THE IMPACT OF AMELOGENESIS IMPERFECTA ON DENTAL HYGIENE AND PSYCHOSOCIAL WELL-BEING: A PILOT STUDY

Original article

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SUMMARY

Introduction and aim: Amelogenesis imperfecta (AI) is a genetic disorder of enamel development that negatively affects the structure and appearance of teeth. The irregular surfaces of affected teeth act as plaque-retentive sites and may adversely impact periodontal health. The well-being of patients with AI is often compromised due to dental hypersensitivity and aesthetic deficits, particularly in the visible regions of the dentition. The aim of this pilot study was to evaluate the impact of AI on periodontal health, oral hygiene status, dental hypersensitivity, and patients' quality of life.

Methods: The pilot study included 20 patients (10 with AI, 10 without dental impairment), matched for age and gender. Periodontal health and oral hygiene were assessed using the Papilla Bleeding Index (PBI) and the Quigley-Hein Index (QHI). Quality of life and dental hypersensitivity were evaluated using a PROM questionnaire. Statistical analysis was performed using the Mann-Whitney U test with a significance level at 5% ($p < 0.05$).

Results: Patients with AI showed a significantly higher PBI ($p = 0.02$) and lower oral health-related quality of life according to the PROM questionnaire ($p = 0.004$). Dental hypersensitivity was markedly higher in AI patients ($p = 0.005$). Descriptive statistics indicated poorer oral hygiene levels in AI patients; however, the difference in QHI did not reach statistical significance ($p = 0.14$).

Conclusion: The study confirmed that AI negatively affects oral hygiene status and increases dental hypersensitivity, which in turn results in poorer quality of life. Although the difference in QHI between groups was not statistically significant, regular professional oral hygiene and psychosocial support remain essential. The findings highlight the need for comprehensive care in patients with AI.

Key words: amelogenesis imperfecta, dental enamel, gingivitis, dental plaque, dental hypersensitivity, OHRQoL

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INTRODUCTION

Amelogenesis imperfecta (AI) represents a group of genetically determined enamel development disorders affecting the structure and clinical appearance of all or nearly all teeth. In some forms, AI may also be associated with morphological or biochemical abnormalities in other organ systems [1]. Its prevalence varies depending on the diagnostic criteria used and the population studied, ranging from 1:700 to 1:14,000 [2, 3]. Over the years, several classifications based on phenotype or inheritance pattern have been proposed. Despite advances in molecular genetic methods and the possibility of identifying the causative gene, Witkop's classification from 1988 remains the most widely used in clinical practice [4]. This classification divides AI into four major types: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic with taurodontism. Clinical manifestations of individual forms **(Figs. 1–4)** frequently overlap, and the phenotype may significantly vary even among

members of the same family [1, 5], making accurate classification difficult in clinical practice. A summary of the clinical and radiographic findings of the individual forms is presented in **Table 1.**

Although the literature does not indicate an increased prevalence of dental caries in patients with AI [6], professional dental hygiene care and early therapeutic intervention are key components of patient management. The irregular enamel surface frequently observed in patients with AI may promote plaque retention and increase susceptibility to plaque-induced gingival inflammation **(Figs. 5 and 6)** [7, 8]. Impaired oral hygiene is further exacerbated by increased dental sensitivity, which is most pronounced in the hypocalcified form [9, 10]. Regarding periodontal health, this form compares unfavourably with the hypoplastic and hypomaturation forms [10]. Oral hygiene instruction therefore represents a fundamental component of AI patient management. Early restorative intervention, including the placement of full-coverage restorations, may further support periodontal health by providing smoother tooth surfaces, facilitating plaque control, and reducing dental hypersensitivity [11].

In addition to dental hypersensitivity, aesthetic concerns are among the primary reasons why patients with AI seek dental care. According to the literature and the authors' clinical experience, visible dental abnormalities can have a substantial psychosocial impact [12]. Patients frequently experience negative comments and unwanted attention from others, which may contribute to social withdrawal, particularly among children and adolescents, and lead to reduced self-esteem. However, these psychosocial consequences are not limited to younger individuals and may persist into adulthood. As a result, AI can become a source of stigmatization and significantly impairs the quality of life of affected individuals [12, 13].

The relationship between AI and periodontal health has not yet been sufficiently investigated. Based on the available literature, the following null and alternative hypotheses were formulated for this pilot study:

$H0_1$: There is no difference in periodontal health between patients with AI and the control group.

$H1_1$: Patients with AI have poorer periodontal health than the control group.

$H0_2$: There is no difference in dental hypersensitivity between patients with AI and the control group.

$H1_2$: Patients with AI have higher levels of dental hypersensitivity than the control group.

$H0_3$: There is no difference in oral hygiene levels between patients with AI and the control group.

$H1_3$: Patients with AI have poorer oral hygiene levels than the control group.

$H0_4$: There is no difference in oral health-related quality of life between patients with AI and the control group.

$H1_4$: Patients with AI have poorer oral health-related quality of life than the control group.

$H0_5$: Attendance at professional dental hygiene appointments has no effect on periodontal health or oral hygiene levels in patients with AI.

$H1_5$: Attendance at professional dental hygiene appointments improves periodontal health and oral hygiene levels in patients with AI.

MATERIALS AND METHODS

The pilot study was conducted in accordance with the Declaration of Helsinki (1964), as revised in 2013, at the Department of Stomatology, Third Faculty of Me-

Tab. 1 *Witkop's classification of amelogenesis imperfecta.*

Amelogenesis imperfecta form	Clinical findings	Radiological findings
Hypoplastic	Hard but thin enamel; uneven enamel thickness with pits and grooves; spacing is common.	Thin enamel with normal radiodensity; distinct dentin-enamel junction.
Hypomaturation	Porous enamel; chalky to mottled appearance (white to yellow-brown); enamel flakes off easily.	Normal enamel thickness, density similar to dentin.
Hypocalcified	Soft enamel, rapid enamel loss after eruption, teeth subsequently yellow-brown to amber in color.	Normal enamel thickness, low density, often lower than dentin.
Hypomaturation-hypoplastic with taurodontism	Combination of the clinical appearance of the hypoplastic and hypomaturation forms.	Non-homogeneous appearance. Enamel thin and dense in some areas, lower density and greater thickness in others. Taurodontism.

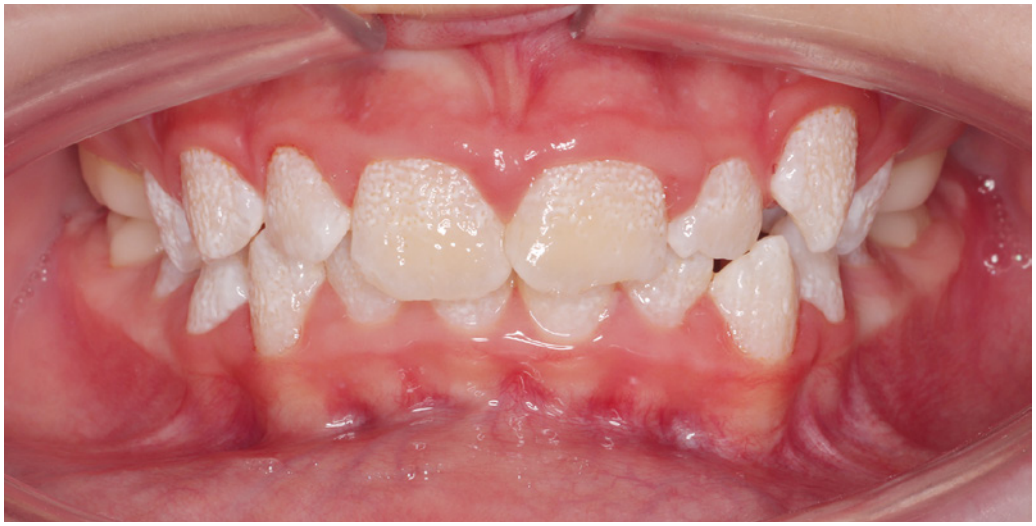

Fig. 1
Intraoral image of a patient with the hypoplastic form of amelogenesis imperfecta. The image shows a generalized distribution of pits resulting from uneven enamel formation. Archive of Department of Stomatology, Third Faculty of Medicine, Charles University, and University Hospital Kralovske Vinohrady, and Cleft Centre Prague, University Hospital Kralovske Vinohrady.

dicine, Charles University and University Hospital Královské Vinohrady. The study was approved by the Ethics Committee for Multi-Centric Clinical Trials of University Hospital Královské Vinohrady (reference number EK-VP/58/00/2025).

Study population

The study included a total of 20 participants divided into two groups. The study group consisted of patients diagnosed with amelogenesis imperfecta, regardless of the clinical subtype. Inclusion criteria were the ability to perform daily oral hygiene independently without parental assistance and the absence of dental caries. Patients with fixed full-mouth prosthetic rehabilitation and those with persistently poor oral hygiene who were unwilling to cooperate in improving their oral hygiene practices were excluded from the study. The initial AI cohort comprised 12 participants. Following the application of the exclusion criteria, the final study group consisted of 10 patients. The control group (N = 10) consisted of patients without a diagnosis of amelogenesis imperfecta treated at the same clinical institution who were independently responsible for their oral hygiene. Control participants were randomly selected from patients within the same age range as the study group.

Methods

Interdental papillary bleeding was assessed using the Papilla Bleeding Index (PBI) according to Saxer and Mühlemann [14], which evaluates the inflammatory response of gingival tissue following gentle stimulation with a WHO periodontal probe (Aesculap, Tuttlingen, Germany). Scores were recorded on a 5-point Likert scale ranging from 0 to 4, where: 0 = no bleeding; 1 = a single bleeding point; 2 = a bleeding line or several bleeding points; 3 = the interdental papillary space

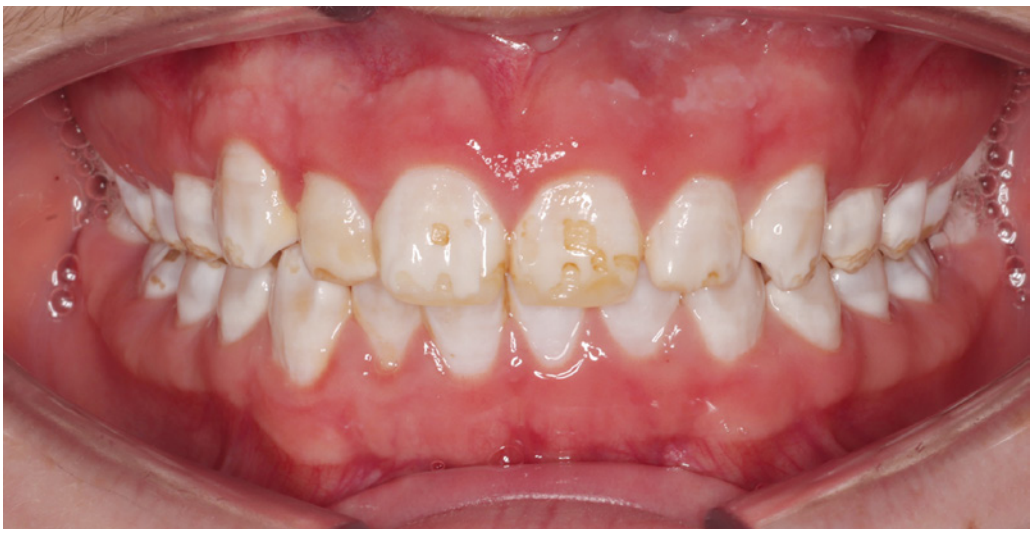

Fig. 2
Intraoral image of a patient with the hypomaturation form of amelogenesis imperfecta. A generalized opaque discoloration of the enamel with localized enamel delamination is observed. Archive of Department of Stomatology, Third Faculty of Medicine, Charles University, and University Hospital Kralovske Vinohrady, and Cleft Centre Prague, University Hospital Kralovske Vinohrady.

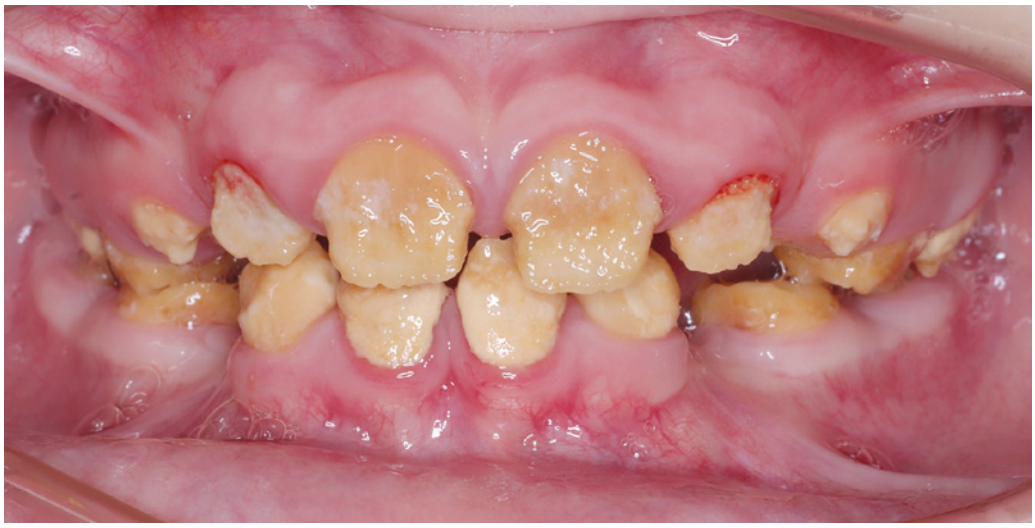

Fig. 3
Intraoral image of a patient with the hypocalcification form of amelogenesis imperfecta. Soft enamel is present, particularly in cervical regions, and is also visible on the cusp of the erupting tooth 24. The rapid loss of enamel results in amber discoloration of the teeth. Archive of Department of Stomatology, Third Faculty of Medicine, Charles University, and University Hospital Kralovske Vinohrady, and Cleft Centre Prague, University Hospital Kralovske Vinohrady.

filled with blood; and 4 = profuse bleeding extending beyond the interdental papilla.

The presence of dental plaque on the vestibular and oral tooth surfaces after staining with a plaque-disclosing agent (Curaprox PCA 260 liquid, Kriens, Switzerland) was assessed using the Quigley-Hein Plaque Index [15]. Scores were recorded on a 6-point Likert scale ranging from 0 to 5, where: 0 = no plaque; 1 = isolated flecks of plaque; 2 = a continuous band of plaque at the gingival margin; 3 = plaque covering the cervical third of the crown; 4 = plaque extending into the middle third of the crown; and 5 = plaque extending into the occlusal third of the crown.

Oral health-related quality of life (OHRQoL) and dental hypersensitivity were assessed using a Patient-Reported Outcome Measure (PROM) questionnaire. The methodology was based on the study published by Lyne et al. [16]. The PROM questionnaire consisted of ten items. Eight items were scored using a Likert scale ("often" = 0 points, "sometimes" = 1 point, "never" = 2 points), one item was dichotomous (yes = 1 point, no = 0 points), and one item was open-ended and was not scored. A complete list of questionnaire items is presented in **Table 2**. The maximum attainable score was 17 points, with higher scores indicating better self-perceived quality of life. Dental hypersensitivity was evaluated using the first questionnaire item: "Do your teeth cause you pain or sensitivity?" Lower scores for this item corresponded to greater dental hypersensitivity. The questionnaire was completed digitally using Google Forms (Google LLC, Mountain View, CA, USA). Participants completed the questionnaire in a quiet environment and were given the opportunity to ask additional questions if any item required clarification.

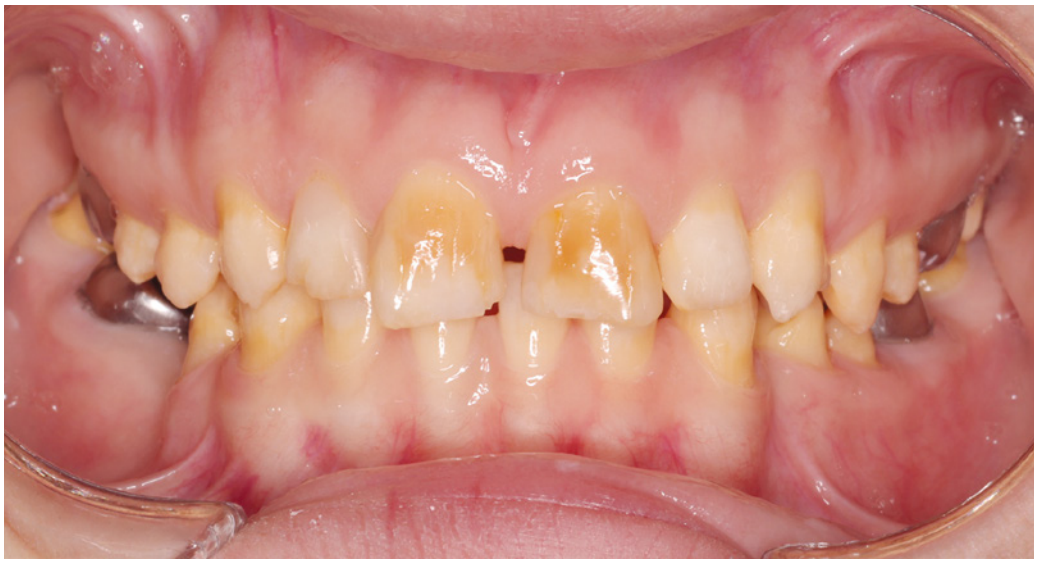

Fig. 4
Intraoral image of a patient with the combined form of amelogenesis imperfecta. The teeth are smaller and enamel discoloration consistent with hypomaturation defects is visible. Archive of Department of Stomatology, Third Faculty of Medicine, Charles University, and University Hospital Kralovske Vinohrady, and Cleft Centre Prague, University Hospital Kralovske Vinohrady.

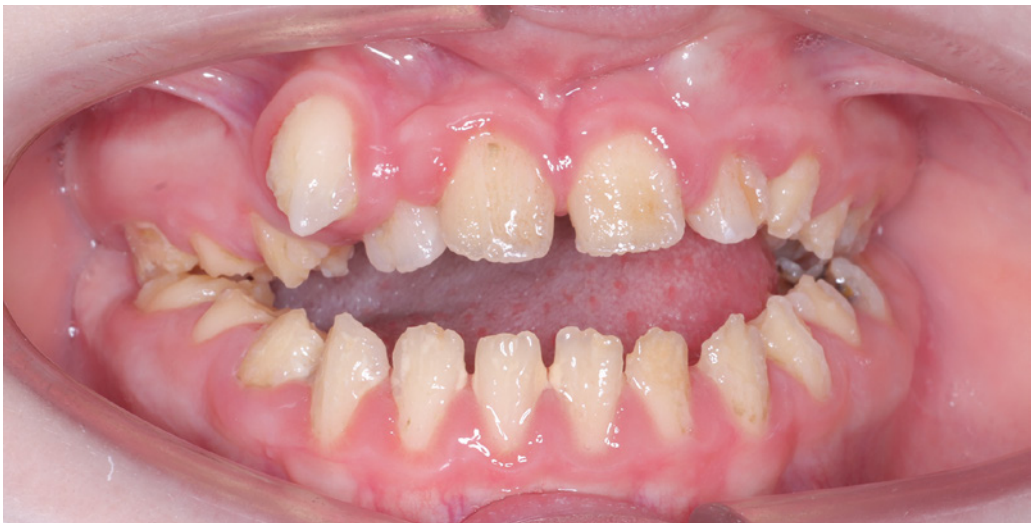

Fig. 5
Intraoral image of a patient with amelogenesis imperfecta, showing dental calculus and gingivitis in the mandibular anterior region.
Archive of Department of Stomatology, Third Faculty of Medicine, Charles University, and University Hospital Kralovske Vinohrady, and Cleft Centre Prague, University Hospital Kralovske Vinohrady.

Statistical analysis

Data were analysed using JASP software (version 0.18.3; University of Amsterdam, the Netherlands). Due to the small sample size and the violation of the assumptions of normal data distribution, comparisons between the two independent groups (patients with AI and the control group) were performed using the non-parametric Mann–Whitney U test.

Results are presented as medians (Mdn), means (M), and standard deviations (SD). Statistical significance was set at $\alpha = 0.05$. Effect size was expressed using the rank-biserial correlation coefficient (r), where values of approximately 0.1 indicate a small effect, approximately 0.3 a medium effect, and ≥ 0.5 a large effect.

RESULTS

Sociodemographic characteristics

The study population consisted of 20 participants aged 11 to 31 years (M = 16.40; SD = 5.21). Of the total sample, 13 were female and seven were male. The control group comprised 10 individuals (age: M = 16.60 years; SD = 4.99), including seven females and three males. This group consisted of nine students and one employed individual. The AI group also comprised 10 individuals (age: M = 16.20 years; SD = 5.67), including six females and four males. Within the AI group, eight participants were students and two were employed. During the six months preceding the examination, five participants in the AI group and one participant in the control group had attended professional dental hygiene appointments. A summary of all sociodemographic characteristics is presented in **Table 3**.

All major clinical forms of AI were represented in the AI group **(Table 4)**. Owing to the small number of participants within each AI subtype, separate statistical analyses of these subgroups were not feasible.

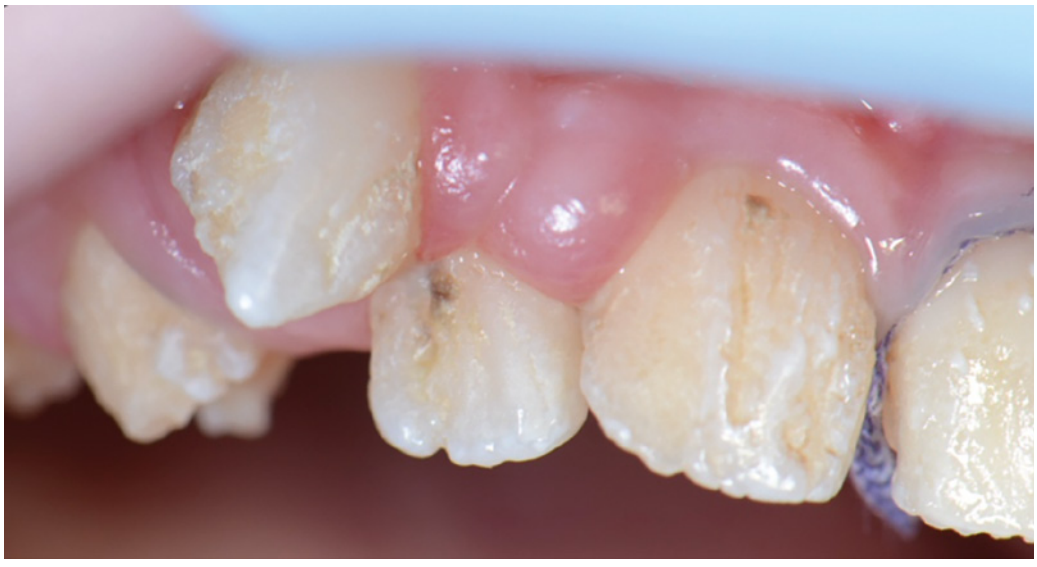

Fig. 6
Detailed image confirming the presence of dental calculus and plaque accumulated in surface irregularities.
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Tab. 2 *PROM questionnaire for assessing OHRQoL.*

Question	Answers		
Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never
Do you have difficulty eating foods you would like to eat because of your teeth?	Often	Sometimes	Never
Does it hurt when you brush your teeth?	Often	Sometimes	Never
Do you miss school or work because of your teeth (excluding dental appointments)?	Often	Sometimes	Never
Do you feel unhappy with the way your teeth look?	Often	Sometimes	Never
Do your teeth affect your confidence to smile?	Often	Sometimes	Never
Do you get teased or bullied because of your teeth?	Often	Sometimes	Never
Do you feel scared or anxious about having dental treatment?	Often	Sometimes	Never
Are you happy with your teeth?	Yes		No
Is there anything else you would like us to know about your teeth and how they affect you?			

Papilla Bleeding Index (PBI)

Patients with AI exhibited significantly higher PBI scores than the control group (MdnAI = 1.24; MdnCo= 0.38; p = 0.02, r = −0.54).

Quigley-Hein Index (QHI)

Patients with AI showed a slightly higher median QHI score. However, assessment of oral hygiene using the QHI revealed no statistically significant difference between the AI and control groups (MdnAI = 1.71; MdnCo= 1.49; p = 0.14, r = −0.30).

Effect of professional dental hygiene in the AI group

Secondary data analysis suggested that patients with AI who had attended professional dental hygiene appointments within the previous six months demonstrated lower PBI values (MDH = 0.92 vs. M_{NoDH} = 1.68) and lower QHI scores (M_{DH} = 1.70 vs. M_{NoDH} = 2.07) than those who had not attended such appointments. However, these differences did not reach statistical significance (PBI: p = 0.08; QHI: p = 0.21).

Oral Health-Related Quality of Life (OHRQoL)

Assessment using the PROM questionnaire demonstrated that patients with AI reported significantly poorer oral health-related quality of life than the control group (Md_{nAI} = 9.5; Md_{nCo} = 14.5; p = 0.004, r = 0.71).

Dental hypersensitivity (DHS)

Patients with AI demonstrated a significantly lower median DHS score (Md_{nAI} = 0.0) than the control group (Md_{nCo} = 0.5; p = 0.005, r = 0.62).

DISCUSSION

The aim of this study was to determine whether amelogenesis imperfecta affects periodontal health, oral hygiene status, dental hypersensitivity, and oral health-related quality of life.

The effect of AI on periodontal health and oral hygiene status was assessed using the widely accepted PBI and QHI indices. The results demonstrated a significantly higher prevalence of gingival inflammation in patients with AI compared with the control group. This finding is consistent with a previously

Tab. 3 *Sociodemographic profile of study participants.*

Characteristics	AI group (N = 10)	Control group (N = 10)	Total (N = 20)
Gender			
Female	6 (60.0 %)	7 (70.0 %)	13 (65.0 %)
Male	4 (40.0 %)	3 (30.0 %)	7 (35.0 %)
Employment			
Student	8 (80.0 %)	9 (90.0 %)	17 (85.0 %)
Employed	2 (20.0 %)	1 (10.0 %)	3 (15.0 %)
DH appointment*	5 (50.0 %)	1 (10.0 %)	6 (30.0 %)

*Attendance at a professional dental hygiene appointment within the previous six months.

Tab. 4 *Distribution of amelogenesis imperfecta types.*

AI form	Hypoplastic	Hypomaturation	Hypocalcified	Mixed
Number	2	2	3	3

published study in which Quandalle et al. [10] additionally reported poorer oral hygiene among patients with the hypocalcified form of AI. No statistically significant difference was observed in the QHI, which assesses dental plaque accumulation, although descriptive statistics indicated poorer oral hygiene in the AI group. One possible explanation for these less favourable findings is the irregular enamel surface commonly observed in AI, which may serve as a plaque-retentive factor. Lundgren et al. [11] reported a reduction in gingival bleeding following the placement of protective full-coverage restorations, which provide smoother tooth surfaces and reduce dental hypersensitivity. The lack of a statistically significant difference in QHI between the study and control groups may be explained by the fact that some patients with AI had already undergone partial restorative rehabilitation using composite build-ups or full-coverage restorations. Ceyhan et al. [17] demonstrated better oral hygiene among patients who attended dental appointments more frequently. The present study supports these findings, as patients with AI who had attended professional dental hygiene appointments within the previous six months exhibited lower PBI and QHI values, although these differences did not reach statistical significance due to the limited sample size.

Dental hypersensitivity and oral health-related quality of life were assessed using the PROM questionnaire. Dental hypersensitivity, which has frequently been reported in association with AI [5, 9, 10, 16, 18, 19], may reduce the effectiveness of oral hygiene practices and thereby contribute to poorer periodontal health. Furthermore, hypersensitivity may adversely affect patients' quality of life. The findings of the present study demonstrated significantly greater dental hypersensitivity among patients with AI compared with controls, which is consistent with previously published literature. The psychosocial consequences of AI, including feelings of social isolation and reduced self-esteem, were also confirmed [16]. This aspect is important for comprehensive patient care, as quality of life may be substantially affected not only by aesthetic concerns but also by psychological factors. Moreover, some studies have reported negative experiences among patients whose healthcare providers lacked sufficient knowledge of AI and failed to understand the challenges associated with the condition [20]. Therefore, awareness of AI among dental professionals may represent an important factor influencing oral health-related quality of life.

Based on the results of this pilot study, the individual hypotheses may be evaluated as follows. The hypothesis that AI adversely affects periodontal health ($H1_1$) was confirmed. The hypothesis that patients with AI experience increased dental hypersensitivity ($H1_2$) was also confirmed. The hypothesis regarding poorer oral hygiene status in patients with AI ($H1_3$) was not confirmed, as the difference in QHI scores between groups did not reach statistical significance, although descriptive statistics suggested less favourable oral hygiene among patients with AI.

Tab. 5 *Summary of Mann-Whitney U test results and descriptive statistics.*

Measurement tool	AI group		Control group		W	p	r
	M (SD)	Mdn	M (SD)	Mdn			
PBI	1.30 (0.86)	1.24	0.56 (0.53)	0.38	23.00	0.02 *	–0.54
QHI	1.88 (0.56)	1.71	1.64 (0.53)	1.49	35.00	0.14	–0.30
PROM	9.70 (3.83)	9.50	14.40 (2.17)	14.50	85.50	0.004 *	0.71
DHS	0.30 (0.48)	0.00	0.50 (0.53)	0.50	81.00	0.005 *	0.62

Note: PBI – Papilla Bleeding Index; QHI – Quigley-Hein Index; PROM – Patient-Reported Outcome Measure; DHS – Dental hypersensitivity. r – rank biserial correlation coefficient; *p < 0.05 statistically significant

The hypothesis concerning the negative impact of AI on oral health-related quality of life ($H1_4$) was confirmed. The hypothesis regarding the effect of professional dental hygiene attendance on the evaluated parameters ($H1_5$) could not be statistically confirmed because of the limited sample size, although the results indicated a trend towards improved oral hygiene and periodontal parameters among patients who had attended professional dental hygiene appointments.

The limitations of this pilot study include the small sample size and the potential for bias associated with self-reported questionnaire data. This limitation was partially mitigated by allowing participants to ask additional questions whenever clarification was needed. Another limitation is the inability to analyse individual AI subtypes separately because of the limited number of participants in each subgroup. A further limitation concerns the assessment of the relationship between professional dental hygiene attendance and the evaluated parameters. The analysis considered only whether participants had attended a professional dental hygiene appointment within the previous six months, without accounting for the exact timing of the visit or the frequency of recall appointments. This variability may have influenced the interpretation of the results.

Future research should include larger study populations and longer follow-up periods, complemented by an evaluation of the effects of therapeutic interventions. Furthermore, the study methodology should be refined to address the limitations identified in the present study, particularly by recording the frequency and timing of professional dental hygiene visits more precisely and by enabling separate analyses of individual AI types.

CONCLUSION

This pilot study suggests that patients with amelogenesis imperfecta may exhibit a higher prevalence of gingival inflammation, increased dental hypersensitivity, and poorer oral health-related quality of life compared with healthy controls. Although differences in plaque accumulation measured using the Quigley–Hein Index were not statistically significant, descriptive statistics indicated poorer oral hygiene among patients with AI.

Given the small sample size and the heterogeneity of the included AI subtypes, these findings should be considered preliminary. The results highlight the need for larger studies to improve understanding of the relationship between individual forms of AI, periodontal health, and the psychosocial impact of the condition, as well as to confirm the observed trends in a broader population of patients with AI.

Abbreviations

AI	Amelogenesis imperfecta
DHS	Dental hypersensitivity
OHRQoL	Oral Health-Related Quality of Life
PBI	Papilla Bleeding Index
PROM	Patient-Reported Outcome Measure
QHI	Quigley-Hein Index

Conflict of interest declaration

The authors declare that they have no conflict of interest regarding the issues investigated in this original article.

Author Contributions

DV: Conceptualization, Writing – original draft.
KV: Investigation, Data curation.
WU: Methodology, Validation.
JV: Formal analysis, Writing – review & editing.
JB: Formal analysis, Validation.
PP: Writing – review & editing, Supervision.

Declaration on the use of artificial intelligence

During the preparation of this manuscript, the artificial intelligence tool ChatGPT (OpenAI, San Francisco, CA, USA) was used exclusively for language editing and stylistic refinement of the text. No part of the manuscript content, including the text, figures, or tables, was generated by artificial intelligence. The authors take full responsibility for the content of the manuscript.

Data availability

The data are available from the corresponding author upon justified request.

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