

Case Report: Over-retained Primary Dentition in CHARGE Syndrome Pediatric Patient

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Abstract

CHARGE syndrome is a complex medical condition, typically diagnosed at the prenatal or neonatal period. CHARGE syndrome is characterized by multiple organ system defects and is an acronym for (C) coloboma of the eye, (H) heart defects, (A) atresia of the choanae, (R) retardation of growth and development, (G) genital or urinary defects, and (E) ear anomalies. CHARGE syndrome often presents as a result of de novo mutations.

Case Report: We present a case of a 12-year, 1-month-old Hispanic male pediatric dental patient with CHARGE syndrome. The patient's mother's chief complaint was for over-retained primary dentition present in the patient's mouth. Upon diagnosis of over-retained primary dentition, a treatment plan to extract them was implemented. Additionally, a medical clearance was requested by the patient's pediatricians to recommend any prophylactic antibiotics or relevant precautions to take in the dental setting due to the patient's condition. Informed consent, pre-, and post-operative instructions were delivered utilizing a video language interpreter. Dental treatment was performed in a traditional dental clinic setting to successfully remove all over-retained primary dentition. The patient was placed on a higher frequency of examination visits to monitor oral hygiene and care.

It was concluded that dental and health professionals should be familiar with CHARGE syndrome, because the complexity of the condition can limit attention to a patient's oral health due to the frequency of other medical visits. This report will include clinical findings, treatment rendered, and recommendations for patients with CHARGE syndrome.

Introduction

CHARGE syndrome was first described in 1979 in 17 children with multiple congenital abnormalities and formally diagnosed in 1981 as a syndrome of non-random association of anomalies occurring together more frequently than they would based on chance¹. CHARGE syndrome is a complex medical condition, typically diagnosed at the prenatal or neonatal period. The incidence of CHARGE syndrome can range from 0.1-1.2/10,000 live births and must be professionally diagnosed¹. It can be characterized by either of four major defining characteristics known as the classical 4Cs: Choanal atresia, coloboma, characteristic ears, and cranial nerve anomalies; or three major and three minor characteristics^{1,9}. The minor characteristics include cardiovascular malformations, genital hypoplasia, cleft lip/palate orofacial cleft, distinctive CHARGE facies (sloping forehead, flattened tip of nose), growth deficiency, and developmental delay¹.

Case Description

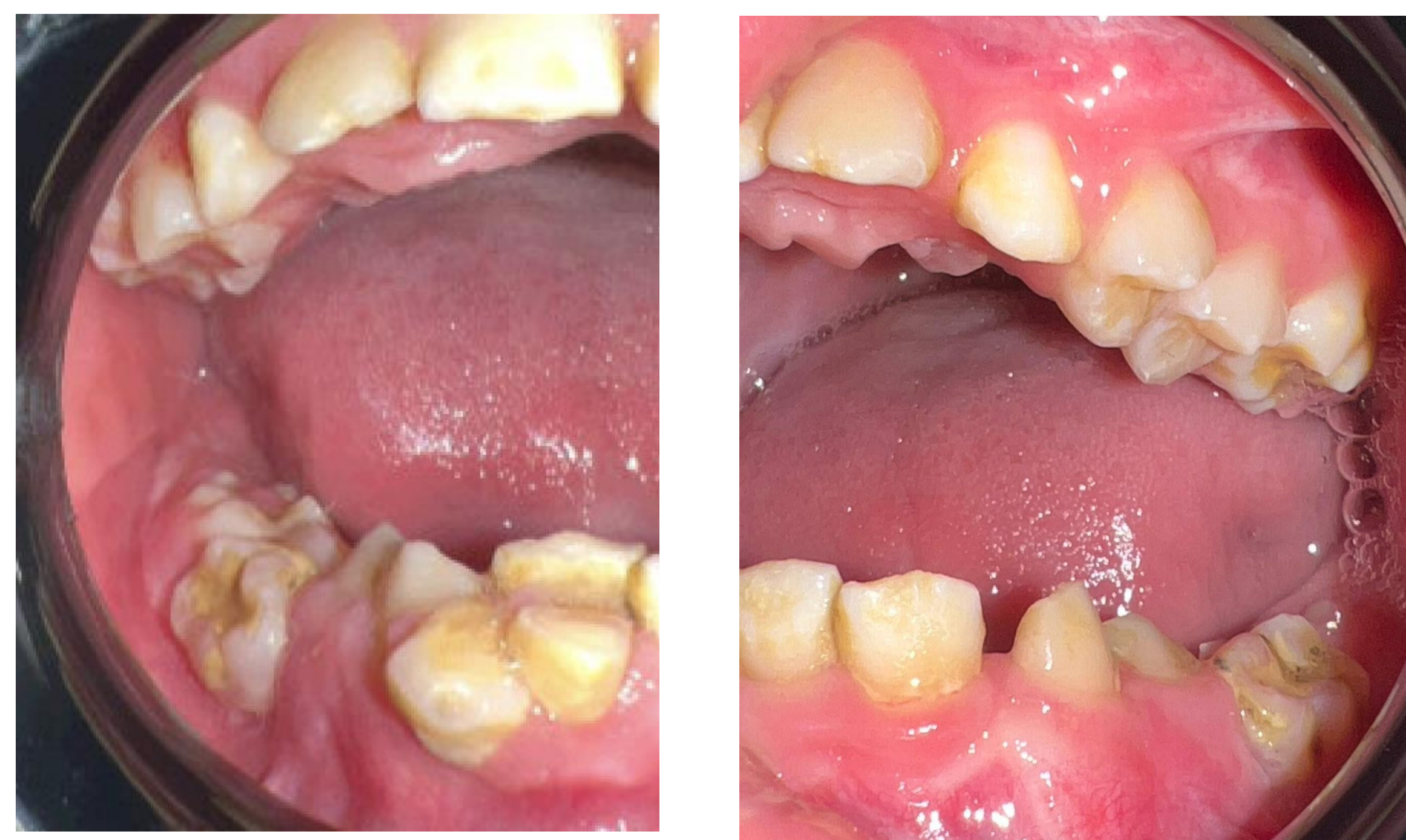
A 12-year, 1-month-old Hispanic male patient presented to the dental clinic at Jamaica Medical Hospital Center (JHMC), Queens, New York, with a chief complaint (as presented by his mother) that "some of his baby teeth are stuck" (Figures 1, 2, 3, 4). His past medical history is significant for premature birth at 36 weeks via Cesarean section, where he was transferred from JHMC to the NICU at Columbia University Hospital and received an eventual CHARGE syndrome diagnosis. He presently feeds primarily through a gastronomy tube (G-tube), which can often cause dysphasia^{2,8}. Since his birth, patient has maintained frequent and routine visits to the hospital to monitor conditions related to his CHARGE syndrome diagnosis. Specifications of patient's CHARGE syndrome diagnosis includes:

- Coloboma of the eye
- Heart defects - Ebstein anomaly of tricuspid valve, congenital pulmonic stenosis
- Atresia of the choanae
- Retardation of growth and development
- Genital or urinary defects - left side hydronephrosis
- Ear anomalies - deafness with hearing loss, uses sign language
- Patient had dysphagia and patient uses a feeding tube

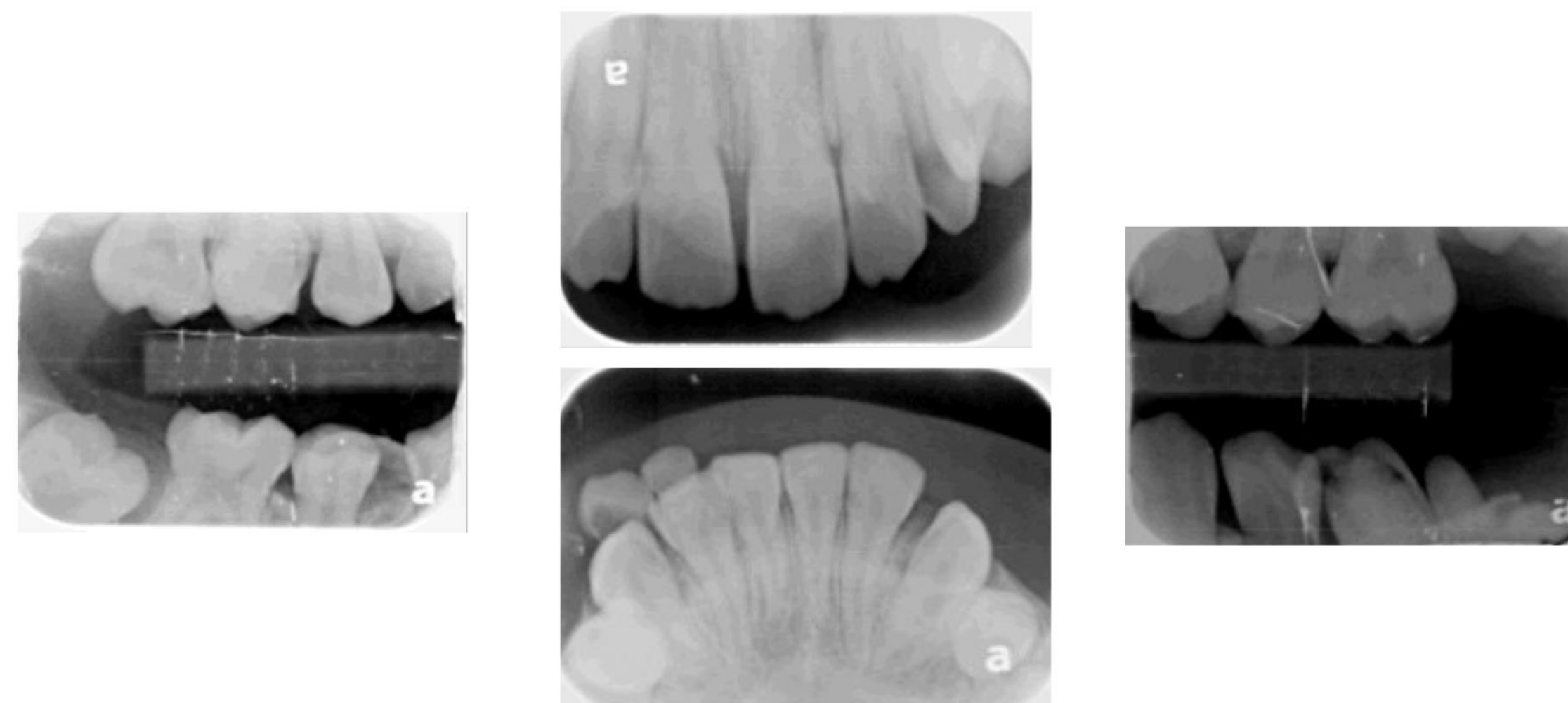
As the patient is frequently being monitored in a medical setting for his CHARGE syndrome diagnosis, he only first presented to the dental clinic at the JHMC in 2021 when he was 9 years old, where he was anxious and required active restraint with his mother to complete a dental consultation. After several missed visits, patient returned in 2024 for periodic examination with mother where he was more combative, a common behavior in CHARGE syndrome patients⁴. Medical clearance was requested from the patient's cardiologist before dental procedure to proceed with dental extractions.



Figure 1. Patient presentation upon visit to dental clinic after 14-month absence, August 2024



Figures 2 and 3. Presentation of right and left side dentition at August 2024 visit. Over-retained primary teeth #K, #T, #C, #Q, #R visible intraorally.



Figures 4. Radiographic presentation of patient's dentition, August 2024.

Discussion

After receipt of medical clearance, patient was treated with active and passive stabilization in the traditional dental setting to remove over-retained primary dentition. Extractions were performed with nitrous oxide gas, local anesthesia, and advanced behavioral management techniques, such as utilization of papoose board and tell-show-do. Post-operative instructions were reviewed with the parent utilizing a video interpreter including monitoring of patient until local anesthesia effects were absent. Furthermore, oral hygiene instruction was reviewed again. Patient utilizes a G-tube and parent has been working on upkeep of oral hygiene for patient by brushing his teeth for him twice a day as he can tolerate.

Patient presented again in March 2025 for periodic examination with healing of gingiva after extraction of over-retained teeth but with presentation of localized moderate calculus presentation in mandibular dentition (Figure 5). It has been documented that non-ambulatory, multi-handicapped tube-fed populations typically present with a significantly larger number of calculus (71%) than non-ambulatory, multi-handicapped non-tube-fed over a thirty-day period⁶.



Figure 5. Patient presentation at subsequent recall visit in March 2025. Intraorally visible are gingival operculums over permanent mandibular first molars and localized moderate calculus presentation in mandibular dentition.

Conclusion

Because CHARGE syndrome has been categorized as a diagnosable condition only in the past few decades, its dental effects and characteristics have yet to be fully documented³. Clinicians should be aware that dental care can be overlooked, due to the need to monitor multiple aspects of the condition. It is the role of the dental provider to ensure that attention is provided to the dental care of CHARGE syndrome patients, and to refer as necessary to optimize the oral health of CHARGE syndrome patients.

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