CHARGE Syndrome: A case report

Abstract

This presentation explores the dental management of patients with CHARGE syndrome, a rare congenital disorder characterized by multisystem involvement. Dental complications associated with CHARGE syndrome include delayed eruption of permanent teeth, congenital tooth absence (e.g., central incisors, premolars, and canines), ectopic tooth eruptions, submergence of primary molars, and taurodontism.

A 14-year-old female with CHARGE syndrome has been under care at the Pediatric Dentistry Clinic at Tufts University School of Dental Medicine since age 5. During routine visits, heavy calculus deposits were consistently observed on all teeth, attributed to her reliance on a gastrostomy (G) tube for feeding, which increases susceptibility to calculus accumulation. Due to behavioral challenges, dental treatment was conducted in an operating room under general anesthesia.

Dental complications in CHARGE syndrome are diverse and often severe, necessitating comprehensive radiographic monitoring and individualized care. These challenges are compounded by the use of feeding tubes, which exacerbate calculus buildup. Early identification and tailored interventions are essential for effective management, ultimately enhancing the quality of life for patients with CHARGE syndrome.

Background

- Rare autosomal dominant genetic disorder.
- Males and females are equally affected
- Incidence of approximately 1: 10,000 births.
- Mutations in CHD7 gene is the most common cause of charge syndrome
- CHD7 gene controls the creation of a protein that regulates developmental pathways though chromatin organization
- Mutations lead to making of an abnormally nonfunctional short CHD7 protein which interrupts the regulation of gene expression and disorded neural crest development
- Most cases arise from de novo mutations, meaning that they are not inherited but occur sporadically





Fig. 1: Facial profile picture







Radiographs obtained in OR: Calculus noted on occlusal and interproximal surfaces.

Clinical Presentation

A 14-year-old female patient is a patient of record at Tufts School of Dental Medicine Pediatric Dentistry department since she was 5 years old. The patient's medical history is significant for patent ductus arteriosus, atrial septal defect, asthma, choanal atresia, coloboma of both optic discs, developmental dysplasia of hip, feeding difficulties, GERD, defective swallowing, microphthalmos, scoliosis, bilateral sensorineural hearing loss, sleep disturbances, right midface hypoplasia, bruxism. Patient is allergic to latex. Past surgical history : choanal dilation, cochlear implant, gastrostomy tube placement, bilateral leg amputation, PDA device occlusion, tonsillectomy, adenoidectomy, bilateral myringotomy and tubes.

Clinical examinations showed heavy calculus deposits on buccal, lingual and occlusal surfaces of teeth.

Patil R, BDS; Loo C.Y., BDS, PhD, MPH, DMD, FAAPD; Laskou M. DDS, DMD, FAAPD (Tufts University School of Dental Medicine, Boston, MA)





Fig.2 Facial front picture



Management

Treatment planning phase:

Due to the heavy calculus deposits noted during periodic dental visits and limited patient co-operation it was decided to treat patient in an operating room setting. Patients' dental treatment was completed in conjunction with the ENT team.

Treatment phase:

- Comprehensive examination.
- Scaling and root planning.
- 4 5 mm deep periodontal pockets noted on #3, #4, #5, #6, #10, #11, #12, #13, #14, #15,#18, #19, #20, #21,#27, #28, #29, #30, #31.
- Fluoride varnish application.
- ENT: Botox injections in parotid glands to help with drooling (excessive salivation).

Maintenance phase:

Recommended periodic exam, caries risk assessment, topical fluoride varnish application and anticipatory guidance every 3 months.

Conclusion

This case highlights the complex oral and systemic challenges associated with CHARGE syndrome and underscores the importance of a multidisciplinary approach in managing patients with special healthcare needs. Given the patient's extensive medical history, including congenital heart defects, choanal atresia, hearing loss, and developmental anomalies, comprehensive dental treatment required collaboration with medical specialists to ensure safe and effective care. Long-term oral health maintenance remains a priority, with a structured follow-up plan involving frequent recall visits, caries risk assessment, and

preventive strategies tailored to the patient's unique needs. This case reinforces the critical role of early intervention and interdisciplinary coordination in improving oral health outcomes and overall quality of life for patients with CHARGE syndrome.

References

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