www.sada.co.za / SADJ Vol. 80 No.9
https://doi.org/10.17159/sadj.v80i09.21352

Multidisciplinary Management is crucial in Pierre-Robin syndrome: A Case Report

SADJ OCTOBER 2025, Vol. 80 No.9 P498-P502
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ABSTRACT

Background

Pierre-Robin syndrome (PRS) is a congenital disorder in which affected individuals present with micro/retrognathia, difficulty feeding, cleft lip/palate, glossoptosis and difficulty breathing. PRS affects between 1 in 8 500 to 1 in 20 000 births. The nature of the congenital defects results in patients presenting with numerous complications including malnutrition, aspiration pneumonia, recurrent local infections, dental abnormalities, respiratory problems as well as aesthetic and functional defects.

Case report

A 3-year-old female patient who was diagnosed with PRS at 6-months of age was referred to the Department of Operative Dentistry by the Department of Orthodontics with a main complaint of dental pain as well as the existing cleft palate.

Discussion

The clinical needs for the alleviation of pain, surgical closure of clefts to facilitate nutritional intake, surgical intervention to prevent respiratory obstruction as well as the dental abnormalities which need to be managed and restored are all highlighted in this case presentation. Furthermore, the need for lifelong therapy, encouragement, monitoring and nurture within a supportive, stimulating environment are essential to improve the overall quality of life for patients with PRS.

Conclusion

The objective of documenting this case report is to demonstrate the diverse scope and nature of the health care team responsible for providing adequate treatment, rehabilitation and surgical intervention in the patient who has PRS. The need for holistic management through multidisciplinary collaboration is emphasised for patient and practitioner education.

Keywords

Pierre-Robin syndrome, congenital, micrognathia, glossoptosis, enamel hypoplasia, rampant decay, malocclusion, airway obstruction.

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Authors contributions

1. Dr J Bull – Consulting clinician who retrieved the case information, obtained photographs, contributed to conceptualisation, prepared treatment plan and assisted with the writing and editing (30%).
2. Dr BK Bunn - Conceptualisation, writing, editing and submission (30%).
3. Dr P Gwengu – Assistance with development of treatment planning, writing and editing (30%).
4. Dr CS Lebaka – Surgical closure of palatal cleft, patient consultation and editing (10%).

INTRODUCTION

Pierre-Robin Syndrome (PRS) is a congenital disorder with an incidence of between 1 in 8 500 to 1 in 20 000 births.[1] Individuals afflicted with this disorder characteristically present with retro-or micrognathia, cleft palate, glossoptosis, difficulty feeding and recurrent respiratory infections.[1] This may often lead to malnutrition and underdevelopment. Cleft palate increases the risk of food aspiration which is frequently complicated by pneumonia.[1, 2]

Embryologically, defective caudal hind brain development has been observed in patients with PRS. Recent studies have shown dysregulation of the genes SOX9 and KCNJ2 as a result of a familial translocation between these two genes, the outcome of which is under expression of the respective gene products.[2, 3] The gene products produced by these genes are functionally involved in craniofacial development.[4] The defect is not routinely detected on ultrasonography prior to birth and may not be initially diagnosed in the first few days of the child's life, however, the diagnosis becomes apparent shortly thereafter following failure to latch and difficulty feeding as a result of the cleft palate. Furthermore, many affected babies will begin to struggle with respiration or develop bronchopneumonia following aspiration.[1] Many babies are successful at breastfeeding small volumes on demand. This may delay diagnosis but also critically lowers the pH of the Stephan curve rendering the patient high risk for the development of caries. Enamel hypoplasia has frequently been documented in patients with PRS.[5]

Retro-micrognathia is clinically identified by an underdeveloped mandible located in a retruded position. Clefting of the palate most often presents as a wide u-shaped defect which may involve both the soft and hard palates. This results in a visible oro-antral communication and nasal septum. Glossoptosis refers to upper airway obstruction due to the posterior displacement of the tongue because of the underdeveloped mandible.[3] Despite the physical, psychological and functional problems in patients with PRS, in most cases there appears

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to be intact intellectual ability.[3] The treatment journey is prolonged, and patients are susceptible to a multitude of complications along the way. These include poor aesthetics, functional defects (dental, speech, swallowing, recurrent upper aerodigestive infections), malnutrition, psychosocial effects, psychomotor delay, loss of autonomy as well as postoperative complications including the possibility of hearing loss.[1-3, 6-8] Therapeutic intervention requires the involvement of a diverse range of health care professionals from the earliest treatment regimen to lifelong management. This case report aims to facilitate awareness for the need of a diverse multidisciplinary approach in the management of PRS as well as stability and support at home. Patients require a lot of attention and a stimulating environment to thrive. Successful management of patients with PRS may be evaluated by their overall quality of life and most are able to attain relatively normal functionality as well as being able to establish positive social relationships in later life.[3]

CASE REPORT

A 3-year-old female patient was referred to the Department of Operative Dentistry by a clinician in the Department of Orthodontics. The patient had been diagnosed previously with PRS at the age of 6-months due to both micro-retrognathia and a cleft palate (Figure 1A). The patient was referred for oral prophylaxis and dental assessment prior to scheduled cleft palate repair surgery.

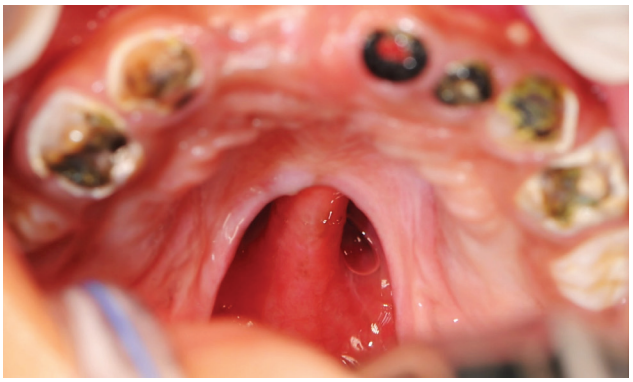

Figure 1A: Intra-oral clinical photograph in which extensive enamel hypoplasia and rampant dental decay of the maxillary deciduous teeth are identified. Furthermore, the u-shaped cleft involving both the hard and soft palate is conspicuous. The cleft is associated with an oro-antral communication with visibility of the nasal septum centrally.

The patient is the only child that is cared for by a single parent (mother) at home. She has not yet attended any preparatory school. She was recently referred to a dietitian as well as to a speech therapist. She appears to be cognitively unimpaired although she rarely interacts and, seldom speaks. She communicates with limited speech in her home language with her mother and grandmother and she can fully comprehend conversations and follows instructions, however, she would benefit from a more socially stimulating environment. Her main complaint was pain in the upper anterior region as well as the existence of “a hole in the upper back part of the mouth”.

Medical History

The patient was confirmed to be healthy, and she was reportedly able to be breastfed normally with no reported incidences of milk aspiration into the nasal cavity. The mother confirmed that there was no familial history of PRS or of isolated cleft palate. The mother informed the clinicians that the patient consumes the same meals as the other family members including porridge and protein such as beef, chicken and eggs.

Dental History

Dental extractions were performed in the past with no complications. Teeth 51 and 52 were previously extracted (Figure 1 B). This dental history is quite relevant for the case as the possibility of hazardous in-office tooth extractions was anticipated due to the presence of an oro-antral communication associated with clefting of the palate in addition to breathing difficulties because of glossoptosis. Other precautionary measures included consideration of complications associated with the administration of general anaesthesia which could be due to airway limitations in PRS cases.[9] There is no history of thumb sucking or of dependence on a pacifier.[10]

Due to the lack of co-operation during examination in the dental chair, a more comprehensive dental examination was scheduled intra-operatively under general anaesthesia in March 2024.

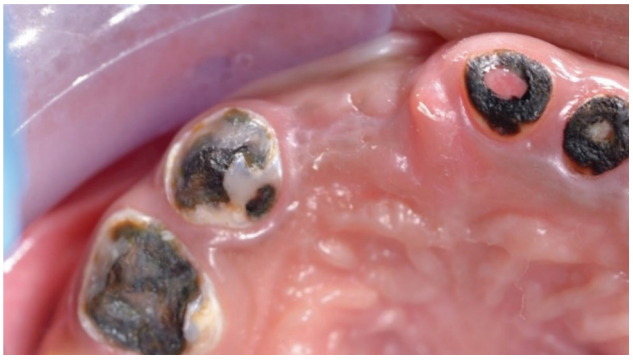

Figure 1B: Intra-oral clinical photograph highlighting the extensive enamel hypoplasia and rampant dental decay of the maxillary deciduous teeth. The site of the previously extracted 51 and 52 has healed adequately.

Treatment procedures and planning

At the patient’s first consultation, a work-up for the surgical repair of the cleft palate under general anaesthesia was discussed. This was a joint sitting involving the maxillofacial surgeon and a dental clinician. A comprehensive examination of the dentition and the cleft palate was performed to facilitate surgical planning.

Extra-oral examination

General appraisal showed the patient to be of small posture yet was well nourished. There was distinct retro-micrognathia. There was no evidence of temporomandibular joint abnormalities or of lymphadenopathy. There was no clinical evidence of jaundice, anaemia, cyanosis, clubbing of the fingernails or of oedema. Radiological examination was not undertaken due to the extensive radiation dose and the difficulty for such a young patient to maintain a still position in addition to the patient being unco-operative.

Intra-oral examination

A fistula distal to tooth 62 was identified (Figure 2A and Figure 2B). In addition, carious involvement of all teeth was evident. The entire dentition was coated with soft adherent plaque. The patient had poor oral hygiene and a low dental IQ which was also facilitated by her mother who reported that she was unable to brush her daughter’s teeth because of discomfort due to the palatal cleft. An underdeveloped mandible was also observed which will likely result in future malocclusion and

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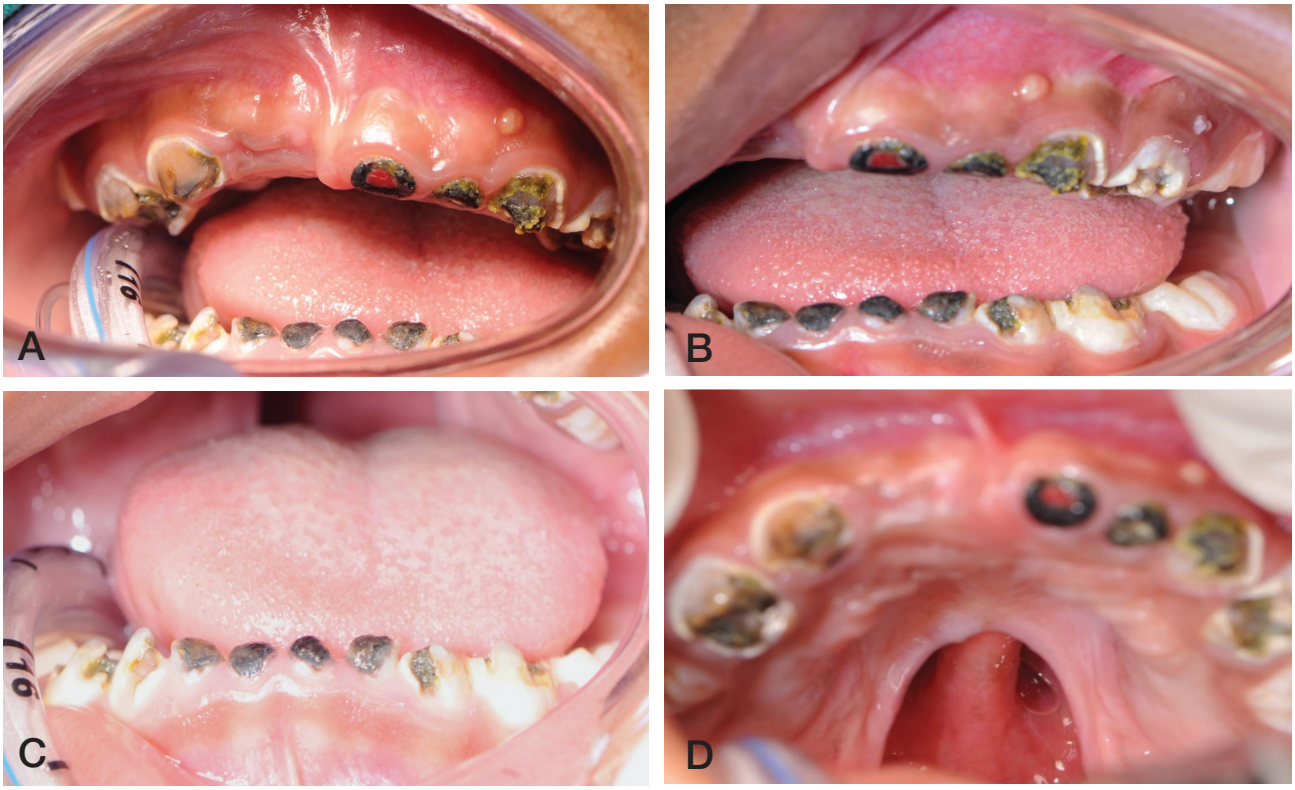

Figure 2A: A clinical intra-oral photograph in which the rampant decay of the maxillary and mandibular dentition can be seen. The labial frenum is also prominent. A parulis (gumboil) is in the region of the apex of tooth 62. **Figure 2B:** A clinical intra-oral photograph in which the maxillary and mandibular teeth are visualised from the left aspect. The friability of the enamel due to hypoplasia and the severity of the caries is also highlighted. The parulis associated with tooth 62 is present on the buccal aspect. **Figure 2C:** Clinical intra-oral mandibular anterior occlusal photograph demonstrating the marked glossoptosis in this patient as well as the carious involvement of the mandibular teeth. **Figure 2D:** Intra-oral maxillary occlusal photograph in which the carious maxillary teeth and cleft palate are depicted.

the need for interceptive orthodontic treatment. Furthermore, the patient exhibited mouth breathing.[9]

The intra-oral findings in this patient were photographed by the Audio-Visual Department with the written consent of the patient's mother. Examination showed the presence of multiple carious teeth, poor oral hygiene, visible soft plaque and fiery red gingiva (Figures 2A to 2C). Clefting of the palate was noted to extend posteriorly from the incisive foramen with partial involvement of the hard palate and more extensive involvement of the soft palate. The cleft corresponds with a "Kernahan striped-Y" classification of 7-8-9.[10] A conspicuous oro-antral communication with visibility of the nasal septum was present (Figure 2D). The documented dental features described in association with PRS include delayed eruption, hypodontia, enamel hypoplasia, abnormal tooth morphology, missing teeth, supernumerary teeth and malocclusion.[11]
Surgical closure of the cleft palate was performed by the maxillofacial surgeon in order to restore anatomy, minimise growth disturbances, to facilitate feeding and speech whilst preventing food aspiration and upper aerodigestive tract infections.[6] It was acknowledged that this was a delayed surgical repair as it is recommended in the literature that surgical repair of the hard and soft palate be performed between the ages of 9 and 12-months by means of an intravelar veloplasty procedure.[6]

The advantages of this clinical procedure when performed at an earlier stage of life include minimisation of growth disturbances whilst being relatively easy to perform and is associated with fewer postoperative risks for fistula development.[6]
It was also anticipated that this surgical procedure may, however, be more complicated when wide clefts are repaired and with loss of palatal length due to this late intervention.[12] The surgery was also planned over two phases because of the long duration of treatment, high volume of blood loss and extensive trauma which resulted from the procedure. It was therefore decided to perform dental extractions and treatment in a subsequent procedure under general anaesthesia.

The first phase of the surgical cleft repair was reportedly successful. The patient had good muscle strength and a good prognosis (Figure 3 and Figure 4). A small palatal communication might remain after surgical healing, and this is often a complication which is noted to occur in 9,7% of cases.[6]

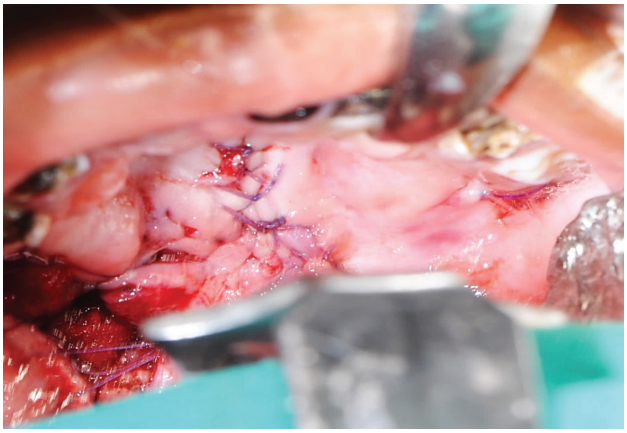

Figure 3: Intra-operative clinical photograph depicting cleft palate repair surgery using the intravelar veloplasty repair technique.

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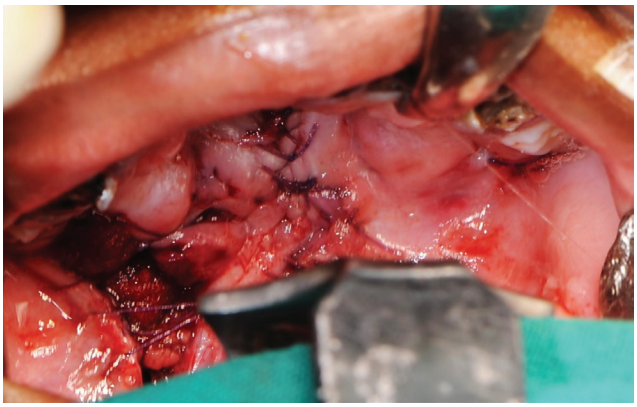

Figure 4: Post-operative clinical photograph showing closure of the palatal cleft.

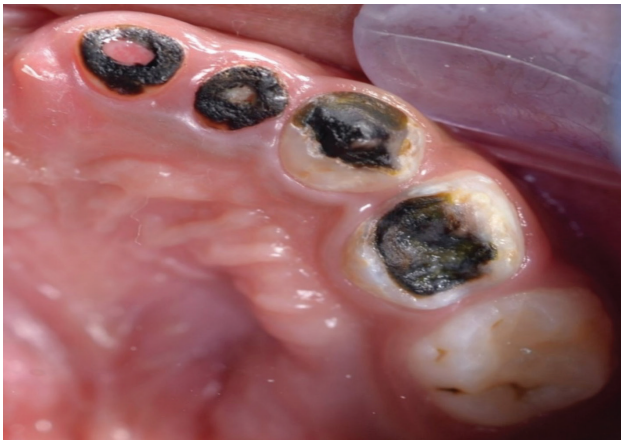

Figure 5A: The pre-operative view of the carious left maxillary teeth.

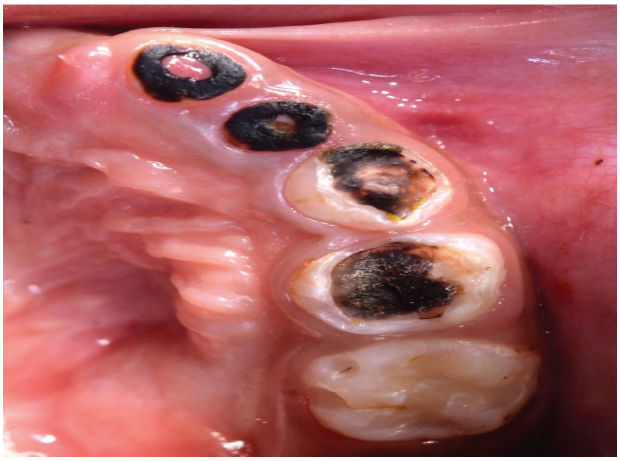

Figure 5B: Intra-operative view of the same teeth. Figure 5C: Post-operative clinical photograph showing extraction sockets of tooth 61 and tooth 62 as well as fissure sealant application on tooth 65.

A team approach was used to decide on the second phase surgery following appropriate healing at which time the dental management will be performed. The dental treatment plan included extraction of tooth 61 and tooth 62 (Figure 5A to 5C). Composite restoration (3M Composite) of carious teeth was performed provided there was no evidence of pulpal involvement (Figures 6A to 6C and Figures 7A to 7C). These teeth were also covered with flowable composite without raising the occlusal surfaces. Fissure sealants were also placed on the occlusal surfaces of all d's ("Twinky Star") as seen in Figure 5C. Dental prophylaxis was undertaken to remove all adherent plaque. The use of fluoride varnish was applied to strengthen and support the remaining enamel.[13] Topical fluoride application was done where possible with re-application at future in-chair follow-up appointments for better results. The placement of stainless-steel crowns on the d's is preferable but in this case, it was not performed due to time constraints.

Close regular monitoring and follow-up is crucial for evaluation of the permanent dentition for early recognition of enamel hypoplasia. There is a dire need for parental education and support. The mother of the patient must be able to assist with tooth brushing and to be vigilant about good oral hygiene. Psychomotor delays should be evaluated at follow-up visits to facilitate early referral if required.[14]

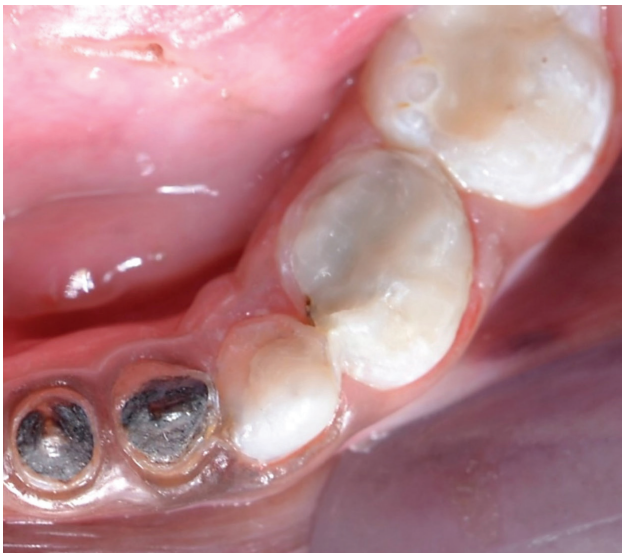

Figure 6A: Pre-operative photograph demonstrating the carious involvement of the teeth in the third quadrant.

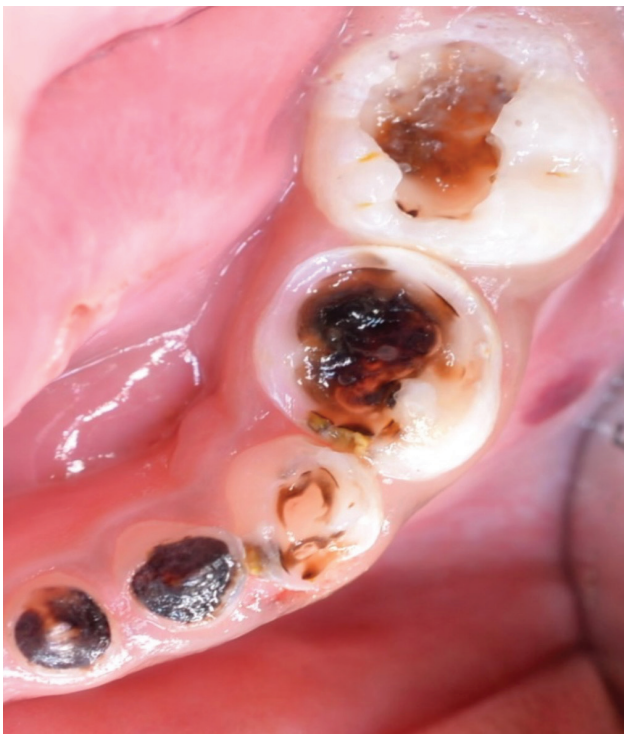

Figure 6B: Intra-operative photograph of the caries removal in teeth 73, 74 and 75. Figure 6C: Post-operative photograph showing composite restorations on the lower left teeth 73,74 and 75.

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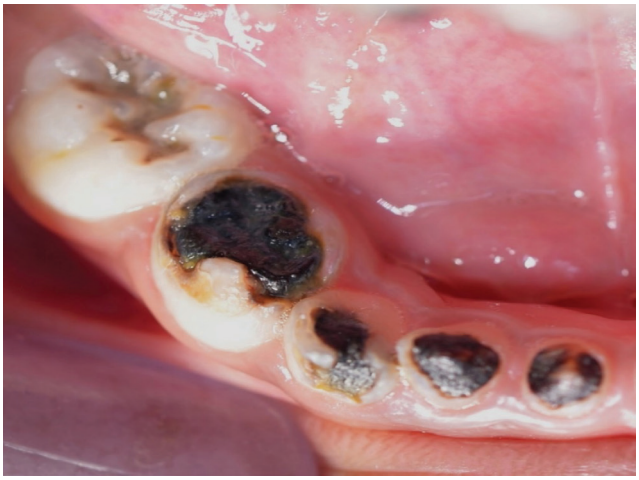

Figure 7A: Pre-operative photograph showing the extent of the decay involving the teeth in the lower right quadrant.

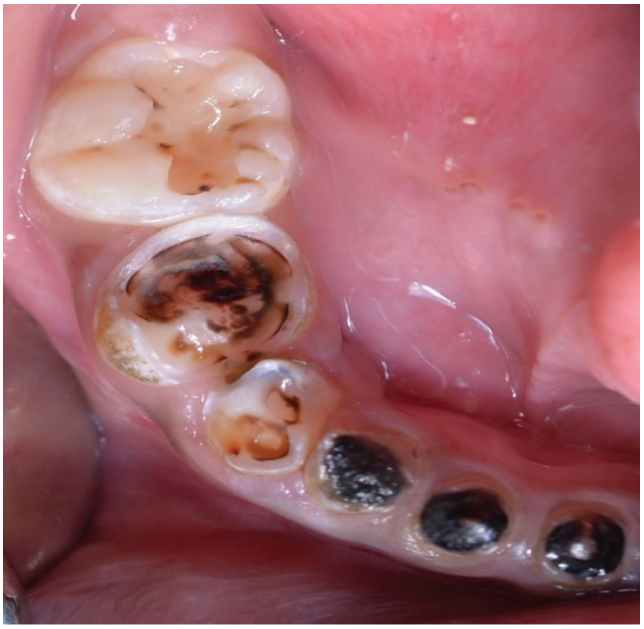

Figure 7B: Intra-operative photograph demonstrating caries removal from teeth 83, 84 and 85. **Figure 7C:** Post-operative clinical photograph showing composite restorations on the lower right teeth 83, 84, and 85.

DISCUSSION

PRS is a rare congenital craniofacial deformity characterised by retro-micrognathia, glossoptosis and cleft palate.[1-4, 12, 13] The disorder may occur in isolation or as part of a wider Mendelian syndrome such as Stickler's Syndrome, Velocardiofacial Syndrome, Marshall's Syndrome, Foetal alcohol Syndrome or Treacher-Collins Syndrome.[1, 2, 4] Such cases are not always hereditary. Recent studies have shown the genes SOX9 and KCNJ2 are dysregulated as a result of a familial translocation between the two genes which may interfere embryologically with caudal hind brain development.[1, 3] The defects associated with PRS occur aetiologically during the 8th to 11th weeks of intra-uterine life. At this stage, the relatively large tongue in these patients fails to descend adequately which prevents the lateral palatine shelves from fusing together in the midline. Patients with PRS are diagnosed with the condition shortly after birth and experience lifelong complications. The most immediate concerns are difficulty breathing and difficulty feeding. Immediate intervention regimens include supine positioning of the patient for feeding and to minimise the potential for airway obstruction by posterior displacement of the tongue or aspiration of food. Patients may require insertion of a nasogastric or orogastric tube or placement of a surgical gastrotomy.[8, 14] The use of an obturator or modification of the nipple for bottle feeding may also be required. There is significant risk of death due to bronchial aspiration or respiratory difficulties. This may be minimised by use of a laryngeal mask, prolonged intubation, a tongue-lip adhesion surgery, mandibular distraction osteogenesis and tracheotomy[1, 8, 14] Recurrent local infections complicate nutritional status which is further exacerbated by a cleft palate. Contemporary therapy combining tongue-lip adhesion and mandibular advancement is thus the most recommended surgical intervention. Surgical intervention is generally postponed until after 18-months of age and in consultation with the entire health care team. Most patients with PRS do not have impairment of intellect and can easily grow up to live relatively normal lives. They do, however, require continuous support as well as psychological follow-up for the remainder of their lives. The only residual complaints in most patients is slight hypernasality although the overall quality of life of a patient may be dramatically affected by the aesthetic outcome of all therapeutic interventions as this may affect the patients autonomy, self esteem and ability to socialise.[1, 3] There remains a need for patient and parent education to ensure that there is continuity of therapy with members of a diverse multidisciplinary health care team to optimise the aesthetic, functional, social and psychological outcomes for each individual patient.

CONCLUSION

The aim of this case report is to emphasise the diversity of the multidisciplinary treatment team needed in the adequate management of a patient diagnosed with PRS. The multitude of complications associated with PRS are both life threatening and may be lifelong. Caretakers and parents must be made aware of the need to be diligent, ensure a stimulating and nurturing environment and to continually provide support of both a physical and psychosocial nature.

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