Abstract

Background: Osteogenesis imperfecta (OI) is a rare hereditary condition linked to mutations affecting type I collagen, leading to increased bone fragility and a tendency toward recurrent fractures. Individuals with OI frequently present with various dental abnormalities, including dentinogenesis imperfecta (DI), hypodontia, and atypical tooth eruption, that complicate the management of oral health. The literature reports structural alterations in dentin; however, findings are conflicting regarding the presence of such abnormalities in individuals without a clinical diagnosis of DI. Similarly, there are inconsistencies in the reported changes affecting enamel and the dentino-enamel junction (DEJ). Optical coherence tomography (OCT) is a non-invasive imaging technique used to visualize biological structures, and in recent years, it has been increasingly applied in the dental field. To date, no research has used OCT to analyse enamel and dentin in OI patients. The aim of this project was to create a database of paediatric OI patients to evaluate dental anomalies, including enamel and dentin changes assessed by OCT imaging, comparing these findings with an age- and sex-matched healthy control group

Material and Methods: Children aged 3 to 15 years with OI and age- and gender-matched healthy controls were examined at the Paediatric Dentistry Section of the Dental School in Turin. Dental examination and panoramic radiographs were both used to identify dental anomalies. OCT sagittal scans of the maxillary central incisors were performed to evaluate enamel thickness and mean gray value and integrated signal intensity of both enamel and dentin in two anatomical in both cervical and middle third regions of the tooth. An additional descriptive analysis was performed to assess the visibility of the DEJ.

Results: A total al of 25 OI patients (mean age: 10.7 ± 2.4 years) and 25 age- and gender-matched healthy controls (mean age: 10.3 ± 2.3 years) were consecutively enrolled.

Dental anomalies were significantly more common in OI patients than controls. Malposition, taurodontism, crown narrowing, intrinsic discoloration, and root abnormalities occurred almost exclusively in OI, showing strong statistical significance ($p \leq 0.0223$).

A total of 288 OCT images of the first maxillary incisor were analysed for 16 patients affected by OI and 306 images for 17 healthy controls. In patients with OI, the cervical third of the crown showed a significant reduction in enamel thickness (0,464 mm vs 0,528 mm; Wilcoxon $p \approx 0,018$) and , accompanied by a marked increase in dentin optical density (6605 vs 4961; Wilcoxon $p < 0,001$) and signal intensity (264731 vs 240341; Wilcoxon $p = 0,0026$), indicating structural alterations associated with the pathology. In the middle third of the crown, enamel thickness was comparable to controls, but optical properties remained reduced; differences in dentin were less pronounced. Among OI patients, the presence of DI was associated with a significant increase in dentin optical density in the cervical region. Finally, the visibility of the DEJ did not differ between groups but was correlated with dentin signal intensity.

Conclusions: Dental anomalies were significantly more prevalent in children with OI compared to healthy controls. OCT imaging revealed microstructural alterations in both enamel and dentin, with dentin showing higher optical intensity values and enamel appearing thinner with reduced signal, particularly in the cervical region. These changes were evident even without a clinical diagnosis of DI, suggesting subclinical structural compromise. Alterations at the enamel–dentin interface may impair adhesion of restorative materials, complicating dental treatment. These findings support the need for early diagnosis, regular oral health assessments, and long-term dental monitoring to optimize care and improve quality of life in OI patients.