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CASE REPORT

A RARE CASE OF PIERRE ROBIN SYNDROME WITH UNILATERAL TEMPOROMANDIBULAR JOINT ANKYLOSIS AND OBSTRUCTIVE SLEEP APNOEA

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ABSTRACT

Pierre robin syndrome with unilateral ankylosis and obstructive sleep apnoea is uncommon. Pierre robin syndrome is characterised by a triad of micrognathia,obstructive sleep apnoea and glossoptosis. Child usually presents with hypoplastic mandible which displaces the tongue posteriorly and causes difficulty in breathing. Pierre robin syndrome is not a syndrome as such rather it is a sequence of a collection of disorders, with an abnormality presenting in other events to occur. It is usually associated with U- shaped cleft palate. It may also be associated with several other craniofacial anomalies like velocardiofacial and stickler syndrome. We hereby present a case of 18-year-old female who presented with complaints of inability to open mouth and difficulty to eat since childhood. The orthopontogram showed fusion of the left temporomandibular joint and plain computed tomography showed severe sclerosis and expansion of the left condylar process with the left temporomandibular joint's glenoid fossa flattening with irregularities and ankylosis/fusion with loss of symmetry in the skull base and smaller left hemimandible with misalignment of dentition and a pushed back tongue with a narrow airway.

INTRODUCTION

Pierre robin syndrome is a cluster of clinical features that occur consistently hand in hand, varying in degree and are characteristic of a medical condition. It can be isolated or syndromic. There is no gender predilection with a prevalence of 1 in 8,500- 14,000 births. Mutations in chromosome 2, 4, 11, 17 are associated with non-syndromic type. There is evidence of SOX9 and KCNJ2 mutations effecting the facial structural development along with cartilage development leading to Pierre robin syndrome. Stickler syndrome which is the most commonly associated with Pierre Robin syndrome is associated with COL gene mutation affecting collagen formation resulting in retinal detachments, flat midface, cataracts, epicanthal folds, joint hypermobility and sensorineural hearing loss in addition to typical features of Pierre robin syndrome. Other associations are trisomy 11q syndrome, trisomy 18 syndrome, hypochondroplasia, rheumatoid arthropathy, mobius syndrome and CHARGE associations.

CASE REPORT

An 18-year-old female patient presented with complaints of inability to open mouth and inability to eat since childhood (18 years). Complaints of snoring was given by her parents. Patient had history of trauma to mandible 14 years ago. On clinical examination temporomandibular joint movement was restricted and tenderness elicited in the left pre-auricular region. Grossly hypoplastic mandible was noted with facial asymmetry. The patient was subjected to orthopontogram and computed tomography for further evaluation.

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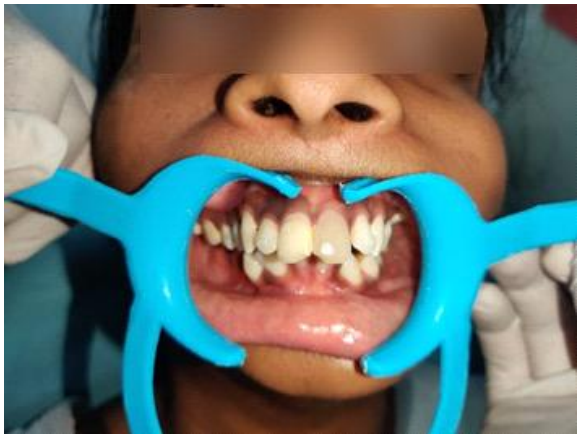
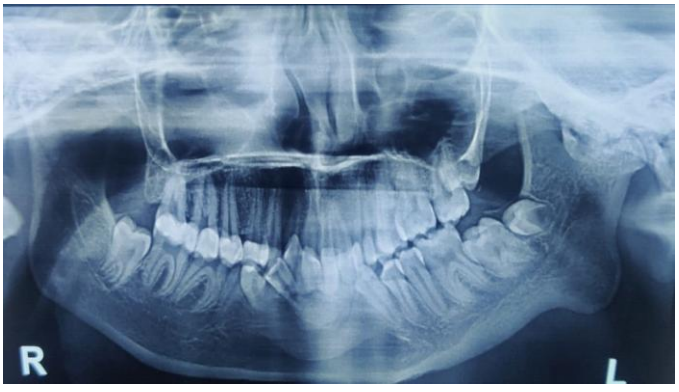


IMAGING FINDINGS

Orthopontogram: An orthopontogram was done which showed the fusion of left temporomandibular joint but clear view of the temporomandibular joint with associated structures could not be evaluated. Thus, computed tomography scan was advised for detailed visualization of the joint.



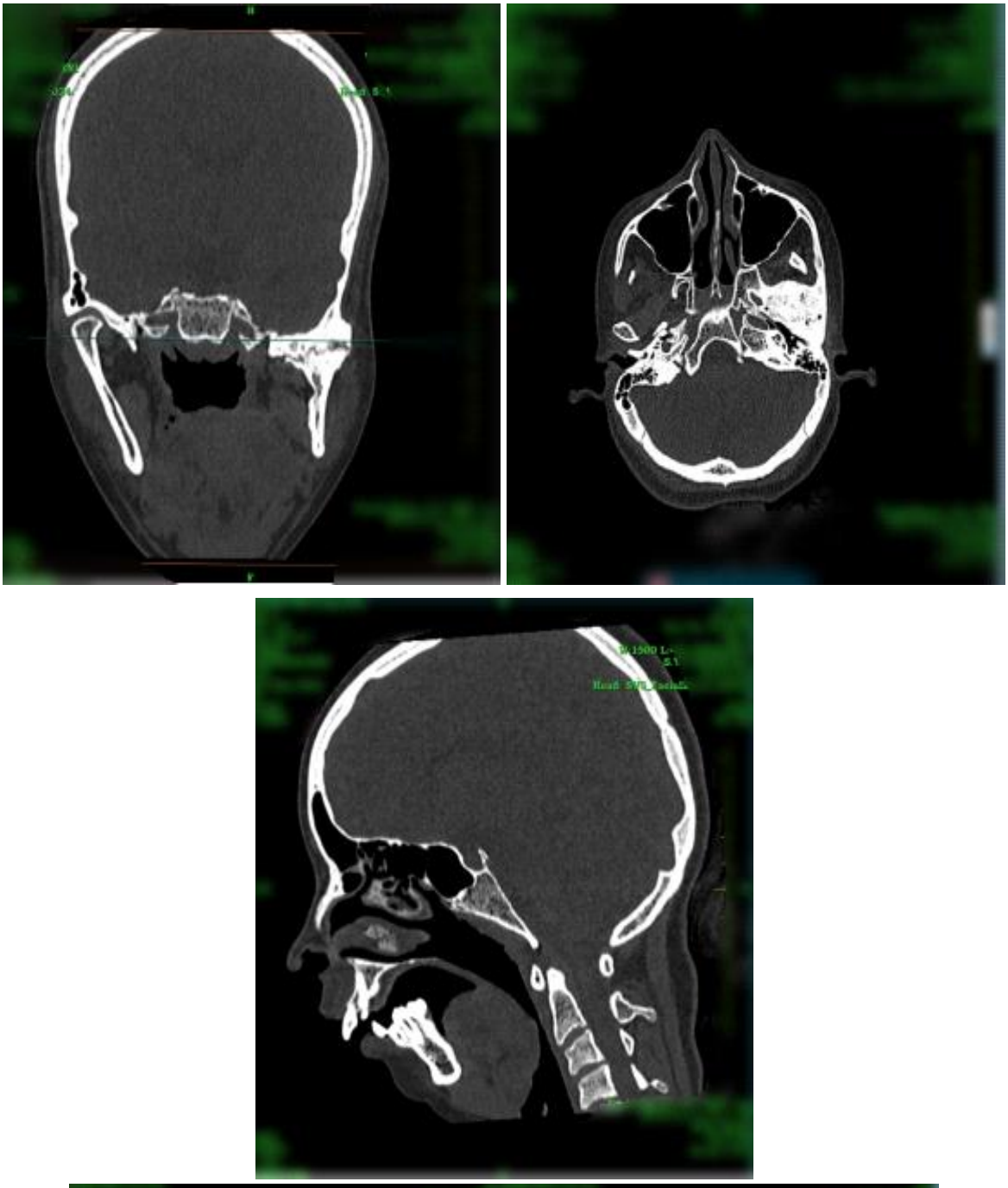
Plain radiography: Lateral plain radiography showed small sized under developed mandible with narrowing of the oropharyngeal airway.

Plain computed tomography: Outline of the normal condylar process is lost with severe sclerosis and expansion of the condylar process of left hemimandible. The condylar process measures approximately 2.5 x 2.5 cm in AP and TR dimensions. Glenoid fossa of the left temporomandibular joint shows loss of normal concavity and appears flattened. Opposing surfaces of glenoid fossa and expanded condylar process show irregularity with areas of ankylosis/ fusion. Additionally, loss of symmetry noted in the base of the skull, anterior and middle cranial fossa (likely due to a long-standing cause). Left hemimandible is smaller in size when compared to the right side with improper alignment of superior and inferior dentition. The tongue is displaced posteriorly abutting the soft palate causing moderate narrowing of the oropharyngeal airway.

DISCUSSION

Pierre Robin Syndrome (PRS) presents as a rare congenital disorder marked by micrognathia, glossoptosis, and obstructive sleep apnoea. While PRS primarily influences mandibular and airway development, its correlation with temporomandibular joint (TMJ) ankylosis is infrequent. Moreover, the emergence of obstructive sleep apnea (OSA) in individuals with PRS and TMJ ankylosis poses additional complexities in airway management. The identification of TMJ ankylosis and its impact on airway obstruction through radiological imaging is pivotal for accurate diagnosis and treatment planning.

Exploration of TMJ ankylosis prevalence in PRS remains limited, with scant literature addressing this connection. Our case study highlights an unusual manifestation of PRS accompanied by unilateral TMJ ankylosis, exacerbating airway compromise and leading to OSA.



RIGHT AND LEFT TEMPOROMANDIBULAR JOINTS

Radiological investigations play a crucial role in diagnosing PRS with TMJ ankylosis and OSA, offering valuable insights into the extent of craniofacial anomalies and their effects on airway patency. Computed tomography (CT) scans provide detailed anatomical visualization, highlighting micrognathia, retrognathia, and the degree of TMJ ankylosis. Magnetic resonance imaging (MRI) further aids in assessing soft tissue structures, including the tongue position and airway dimensions, contributing to a comprehensive evaluation of airway obstruction severity. In cases of PRS with TMJ ankylosis, characteristic radiological findings such as mandibular hypoplasia, abnormal TMJ morphology, and restricted mandibular movement on imaging studies are noted. Additionally, identification of associated complications such as airway compression or displacement secondary to TMJ ankylosis is crucial for guiding treatment decisions.

The management of PRS along with TMJ ankylosis and OSA demands a multidisciplinary approach involving radiologists, otolaryngologists, maxillofacial surgeons, orthodontists, and sleep specialists. Surgical interventions like distraction mandibular osteogenesis for micrognathia correction and TMJ ankylosis release, alongside continuous positive airway pressure (CPAP) therapy for OSA, may be contemplated to mitigate airway obstruction and enhance respiratory function. CT and MRI assessments aid in surgical planning for TMJ ankylosis release and mandibular distraction osteogenesis to address micrognathia and improve airway patency.

CONCLUSION

In conclusion, radiological evaluation plays a central role in diagnosing and managing Pierre Robin Syndrome with unilateral temporomandibular joint ankylosis and obstructive sleep apnoea. Diagnosis requires a comprehensive assessment involving clinical, radiographic, and polysomnographic evaluations, while treatment demands a collaborative approach integrating surgical interventions and continuous positive airway pressure (CPAP) therapy. Close collaboration between radiologists, otolaryngologists, maxillofacial surgeons, and other healthcare providers is essential for comprehensive patient care and treatment optimization.

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