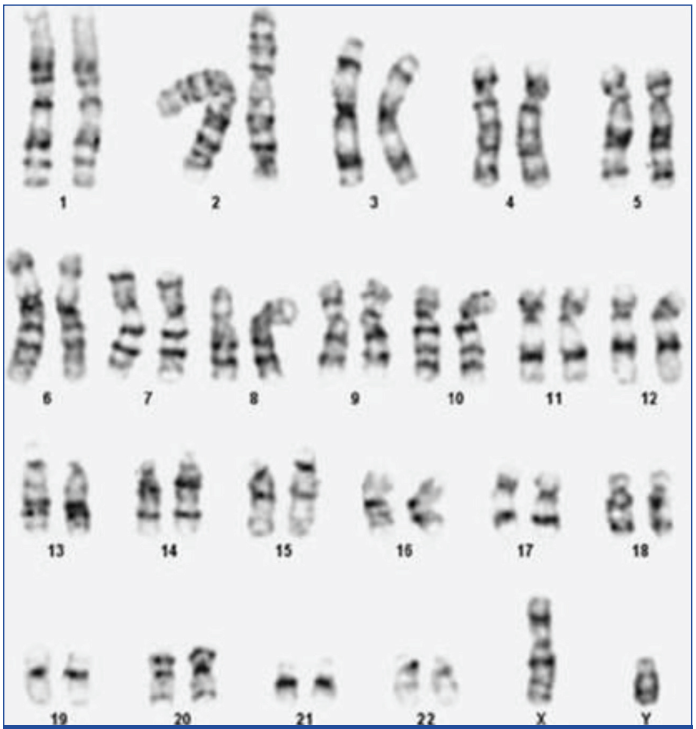
[Table/Fig-1]: Clinical examination – (a) Frontal view (b) profile view (c) intraoral view (d) Limb deformity



[Table/Fig-2]: Radiographic examination: (a) Lateral Cephalogram (b) Chest X-ray. (c) orthopantamograph (d) Pelvic radiograph reveals hypoplasia of left femur and aplasia of the left caput femoris and collum femoris



[Table/Fig-3]: 3D reconstruction image a) Ankylosis of right condyle b) Aplasia of left condyle



[Table/Fig-4]: Chromosomal karyotyping which revealed normal male karyotype

The upper airway was constricted. But, Chest x-ray of the patient was normal [Table/Fig-2b]. The panoramic radiograph revealed short and underdeveloped condyles bilaterally. The condyle on right side was fused with the glenoid fossa. Absence of condyle on left side was seen [Table/Fig-2c].

A pelvic radiograph was taken which revealed hip hypoplasia of the left side. Additionally, absence of femur head on left side hip was detected [Table/Fig-2d].

Three-dimensional (3D) CT reconstruction revealed fusion of condyle with glenoid fossa on right side [Table/Fig-3a] & condylar aplasia of the left side [Table/Fig-3b].

GENE Mapping

Chromosomal karyotyping was done in order to rule out if there was any chromosomal abnormality. This was performed using G banding at trypsin & giemsa technique & banding resolution at 450-550, which revealed normal male karyotype. No numerical or structural chromosomal anomalies were detected at 450-550 banding resolution [Table/Fig-4].

On the basis of history, clinical and radiographic examination and gene mapping, a diagnosis of congenital unilateral TMJ ankylosis of right side and condylar aplasia of left side with an associated femoral hypoplasia was made. The patient was referred to department of oral and maxillo-facial surgery for correction of the cranio-facial deformity, but the patient refused for getting any surgical treatment done.

DISCUSSION

The TMJ is among the intricate joints of the human body and appears first during eight week of gestation [1]. Growth disturbances of mandibular condyle in the first trimester can lead to hypoplasia or aplasia. Condylar aplasia is a rare condition and it means “failure of development” and can be unilateral or bilateral [2] Condylar aplasia without any additional facial malformations is exceedingly rare [3]. The incidence is estimated to be 1 in 5,600 births [2,3]. Congenital cases are very rare, only 20%, as reported by Converse and Tideman and Doddridge [4].

Developmental mandibular condylar anomalies are classified as hypoplasia, hyperplasia, aplasia and bifidity. Condylar aplasia means failure to develop and is mostly associated with craniofacial abnormalities. These are congenital or acquired [5]. Congenital causes are due to first and second branchial arch defects, such as Hemifacial microsomia, Treacher Collin syndrome, Hurler’s syndrome, Goldenhar syndrome, Proteus syndrome, Auriculocondylar syndrome, Hallerman-Streiff syndrome and Morquio syndrome [6]. But in these conditions, as a rule some soft tissue manifestation is also present along with condylar malformation. Krogstad reported a case with condylar hypoplasia and claimed it to have an acquired origin because the condition obviously started after the age of six [5]. In our case, besides aplasia of the mandibular condyle, there were no other commonly found syndromes or soft tissue manifestations. The rarity of the present case is highlighted by the fact that the condylar aplasia is not associated with any known syndrome. Acquired causes can be due to trauma (before age of two years) or infection (middle ear infection, rheumatoid arthritis) and parathyroid hormone associated protein deficiency which affect formation & differentiation of bone & cartilage leading to developmental arrest [3,7]. The present case had no history of infection or trauma and patients’ blood report revealed normal parathyroid hormone level eliminating the acquired origin.

TMJ ankylosis is the result of damage to joint structures that can lead to fibrosis. Causes are mainly trauma and infection (otitis media or hematogenous infection, rheumatoid arthritis). Congenital cases are very rare [8,9]. The present case is true congenital ankylosis of the TMJ, as no underlying etiological factors were identified in the history. The fact that restricted mouth opening was not noted until six months does not exclude a congenital defect as little mobility of the mandible in congenital ankylosis has been observed before the time of fusion of the cranial sutures [9].

Among the presentation of clinical features, retarded mental development and short stature with associated orthopedic deformity signify various defects of unknown basis. To our knowledge, no such case of above mentioned features has been reported in the literature previously.

It seems to be a new variant of TMJ ankylosis, resembling a syndrome rather than a single defect of the joint, which has not been classified. Detailed descriptions of clinical and radiographic findings are important to establish pathognomonic symptoms which may help in identifying true congenital TMJ ankylosis associated with other manifestations representing a new syndrome.

Various treatment modalities have been proposed for treating TMJ ankylosis and condylar aplasia and possibilities for influencing mandibular growth. This involves the help of oral surgeon, plastic surgeon and orthodontist. The timing and schedule of surgery is a question to be resolved. A costochondral rib graft can be used in order to establish an active growth center. Condylectomy and reconstruction with either an autogenous material, for example, sternoclavicular grafts, or alloplastic materials, are other treatment modalities. Early and aggressive surgery is the critical part of the treatment [10]. Unfortunately, in the present case, probably due to their poor socio-economic status and illiteracy, patient's family refused for any surgical intervention.

CONCLUSION

In conclusion, a rare case of congenital condylar aplasia of the left side and TMJ ankylosis on the right side, with associated orthopedic abnormality of unknown etiology has been reported. Cases of nonsyndromic condylar aplasia and congenital TMJ ankylosis are extremely rare and not many case reports have been published so far. Our case is a significant contribution to the literature as no case with condylar aplasia and ankylosis with associated orthopedic abnormality has been reported so far. Further studies need to be done to discover whether this case is a new unidentified syndrome not yet recognized. Early detection and appropriate treatment should be done to provide psychological & esthetic benefits to these patients.

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