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Case Report

NON SYNDROMIC 'APLASIA' OF MANDIBULAR CONDYLE

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ABSTRACT

Mandibular condyle aplasia is an anomaly which usually manifests in association with various syndromes. When not seen in conjunction with any other developmental anomalies, it is an extremely rare condition. Only a few cases of Non syndromic condylar aplasia have been reported in literature till date. Proper diagnosis along with differentiation from the syndromic cases is important as treatment plan and prognosis for each varies. The aim of this article is to present a peculiar type of mandibular asymmetry with non syndromic condylar aplasia; frequently misdiagnosed as Hemi facial microsomia or Treacher Collins syndrome (syndromic aplasia).

Keywords: Condylar Aplasia, Mandibular Condylar Aplasia, Non Syndromic, Condylar Agenesis, Anomaly.

INTRODUCTION

Mandibular condyle aplasia is a term used to describe total absence of the condyle. Earlier the term 'agenesis' was used, signifying absence of an organ but it has now been replaced by the term aplasia i.e. lack of development of a tissue, as the condylar cartilage is considered a tissue rather than an organ¹. Abnormality during the development and growth of TMJ may lead to condylar aplasia. The incidence is estimated to be 1 in 5,600 births when presenting as facial manifestations of syndromes such as Hemifacial microsomia, Treacher Collins syndrome and Goldenhar's syndrome¹, but is extremely uncommon when not associated with any syndrome. Here we aim to present a rare case of non- syndromic condylar aplasia laying emphasis on differentiation between non syndromic and syndromic aplasia as this may significantly affect the treatment planning and prognosis.

CASE HISTORY

A 30 year old woman reported at our centre with the chief complaint of impaired facial aesthetics and chin deviation towards the right side. The patient was in good general health and did not give any history of auricular infection or trauma to the craniofacial region. Family history was unremarkable too. History of presenting illness revealed that the deviation of chin to the right side of face first became apparent during early childhood and progressively worsened thereafter.

Clinically there was marked deviation of the mandibular midline towards the right side of the facial midline both in the mandibular rest position as well as in occlusion. The asymmetry worsened with maximum mouth opening. The range of mandibular movements was however, normal despite the fact that the mandibular condyle was not palpable on the right side. The lower incisors were inclined to the left in such a manner as to attempt to partially offset the skeletal discrepancy.

Orthopantomogram (OPG) revealed total absence of the condylar head and the neck on the right side. The mandibular angle was placed abnormally close to the base of the skull on the affected side; also the head of the condyle on the left side appeared more rounded than normal. Lateral cephalogram showed mandibular deficiency with its lower border sloping steeply downwards to end in a retruded bony chin. A provisional diagnosis of unilateral condylar aplasia along with anatomical facial asymmetry was established on clinical and radiographic findings. (Fig. 1, 2)

Differential Diagnosis:

The patient was then further examined for the presence of any other syndromic features which are usually associated with condylar aplasia, but no syndromic features were apparent. HFM was ruled out as no deficit in the neuromuscular pattern or in the soft tissues was present. Other types of acquired mandibular hypoplasia, such as rheumatoid arthritis and Parry-

Romberg syndrome were also ruled out as no definite clinical history or radiological features were present in our patient. In view of above findings this case was suspected of having a peculiar type of non syndromic condylar aplasia rather than a syndromic one. Though the definitive diagnosis required karyotyping reports, the patient was unfortunately not ready for the same.

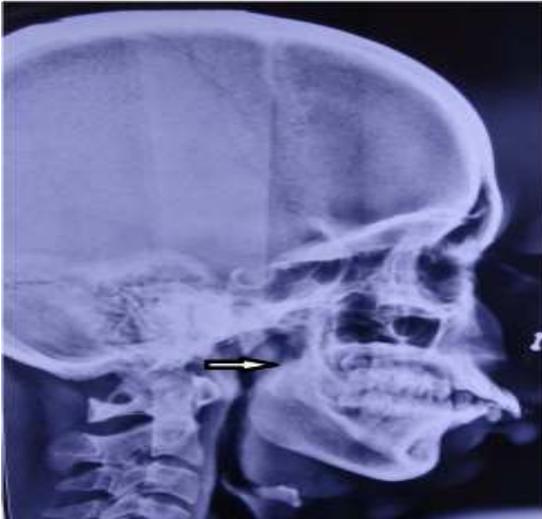


Figure 1: Lateral Cephalogram showing absence of right mandibular condyle.



Figure 2: OPG showing dental and skeletal discrepancies.

Treatment:

Limited growth potential (due to the patient age) ruled out the possibility of growth modification treatment; as there was no neurophysiologic deficit, it indicated a good prognosis. Patient was advised CT scan for proper surgical planning; but the patient refused any further investigation or any surgical intervention. Enhancement of facial aesthetics without any

surgical procedure was her primary and rather, only concern. Hence the patient was referred to Department of Orthodontics for further management. The patient was advised to follow her descendants in order to report of early similar manifestations in her family.

DISCUSSION

The temporomandibular joint (TMJ) is one of the most unique joints of the human body which starts developing at 7th intrauterine week. The initial functions of mouth opening movements start appearing by the twentieth week during foetal stage, but the development process continues until the twelfth year of life². Growth disturbances in the development of the mandibular condyle may occur in utero; late in the first trimester or may be of acquired nature resulting in disorders such as aplasia or hypoplasia of the mandibular condyle. Congenital (primary) condylar hypoplasia is generally associated with some systemic condition that originate from first and second branchial arches, such as Treacher Collins syndrome, Goldenhar syndrome, Hemifacial microsomia, Hurler’s syndrome, Proteus syndrome, Morquio syndrome and Auriculocondylar syndrome³. Acquired (secondary) condylar aplasia or hypoplasia may occur due to mechanical trauma during active growth. Other causes may include inflammation in the TMJ area, rheumatoid arthritis and radiotherapy⁴. Parathyroid hormone-related protein deficiencies also affect bone formation and chondrocyte differentiation, which consequently affects the condyle formation. Recent reports have shown that various extracellular matrix proteins, such as transforming growth factor- β (TGF- β), play important roles affecting Meckel’s cartilage for normal mandibular development⁵. Whereas some authors affirm that mandibular condyle deficiency can occur with no defined aetiology⁶.

The clinical presentation of condylar aplasia in syndromic cases is governed by the traits of the syndromes it is associated with. Conversely, in non syndromic condylar aplasia there will not be associated developmental defects and the clinical manifestations will include facial asymmetry, deviation of mandible towards the affected side and accentuation of the antegonial notch. Additionally the mandibular angle is usually placed abnormally close to the base of the skull on the affected side; shift of dental and skeletal midline and malocclusion is seen. However the function of the mandible is not affected, as was seen in our case. No apparent soft-tissue involvement was present, the external ear was well-formed, and the facial musculature well developed. Although the chin was deviated to the affected side, the typical flatness of the gonial angle was not present (fig 2). In view of above findings the patient was suspected of having non syndromic condylar aplasia rather than a syndromic one. Though the definitive diagnosis required karyotyping reports, the patient was unfortunately not ready for the same. Through this rare case report, we have summarized the most important clinical, radiologic and possible genetic differences (Tables I)^{7, 8} to aid in differential diagnosis.

As the prognosis for syndromic and non-syndromic condylar aplasia would vary considerably, the collaboration between not only the surgeons and orthodontist, but also the geneticists

should be considered in order to establish the differential diagnosis⁸. This will allow appropriate counselling and guidance, including reconstructive surgery and orthodontia. The radiographic evaluation includes OPG, postero-anterior view of skull, CT (computed tomography) scan; MRI (magnetic resonance imaging) and Cone beam CT. The 3D CT examination enables accurate surgical planning and provides quantitative information from skeletal and muscular parameters.

Various treatment modalities have been proposed for the treatment of condylar aplasia, the timing of treatment and possibility of influencing mandibular growth has been the topic of numerous clinical and experimental studies. Early clinical procedures could prevent facial asymmetries as well as other functional disorders. The surgical treatment methods include restoring the mandibular dental base to its correct relationship with the maxillary dental base by osteotomy and bone-grafting operations in the mixed dentition stage so that the permanent dentition can take up a satisfactory occlusion. Alternatively orthodontic treatment may be carried out in the mixed or permanent dentition, which would be followed by corrective surgery in the form of masking operations such as epithelial inlays or onlay bone grafts⁹. The severity of the damage to the head of the condyle, age of the patient, the potential for facial growth of the patient are some of the factors to be considered when making this decision.

CONCLUSION

Patients with non syndromic mandibular aplasia are a rare subgroup; in which the condition does not seem to be progressive. Integration of surgical treatment with orthodontic treatment is required for a good prognosis and predictable outcome. The timing of the treatment is, however, very important and should start before the pubertal growth spurt, but depends entirely on the age when the patient first reports.

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Table 1: Differential diagnosis of Syndromic /non syndromic- condylar aplasia

	Syndromic-condylar aplasia	Non-syndromic condylar aplasia
History	Generally diagnosed at birth	Usually not diagnosed at birth, history of trauma unusual
Clinical presentation (as identified)	Soft-tissue defects(may be very mild to severe)	No soft-tissue defects
	Masseter muscle hypoplasia	Well developed facial muscles
	Ear defects, pre-auricular tags	Normal external and middle ears
	Facial nerve deficit	No nerve deficit
	Deviation of the chin on the affected side, associated with flatness on the affected cheek	Deviation of the chin on the affected side, associated with fullness on the affected cheek
	Mild deviation to the affected side during opening	Significant deviation to the affected side during opening
Radiographic (as identified)	Hypoplasia of the ramus and condyle and coronoid processes up to absence of the condyle and temporal fossa	Hypoplasia of the ramus and condyle and coronoid processes, the temporal fossa is always present
Karyotyping (as suspected)	OMIM (Online Mendelian Inheritance in Man) 164210: Hemifacial microsomia, OMIM 602483: Auriculo-Condylar Syndrome identified.7	Normal genetic makeup

Adapted from Meazzini MC et al⁸

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