Amelogenesis Imperfecta Prevalence in a Colombian Population: A Retrospective Study*

Prevalencia de la amelogénesis imperfecta en una población colombiana: estudio retrospectivo

Prevalência de amelogênese imperfeita em uma população colombiana: estudo retrospectivo

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*Original research.

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ABSTRACT

Background: Amelogenesis imperfecta (AI) is a hereditary condition that affects the structure of tooth enamel and causes sensitivity, predisposition to cavities, and psychological problems. In

Colombia, its frequency, magnitude, distribution, and behavior are unknown, so it is necessary to carry out prevalence studies to implement preventive actions. **Purpose**: To determine the prevalence of AI in patients who have attended the Pontificia Universidad Javeriana clinics in Bogotá. **Methods**: A retrospective cross-sectional observational study was carried out, whose sample included 1,394 medical records of patients who attended between January 2015 and December 2017. **Results**: The prevalence of AI was 0.6 %, corresponding to 8 people affected, 4 men and 4 women between the ages of 9 and 10 years. The most frequent phenotype was hypoplastic in 7 patients (87.5 %) and one person had a hypocalcified phenotype (12.5 %). Taurodontism was the most frequent anomaly in the 8 patients (100 %). Seven of the eight patients (87.5 %) had a family history of AI. All the individuals had a lower-middle socioeconomic level and came from urban areas. **Conclusions**: This study is the first approximation to determine the prevalence of AI in a group of the Colombian population. Although the prevalence was low, it is comparable with the findings of other studies.

Keywords

amelogenesis imperfecta; Colombia; dental anomalies; dentistry; enamel defects; epidemiology; genetics; prevalence; retrospective

RESUMEN

Antecedentes: La amelogénesis imperfecta (AI) es una condición hereditaria que afecta la estructura del esmalte dental y ocasiona sensibilidad, predisposición a la caries y problemas psicológicos. En Colombia, se desconoce su frecuencia, magnitud, distribución y comportamiento, por lo que es necesario realizar estudios de prevalencia para implementar acciones preventivas. **Objetivo:** Determinar la prevalencia de AI en pacientes que han asistido a las clínicas de la Pontificia Universidad Javeriana de Bogotá. **Métodos:** Se realizó un estudio observacional descriptivo de corte transversal retrospectivo, cuya muestra incluyó 1.394 historias clínicas de pacientes que acudieron entre enero de 2015 y diciembre de 2017. **Resultados:** La prevalencia de AI fue del 0,6 %, correspondiente a 8 personas afectadas, 4 hombres y 4 mujeres con edades entre 9 y 10 años. El fenotipo más frecuente fue el hipoplásico en 7 pacientes (87,5 %) y una persona con fenotipo hipocalcificado (12,5 %). El taurodontismo fue la anomalía más frecuente en los 8 pacientes (100 %). Siete de los ocho pacientes (87,5 %) tenían antecedentes familiares de AI. Todos los individuos tenían un nivel socioeconómico medio-bajo y procedían de zonas urbanas. **Conclusiones:** Este estudio es la primera aproximación para determinar la prevalencia de AI en un grupo de la población colombiana. Aunque la prevalencia fue baja, es comparable con los hallazgos de otros estudios.

Palabras clave

amelogénesis imperfecta; anomalías dentarias; Colombia; defectos del esmalte dental; epidemiología; genética; odontología; prevalencia; retrospectivo

RESUMO

Antecedentes: Amelogênese imperfeita (AI) é uma condição hereditária que afeta a estrutura do esmalte dentário e causa sensibilidade, predisposição a cáries e problemas psicológicos. Na Colômbia, sua frequência, magnitude, distribuição e comportamento são desconhecidos, por isso é necessário realizar estudos de prevalência para implementar ações preventivas. **Objetivo**: Determinar a prevalência de AI em pacientes atendidos nas clínicas da Pontificia Universidade Javeriana de Bogotá. **Métodos**: Foi realizado um estudo transversal observacional descritivo retrospectivo, cuja amostra incluiu 1.394 prontuários de pacientes atendidos entre janeiro de 2015 e dezembro de 2017. **Resultados**: A prevalência de AI foi de 0,6 %, correspondendo a 8 afetados, 4 homens e 4 mulheres com idade entre 9 e 10 anos. O fenótipo mais frequente foi hipoplásico em 7 pacientes (87,5%) e uma pessoa com fenótipo hipocalcificado (12,5 %). O taurodontismo foi a anomalia mais frequente nos 8 pacientes (100 %). Sete dos oito pacientes (87,5 %) tinham história familiar de AI. Todos os indivíduos possuíam nível socioeconômico médio-baixo e procediam de áreas urbanas. **Conclusões**: Este estudo é a primeira aproximação para determinar a prevalência de AI em um grupo da população colombiana. Embora a prevalência tenha sido baixa, é comparável aos achados de outros estudos.

Palavras-chave

amelogênese imperfeita; anormalidades dentárias; Colombia; defeitos do esmalte dentário; epidemiologia; genética; odontologia; prevalência; retrospectivo

INTRODUCTION

Enamel is a highly mineralized dental tissue, organized, and structured in hydroxyapatite crystals. It is secreted by ameloblasts in a protein-containing organic matrix. Because it is an acellular tissue, enamel does not have the capacity to repair itself when it is altered extrinsically or intrinsically (1). Alterations of enamel development can cause amelogenesis imperfecta (AI), a widespread hereditary condition that affects the structure of dental enamel of deciduous and permanent dentitions in quantity and quality (2).

The color of the teeth in people with AI varies according to the phenotype and occurs in different shades from light yellow to brown. Patients experience high sensitivity to thermal changes during the intake of beverages and food, which does not allow them to have adequate oral hygiene habits, causing a greater predisposition to cavities, dental fractures, and tooth loss (3). AI patients, due to aesthetic alterations, may suffer from low self-esteem (2). AI is caused by mutations in genes such as *AMEL* (amelogenin), *ENAM* (enamelin), *MMP20* (matrix metalloproteinase-20), and *KLK4* (kallikrein-4), which code for proteins involved in the formation of tooth enamel (4-8), and *FAM83H*, a gene that codes for a protein apparently involved in the organization of ameloblast desmosomes (9). AI has also been associated with other clinical alterations such as dental agenesis, taurodontism, and anterior open bite, and systemic alterations such as nephrocalcinosis, tricho-dento-osseous syndrome and Jalili syndrome (10-13).

AI is classified according to its phenotype and mode of inheritance into 14 subtypes. Based on the phenotype, it is classified as hypoplastic, hypocalcified, hypomadurative, and hypomadurative-hypoplastic with taurodontism. According to its inheritance pattern, AI is classified as autosomal dominant, autosomal recessive, and linked to the X chromosome (14). Radiographic examination

of some phenotypes may show lack of contrast between enamel and dentin. It is also possible to observe some associated anomalies that are not clinically visible, such as pulp calcifications, dental agenesis, and apical dilacerations (15).

Research on the prevalence of AI is limited. In the international context, studies carried out in Switzerland by Backman and Holm (1986) stand out. They found 1:700 individuals affected with AI (16). The proportion of impacts was 1:14,000-1:16,000 in the United States (14), 1:8,000 in Israel (Jerusalem) (17) and 1:10,000 in Pakistan (18).

In Colombia, clinical and molecular studies of AI in families have been carried out. Different phenotypic, dental, and skeletal characteristics have been found, as well as genes involved with this anomaly (19-23). However, to date, prevalence studies have been limited to describing developmental enamel defects, such as hypoplasia, hypomineralization, and opacities (24-27). Even in the 4th National Oral Health Study (ENSAB IV, 2013-2014) (28), AI was not specifically reported. For this reason, it is relevant to know the frequency, magnitude, and distribution of this pathology and its behavior in the different regions of the country. The research question of this study was: What is the prevalence of AI in Colombian patients who attended the dental clinics of the Pontificia Universidad Javeriana from Bogotá, between January 2015 and December 2017? This prevalence study is the first one carried out in Colombia on AI. The findings provide initial evidence to plan preventive actions for caries, periodontal disease, and associated complications, and corrective actions to improve functionality and aesthetics (16). They also represent the basis to carry out similar studies in other Colombian regions.

MATERIALS AND METHODS

This is a descriptive study with retrospective cross-sectional observational design. The study was approved by the Ethics and Research Committee of the Pontificia Universidad Javeriana's Dental School, through Resolution OD-0218 of 28 July 2017, minute 010B. It adheres to the standards of Resolution 008430 of 1993 and Resolution 2378 of 2008, according to which it was classified as a risk-free study.

Population and Sample

The reference population was taken from the medical records of patients treated at the undergraduate and graduate clinics of the Pontificia Universidad Javeriana's Dental School, between January 2015 and December 2017. The sample size was determined with a confidence of the 95 % and 80 % power. It started by assuming an unknown prevalence (50%), since the reported prevalence of the event of 1:14,000 comes from an Anglo-Saxon population (14). In accordance with this, a sample of 1,394 medical records was taken.

Clinical records of patients 5 years of age and older were included, taking into account that AI occurs in both dentitions. Likewise, criteria such as completion of the clinical records, their approval by the instructor in charge, informed consents filled out and signed, and radiographic examinations were included. The medical records of patients with syndromes and those of patients diagnosed with total absence of teeth (anodontia) were excluded.

AI-related variables analyzed as dependent variables, in terms of presence and absence, followed Witkop (1988) (14) classification criteria (Table 1). Sociodemographic factors such as age, sex, and origin were considered as independent variables. Similarly, radiographic findings related to taurodontism, enamel-dentin contrast, dental agenesis, presence of microdontia, macrodontia and pulp calcifications were included. The presence or absence of a family history of AI and other alterations of tooth structure such as hypoplasias were evaluated as independent variables. The genetic factor and the consumption of any antibiotic during childhood were taken into account as confounding variables.

Table 1
AI Classification according to Witkop & Sauk, 1988*

Type	Factors
I	**Hypoplastic**
IA	Hypoplastic, fossae, autosomal dominant
IB	Hypoplastic, localized, autosomal dominant
IC	Hypoplastic, smooth, autosomal dominant
I-D	Hypoplastic, smooth, linked to X
IE	Hypoplastic, rough, autosomal dominant
IF	Enamel agenesis, autosomal recessive
II	**Hypomaturative**
IIA	Hypomaturative, pigmented, autosomal recessive
II B	Hypomaturative, X-linked, recessive
II C	Mottled (snow covered tooth), autosomal dominant
III	**Hypocalcified**
IIIA	Hypocalcified, autosomal dominant
IIIB	Hypocalcified, autosomal recessive
IV	**Hypomadurative-hypoplastic with taurodontism**
IVA	Hypomadurative-hypoplastic with taurodontism, autosomal dominant
IVB	Hypomadurative-hypoplastic with taurodontism, autosomal recessive

* This classification system is the most widely accepted to date, which considers the inheritance pattern of the disorder, as well as its specific phenotypic characteristics.

Training Process for Diagnostic Unification

With prior authorization of access to reviewing clinical records and respecting the confidentiality of diagnoses and paraclinical examinations, medical records were randomly selected, using the Microsoft Excel 2016® selection program. The examiner (graduate students) training process in identifying the diagnosis of AI was carried out in three stages: homogenization of the theoretical criteria; observation; and discussion of them. This process was carried out using digital photographs and panoramic radiographs of children and adolescents with AI, on which each phenotype and mode of inheritance of this alteration was analyzed and described based on the Witkop 1988 classification (14) (Table 1). Subsequently, the homogeneity of the observation was determined by means of files selected by the main researcher (training, Universidad de Chile, 2010), in which 50 panoramic images and radiographs of patients with AI were analyzed, thus evaluating the similarity and variability from what was observed among the researchers in this study. Of the 1,394 records reviewed for this study, 5 patients with a previous diagnosis of AI were reevaluated.

Data Collection from Medical Records

Data collected from the medical records were based on both the odontogram and the final diagnosis and was recorded in tailored formats designed for this study (Figures 1 and 2). Afterwards, the data obtained were digitized into a database designed in Excel 2016. The data analysis was performed through the IBM® SPSS Statistics® V22 program. To determine the prevalence of AI, as well as to estimate the percentage of occurrence of other dental alterations, a descriptive analysis was carried out. An inferential analysis according to sex, age by decade, and origin in relation to the presence of other dental anomalies was carried out, using the Chi-square test / Fisher's exact test ($p < 0.05$).

Figure 1
Clinical Record Format for AI designed for this Study

Code _____

Name _____
Identification _____
University _____
Date of Birth _____
Sex _____
Socioeconomic level _____
Origin _____
Address _____
Phone _____
AGE _____

Enamel structure alteration

_____
Antibiotic intake during childhood

_____

Family background of AI?

_____
Diagnosis of AI YES NO
Phenotype of AI _____

Intraoral photographs SI NO

Figure 2
Radiographic Assessment Form for AI (designed for this study)

Code _____

Name _____
Identification _____
University _____

	Presence	Absence	Location
Enamel-Dentin Density			

Tooth Agenesis			
Taurodontism			
Microdontia/macrodontia			
Pulp calcification			

RESULTS

Clinical Record Data Analysis

Prevalence. The prevalence of AI in patients who attended the dental clinics of Pontificia Universidad Javeriana from Bogotá was assessed between January 2015 and December 2017. Of the total medical records (n = 1,394), 8 were found with a diagnosis of AI (0.6 %) (Table 2). Of the 8 people diagnosed with AI, 7 had a hypoplastic phenotype and 1 had a hypocalcified one.

Table 2
Prevalence of AI and Associated Dental Anomalies in the Population Studied (Guide Chart)

Dental Anomalies	**n**	**%**
AI	8	0,6
Fluorosis-like hypomineralization	6	0,4
Hypocalcification	3	0,2
Enamel-dentin contrast	11	0,8
Dental agenesis	8	0,6
Taurodontism	59	4,2
Microdontia	11	0,8
Macrodontia	8	0,6
Pulp calcification	0	0,0
Incisor-molar hypomineralization syndrome	0	0,0
Total (N)	1.394	100,0

Source: the authors.

Sociodemographic Characteristics

Among the sociodemographic characteristics of the analyzed population, 55.31 % were female (n = 771) and 44.69% (n = 623) male. Regarding patients diagnosed with AI, based on the Witkop classification (1988) (14) (table 1), no statistically significant differences or trends were observed with respect to sex (1:1 ratio) (Table 3).

Table 3
Distribution by Sex of Dental Anomalies associated with AI

Alteration	**Presence**	**Sex**				**p**
		Female		**Male**		
		n	**%**	**n**	**%**	
Fluorosis	No	770	55,5	618	44,5	0,056
	Yes	1	16,7	5	83,3	
Hypoplasia	No	736	56,0	579	44,0	0,043*
	Yes	35	44,3	44	55,7	

Hypocalcification	No	769	55,3	621	44,7	0,444
	Yes	1	33,3	2	66,7	
AI	No	766	55,3	618	44,7	0,762
	Yes	4	50,0	4	50,0	
Enamel-dentin contrast	No	764	55,4	616	44,6	0,207
	Yes	4	36,4	7	63,6	
Dental agenesis	No	765	55,4	617	44,6	0,085
	Yes	2	25,0	6	75,0	
Taurodontism	No	739	55,5	592	44,5	0,223
	Yes	28	47,5	31	52,5	
Microdontia	No	763	55,4	615	44,6	0,207
	Yes	4	36,4	7	63,6	
Macrodontia	No	765	55,4	616	44,6	0,085
	Yes	2	25,0	6	75,0	

Regarding the origin, 7 patients (87.50 %) who presented AI came from the urban area and one from the dispersed rural area. Regarding the age distribution, the subjects affected by AI were in the age range of 10-19 years (87.5 %; n = 7) (Table 4).

Table 4
Distribution of Dental Pathologies analyzed by Age

Age (years)	Fluorosis		Hypoplasia		AI		Enamel-dentin contrast		Dental agenesis		Taurodontism	
	n	%	n	%	n	%	n	%	n	%	n	%
< 9	0	0,0	12	15,4	1	14,3	1	10,0	1	12,5	24	41,4
10 a 19	3	50,0	34	43,6	7	87,5	6	60,0	5	62,5	22	37,9
20 a 29	3	50,0	25	32,1	0	0,0	1	10,0	2	25,0	8	13,8
30 a 39	0	0,0	5	6,4	0	0,0	2	20,0	0	0,0	4	6,9
40 a 49	0	0,0	1	1,3	0	0,0	0	0,0	0	0,0	0	0,0
50 a 59	0	0,0	1	1,3	0	0,0	0	0,0	0	0,0	0	0,0

Dental Anomalies

In the population analyzed, the 8 patients with a diagnosis of AI (0.6 %) had a higher frequency of dental anomalies related to enamel hypoplasia (n = 7; 87.50 %), according to which a lack of contrast between enamel and dentin in 6 (75 %) and taurodontism in all (100 %). Alterations such as dental agenesis, microdontia, macrodontia, or pulp calcifications were not described (Table 5). On the other side, the population had a higher frequency of dental anomalies related to enamel hypoplasia in 5.7 % and taurodontism in 4.2 %.

Table 5
Distribution of Dental Conditions in Presence of AI

Characteristic	Presence	Absence		Presence		p
		n	%	n	%	
Hypoplasia	No	1312	94,80	1	12,50	0,001*
	Yes	72	5,20	7	87,50	
Hypocalcification	No	1380	99,78	8	100,00	0,895

	Yes	3	0,22	0	0,00	
Enamel-dentin contrast	No	1376	99,64	2	25,00	0,001*
	Yes	5	0,36	6	75,00	
Dental agenesis	No	1372	99,42	8	100,00	0,829
	Yes	8	0,58	0	0,00	
Taurodontism	No	1329	96,30	0	0,00	0,001*
	Yes	51	3,70	8	100,00	
Microdontia	No	1368	99,20	8	100,00	0,800
	Yes	11	0,80	0	0,00	
Macrodontia	No	1371	99,42	8	100,00	0,829
	Yes	8	0,58	0	0,00	

*Statistically significant differences.
Source: the authors.

Radiographic Characteristics

The analysis of radiographic characteristics in patients with AI showed lack of contrast between enamel and dentin in 6 patients (75 %) and taurodontism in 8 (100 %). No dental agenesis or alterations in tooth size (microdontia or macrodontia) were observed (Table 6). In the population of study, taurodontism was the most frequent anomaly (3.7%), although it was not statistically significant.

Table 6
Distribution of Radiographic Characteristics Assessed Regarding Presence of AI

Characteristic	Presence	Absence		Presence		p
		n	%	n	%	
Enamel-dentin contrast	No	1376	99,64	2	25,00	0,001*
	Yes	5	0,36	6	75,00	
Dental agenesis	No	1372	99,42	8	100,00	0,829
	Yes	8	0,58	0	0,00	
Taurodontism	No	1329	96,30	0	0,00	0,001*
	Yes	51	3,70	8	100,00	
Microdontia	No	1368	99,20	8	100,00	0,800
	Yes	11	0,80	0	0,00	
Macrodontia	No	1371	99,42	8	100,00	0,829
	Yes	8	0,58	0	0,00	

*Statistically significant differences
Source: the authors.

Other findings observed among people diagnosed with AI included one (12.5 %) with a history of antibiotic intake in their first year of life. Likewise, 62.5 % reported having a family history of AI (Table 7).

Table 7
History of Antibiotic Intake and AI in the Family Regarding Present Pathologies (Percentages)

Condition	History of Antibiotics	History of AI
Hypoplasia	1,3	5,06
AI	12,5	62,50
Enamel-dentin contrast	9,1	45,45

Taurodontism	1,7	10,17

Source: the authors

DISCUSSION

This study aimed to determine the prevalence of AI in patients who attended the clinics of the Pontificia Universidad Javeriana' Dental School from Bogotá, between January 2015 and December 2017. 1,394 medical records were analyzed. To date, few studies describe the prevalence of AI in some populations (14,16-18). Shokri et al. (2014) (18) evaluated the prevalence of all types of dental anomalies in 1,640 Iranians, including AI, aged between 7 and 35 years, by means of panoramic radiographs. They found a prevalence of AI of 0.24 %, that is, 4 individuals.

Gupta et al. (2011) (29) reported a prevalence of 0.27 % in 3 people in the Indian population. More recently, Harini & Don (2019) (30), in the same population, reported a prevalence of 0.6 %. In the present study, the prevalence of AI in the Colombian population was also 0.6 %. Such data coincide with those found by Witkop et al. (1988), Shokri et al. (2014), and Gupta et al. (2014) (14,18,29). However, the prevalence is lower than that described in studies from Switzerland (16) and Jerusalem (17).

Regarding age, it was found that AI was diagnosed in the group aged 10 to 19 years, n = 8 (Table 4). This finding coincides with that described in studies of populations from Switzerland (16), Michigan (14), and Jerusalem (17). This could suggest that, at these ages, since permanent teeth are erupting or already erupted, the AI is more visible since this pathology is more serious in permanent teeth than in temporary ones. This aspect could suggest that AI in primary teeth sometimes goes unnoticed and may be underdiagnosed. On the other hand, it is also possible that a differential diagnosis has not been made with other enamel defects such as incisor-molar hypomineralization and fluorosis.

In this study, 7 patients presented hypoplastic phenotype (87.50 %) and one hypocalcified (Table 2). This suggests that the hypoplastic phenotype could influence its mainly autosomal dominant segregation, in which the enamel formation process (amelogenesis) is interrupted in the first stage (secretory). Likewise, the presence of a family history of AI was understood as an important sociodemographic factor since it is a hereditary disorder (2).

On the other hand, the 8 individuals affected by AI came from the lower-middle socioeconomic background and their origin was mainly urban. This could suggest that environmental aspects could be part of the etiology of AI. For example, taking antibiotics was found in 12.5 % of cases (Table 7). More specific studies must be carried out to clarify whether antibiotics play a role in the etiology of AI.

The Colombian population analyzed in this study was represented in its majority (55.31 %) by the female sex (n = 771), although the reported cases of AI showed a 1:1 relationship in both sexes (Table 3). The same was observed in clinical and molecular studies carried out in Colombians (19,22), in which AI has presented indistinctly in men and women, especially the hypoplastic phenotype, which was one of the most frequent in these studies. These findings also coincide with those of Gupta et al. (2011) (29) in Indian population, who evaluated 1,123 patients, 572 men and 551 women. They found that two men and one woman were affected by AI. Similarly, Shokri et al. (2014) (18) and Harini & Don (2019) (19), in Indian population, found a total of 4 people (2 men and 2 women) affected by AI.

Regarding the contrast between enamel and dentin, in the present study an absence of said contrast was evidenced in patients with hypoplastic AI (75 %), a characteristic that is more marked in the hypocalcified phenotype that was identified in 1 of the 8 individuals who presented AI. Another relevant radiographic characteristic was taurodontism, which was observed in all patients affected by AI. However, despite taurodontism being described more frequently than other abnormalities studied (3.7 %), the differences were not statistically significant (Table 6). These results coincide with those of Collins et al. (1999) (10), who analyzed dental anomalies associated with AI in panoramic radiographs. They found that in people with AI, taurodontism was present in 9 of 23 (39 %), a proportion that is similar to that of individuals without AI (10 of 24 cases, 42%).

Similarly, it was observed that in patients affected by AI with a hypoplastic phenotype, the enamel density was quantitatively lower than in those with a hypocalcified phenotype. This was also reported by Witkop in 1988 (14), who showed that the density in the hypoplastic phenotype is highly variable in its mineral content, while in the hypocalcified one this decrease is more constant.

Finally, this study is the first approximation to determine the prevalence of AI and, in turn, of dental anomalies in a Colombian population of patients from the dental clinics of the Pontificia Universidad Javeriana from Bogotá in a period of two years. The data collected allow us to see a trend in the distribution of AI and provide the basis for further studies with a larger population to implement preventive actions and establish therapeutic programs involving different disciplines.

CONCLUSIONS

A prevalence of 0.6 % (n = 8) of AI was found in Colombian patients who attended the dental clinics of the Pontificia Universidad Javeriana from Bogotá, attended between January 2015 and December 2017. This prevalence is similar to that described in other populations.

This is the first study of the prevalence of AI carried out in Colombia and shows a trend in magnitude, frequency, and distribution, as well as associated characteristics of this pathology. Taurodontism was the most frequent associated characteristic (100 %).

No predilection for AI was identified with respect to the sex variable.

Family history was important as AI presented more frequently in an autosomal dominant form, closely related to the hypoplastic phenotype.

RECOMMENDATIONS

Continue conducting AI prevalence studies with a larger population that includes different regions of Colombia.

Include this pathology in the National Oral Health Studies of Colombia.

Perform updates in the unification of diagnostic criteria for AI with respect to phenotype and associated dental and skeletal abnormalities bearing in mind this pathology can often be underdiagnosed.

Conduct trainings on differential diagnosis criteria with other enamel defects (hypomineralization, hypoplasia, incisor-molar syndrome and fluorosis).

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