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Case Report

The Robin Anomalad: A case report of genetic disorder with rare oral manifestations

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ABSTRACT

This case report describes a case of non-syndromic Pierre Robin Syndrome (ns-PRS) in a 17-year-old male patient with unique oral characteristics, adding to the body of knowledge on unusual PRS presentations. There was enough literature to support all of the unusual findings reported in this instance leading to a precise diagnosis of Grade I ns-PRS. Some individuals with PRS might show all the typical signs, whereas others may appear with only rare signs. Such kind of more rare manifestations should be recognized and should be reported further to add credence to the literature on unusual manifestations of PRS.

1. Introduction

In 1923, Pierre Robin identified a trifecta of clinical indicators in a group of newborns which includes: micrognathia, glossoptosis, and upper airway obstruction. He described a common relationship of PRS with palatal cleft in 1934 and named this clinical condition as Pierre Robin syndrome (PRS). Breugem and Mink Van Der Molen pointed out in 2009 that Pierre Robin's triad is clinically manifested as an effective series of pathogenic occurrences.[1] PRS can occur solely known as isolated or non-syndromic PRS (ns-PRS) and can occur in association with other syndromes known as Syndromic PRS (s-PRS).[2] According to epidemiological data, PRS is a rare condition with a variable incidence ranging from 1 to 8,500 to 1 to 30,000 neonates in India.[3] Because of the numerous comorbidities associated with PRS and their varying manifestations, a precocious diagnosis of PRS is required to analyze the patient's growth alterations and to choose the timing and mode of intervention.

2. Case Presentation

A 17-year-old, male patient, reported to the Department of Oral Medicine and Radiology of Swargiya Dadasaheb Kalmegh Smruti Dental College and Hospital, Nagpur with the chief complaint of missing teeth in the lower front region of the jaw and loosening of teeth in the same region since, 28^{th} May 2023.

Patient give history that, on exfoliation of deciduous teeth no permanent teeth erupted in lower front region of jaw due to which spacing was present in same region and there was no history of any respiratory distress or altered speech.

On extra oral examination, his facial profile was convex and face was bird like (Figure 1). There was marked

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micrognathia with mandible and downward slant with right eye (Figure 2). Intra oral examination revealed high arched palate (Figure 3) and bifid tongue with ankyloglossia (Figure 4). On hard tissue examination, teeth 31, 33, 41, 43 and 24 were missing (Figure 5).

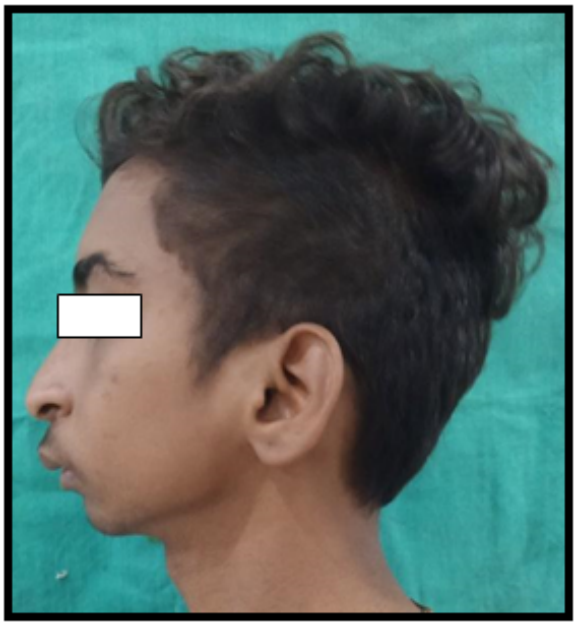

Figure 1: Left side showing convex facial profile and bird face

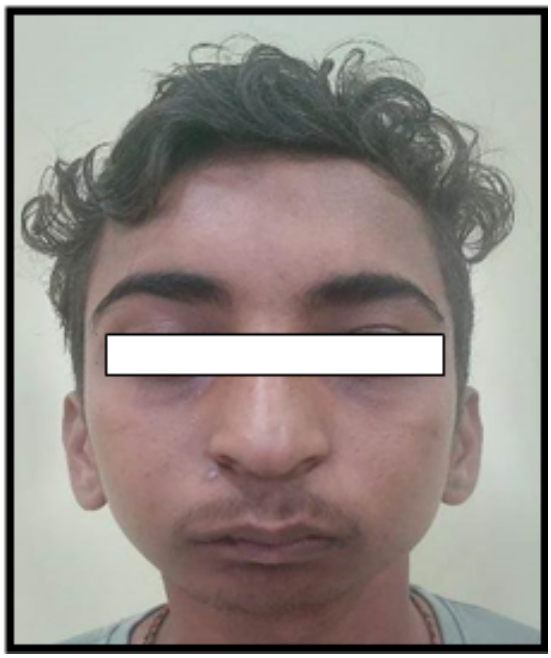

Figure 2: Straight profile of patient showing right eye with downward slant

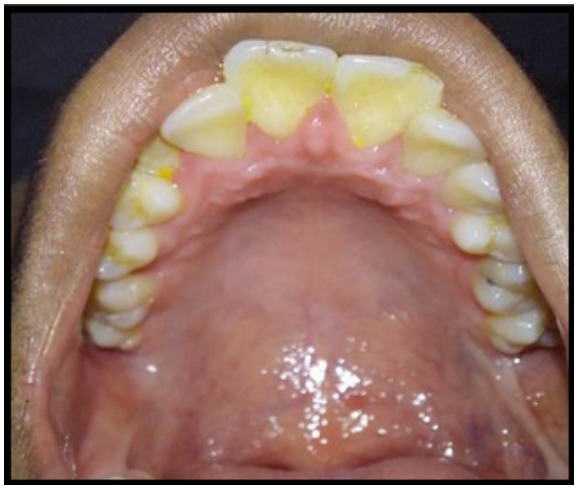

Figure 3: High arched palate

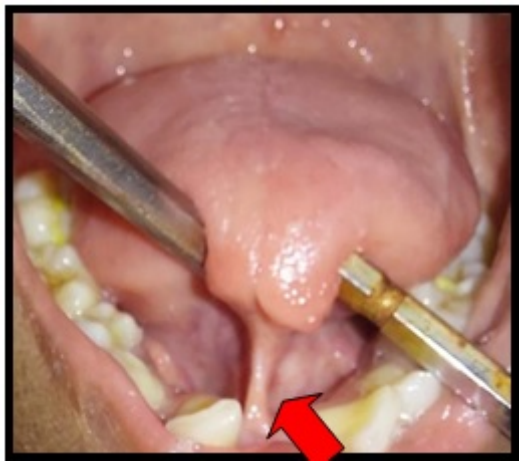

Figure 4: Bifid tongue with ankyloglossia

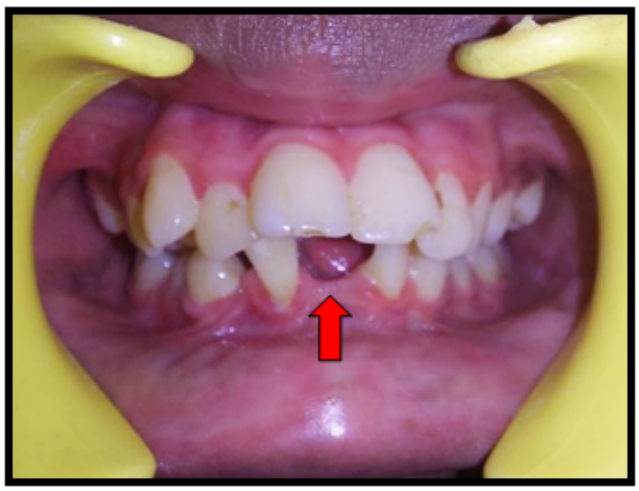

Figure 5: Missing teeth 31, 33, 41 and 43

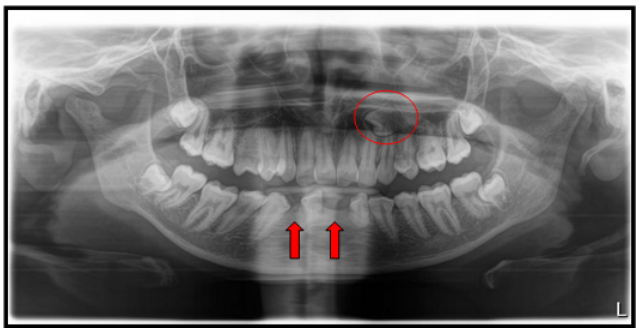

Figure 6: Orthopantomogram showing hypodontia with teeth 31, 33, 41, 43 andhorizontal impaction with tooth 24

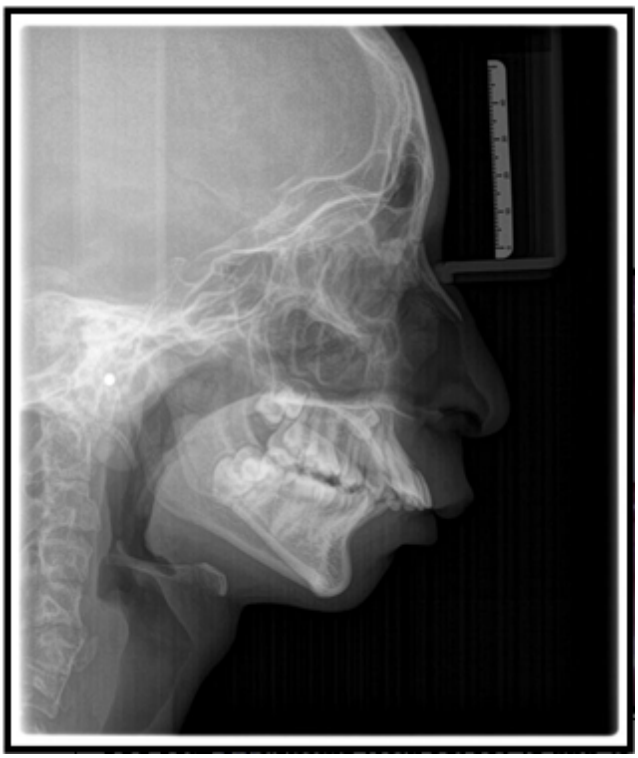

Figure 7: Lateral cephalogram showing micrognathicmandible and prognathic maxilla, proclination with upper and lower anteriors,increased overjet, end on molar relation, class II skeletal base, average airspace, acute nasolabial angle and deep mentolabial angle

There was grade I mobility with teeth 32 and 42. Thus, the patient was advised of necessary investigations i.e. the Orthopantomogram (OPG) and Lateral Cephalogram (Lat Ceph). OPG revealed hypodontia with teeth 31, 33, 41, 43, and horizontal impaction with tooth 24 (Figure 6). Lat Ceph revealed micrognathic mandible and prognathic maxilla, proclination with upper and lower anteriors, increased overjet, end on molar relation, class II skeletal base, average air space, acute nasolabial angle and deep mentolabial angle (Figure 7). Cone Beam Computed Tomography (CBCT)

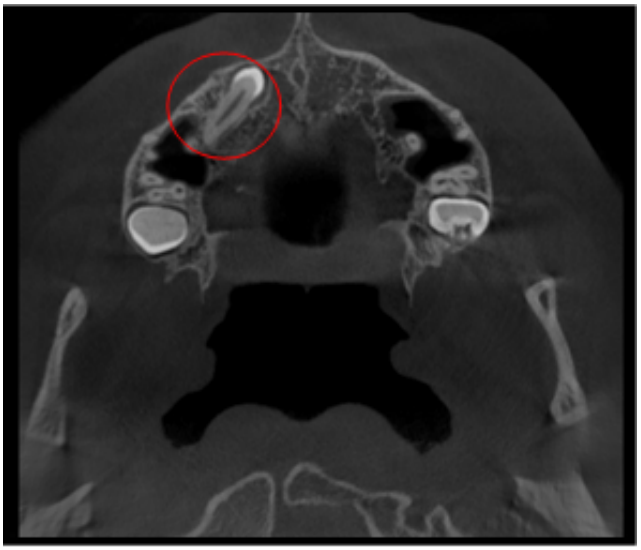

Figure 8: Axial view of Cone Beam Computed Tomographyshowing horizontal impaction with 24

was advised for proper localization of tooth 24 (Figure 8). No other abnormality was detected in CBCT. Finally, based on the clinical and radiographic examination a conclusive diagnosis of Localized periodontitis with teeth 32 and 42; Hypodontia with teeth 31, 33, 41, and 43; Horizontal impaction with tooth 24 and Grade I Non-Syndromic Pierre Robin Syndrome was given. The final grading for this case was given according to Annie Cole, Patricia Lynch, and Rona Siator.[4] The patient was counseled regarding necessary treatment and has been recalled every 6 months for follow-up.

3. Discussion

PRS is characterized by a triad which includes micrognathia, cleft palate, and glossoptosis, which can lead to respiratory obstruction. There are two types of PRS: non-syndromic PRS (nsPRS) and PRS linked with other syndromes (s-PRS).[5] In 2009, Benko et al. used genetic linkage analysis to locate an autosomal dominant and highly penetrant PRS locus to chromosome 17q24.3-25.1.[6] As SOX9, KCNJ2, KCNJ16, and MAP2K6 genes are engaged in mandibular development molecular pathways, their changes may be linked to the isolated PRS etiology.[7] Various syndromes associated with PRS are Stickler syndrome, Velocardiofacial syndrome, Treacher Collins syndrome, Campomelic dysplasia, Marshall syndrome, Nager syndrome, Miller syndrome, Kabuki syndrome, Catel-Manske syndrome, Congenital myotonic dystrophy, Carey-Fineman-Ziter syndrome, Fetal alcohol syndrome, Maternal diabetes, Spondyloepiphyseal dysplasia congenita, Hemifacial microsomia, Glass syndrome and Mandibulofacial dystosis.[1]

In PRS patients most frequent clinical findings are micrognathia (91.7%), Glossoptosis (70-85%), and cleft palate (14-91%). Other uncommon findings include macroglossia and ankyloglossia (10-15%), nasal deformities, dental and philtral malformations (33%), auricular malformations and otitis media (75-80%), gastroesophageal reflux with esophagitis (10-15%) and laryngomalacia (10-15%).[8] Apart from these findings, Han Hyun Ho et al. in 2014, reported a patient with PRS with a bifid tongue, as present in this case.[9] Merza Ahmed Maki and Salih Haydar Munir reported a patient with PRS who had a tri-lobed tongue in 2023.[10] In 2013, Ophir Klein et al. reported that the prevalence of hypodontia in PRS is 50%.[11] According to a systematic review and meta-analysis by G.S. Antonarakis, P.K. Palaska, and S. Suri in 2017, tooth agenesis in ns-PRS patients is mostly seen with upper and lower anteriors and premolars, as seen in this case.[12] Murgae Kariuki et al. revealed in a 2013 retrospective review that 8 of 50 PRS patients presented without a cleft palate, as seen in this case.[13]

To manage the complex features associated with PRS a multidisciplinary approach is essential. Patients who are asymptomatic and are concerned only regarding aesthetics should be properly counseled for the same and should be recalled every 6 months for follow-up to investigate any changes in growth. In a symptomatic patient, the severity of airway impairment is prioritized, followed by the level of feeding issues. Infants with severe micrognathia may suffer from severe respiratory distress in such circumstances, surgical intervention is required.

4. Conclusion

PRS comprises a heterogeneous group of patients. Some may present with all common findings, whereas others may appear with only rare signs. There was enough evidence in the literature to back up all of the unusual symptoms that were recorded in this instance and to establish a definitive diagnosis of Grade I ns-PRS. Such kind of more rare manifestations should also be reported further, to add and to credence to the literature on unusual manifestations of PRS.

5. Source of Funding

None

6. Conflict of Interest

None

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