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CASE REPORT

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Case report: dental care approaches for a pediatric patient with Joubert syndrome

Rana A.S. Alamoudi[1,*], Jawan Shaker[2], Raghad Alsalim[3], Rahaf Bakheet[2]

[1]Pediatric Dentistry Department, Faculty of Dentistry, King Abdulaziz University, 21589 Jeddah, Saudi Arabia
[2]Pediatric Dentistry Department, Jeddah Medical Health Center, 21452 Jeddah, Saudi Arabia
[3]Pediatric Dentistry Department, Ministry of Health, 65525 Al Bahah, Saudi Arabia

***Correspondence**
rasalamoudi@kau.edu.sa
(Rana A.S. Alamoudi)

Abstract

Background: Most dental professionals, including students, find it challenging to provide care to patients with particular medical needs, particularly in cases of rare syndromes. **Methods**: A 6-year-old girl with Joubert syndrome (JS) scheduled an appointment at our pediatric dentistry clinic. JS is a rare, genetically heterogeneous autosomal recessive disorder, which manifests as a neuropathological illness of the cerebellum and brainstem, along with the involvement of several organs. Various nonpharmacological interventions for behavioral management were offered by our dental office to meet her dental needs, including early morning appointments and minimizing the treatment duration to tackle her short attention span. The "tell, show and do" technique was advocated through the use of simple words and short sentences to facilitate slow language development. Due to her mild hyperactivity, she was appreciated for good behavior and rewarded with short breaks during dental treatment session. **Results**: These interventions resulted in a positive outcome, facilitated by the use of potent local anesthetics and strategies to maintain her attention, coupled with rewarding tactics. Maintaining the patient under local anesthesia, we achieved a positive outcome in the form of complete dental care through adequate placement of stainless-steel crowns, composite restoration, pulp therapy, and extraction. **Conclusions**: Managing patients with specific healthcare needs requires collaboration and consent from the patient's physician, as well as a multidisciplinary approach. Recognizing oral health issues and emergency protocols before initiating dental treatment is crucial.

Keywords

Dental treatment; Joubert syndrome; Molar tooth appearance; Bat wing appearance; Case report

1. Introduction

In 1969, the French pediatric neurologist Marie Joubert first described Joubert syndrome (JS), a rare inherited autosomal recessive disorder, in Montreal, Canada. JS affects approximately 1/80,000–100,000 live infants globally [1]. In most Arab countries, the prevalence of JS is unclear; however, the United Arab Emirates has a reported incidence of 1:5000 births, which is significantly higher than that reported in the United States [2]. This disorder can be caused by more than 35 genes that affect various organs and create neuropathological abnormalities in the brainstem and cerebellum [3].

The term "Joubert syndrome related disorders" (JSRD) refers to a group of pleiotropic disorders that share similarities with JS but also affect other body systems [4]. Because primary cilia are required for the growth and function of various cell types, these diseases are classified as ciliopathies [5]. Magnetic resonance imaging (MRI) is required for a definitive diagnosis. The radiographic findings include (1) elongation of the isthmus (the region of the brain stem between the pons and inferior colliculus), (2) thinning of the deep interpeduncular fossa and pontomesencephalic junction, and (3) thickening of the superior cerebellar peduncles. The molar tooth sign (MTS) is a radiological finding associated with JS, characterized by thick and horizontally oriented superior cerebellar peduncles, an exceptionally deep interpeduncular fossa, and cerebellar vermis hypoplasia or dysplasia. Vermian hypoplasia is characterized by inadequate lobulation or fusion of the halves of the vermis, resulting in a sagittal vermis cleft, seen on axial or coronal MRI planes [6].

The neurological hallmarks of JS include hypotonia, ataxia, abnormal eye movements, newborn breathing dysregulation, intellectual impairment, and developmental delay [7]. Retinal dystrophy affects 25% of cases, and polydactyly affects 8–16% of cases [8]. Seizures, cleft lip or palate, tongue anomalies, and other deformities may also be present [9]. During the first 6 months of life, aberrant breathing patterns, such as hyperpnea alternating with apnea or hyperpnea, often resolve. The co-occurrence of these disorders strengthens the clinical suspicion of the disease. Prognosis soon after birth is determined based

on the extent and severity of respiratory dysregulation, which can be fatal and requires assisted ventilation. Thereafter, the prognosis depends on renal complications, as they are the main cause of death if not detected and treated promptly [10].

Individuals with JS have vision impairment, poor motor imbalance, and hyperactivity, which lead to less effective plaque removal and consequently an increased susceptibility to carious lesions and periodontal disease. Pediatric dentists play a crucial role in the care of these patients by implementing primary prevention measures, such as oral prophylaxis, that help mitigate the detrimental effects on the patient's oral and perioral tissues, because these individuals typically have poor oral hygiene and lack the manual dexterity needed for tooth brushing. Behavioral modification can be safely used to manage dental problems in patients with JS, thereby reducing the chance of an apneic episode. When a patient has co-occurring cleft lip and palate, the pediatric dentist plays a critical role as a part of the multidisciplinary team managing the patient's rehabilitation [11, 12].

2. Patient information

A 6-year-old girl presented with tooth pain caused by multiple carious lesions at the outpatient pediatric dentistry department of the Jeddah Medical Health Center in Jeddah, Saudi Arabia (Fig. 1). This was the patient's first dental appointment. Her parents provided a complete medical history, indicating a successful full-term cesarean section delivery in 2016. Significant medical concerns arose when her mother noticed unusual behaviors in the newborn, including quiet sobbing, feeding difficulties, muscle weakness, and jerky eye movements. dental treatment posed challenges, such as mild hyperactivity, speech and language delays, and short attention span. In addition, the patient exhibited an abnormal breathing pattern, alternating between hyperpnea intervals and apnea or hyperpnea periods, and required home breathing support for the first 6 months of life, after which the problem was resolved. The patient had significant delays in her development, such as sitting independently at 1 year and 9 months and beginning to walk at 3 years. As a child, the patient suffered many traumas attributed to balance disorders, resulting in tooth loss and leg fractures, before being admitted to the intensive care unit (ICU) due to hypovolemic shock resulting from hemorrhage (Fig. 2).

At 2 years of age, the patient underwent her first MRI in 2018, which confirmed the diagnosis of JS. Vision impairment and communication difficulties were subsequently detected. Based on the reports from her social worker, psychologist, and speech therapist, the patient appeared to have low-average intelligence, with a developmental quotient score of 80–89. She also exhibited mild hyperactivity, impulsivity, and distractibility, along with nervous speech disorders, delayed expressive language development, and struggle with voice modulation, fluency, and smooth flow of speech. Renal involvement was presented as hepatic fibrosis, whereas ocular disease manifested as oculomotor amblyopia. According to the parents, the patient was the eldest of four siblings and had no significant medical history. Genetic testing will be performed on the patient and both parents to determine whether the cause is a new mutation or inherited, for future pregnancies.

3. Results

3.1 Clinical findings

During the patient's initial physical examination at the dental clinic, her vital signs were normal, and her weight (20 kg) and height (108 cm) fell within the 50 percentiles for her age, according to The Growth Charts for Saudi Children and Adolescents [13]. She exhibited mild hyperactivity; linguistic difficulties, manifesting as abnormalities in voice modulation, speech flow, and fluency; attention deficit disorder; and below-average intellectual abilities. Evidence of decreased mental ability and cognitive impairment was also noted. The patient wore spectacles, had an ataxic gait, and required walking assistance. Her facial profile was convex, with an elevated mandibular plane angle. Intraoral examination revealed U-shaped upper and lower arches and early phases of mixed dentition. Multiple proximal and occlusal carious lesions were observed, indicating early childhood caries (Figs. 3,4) The upper right first central incisor (#51) was lost early because of earlier avulsion trauma (Figs. 3,4). Teeth #55 and #65 had mild occlusopalatal caries, whereas tooth #64 had severe occlusodistal caries, approximating the pulp horn. All lower primary molars had occlusolingual caries, and tooth #61 had mesial caries. The Oral Hygiene Index-Simplified (OH-S) indicated low plaque index, mild gingival irritation, and poor oral hygiene [14]. Black chromogenic bacterial strains were evident on the lower incisors and canines (Fig. 3).

3.2 Diagnosis

In 1992, Saraiva and Baraitser established the following diagnostic criteria for JS: hypoplasia of the cerebellar vermis and developmental delay with at least one of the following signs and symptoms: (1) atypical eye motions and (2) an irregular breathing pattern with apnea and/or hyperpnea [15]. Following the diagnosis of JS, the patient underwent additional tests to determine the presence of multiorgan involvement. The MRI scans of the brain displayed a dysplastic or absent cerebellar vermis with enlarged superior cerebellar peduncle, indicative of the characteristics MTS. The third ventricle appeared larger than usual and resembled a bat wing (Fig. 5).

3.3 Therapeutic intervention

The patient underwent regular visits to multiple departments, including urology, pediatric neurology (every 6 months), pediatric ophthalmology (annually), weekly speech therapy, and physiotherapy. Apart from over-the-counter vitamins provided by her mother, she did not receive any medical treatment for her medical condition.

At our dental clinic, behavioral management techniques were used to successfully complete the dental treatment over several sessions (Fig. 6). Considering that emotional stress can lead to breathing problems, such as tachypnea or apnea, we implemented a stress-reduction protocol during treatment.

Various nonpharmacological behavior management techniques were used at our dentist office to meet all her dental needs and to give her a pleasant dental experience. She was introduced to the clinic at the first dental appointment so that

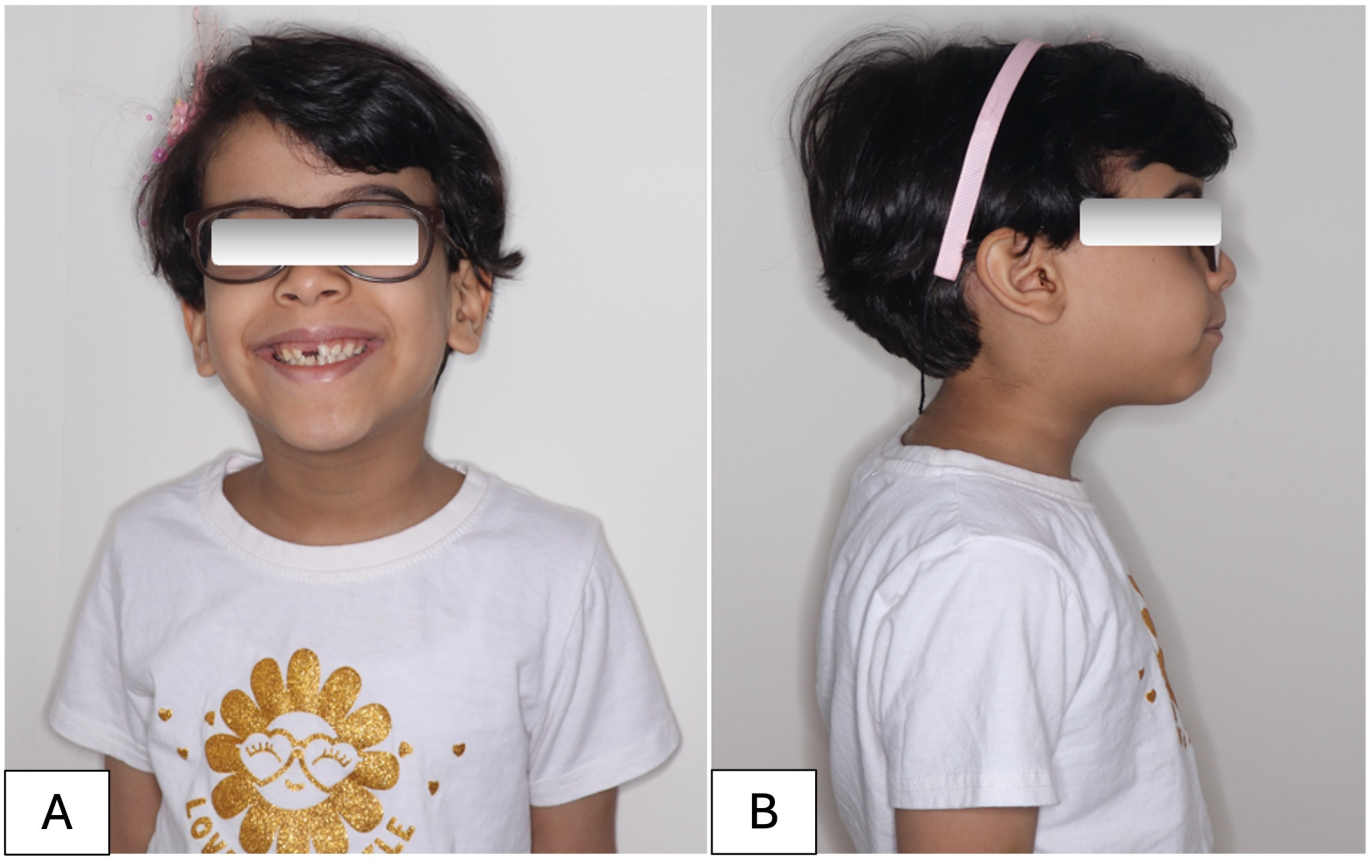

FIGURE 1. The 6-year-old girl extraoral clinical appearance shows ocular impairment. (A) Frontal view. (B) Lateral view.

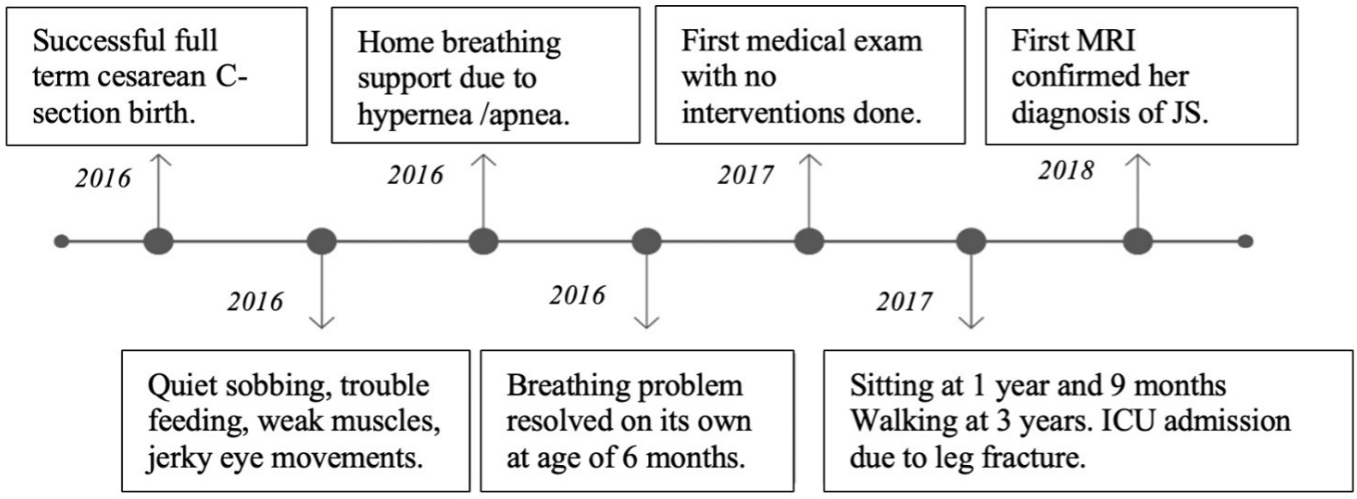

FIGURE 2. Timeline of the patient's significant medical history. MRI: Magnetic resonance imaging; JS: Joubert syndrome; ICU: intensive care unit.

she could become acquainted with the dental personnel and environment.

Before commencing any dental procedure, we familiarized her with the dental chair and equipment. She was given the option of undergoing one or two minor dental procedures at a time after establishing rapport. To address her short attention span, appointments were scheduled early in the morning with limited duration. Considering her limited language development, we adopted the "tell, show and do technique" using simple words and brief sentences. The child was given many brief pauses during the dental treatment session as a reward for her good behavior, considering her mild hyperactivity.

Furthermore, by directing the patient's attention, using strong local aesthetics, and providing positive and pleasant reinforcement, the dental team established a good rapport with the patient, who was receptive to treatment without any limitation. Throughout dental treatment, a nearby source of oxygen was readily available to prevent any potential apneic

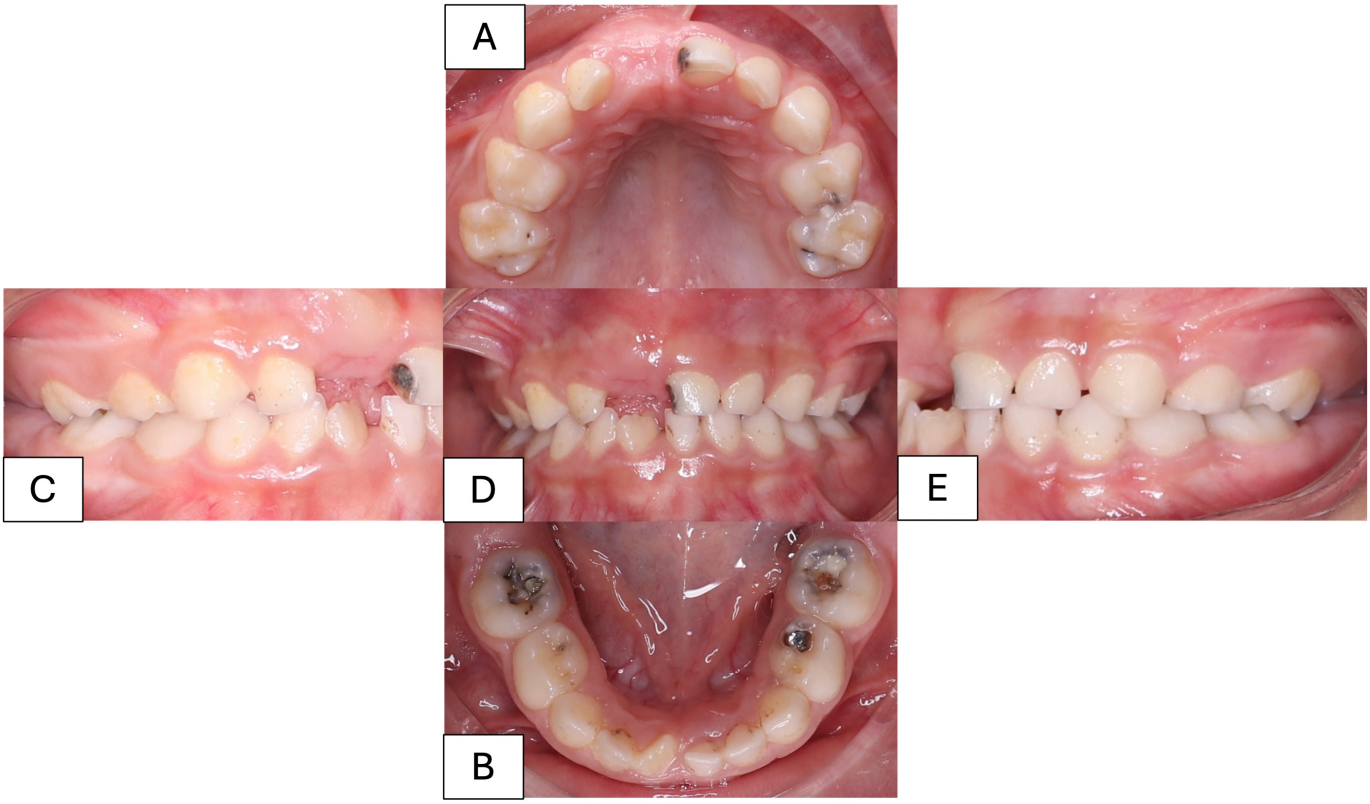

FIGURE 3. The intraoral clinical appearance of the patient before receiving dental treatment. (A) Intraoral upper occlusal view, showing clinical exfoliation of tooth #51, incipient carious lesion on tooth #55 occlussaly, mesial carious lesion on tooth #61, distal carious lesion on teeth #52 and #64, occluso-palatal carious lesion on tooth #65. (B) Intraoral lower occlusal view, showing occlusal carious lesion on teeth #75 and #74, mild lower anterior crowding, occlusal carious lesion on tooth #85. (C) Intraoral Right lateral view, showing mesial step primary molar relationship, class I primary canine relationship. (D) Intraoral frontal view, showing 10% overbite. (E) Intraoral left lateral view, showing mesial step primary molar relationship, class I primary canine relationship.

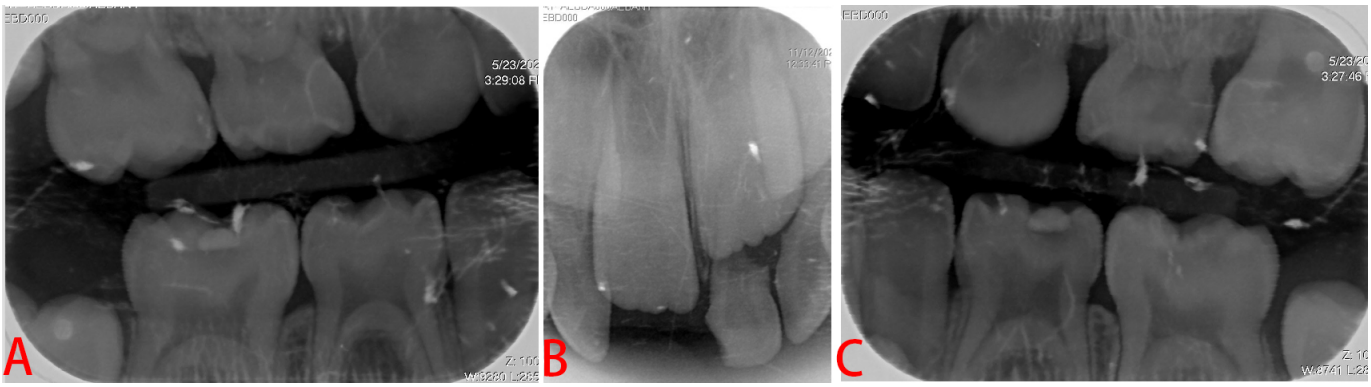

FIGURE 4. The intraoral bitewing radiographs and upper anterior periapical radiograph of the patient before receiving dental treatment. (A) Right intraoral bitewing radiograph, showing normal bone level, mesial carious lesion on tooth #84, occlusal carious lesion on tooth #85. (B) Intraoral periapical radiograph, showing erupting permanent tooth #11, physiologic root resorption of tooth #61. (C) Left intraoral bitewing radiograph, showing distal carious lesion on tooth #64, occlusal and mesial carious lesion on tooth #74, occlusal carious lesion on #75.

episode, providing oxygenation (3 L/min) in advance. The treating physician advised against using oral sedatives due to the risk of respiratory depression leading to an apneic episode. Moreover, because JS is associated with airway problems, not all individuals with JS are suitable candidates for general anesthesia. Micrognathia, a broad, protruding tongue, and a high-arched palate can make endotracheal intubation more challenging for these patients [16]. Although volatile anesthetics and opioids are believed to contribute to postoperative respiratory issues, only few reports in the literature are available regarding anesthesia in patients with JS. Therefore, a recommendation for either volatile or

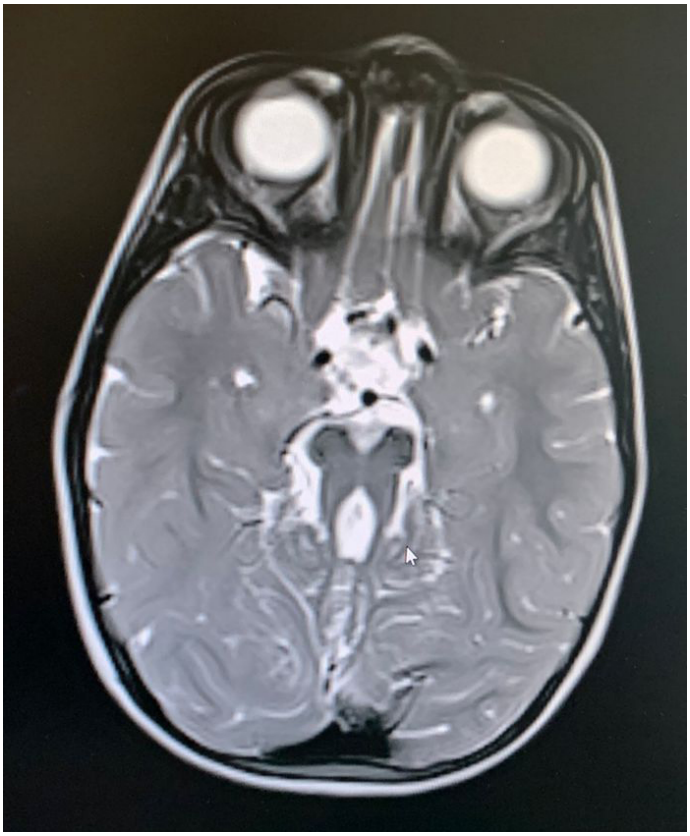

FIGURE 5. The patient's brain magnetic resonance scan (MRI) displays the molar tooth appearance.

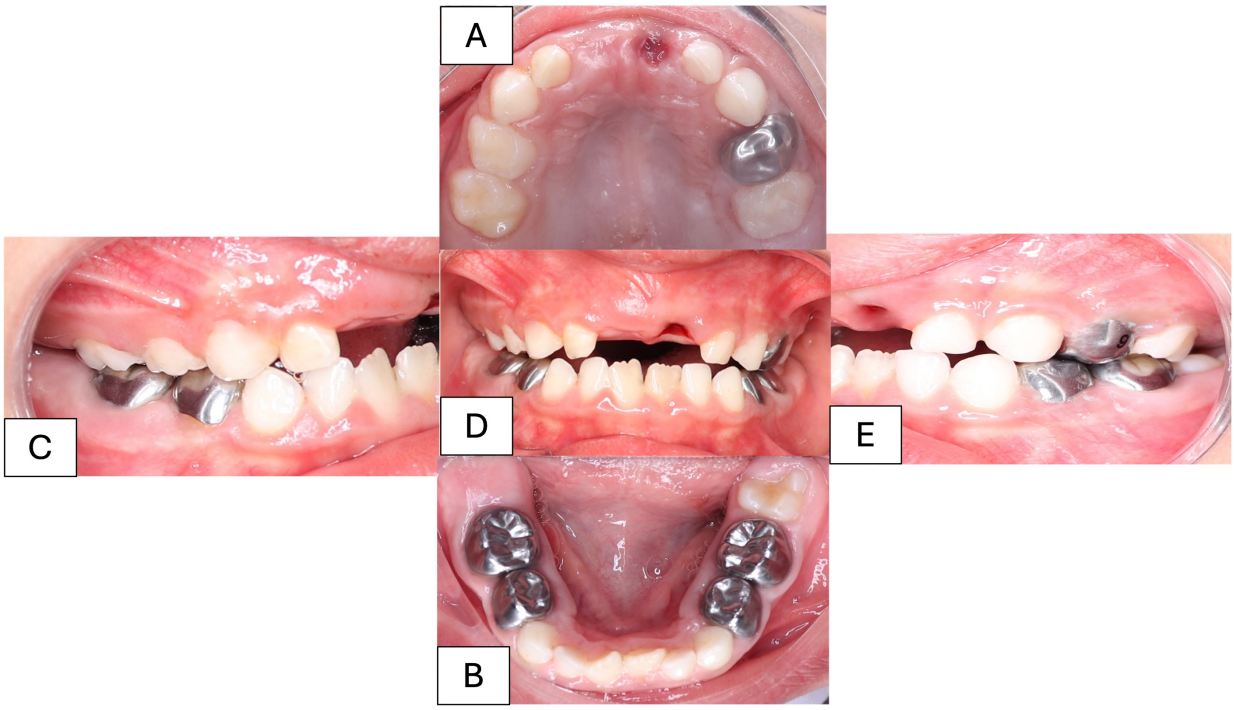

FIGURE 6. The intraoral clinical appearance of the patient after dental treatment. (A) Upper intraoral occlusal view, showing CI composite restoration on tooth #55, extraction socket of tooth #61, Stainless steel crown (SSC) on tooth #64, Compound CI composite restoration on tooth #65. (B) Lower intraoral occlusal view, showing SSC on teeth #74, #75, #84, #85, partially erupted tooth #36. (C) Right intraoral lateral view, showing class I canine relationship and SSC on teeth #84, #85. (D) Intraoral frontal view, showing eruption of lower permanent central incisors. (E) Intraoral left lateral view, showing class I canine relationship, SSC on teeth #64, #84 and #85.

intravenous anesthetic drugs cannot be made at this time [17]. Short-acting medications are preferred, and the utilization of α-2-agonists has been documented. These individuals are vulnerable to drugs, such as opiates and nitrous oxide, that induce respiratory depression; therefore, medication must be prescribed carefully with the consent of the patient's physician.

3.4 Follow-up and outcomes

Dental treatments are performed to maintain tooth structure, arrest the progression of dental caries, reduce pain and discomfort, restore the tooth to its functional and aesthetic state, enhance oral health, monitor the development of the patient's occlusion and dentition, and minimize cariogenic diet. Following oral prophylaxis using a fluoridated paste, complete removal of carious lesions was performed (Fig. 7), followed by the placement of stainless-steel crowns (SSCs) in teeth #64, #74, #84 and #85. Pathologic pulp exposure occurred during caries removal from tooth #64; therefore, MTA pulpotomy was performed followed by SSC placement. Mild carious lesions in teeth #55 and #65 were excavated and permanently restored with composite. Because tooth #61 was carious and near exfoliation, extraction was performed instead of restoration to facilitate eruption of the permanent successor. SSCs are the treatment of choice for extensive carious lesions involving more than one tooth surface in patients at high risk of caries. Moreover, full-coverage crowns are the best option for prolonging the durability of restorations. Oral hygiene instructions were provided, including scrub brushing technique with a youth multitoothed soft nylon toothbrush and 1450 ppm fluoridated toothpaste to be performed twice a day (in the morning after breakfast and before going to school and before bedtime), along with floss once a day to remove plaque from between the teeth and at tight interproximal areas. The patient was advised to rinse with water after meals and snacks. Parents were advised to supervise oral hygiene measures. Diet counseling involved assessing the patients' usual dietary intake and identifying areas where changes were needed. A plan to reduce snack frequency and fermentable carbohydrate ingestion was developed through the completion of a diet survey form and repeated discussions with the child and his mother regarding the etiology of dental caries and the role of fermentable carbohydrates. The patient's complaints had been addressed, and her treatment was completed. The oral cavity was free of dental caries, and the gingival soft tissues were healthy. The debris index was used to determine the acceptable level of oral hygiene, which ranged from 2 at the first appointment to 0.1 at the last visit. During dental treatment, the patient behaved well, assessed using Frankl rating scale (positive + ve). The challenge that we faced during her dental treatment was that she needed multiple exploratory visits before initiating dental treatment and her short tolerance span required additional dental visits to successfully complete all treatment. The prognosis is favourable with regular maintenance visits and appropriate oral hygiene. In addition to her dental follow-up appointments, the patient will be expected to be followed up by a multidisciplinary team to maintain her general health and improve her prognosis.

4. Discussion

Joubert's is a rare autosomal recessive disorder. Approximately half of the patients with JS have *CEP290* gene mutations [18]. Recent studies have mapped one position to chromosome 9q [19]. Ben-Salem *et al*. [20] examined 70 families from all Arabic countries known to have JS and JSRDs at the molecular level and compiled the discovered mutations. They found 53 mutations in 15 genes, demonstrating the genetic heterogeneity of JS and associated JSRDs in Arabs. Arab patients with JS have been reported to have mutations in 15 out of 23 genes, with Inositol Polyphosphate-5-Phosphatase E (*INPP5E*), Abelson Helper Integration Site1 (*AH1*), Centrosomal protein 290 (*CEP290*), coil and C2 domain containing 2A (*CC2D2A*), Transmembrane Protein 138 (*TMEM138*), being the most frequently mutated genes, all believed to have founder mutations. Mutations in Kinesin Family Member 7 (*KIF7*) and Transmembrane Protein 67 (*TMEM67*) have been identified primarily in North African Arab countries, whereas those in *CPLANE1* (previously known as C5orf42) and *RPGRIP1L* gene—responsible for Protin coding—have only been detected

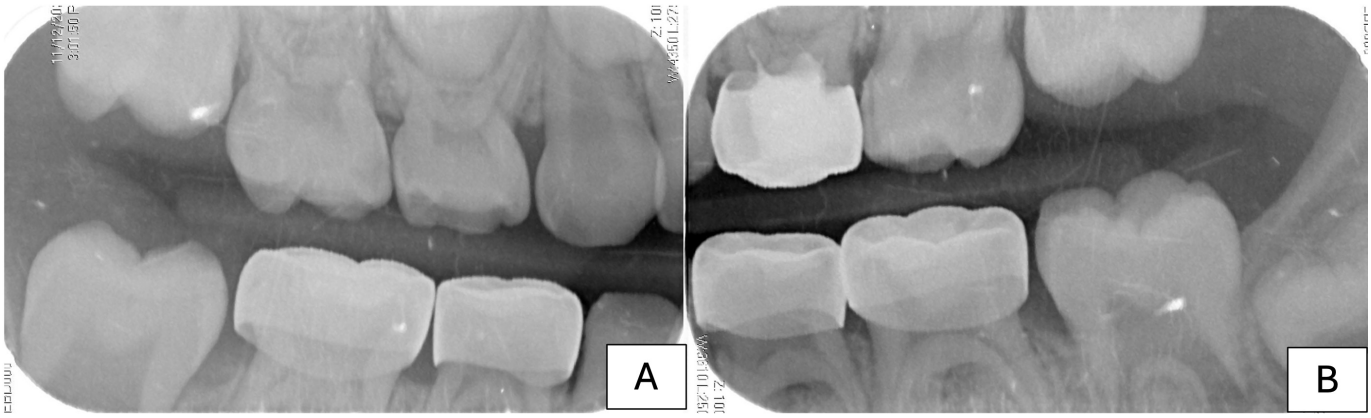

FIGURE 7. The intraoral bitewing radiographs of the patient after receiving dental treatment. (A) Right intraoral bitewing, showing SSC on teeth #84 and #85. (B) Left intraoral bitewing, showing pulpotomy and SSC on tooth #64, and SSC on teeth #74 and #75.

in Saudi Arabia. Mutations in *AHI1* and *CEP290* predominantly occur in the Arabian Gulf.

JS is characterized by a partial or complete loss of the cerebellar vermis, resulting in hypotonia, ataxia, erratic eye movements, respiratory problems, and mental retardation. Several clinical features of JS are evident during infancy [21]. In a study conducted by Elhassanien and Alghaiaty, the onset of symptoms, usually apnea or hyperpnea, occurred in the first 40 days (range: 10 days to 5 months) [22]. While MTS is a feature of JS, it should be considered a differential diagnosis as it can also be present in other disorders such as Senior-Løken syndrome, COACH syndrome (also known as Joubert syndrome with hepatic defect), Dekaban-Arima syndrome and Varadi-Papp syndrome [1]. Vermis hypogenesis results in a superior fourth ventricle resembling bat wings and a mid-fourth ventricle with a triangular form. Cortical dysplasia, gray matter heterotopia, ventriculomegaly, and corpus callosum agenesis are other brain abnormalities associated with this condition [23]. Short, alternating episodes of hyperpnea and apnea or episodic hyperpnea alone are indicative of respiratory anomalies in patients with JS. These episodes often start soon after birth, improve with age, and eventually resolve at approximately 6 months of age. Their severity may vary from sporadic, brief episodes occurring every few days to frequent, protracted episodes happening daily [4]. Almost all patients with JS have hypotonia during the early neonatal or infant stages, which is typically regarded as a primary symptom. Maria *et al*. [24] reported newborn hypotonia in 59 cases they examined. Nystagmus and oculomotor apraxia often occur at birth and improve with age. Nystagmus can be torsional, vertical or horizontal, and usually has a pendular or occasionally a seesaw pattern. Other common visual abnormalities include strabismus, decreased vestibulo-ocular reflexes, ptosis, pigmentary changes in the fundus, and ocular colobomas [23–25]. Renal dysfunction is prevalent in 25% of patients with JS. Renal dysplasia and juvenile nephronophthisis, a kind of chronic tubulointerstitial nephropathy, are additional renal issues that can be present [26]. Patients with JS experience intellectual and developmental disabilities that affect their motor abilities, behavior, language and general growth [27]. Although ataxia and balance issues are not exclusive to JS, they are frequently observed in patients with cerebellar vermis hypoplasia. Polydactyly, which affects 8–16% of individuals, has been associated with this condition. When a patient is suspected of having JS, MRI, ocular examination, renal ultrasonography, electroretinography and karyotyping should all included in the evaluation process. Fetal MRI scans obtained between weeks 20 and 22 of gestation have been shown to be valuable a diagnostic tool for prenatal care. In this case study, clinical evidence, including abnormal breathing patterns, ataxia, oculomotor apraxia, dysphagia, muscle hypotonia, grasp inefficiency, and intellectual disability, supported the diagnosis of JS, which was later confirmed through MRI, which revealed the MTS.

Providing dental care to patients with JS is challenging owing to their noncooperative behavior, and despite being a skilled and competent dentist, various challenges and hurdles appear during treatment sessions. Our dental office provided various nonpharmacological strategies to address the patient's dental needs, including early morning appointments and shorter treatment duration to accommodate her short attention span. We employed the "tell, show, and do" technique, using simple language to facilitate her language development gradually. Praising her good behavior and providing short breaks during dental sessions as a reward helped manage mild hyperactivity. These interventions, combined with potent local anesthetics, maintained her attention and yielded positive outcomes. Under local anesthesia, we successfully completed her dental care, including SSC placement, composite restorations, pulp therapy, and extractions.

To supplement our report, there is a comparable case of a 7-year-old girl reported in New Delhi [28]. Similar to our case, the parent reported the infant's peculiar behavior, such as slow movements and lack of crying. The patient exhibited an ataxic gait and inward bending the legs and required assistance to walk. The results of the fine motor skills test showed no established grasp. In addition to hypertelorism, the patient also had low-set ears, a flattened nasal bridge, polydactyly and syndactyly in all four limbs, partial midline clefting of the lip and palate, and an enlarged tongue, distinguishing her condition from that of our patient.

This case report highlights the dental management of a pediatric patient with Joubert syndrome; however, it has several limitations. First, as a single case report, the findings may not be generalizable to all patients with Joubert syndrome, given the variability in clinical manifestations and severity of the disorder. Second, while the study details the behavioral management techniques used, it does not provide comparative data on the effectiveness of alternative approaches, such as pharmacological sedation or general anesthesia, which may be necessary for other patients with more severe behavioral or medical complications. Additionally, the long-term outcomes of the dental interventions were not assessed, limiting the ability to evaluate the durability of treatments and the patient's oral health maintenance over time. Further research, including longitudinal studies and case series, is needed to establish evidence-based guidelines for the dental care of individuals with Joubert syndrome.

5. Conclusions

When a genetic disorder, such as JS, is diagnosed, special consideration should be given to promptly refer the patient to a pediatric dentist, as these patients are more susceptible to dental caries and periodontal diseases compared with the general population. To effectively treat patients with JS who have poor oral hygiene and insufficient manual dexterity, pediatric dentists must provide a comprehensive preventive regimen. Early implementation of the main preventive program minimizes adverse effects on patients' perioral and oral structures. To avoid negative consequences on the child's oral health, pediatric dentists should routinely remind their parents of the importance of dental hygiene at every appointment. In addition to using appropriate behavioral management techniques to facilitate communication, pediatric dentists should offer supportive counseling to parents. Since JS is associated with airway problems, not all patients are suitable candidates for oral sedation or general anesthesia. Such individuals may

present with a high-arched palate, micrognathia, and a protruding tongue, making endotracheal intubation challenging. Hence, whenever feasible, chairside dental care is preferred. Prescriptions for these patients require a doctor's order due to their susceptibility to drugs causing respiratory depression, such as nitrous oxide and opioids. For patients with JS with cleft lip and palate, the pediatric dentist should be a part of the multidisciplinary team managing cleft cases. Pediatric dentists must be active members of the multidisciplinary team treating patients with JS. Early referral of such patients to a pediatric dentist is crucial for addressing their dental problems with comprehensive preventive and treatment plans that will lead to consistent long-term results.

AVAILABILITY OF DATA AND MATERIALS

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

AUTHOR CONTRIBUTIONS

RASA and RA—designed the case report; performed the case report. RB—provided help and advice on record acquisition and interpretation. RA—analyzed the data. RASA, JS, RB and RA—wrote the manuscript. All authors contributed to editorial changes in the manuscript. All authors read and approved the final manuscript.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

As per our Institutional Review Board (IRB) we don't need an ethical approval for case report publication. However, a consent form was obtained by the patient's legal guardian. The parents authorized us to publish this case study of their child by signing an official informed consent form. We appreciate the patient's and parent's cooperation.

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CONFLICT OF INTEREST

The authors declare no conflict of interest.

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