



PROSTHODONTIC CHALLENGES IN BLEPHAROCHEILODONTIC SYNDROME PATIENTS

Marta Flores, BDS, MS, PhD; Amara Abreu, DDS, MSD, FACP

Adams School of Dentistry, University of North Carolina, Chapel Hill, NC



Introduction

Blepharocheilodontic (BCD) syndrome is a rare congenital disorder caused by mutations in the CDH1 and CTNND1 genes, with an estimated prevalence of less than 1 in 1,000,000. This condition is characterized by eyelid malformations, ectodermal dysplasia, and other systemic conditions such as hypothyroidism and increased cancer risk. Affected individuals also experience dental anomalies, such as bilateral cleft lip and palate (BCLP), maxillary hypoplasia, oligodontia, and irregular tooth morphology. These features, along with craniofacial skeletal defects, present unique challenges for prosthodontic oral rehabilitation.

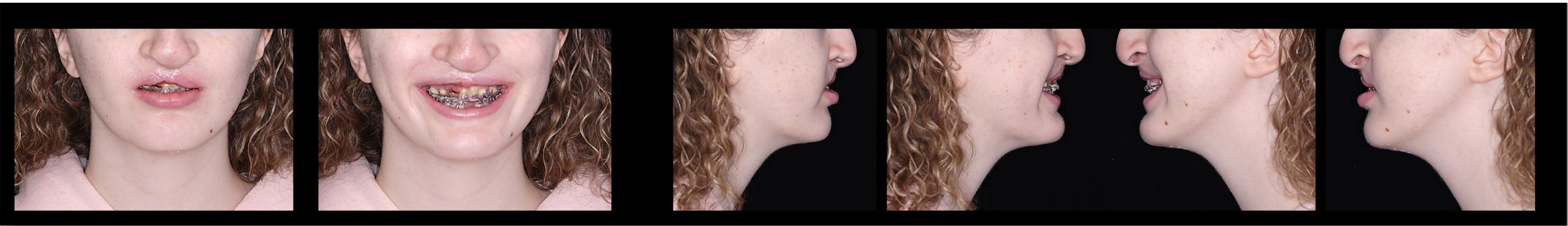


Fig 1. S.G.H., a 20-year-old female presented to the Graduate Prosthodontic Program at UNC, Chapel Hill, North Carolina, for oral rehabilitation after multiple clinical and surgical treatment. The patient was using fixed orthodontic appliances and exhibited the following findings: ectropion of the lower eyelids; bilateral skin graft repairs on both the left and right eyelids; rounded lateral canthi. Midface hypoplasia with poorly defined malar eminences bilaterally, widened nasal tip and alar bases; cleft lip repair incision; gingival display; straight profile; dental midline not aligned with the facial midline; even facial thirds; wider lateral fifths; and lip incompetence was present. The FMA measured 26 degrees, and nasolabial angle was within the normal limits.



Figs 2. Intraoral photos. Angle Class I occlusion, uneven gingival margins, oligodontia, mixed dentition, residual root on tooth #C, non-coincident maxillary and mandibular midline, gingival recession on tooth #25 (Miller's Class III). There was a high mandibular labial frenum attachment, dental fluorosis, canted occlusal plane, cleft palate surgery scar, and the palatine raphe was not aligned with the midline. Abnormalities in tooth morphology, generalized plaque accumulation and Stage II Periodontitis Grade A were present.

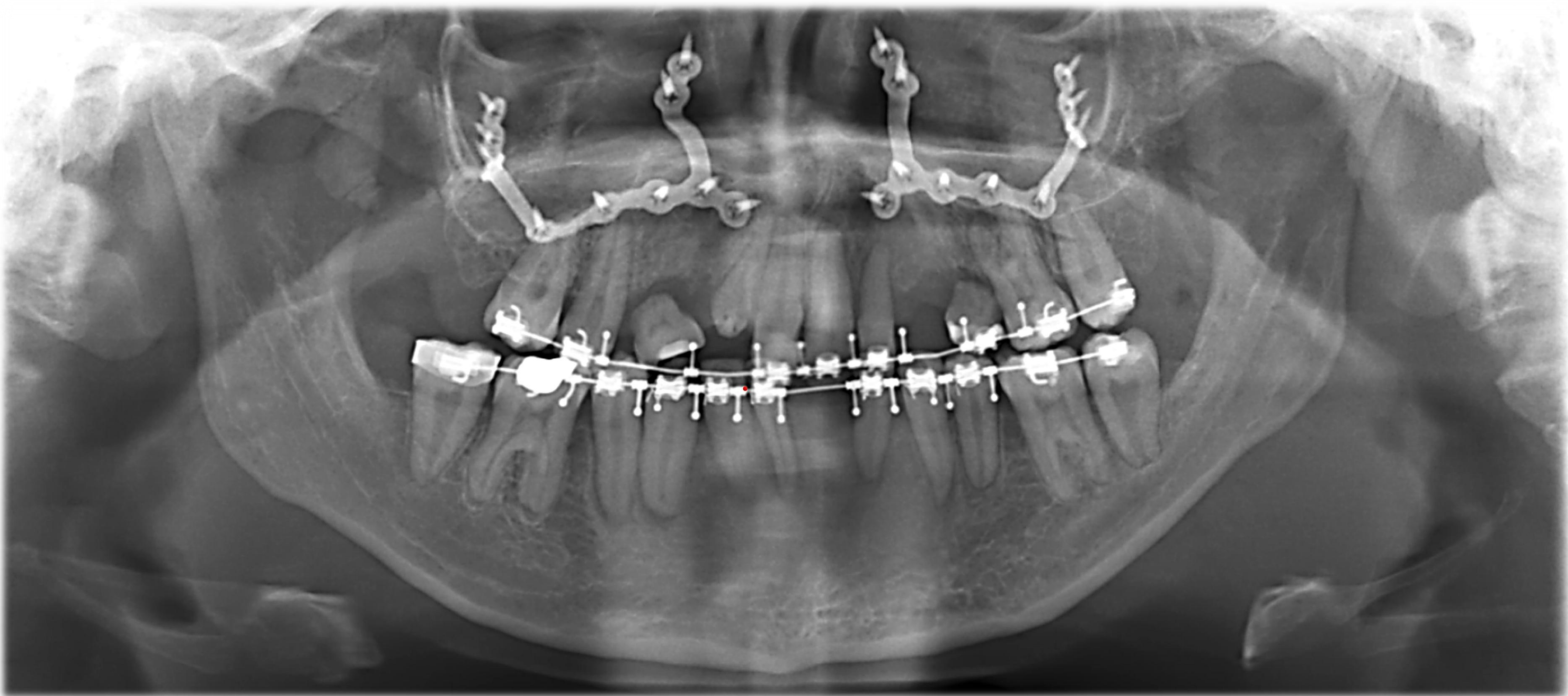


Fig 3. Panoramic radiograph. Atrophic maxillary and mandibular bones, enlarged periodontal ligament, primary teeth, *dens invaginatus* on tooth #15, taurodontism on teeth #15 and #18, unfavorable crown-to-root ratio, and evidence of rigid fixation after Le Fort I osteotomy.

Discussion

Despite BCD low prevalence, this syndrome shares characteristics with other ectodermal dysplasias and craniofacial disorders. Understanding these similarities can assist in tailoring treatment for related conditions. Prosthodontists must be prepared to address several significant challenges when treating these patients, including the presence of unfavorable root-to-crown ratios; malocclusion; poor bone quantity and uncertain quality; gingival recession; insufficient keratinized tissue; variations in tooth morphology; velopharyngeal incompetency; elevation of soft palate during phonations; and deficiencies in lip anatomy and function. When considering treatment options, removable prostheses reveals to be the primary treatment option for young patients, while implants are generally reserved for after growth has finished. A multidisciplinary approach, early intervention, and regular monitoring are crucial to optimizing functional, aesthetic, and psychosocial outcomes.

Conclusion

Managing dental prosthetics for patients with BCD syndrome is challenging. However, tailored prosthetic solutions can greatly improve the quality of life for those affected by BCD syndrome.

References

Awadh VV, Kiukkonen A, Nieminen P, Arte S, Hurmerinta K, Rice DP. Blepharocheilodontic (BCD) syndrome: New insights on craniofacial and dental features. *Am J Med Genet A*. 2017;173(4):905-913.
Ababneh FK, Al-Swaid A, Elhag A, Youssef T, Alsaif S. Blepharo-cheilo-dontic (BCD) syndrome: expanding the phenotype, case report and review of literature. *Am J Med Genet A*. 2014;164A(6):1525-1529.
Ghoumid J, Stichelboud M, Jourdain AS, et al. Blepharocheilodontic syndrome is a CDH1 pathway-related disorder due to mutations in CDH1 and CTNND1. *Genet Med*. 2017;19(9):1013-1021.
Ramos-Montiel RR, Reinoso-Quezada SJ, Astudillo-Carrera AS, Guamán-Roldán HX, Astudillo-Carrera CX, et al., (2025), Current Overview of Blepharocheilodontic Syndrome (bcd). A Systematic Review and Case Report, *J Clinical Research and Reports*. 2025;20(2).
LeBlanc S, Naveen D, Haan E, et al. CDH1-related blepharocheilodontic syndrome is associated with diffuse gastric cancer risk. *Am J Med Genet A*. 2020;182(7):1780-1784.