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Original Article

Popliteal Petrygium Syndrome: A Rare Case Report with Hypodontia

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ABSTRACT:

Popliteal Petrygium Syndrome is a rare genetic condition. This syndrome is responsible for defects in craniofacial, digital, extremities and genital region. It consists of popliteal petrygium, clefting of lip and palate, labial pits, syngnathia, syndactyly, oligodontia, fusion of eyelid margins and genitourinary abnormalities. Overlapping features of this syndrome with other craniofacial disorders make it a diagnostic challenge; hence awareness concerning this is vital for its prompt diagnosis and management.

Key words- craniofacial anomaly, popliteal petrygium, hypodontia, cleft lip, cleft palate, labial pits, syndactyly.

Key message- Popliteal Petrygium Syndrome is an infrequent craniofacial anomaly affecting extremities, maxillofacial structures and genitourinary region. Our aim is to create awareness among dental and medical professionals regarding this condition for prompt diagnosis and treatment of these patients.

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INTRODUCTION

Popliteal Petrygium Syndrome (PPS) is a rare genetic disorder affecting 1in every 300000 live births.[1] This syndrome is a complex of malformations in craniofacial region, extremities and genitalia. It was first reported in literature in 1869 by Trelat and was termed by Gorlin in 1968.[2]

Craniofacial anomalies are associated in majority of reported cases. These findings include cleft lip, cleft palate, presence of labial pits with salivary ducts, syngnathia where maxilla and mandible are connected by band of tissue, missing teeth and presence of ankyloblepharon filiforme adnatum in which eyelid margins are fused by minute threads of abnormal tissue.

Syndactyly of digits and presence of popliteal webs are pathogomonic features of this disorder. Crease of skin over the haullx of nail is also seen in 30% of the cases.

Genitourinary region is affected in both females and males. Hypoplasia of labia majora, vagina and uterus is a frequent finding in females whereas cryptorichidism with absent or bifid scrotum is seen in males.

Other allied deformities include bifid rib, spina bifida, small sternum and clubfoot. Inspite of considerable developmental anomalies, IQ and growth of these individuals is observed to be at parity with their healthier counterparts.[3]

This case report presents general and orofacial findings noted in a Popliteal Petrygium Syndrome patient.

CASE REPORT

A 16 year old male patient reported in the department with chief complaint of forward placed upper front teeth. Patient gave history of removal of popliteal webs followed by repair of cleft palate at the age of 10 months. He gave no history of any of his family members having similar deformities. On extraoral examination anterior maxilla was proclined [fig.1]. Pits were seen in lower lip with presence of mucous.[fig.2] Intraorally high arched palate with cleft

repair with missing bilateral maxillary lateral incisors was seen[fig.3],uvula was short [fig.4]and tissue band joining tongue to mandible was present leading to ankyloglossia [fig.5]. Syndactyly was seen on 3rd and 4th digit of left hand [fig.6]. Orthopantomogram showed absence of mandibular 1st premolar along with maxillary lateral incisors [fig.7].

On the basis of clinical and radiological findings patient was diagnosed as a case of popliteal petrygium syndrome and was planned for dental correction through orthodontic treatment.

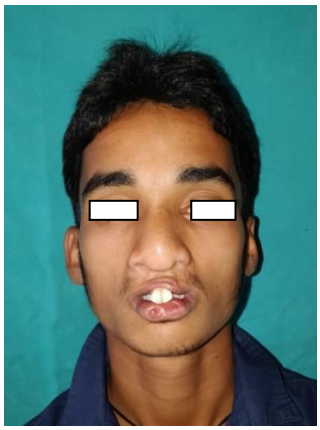

Figure 1: Extra oral photograph showing forward placed premaxilla.

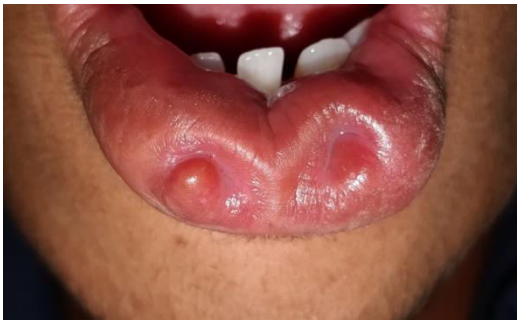

Figure 2: Paramedian pits on lower lip

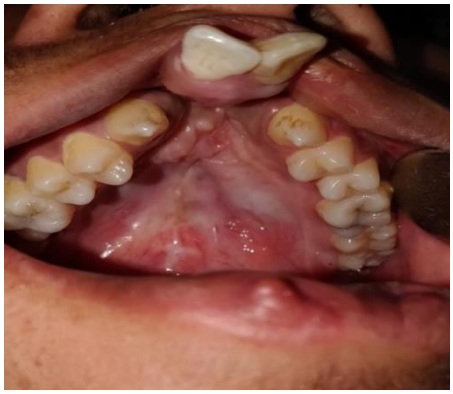

Figure 3: Bilateral cleft with missing bilateral lateral incisors

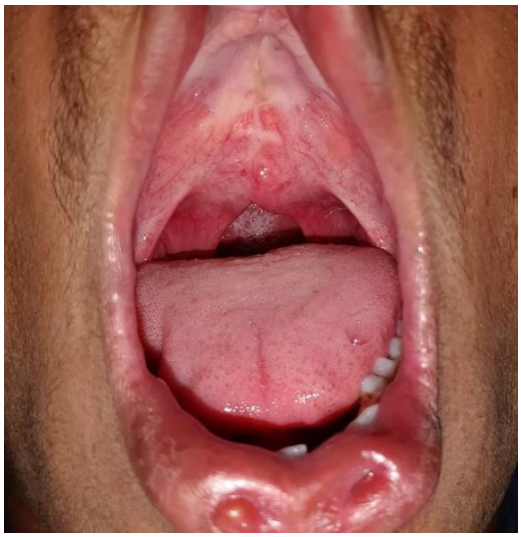

Figure 4: Intraoral image showing shortened Uvula.

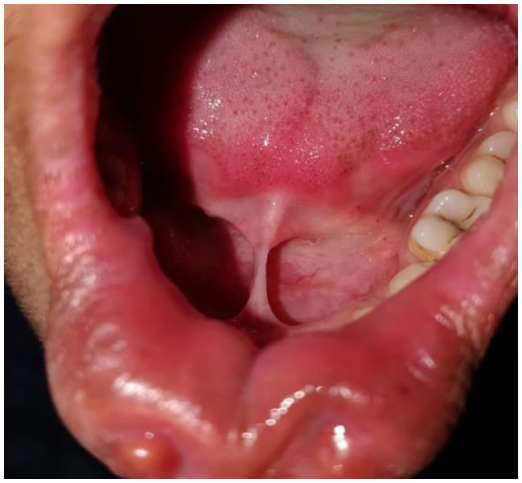

Figure 5: Intraoral image showing Ankyloglossia

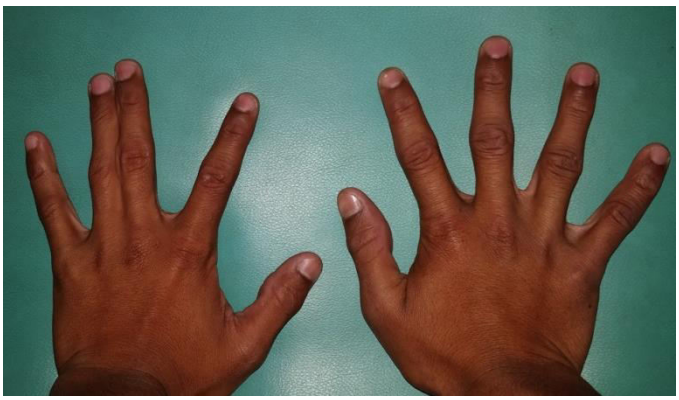

Figure 6: Syndactyly of 3rd and 4th finger of left hand.

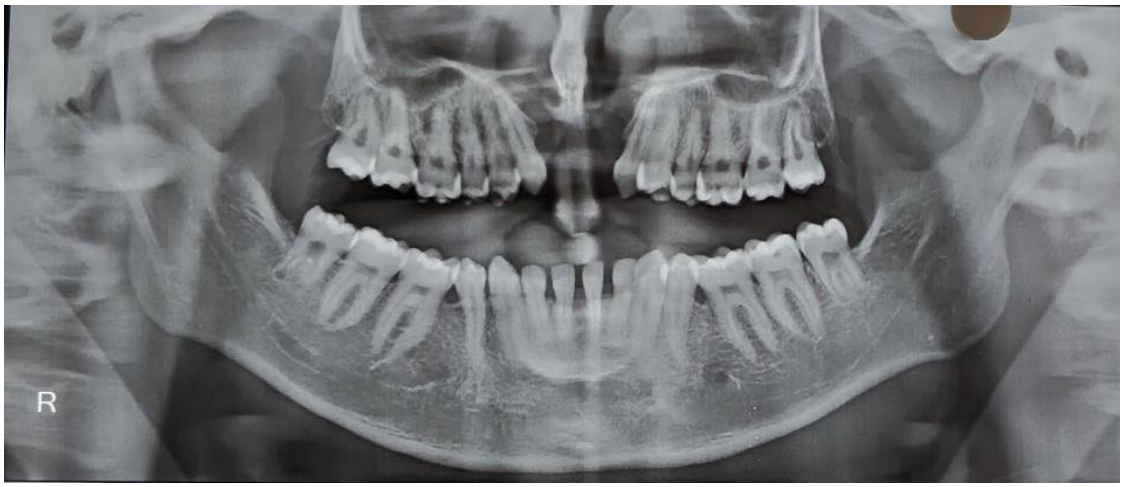

Figure 7: Orthopantomogram showing absent mandibular 1st premolar and maxillary lateral incisors.

DISCUSSION

Popliteal petrygium syndrome is an unusual condition. Its etiology is ambiguous though inheritance following autosomal dominant pattern is most widely accepted,[3] with few cases of autosomal recessive pattern also being reported.[4] Recently interferon regulatory factor-6 gene (IRF6) on 1q32.2 has been identified as a gene accountable for PPS and van der woude.[5]

PPS should be carefully differentiated from analogous conditions having craniofacial anomalies and limb defects. Isolated cases of cleft lip & palate and van der woude have striking similarities with PPS but there is absence of popliteal petrygium in both the above mentioned conditions. Popliteal Pterygium is a band of connective tissue which sometimes involves the popliteal artery and the peroneal nerve and hampers the free movement of legs, leading to morbidity.[6]

In previously documented cases ankyloblepharon filiforme adnatum and genitourinary anomaly were reported as a distinctive trait which were absent in our present case, however hypodontia which was not reported considerably in earlier cases was a noteworthy finding in this case.

CONCLUSION

Popliteal petrygium syndrome presents as a diagnostic challenge. Early detection through prenatal imaging and genetic counseling can help the patients early though its success is still questionable. As PPS affects considerable portion of orofacial region dental surgeons play a vital role in diagnosing and restoring function and esthetic in such patients.

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