Clinical Doctorate in Paediatric Dentistry (DDent)

Exploring the Impact of Amelogenesis Imperfecta on Oral Health Related Quality of Life in Children and Young Persons

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Word Count: 37169

Declaration

This report entitled “Exploring the Impact of Amelogenesis Imperfecta on Oral Health Related Quality of Life in Children and Young Persons” was composed by me and is based on my own work. Where the work of others has been used, it is fully acknowledged in the text and in captions to table illustrations. This report has not been submitted for any other qualification.

Jenan Altaher
December 2024

Acknowledgements

First and foremost, I would like to thank God for His countless blessings and for granting me the strength, perseverance, and ability to complete this course and research project. Without His guidance, none of this would have been possible.

I would like to extend my deepest gratitude to my main supervisors, Professor Susan Parekh, and Dr. Ryan, for their invaluable guidance, encouragement, and unwavering support throughout this project. Your expertise and mentorship have been instrumental in shaping this research, and I am truly grateful for the opportunity to work under your supervision.

I am also deeply thankful to Professor Paul Ashley, whose insightful advice and calming presence have been a constant source of reassurance throughout this journey. During moments when clinical pressures and research-related anxiety felt overwhelming, his support and steady guidance helped me refocus and regain confidence, making an immeasurable difference in my overall postgraduate experience, for which I will forever be grateful for.

A special thank you to my incredible mother, who has been my backbone and source of strength throughout this challenging journey. Your relentless encouragement and belief in me, even while facing your own health and life struggles, have been my greatest source of inspiration to push through. You have taught me resilience and determination, and I dedicate this work to you.

Finally, I would like to thank all my colleagues, mentors, and the wonderful staff at the EDH and UCL. Your kindness, encouragement, and collaborative spirit have made this experience both memorable and fulfilling. I am truly grateful for the sense of community and support you have provided throughout my journey, which has played a significant role in helping me achieve my goals.

Abstract

Introduction

Amelogenesis Imperfecta (AI) can significantly impact the oral health-related quality of life (OHRQoL) of affected children and young persons (CYP). This thesis explores the multifaceted challenges associated with AI and evaluates the efficacy of patient-reported outcome measures (PROMs) and clinician feedback to inform patient-centred care.

Aims and Objectives

To investigate the impact of AI on OHRQoL from both patient and clinician perspectives. This study includes a scoping review to explore existing research on the relationship between developmental enamel defects (DED) and OHRQoL in children and young persons (CYP). Other key objectives of this project included evaluating the utility and limitations of the AI PROM in clinical practice, identifying age- and treatment-specific trends in patient-reported outcomes, and gathering clinician recommendations for improving the tool.

Methodology

This mixed-methods research involved a scoping review and a retrospective service evaluation of AI PROM responses from CYP at pre- and mid-treatment stages, as well as a survey capturing paediatric dentists' feedback on the tool. The scoping review followed the Joanna Briggs Institute (JBI) methodology for scoping reviews and aimed to map the breadth of literature on the relationship between DED and OHRQoL in CYP. The retrospective service evaluation analysed AI PROM data collected at EDH, identifying trends and patterns quantitatively, while thematic analysis provided qualitative insights. The clinician survey, detailed in Appendix 2, was analysed to capture feedback on the AI PROM's implementation and effectiveness in clinical practice.

Results

The scoping review identified 25 studies, predominantly focusing on MIH and psychological impacts, while highlighting gaps in research on AI and its impacts on OHRQoL. Analysis of 68 AI PROM responses, collected at EDH, revealed age-specific patterns, with younger children facing functional challenges and adolescents experiencing psychosocial distress. Subgroup analysis highlighted greater challenges for Hypoplastic AI cases. The clinician survey (30.8% response rate) confirmed the AI PROM's utility in improving communication and planning but noted challenges like workflow integration and capturing specific patient concerns. This research underscores the need for a refined AI PROM and multidisciplinary, patient-centred approaches to enhance outcomes and quality of life for CYP with AI.

Impact Statement

This thesis, titled **"Exploring the Impact of Amelogenesis Imperfecta on Oral Health-Related Quality of Life in Children and Young Persons,"** sheds light on a critical yet under-researched area in paediatric dentistry. By examining the profound psychosocial, functional, and emotional challenges experienced by children and adolescents with Developmental Enamel Defects, the study emphasizes the importance of holistic, patient-centred approaches to care. Utilizing Patient-Reported Outcome Measures (PROMs), it highlights the real-world experiences of affected individuals, offering a nuanced understanding of how enamel defects impact their daily lives and overall well-being.

A key component of this research lies in its recognition of the variability of impacts by age, gender, and socioeconomic factors, addressing significant gaps in the literature. Adolescents face heightened psychosocial challenges—such as bullying, embarrassment, and social withdrawal due to the visibility of defects—while younger children experience functional limitations, including dental sensitivity, pain, and difficulty chewing, which disrupt nutritional habits and development. The thesis also underscores the importance of standardized assessment tools to improve research comparability and guide more effective, targeted interventions.

The study advocates for a multidisciplinary approach—bringing together paediatric dentists, psychologists, educators, and policymakers—to deliver actionable strategies that enhance patient outcomes while easing emotional and financial burdens on families. By integrating psychological and educational support into dental care pathways, it ensures treatment plans address not only functional and aesthetic concerns but also prioritize mental health, self-esteem, and social participation.

The findings of this research offer significant contributions to clinical practice, public health policy, and future research. This work lays a strong foundation for developing evidence-based guidelines that advance patient-centred care. By incorporating patient perspectives into treatment planning, it highlights the need for clinical and policy decisions to reflect the voices and lived experiences of affected individuals. Ultimately, this thesis calls for a paradigm shift toward holistic care models, recognizing the broader quality-of-life implications for both patients and their families and fostering more compassionate and comprehensive paediatric dental care.

List of Key Words

Developmental Enamel Defects (DED)

Amelogenesis Imperfecta (AI)

- Hypomature (HM) Amelogenesis Imperfecta
- Hypocalcified (HC) Amelogenesis Imperfecta
- Hypoplastic (HP) Amelogenesis Imperfecta

Patient-Reported Outcome Measures (PROMs)

Quality of Life (QoL)

Oral Health-Related Quality of Life (OHRQoL)

Paediatric Dentistry

Tooth Sensitivity

Aesthetic Satisfaction

List of Abbreviations

Abbreviation	Full Term
AI	Amelogenesis Imperfecta
AICEN	Amelogenesis Imperfecta Clinical Excellence Network
AMELX	Amelogenin Gene
AOB	Anterior Open Bite
CDSR	Cochrane Database of Systematic Reviews
COHIP	Child Oral Health Impact Profile
C-OIDP	Child Oral Impacts on Daily Performance
CPQ	Child Perceptions Questionnaire
CSS	Cross-Sectional Study
CYP	Children and Young Persons
DED	Developmental Enamel Defects
DMFT	Decayed, Missing, and Filled Teeth
EAPD	European Academy of Paediatric Dentistry
EAPD MIH Index	European Academy of Paediatric Dentistry MIH Index
ECOHIS	Early Childhood Oral Health Impact Scale
EDH	Eastman Dental Hospital
EHR	Electronic Health Record
ENAM	Enamelin Gene
FAM83H	Family with Sequence Similarity 83 Member H Gene
FPM	First Permanent Molar
FS	Fissure Sealant
GDPR	General Data Protection Regulation
GIC	Glass Ionomer Cement
HC AI	Hypocalcified Amelogenesis Imperfecta
HM AI	Hypomature Amelogenesis Imperfecta
HP AI	Hypoplastic Amelogenesis Imperfecta
ICON	Resin Infiltration System
IS	Inhalation Sedation
IV	Intravenous Sedation
JBI	Joanna Briggs Institute
KLK4	Kallikrein-4
LA	Local Anaesthesia
MDDE	Modified Developmental Defects of Enamel Index

MCDAS	Modified Child Dental Anxiety Scale
MeSH	Medical Subject Headings
Micro-CT	Micro-Computed Tomography
MIH	Molar-Incisor Hypomineralization
MMP-20	Matrix Metalloproteinase-20
OHRQoL	Oral Health-Related Quality of Life
OPG	Orthopantomogram
PEB	Post – Eruptive Breakdown
PedsQL	Pediatric Quality of Life Inventory
PCC	Population, Concept, and Context
PMC	Preformed Metal Crown
PRISMA	Preferred Reporting Items for Systematic Reviews
PRISMA-ScR	Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews
PROM	Patient-Reported Outcome Measure
QLF	Quantitative Light-Induced Fluorescence
QoL	Quality of Life
RCT	Randomized Controlled Trial
REDCap	Research Electronic Data Capture
RI	Resin Infiltration
SCASS	School Child Assessment Scale
SPPC	Self-Perception Profile for Children
SUMARI	System for the Unified Management, Assessment and Review of Information
TFI	Thylstrup-Fejerskov Index
VAS	Visual Analog Scale

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Chapter One: Understanding Developmental Enamel Defects: A Literature Review

Developmental enamel defects (DEDs) pose significant challenges in paediatric dentistry. These defects affect enamel quality and thickness, resulting in functional limitations, aesthetic concerns, and psychosocial burdens for affected individuals. Despite advances in diagnosis and management, gaps remain in understanding the holistic impact of these conditions, particularly on children's quality of life (QoL). This chapter provides an in-depth background on enamel development, the classification of DEDs, diagnostic tools, and their multifaceted consequences, setting the foundation for the current study.

1.1 Enamel Development

The development of dental enamel, the hardest and most mineralized tissue in the human body, occurs through a highly regulated process known as amelogenesis. This process is carried out by specialized cells called ameloblasts, which coordinate the secretion and mineralization of enamel. Enamel formation is a multi-phased process critical to the integrity and structure of teeth. Any disruptions during this period can result in DEDs, including enamel hypoplasia, hypomineralization, and hereditary disorders such as AI.

1.1.1 Stages of Amelogenesis

Secretory Stage

During the secretory stage, ameloblasts produce and secrete an organic matrix, which is can also be described as a protein-rich scaffold primarily composed of amelogenins, enamelins, and tuftelins. These proteins facilitate the initial formation of the enamel framework and regulate the orderly deposition of hydroxyapatite crystals (Hu et al., 2007). Ameloblasts extend Tomes' processes, specialized cellular extensions, which guide the orientation of enamel prisms, contributing to the unique structural organization of enamel (Nanci, 2017). At this stage, enamel remains soft and immature due to its high organic content.

Mineralisation Stage

The mineralization stage is a critical phase of amelogenesis, where the organic enamel matrix undergoes progressive replacement with minerals, primarily hydroxyapatite, to form hydroxyapatite crystals. This process occurs in **two distinct phases**:

1. **Early Calcification Stage**

In this phase, ameloblasts facilitate the initial deposition of mineral ions (calcium and phosphate) into the enamel matrix to form nascent hydroxyapatite crystals. These crystals provide the foundation for enamel's mineral structure. Disruptions during this phase can result in **hypocalcified enamel**, characterized by poor mineral density and fragility. (Crawford et al., 2007; Smith et al., 2016).

2. **Late Maturation Stage**

During the maturation phase, ameloblasts remove any residual organic material, including proteins like amelogenin and enamelin, through the action of enzymes such as matrix metalloproteinase-20 (MMP-20) and kallikrein-4 (KLK4). Enzymatic activity facilitates the expansion and interlocking of hydroxyapatite crystals, resulting in the hardness and translucency characteristic of mature enamel (Bartlett et al., 2006; Wright et al., 2015). Disruptions at this phase cause **hypomature enamel**, which is structurally weak but retains normal thickness (Seow, 2014; Bekes et al., 2021).

1.1.2 Factors Influencing Amelogenesis

The integrity of enamel development is influenced by a combination of **genetic** and **environmental factors**, both of which can cause DEDs if disrupted.

Genetic Factors

Genetic mutations play a pivotal role in enamel formation by affecting genes critical for amelogenesis. Mutations in genes such as AMELX (amelogenin), ENAM (enamelin), and FAM83H disrupt the production and mineralization of enamel, leading to AI, a hereditary disorder characterized by defective enamel formation (Smith et al., 2017; Wright et al., 2015). For example, mutations in AMELX disrupt protein regulation during the secretory phase, resulting in hypoplastic AI, while mutations in FAM83H interfere with maturation, causing hypocalcified AI. (Crawford et al., 2007). These genetic abnormalities highlight the importance of molecular pathways in enamel development.

Environmental Factors:

In addition to genetics, enamel development is sensitive to environmental influences during tooth formation. Systemic conditions such as high fevers, malnutrition, hypoxia, and metabolic disturbances can impair amelogenesis, particularly during critical developmental windows and lead to abnormalities in enamel, such as enamel hypoplasia (Arrow, 2017; Bekes et al., 2021). Nutritional deficiencies, particularly of calcium, vitamin D, and vitamin A, are also associated with defective enamel mineralization, as these nutrients are crucial for hydroxyapatite formation and ameloblast activity (Silva et al., 2021). Furthermore, exposure to environmental toxins, such as excessive fluoride (fluorosis) or tetracycline antibiotics during tooth development, can result in enamel hypomineralization, further emphasizing the susceptibility of enamel to external factors (Lyaruu et al., 2014).

1.2 Definition and Classification of Developmental Enamel Defects (DEDs)

DEDs refer to structural anomalies in dental enamel that occur due to disruptions during the process of amelogenesis. These defects result in either a quantitative or qualitative alteration in enamel structure, leading to functional and aesthetic consequences.

DEDs are broadly classified into **two main types**:

- **Enamel Hypoplasia:** A quantitative defect involving reduced enamel formation.

- **Enamel Hypomineralization**: A qualitative defect affecting enamel mineralization, resulting in structurally weak enamel.

This section will explore these defects in detail, followed by discussions on specific conditions such as **MIH** and **AI**.

1.2.1 Enamel Hypoplasia and Enamel Hypomineralization

Enamel Hypoplasia

Enamel hypoplasia occurs due to disruptions during the secretory phase of amelogenesis, leading to incomplete enamel matrix formation. This results in visibly thinner enamel or localized pits, grooves, and bands on the tooth surface (Arrow, 2017; Wright et al., 2015). Examples of aetiologies include AI, a genetic condition affecting enamel formation, and chronological hypoplasia, which can arise from systemic conditions such as vitamin D deficiency during critical periods of tooth development. These factors highlight the diverse origins and presentations of enamel hypoplasia.

Clinical Features:

- Enamel is reduced in thickness and may appear rough or irregular.
- Defects may be localized (specific teeth or areas) or generalized across the dentition.
- Teeth are often smaller in size and more prone to caries and sensitivity due to the reduced enamel volume.

Figure 1 A clinical image showing thin enamel with pits and grooves on anterior teeth—an example of enamel hypoplasia (Patel, 2019).

Radiographic Features:

- Radiographs reveal reduced enamel thickness with a clear demarcation between the enamel and underlying dentin.
- In severe cases, the enamel contour appears irregular or absent, particularly in localized defects caused by systemic insults.

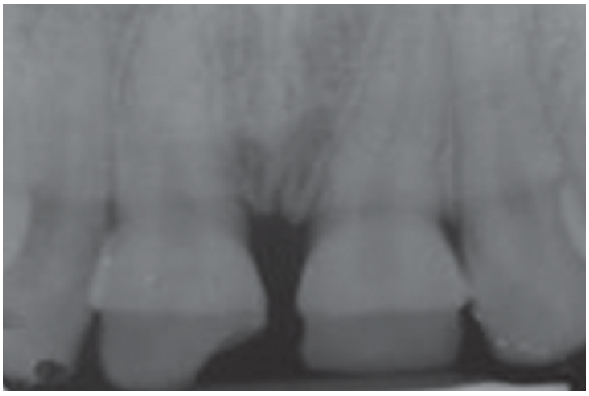


Figure 2 Preoperative intraoral periapical radiograph showing reduced enamel thickness in the central incisors due to enamel hypoplasia from a history of trauma (Gupta et al., 2014).

Enamel Hypomineralization

Enamel hypomineralization arises from disturbances during the mineralization phase of amelogenesis, resulting in enamel that is deficient in mineral content but of normal thickness. It can occur at **two distinct phases**:

During the **calcification phase** resulting in **Hypocalcified Enamel**

Clinical Features: Enamel is soft, poorly mineralized, and prone to fractures. Teeth often appear chalky white or yellow-brown.

Figure 3 A clinical image showing hypocalcified enamel with yellow-brown discolorations (Pediatric Dentistry SF, n.d.).

During the **maturation phase** resulting in **Hypomature Enamel**

Clinical Features: Enamel appears mottled with creamy-white or brown discoloration. Although thicker than hypocalcified enamel, it remains structurally weak and prone to chipping under occlusal forces (Seow, 1993).

Figure 4 A clinical image showing hypomature enamel with creamy-white discolorations (Patel, 2019).

Radiographic Features of hypomineralised enamel:

Radiographs show enamel of normal thickness, but with slightly reduced radiopacity compared to surrounding healthy enamel.

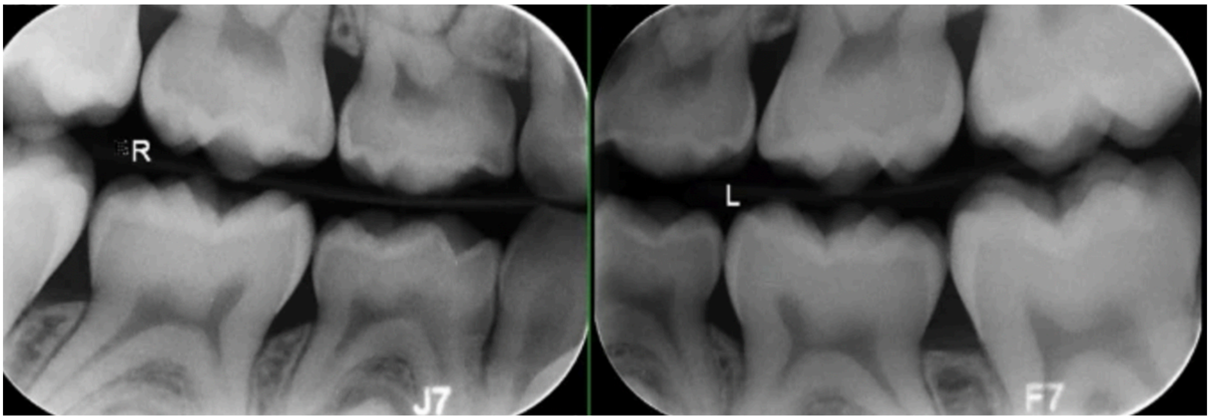


Figure 5 Bitewing radiograph showing a hypomineralized lower left first permanent molar with radiolucent areas and post-eruptive enamel loss, typical of MIH (Humphreys et al., 2021).

1.2.1.3 Comparative Analysis: Enamel Hypoplasia vs. Hypomineralization

The following table summarizes the key differences between enamel hypoplasia and enamel hypomineralization:

Table 1 Comparative Analysis: Enamel Hypoplasia with Enamel Hypomineralization.

	Enamel Hypoplasia	**Enamel Hypomineralization**
Cause	Disruption during the **secretory** stage.	Disruption during the **mineralization** stage.
Clinical Features	Thinner enamel, pits, grooves, or missing areas.	Normal enamel thickness; opaque white/yellow spots.
Radiographic Features	Reduced enamel thickness, irregular contour.	Normal thickness, reduced enamel radiopacity.
Severity	Depends on the extent of reduced enamel formation.	Weak enamel prone to fractures post-eruption.
Functional consequences	Sensitivity due to enamel loss; increased caries risk.	Sensitivity due to compromised enamel integrity; increased risk of PEB and caries; bonding challenges due to the porous and weak nature of enamel.

1.2.2 Molar-Incisor Hypomineralization (MIH)

MIH Definition

MIH is a qualitative enamel defect affecting at least one first permanent molar (FPM), often involving permanent incisors. It arises during the maturation stage of amelogenesis, leading to weakened enamel with reduced mineral content (Weerheijm et al., 2001).

MIH Prevalence

MIH has a global prevalence ranging from 3.6% to 40%, varying with geographic regions and diagnostic criteria, making it one of the most common DEDs (Jälevik, 2010).

MIH Aetiology

The aetiology of MIH is considered multifactorial, involving systemic disturbances during the maturation stage of amelogenesis.

Contributing factors may include:

- **Systemic illnesses:** Early childhood fevers, respiratory infections, and otitis media.
- **Environmental factors:** Perinatal complications, nutritional deficiencies, or exposure to toxins (e.g., dioxins).
- **Genetic predisposition:** While the genetic influence is not fully understood, familial patterns suggest heritability.

(Elhennawy et al., 2022)

MIH Clinical Features

- **Hypersensitivity**, particularly to thermal or mechanical stimuli, due to exposed porous enamel
- **Demarcated opacities** on the occlusal and smooth surfaces of affected teeth, ranging in colour from white to yellow brown.

Figure 6 Clinical presentation of Molar-Incisor Hypomineralization (MIH) showing demarcated opacities on the smooth surfaces of anterior teeth (King's College Hospital NHS, 2023)

- **PEB** caused by reduced mineral content, leading to fractures under normal chewing forces and resulting in a higher risk for caries.

Figure 7 Post-eruptive breakdown (PEB) in lower first permanent molars (FPMs) affected by Molar-Incisor Hypomineralization (MIH), leading to loss of enamel and irregular surfaces (King's College Hospital NHS, 2023)

- **Atypical restorations** are often required due to the challenges in bonding to hypomineralized enamel. The reduced mineral content compromises bonding strength, resulting in poor retention and higher failure rates of restorations.

Consequently, patients may require more invasive restorative procedures at a young age, increasing the risk of long-term dental complications.

MIH Radiographic Features

- Enamel appears normal in thickness but shows **reduced radiopacity** compared to unaffected enamel.
- Areas of **PEB** may appear as irregular or radiolucent patches. They can resemble early carious lesions but lacks distinct cavitation (Humphreys et al., 2021).

Figure 8 Atypical restoration in a lower first permanent molar (FPM) affected by Molar-Incisor Hypomineralization (MIH) (Alfarraj et al., 2022).

Impact of MIH

- **Aesthetic Implications**: The visible defects in anterior teeth caused by MIH can lead to significant psychosocial distress, particularly in school-aged children and adolescents. The discoloration and irregular appearance of affected teeth often result in bullying, anxiety, and reduced self-esteem. Consequently, children may avoid smiling, speaking, or participating in social activities due to embarrassment and aesthetic concerns (Marshman et al., 2009).

- **Functional Implications**: Hypomineralized enamel is prone to sensitivity and PEB, which can cause discomfort during everyday activities such as eating or brushing. The increased enamel porosity also heightens the risk of dental caries. Combined with the difficulty in achieving reliable restorations due to compromised enamel quality, this often leads to repeated dental interventions at an early age (Ghanim et al., 2013). Failed restorations and PEB may necessitate long-term multidisciplinary management, including restorative, orthodontic, and preventive care.

1.2.3 Amelogenesis Imperfecta (AI)

AI Definition and Genetic Aetiology

AI is a group of hereditary enamel disorders caused by mutations in genes essential for enamel development, including AMELX, ENAM, and FAM83H. These mutations interfere with the formation, mineralization, or maturation of enamel, leading to structural defects in both primary and permanent dentitions (Wright et al., 2015). AI can be inherited in autosomal dominant (AD), autosomal recessive (AR), or X-linked patterns, depending on the affected gene.

AI is classified into subtypes based on the specific stage of enamel formation that is disrupted. This results in variations in enamel thickness, quality, and appearance, with significant implications for oral function, aesthetics, and overall QoL.

AI Prevalence

AI affects approximately 1 in 700 to 1 in 14,000 individuals globally, with varying inheritance patterns (autosomal dominant/recessive and X-linked) (Crawford et al., 2007).

AI Subtypes

AI - Hypoplastic (HP) Type

Hypoplastic AI occurs due to disruptions during the secretory phase of amelogenesis, leading to a failure in enamel matrix formation. Clinically, the enamel appears thin and irregular, often presenting with pits, grooves, or smooth surfaces. Teeth may be smaller than normal, discoloured, and prone to surface roughness. The defect is quantitative, meaning enamel thickness is reduced but its mineral content may remain intact (Wright et al., 2015; Crawford et al., 2007). Radiographically, hypoplastic AI is characterized by reduced enamel thickness with a sharp demarcation between the enamel and underlying dentin (Seow, 1993; Smith et al., 2016).

Hypoplastic enamel's rough surface can lead to increased plaque retention, predisposing patients to gingivitis and periodontal issues (Seow, 2014). Functionally, the thin enamel increases the risk of dental caries and tooth sensitivity due to exposed dentin. Aesthetic concerns due to tooth size and irregular morphology significantly impact psychosocial well-being, particularly in children and adolescents (Hasmun et al., 2020). In severe cases, teeth may require comprehensive restorative treatments such as crowns to restore both function and appearance (Mahoney et al., 2003).

Figure 9 Hypoplastic Amelogenesis Imperfecta (HP AI) characterized by reduced enamel volume, smooth surfaces, and the absence of normal enamel curvature. This results in smaller teeth with visible pits and grooves (Front. Physiol., 2017).

AI – Hypocalcified (HC) Type

Hypocalcified AI arises during the early mineralization phase of enamel development, resulting in enamel that is soft, poorly mineralized, and prone to rapid fractures under normal occlusal forces. Clinically, the enamel appears chalky white to yellow-brown, with post-eruptive breakdown (PEB) commonly occurring shortly after eruption (Smith et al., 2017; Wright et al., 2015). Radiographic features include normal enamel thickness but reduced radiopacity due to the enamel's poor mineral content (Silva et al., 2021). The porous enamel often exposes dentin, exacerbating sensitivity (Seow, 1993).

Hypocalcified AI has significant functional implications, including hypersensitivity to thermal and mechanical stimuli, difficulty in chewing, and a higher risk of dental caries due to the compromised enamel structure (Crawford et al., 2007). Aesthetic concerns are profound, as the discolouration and enamel loss can cause embarrassment and low self-esteem (Ghanim et al., 2013). Restorative challenges are frequent, as bonding restorative materials to soft enamel is unreliable, resulting in high failure rates. These patients often require extensive dental interventions, including veneers, crowns, or full-coverage restorations, to manage the condition effectively (Wright et al., 2015; Mahajan & Waingade, 2016).

Figure 10 Hypocalcified Amelogenesis Imperfecta (HC AI) showing generalized enamel discoloration with a yellow-brown appearance. The enamel is poorly mineralized, resulting in significant post-eruptive breakdown, rapid wear, and exposure of underlying dentine, leading to functional and aesthetic impairments (Mahajan and Waingade, 2016).

AI – Hypomature (HM) Type

Hypomature AI results from disturbances during the late maturation phase of amelogenesis, leading to incomplete removal of organic material and insufficient mineralization. Clinically, the enamel appears mottled, creamy-white, or brown, and while it retains normal thickness, it is softer than usual and prone to chipping under occlusal forces (Seow, 1993; Bekes et al., 2021). Radiographically, the enamel shows near-normal thickness but demonstrates slightly reduced radiopacity compared to healthy enamel (Smith et al., 2016).

Hypomature enamel is less durable than normal enamel, leading to occlusal wear, fractures, and enamel sensitivity. The functional impact includes difficulty in maintaining oral hygiene due to the enamel's friability, and in severe cases, occlusal changes may occur due to enamel chipping (Wright et al., 2015). Aesthetic concerns, though milder than hypocalcified AI, can still affect self-esteem and social confidence. Treatment may involve resin-based restorations or crowns to protect the remaining enamel and restore function (Mahoney et al., 2003).

Figure 11 Hypomaturation AI (HM AI) presenting with near-normal enamel thickness but opaque, structurally weak enamel. The enamel fractures easily post-eruption, exposing the underlying dentine (Front. Physiol., 2017).

AI - Combined Type

Combined AI represents cases where features of hypoplastic and hypomineralized enamel defects coexist, often complicating diagnosis and treatment planning. Clinically, teeth exhibit a combination of thin enamel, pits, grooves, and areas of poor mineralization, leading to both structural and aesthetic challenges (Wright et al., 2015). Radiographically, enamel thickness varies, and radiopacity may be inconsistent depending on the extent of mineralization (Silva et al., 2021).

Combined AI presents the greatest clinical complexity due to its mixed presentation. Functionally, teeth are vulnerable to fractures, sensitivity, and rapid wear, while aesthetically, the combination of thin and discoloured enamel compounds the psychosocial impact (Hasmun et al., 2020). Additionally, patients with combined AI may exhibit gingival inflammation due to increased plaque retention and altered tooth morphology, and cases with familial associations may include systemic findings such as anterior open bite (AOB) or malocclusion (Smith et al., 2016; Crawford et al., 2007). Restorative management often requires a multidisciplinary approach, including prosthodontics, paediatric dentistry, and orthodontics, particularly in cases where altered tooth morphology leads to functional and occlusal challenges. Early diagnosis and a tailored treatment plan are crucial to address both functional and aesthetic concerns effectively (Wright et al., 2015).

Figure 12 Combined type of AI demonstrating near-normal enamel thickness with focal pits, variable coloration, and localized opacities. This highlights the overlapping features of multiple AI subtypes (Front. Physiol., 2017).

1.2.3 Diagnosis of DEDs

Accurate diagnosis of DEDs is essential for differentiating between enamel hypoplasia, hypomineralization, MIH, and AI. This involves a multifaceted approach combining clinical examination, radiographic imaging, genetic testing, and standardized assessment tools. The integration of patient-centred outcomes also ensures a holistic evaluation.

Clinical Examination

Clinical examination remains the cornerstone of diagnosing DEDs. A systematic approach involves the integration of visual assessment and standardized diagnostic tools, ensuring accuracy and reproducibility in diagnosis.

- **Visual Assessment**: Teeth are assessed under adequate lighting with the use of magnification tools (e.g., loupes) to detect enamel surface irregularities such as pits, grooves, thinning, and opacities (Clarkson & O'Mullane, 1989). Drying the tooth surface is particularly important, as it enhances the visibility of chalky white or yellow opacities typical of hypomineralized enamel (Jälevik & Klingberg, 2002). Visual assessment provides the first-line identification of DEDs, but its diagnostic accuracy is limited without the inclusion of standardized tools.

- **Developmental Defects of Enamel (DDE) Index**: The DDE Index (Clarkson & O'Mullane, 1989) is widely used to document enamel defects by location, size, and appearance. It is primarily visual and categorizes defects into three types: demarcated opacities, diffuse opacities, and enamel hypoplasia. While comprehensive, it does not account for broader clinical or systemic associations and may be supplemented with other indices for specific conditions like MIH.

- **MIH Index**: The European Academy of Paediatric Dentistry (EAPD) established diagnostic criteria specifically for MIH, which include:
  - Presence of demarcated opacities on the enamel of one or more first permanent molars, with or without involvement of incisors.
  - Post-eruptive enamel breakdown unrelated to caries.

- Atypical restorations in teeth affected by opacities.
- Extraction of permanent molars where MIH was the likely cause (Weerheijm et al., 2003). The MIH index offers a structured approach for MIH diagnosis, emphasizing severity and the functional implications of the defect. It is particularly useful for cases where enamel breakdown significantly affects quality of life and treatment planning.

Other Available Indices: Additional indices are available to evaluate specific enamel defects or general dental anomalies:

- **Modified DDE Index**: Updates the original DDE Index with more precise criteria for documenting enamel defects, particularly diffuse opacities (FDI World Dental Federation, 1992).
- **Enamel Defects Index (EDI)**: Provides a broader framework to categorize enamel defects, including defects associated with systemic conditions or trauma (Seow, 1993).
- **Dean's Fluorosis Index**: Designed to assess fluorosis severity, it is occasionally used in differential diagnoses when hypomineralization due to fluorosis is suspected (Dean, 1934).

Differential Diagnosis Example*:*

Enamel hypoplasia typically presents as localized pits or grooves due to reduced enamel thickness, while hypomineralization manifests as demarcated opacities with potential post-eruptive breakdown (Elhennawy et al., 2022). These features can often be differentiated using the MIH or DDE indices, with the former focusing specifically on hypomineralized lesions in molars and incisors.

By combining visual assessment with the appropriate diagnostic indices, clinicians can achieve a comprehensive evaluation, ensuring accurate diagnosis and effective treatment planning for individuals with DEDs.

Radiographic Imaging

Radiographic imaging plays a critical role in the diagnosis of DEDs, particularly in distinguishing between conditions like hypomineralization and hypoplasia. While clinical examination provides essential information, imaging is invaluable in identifying features that may not be apparent in erupted teeth, especially when differentiating between post-eruptive breakdown (PEB) and hypoplasia.

- **Bitewing Radiographs (BW)**: Bitewing radiographs are ideal for evaluating post-eruptive breakdown (PEB) in hypomineralized enamel, which is commonly observed in conditions like MIH. They are also effective for detecting early caries in MIH-affected teeth, as these areas are more prone to decay due to compromised enamel structure (Humphreys et al., 2021).

- **Intra-Oral Periapical Radiographs (IOPA)**: Intra-oral periapical radiographs provide detailed images of individual teeth, making them particularly useful for assessing enamel thickness in cases of hypoplasia. These images can reveal reduced enamel thickness and irregular contours, which are hallmark features of enamel hypoplasia (Gupta et al., 2014).

- **Orthopantomogram (OPG)**: An orthopantomogram offers a panoramic view of the entire dentition, making it useful for detecting generalized enamel defects, especially in severe forms of AI. It can help identify the extent of enamel anomalies across both erupted and unerupted teeth, which is essential for treatment planning in severe cases (Wright et al., 2015).

Importance of Assessing Unerupted Teeth: Radiographs are particularly important for evaluating unerupted teeth, as they provide critical information on enamel development before eruption. Once a tooth erupts, distinguishing between post-eruptive breakdown (PEB) and enamel hypoplasia can be challenging. PEB typically results from structural weakness in hypomineralized enamel, leading to enamel loss after eruption. In contrast, hypoplasia presents as a developmental defect with reduced enamel thickness from the outset. Radiographic imaging of unerupted teeth allows clinicians to determine whether the defect is developmental (hypoplasia) or acquired (PEB), which aids in formulating an accurate diagnosis and providing ideal management.

***Example of Use in Differential Diagnosis**: Radiographic imaging can help differentiate between enamel hypoplasia and hypomineralization:*

- ***Enamel Hypoplasia**: Radiographs typically show reduced enamel thickness and irregular contours, consistent with a developmental defect (Silva et al., 2021).*
- ***Enamel Hypomineralization**: Radiographs reveal enamel of normal thickness but with reduced radiopacity, reflecting poor mineralization. This feature is most observed in conditions such as MIH.*

By combining clinical and radiographic findings, clinicians can accurately diagnose and differentiate between DED subtypes, ensuring appropriate treatment strategies for the patient.

Genetic Testing for AI and Systemic Associations

Genetic testing is a critical tool in confirming a diagnosis of AI, as it identifies specific causative gene mutations and helps differentiate AI from enamel defects caused by environmental factors. AI is genetically heterogeneous, with multiple genes implicated, each associated with distinct clinical presentations and inheritance patterns.

Key Genes:

- **AMELX**: Linked to X – Linked hypoplastic AI, affecting the regulation of enamel proteins during the secretory phase. Females often exhibit milder manifestations due to X-chromosome inactivation (Wright et al., 2015).

- **ENAM**: Mutations result in both autosomal dominant and recessive hypoplastic AI, affecting enamel matrix secretion and thickness (Smith et al., 2017).

- **FAM83H**: Commonly implicated in autosomal dominant hypocalcified AI causing severe enamel fragility and post-eruptive breakdown (Gadhia et al., 2012).

- **COL17A1:** Mutations disrupt the integrity of the basement membrane, resulting in hypoplastic enamel and potentially causing junctional epidermolysis bullosa (Kim et al., 2019).

- **DLX3:** Associated with Tricho-Dento-Osseous Syndrome, a syndromic AI subtype characterized by hypoplastic enamel, taurodontism, and craniofacial abnormalities (Wright et al., 2015).

- **MMP20 and KLK4:** Both genes are crucial for enamel matrix protein degradation during the maturation stage, and mutations result in hypomature AI with enamel that is softer and prone to chipping (Smith et al., 2017).

- **WDR72:** Associated with hypomature enamel and nephrocalcinosis in some syndromic presentations, highlighting the systemic implications of genetic mutations (Wright et al., 2015).

Genetic testing enables accurate classification of AI subtypes and distinguishes them from environmental causes, such as fluoride exposure or systemic illnesses. It plays a pivotal role in identifying syndromic associations, where AI is linked to broader systemic conditions. For instance, persons with DLX3 mutations often present with a combination of craniofacial abnormalities, hair anomalies, and enamel defects also known as Tricho-Dento-Osseous Syndrome. Understanding the genetic basis of AI facilitates personalized treatment planning. For example, ENAM mutations may permit conservative approaches as the enamel defects are often milder.

Moreover, genetic analysis informs hereditary and family counselling, providing insights into inheritance patterns and aiding in the anticipation of familial and future generational impacts. This approach empowers clinicians to offer tailored guidance and support for affected families, enhancing both medical and psychological care strategies (Crawford et al., 2007).

Advanced Diagnostic Tools

Emerging technologies enhance diagnostic accuracy for complex DED cases:

- **Quantitative Light-Induced Fluorescence (QLF)**: Allows quantification of enamel mineral density, aiding in early detection of hypomineralization (Silva et al., 2021).

- **Micro-Computed Tomography (Micro-CT)**: Provides 3D visualization of enamel thickness and structure, enabling detailed analysis of enamel defects (Elhennawy et al., 2022).

Standardized Assessment Tools: PROMs and QoL measures

DEDs, such as those seen in AI and MIH, impact not only the clinical health of patients but also their psychosocial well-being. The functional, aesthetic, and emotional implications of these defects can lead to challenges such as reduced confidence, anxiety, and social stigma. By incorporating Patient-Reported Outcome Measures (PROMs) and Quality of Life (QoL) assessments, clinicians can adopt a patient-centred approach that addresses both the physical and emotional dimensions of these conditions.

- **Patient-Reported Outcome Measures (PROMs):** PROMs are valuable tools for understanding the impact of DEDs on a patient’s daily life, including their functional abilities, aesthetics, and overall quality of life. Instruments such as the CPQ and the AI - specific PROM (AI PROM) evaluate key domains such as sensitivity, pain, aesthetics, and psychosocial well-being (Hasmun et al., 2020; Bekes et al., 2021). These tools ensure that patient-centred outcomes are integrated into clinical decision-making and research, reflecting the real-world impact of the condition.

1.2.3 Diagnostic Challenges

Accurate differentiation between enamel hypoplasia, hypomineralization, MIH, and AI is challenging due to overlapping features:

A. Chronological Enamel Hypoplasia vs. Hypoplastic AI

- **Chronological enamel hypoplasia** refers to refers to enamel defects that affect all teeth developing at the time of the environmental insult. These defects are systemic in nature and arise from disturbances during the secretory phase of amelogenesis, such as malnutrition, febrile illnesses, or vitamin D deficiency. The defects often appear as linear grooves or pits on the enamel surface, reflecting the developmental timing of the insult.

- **Hypoplastic AI** is a **generalized** enamel defect affecting all teeth, arising from genetic mutations such as in AMELX or ENAM. It is characterized by uniformly thin or pitted enamel across the dentition, highlighting its hereditary basis (Crawford et al., 2007).

B. Enamel Hypomineralization vs. Hypocalcified/Hypomature AI

- **MIH** affects **first permanent molars and incisors** and is linked to systemic childhood disturbances.

- **Hypocalcified/Hypomature AI** presents as a **generalized** defect, involving both dentitions (Jälevik, 2010).

C. Post-Eruptive Breakdown (PEB):

Seen in both MIH and AI, but radiographic localization may help distinguish the two conditions (Humphreys et al., 2021).

In cases of diagnostic uncertainty, genetic testing, and advanced imaging techniques (e.g., Micro-CT) can provide definitive differentiation (Wright et al., 2015); However, these methods are rarely sought out as management, regardless of the diagnosis, is relatively the same.

1.3 Treatment Modalities for DEDs

DEDs such present unique challenges in paediatric dentistry. Treatment decisions are guided by the severity and extent of the defect, functional needs, and aesthetic concerns. The management options range from non-invasive preventive measures to complex restorative and aesthetic interventions, tailored to individual patient needs.

1.3.1 Minimally Invasive Approaches

Minimally invasive treatments aim to enhance enamel strength, alleviate sensitivity, and prevent further deterioration. These are typically indicated for early-stage defects, such as mild MIH or hypomineralized enamel with no significant post-eruptive breakdown (PEB).

Fluoride Therapy

High-concentration fluoride varnishes, gels, or toothpastes are used to promote remineralization and alleviate hypersensitivity by forming a protective layer of fluorapatite crystals. This is particularly effective in hypomineralized enamel with no significant structural loss (Cochrane et al., 2013). Regular fluoride application is essential for children with systemic conditions that predispose them to enamel defects.

Fissure Sealants

Indicated for teeth with deep pits and grooves, sealants provide a protective barrier against plaque accumulation and caries. Resin-based sealants are preferred due to their durability, but glass ionomer sealants may be used when moisture control is a concern (Kotsanos et al., 2005).

Resin Infiltration (RI)

Used for hypomineralized enamel with early lesions or discolorations, RI involves infiltrating porous enamel with low-viscosity resin. It improves both aesthetics and strength by sealing the enamel surface. This technique is minimally invasive and can be particularly effective in managing anterior teeth with visible opacities (Paris et al., 2013).

While non-invasive treatments are painless and preserve tooth structure, they are limited in cases of severe structural loss or advanced PEB, where more durable restorative interventions are required.

1.3.2 Restorative Treatments

Restorative treatments are indicated when defects compromise tooth structure or function, such as in MIH with extensive PEB or AI with generalized enamel hypoplasia. The goals are to restore function, protect remaining enamel, and alleviate sensitivity.

Composite Resin Restorations

Composite resins are a first-line option for localized defects, such as small PEBs or hypoplastic lesions. These restorations bond to enamel and dentin to rebuild tooth structure and improve aesthetics (Wright et al., 2015). However, hypomineralized enamel poses challenges for bonding due to reduced mineral content and porosity, increasing the risk of restoration failure (Bekes et al., 2021).

Glass Ionomer Cement (GIC)

GIC is often used for temporary restorations, particularly in young children or cases with poor moisture control. It releases fluoride, aiding in enamel remineralization, but lacks the durability required for long-term restorations under high masticatory forces (Arrow, 2017).

Prefabricated Metal Crowns (PMCs)

PMCs are ideal for protecting first permanent molars with significant PEB. These crowns provide full coverage, prevent further enamel loss, and require minimal tooth preparation. They are durable and cost-effective but are generally limited to molars due to their metallic appearance (William et al., 2014).

1.3.3 Aesthetic Interventions

Aesthetic treatments are particularly important for anterior teeth with visible discoloration or defects. These procedures address aesthetic concerns while restoring function in cases of moderate to severe defects.

Microabrasion

Microabrasion is a minimally invasive technique indicated for superficial enamel defects or discoloration caused by hypomineralization. The procedure involves the controlled removal of a thin enamel layer, typically up to **100–200 microns**, using a combination of abrasive particles (such as pumice) and hydrochloric acid. This process smoothens the enamel surface and removes surface discolorations, improving the aesthetic appearance (Croll, 1995; Wright et al., 2015). Microabrasion is particularly effective for treating white spot lesions or mild opacities. While conservative, microabrasion is limited to cases where discoloration does not extend deep into the enamel. Post-treatment fluoride application is often recommended to promote remineralization of the treated enamel surface. This technique is conservative and cost-effective, but it may not be suitable for defects with significant enamel loss or structural fragility.

Tooth – Whitening

Tooth whitening, which utilizes hydrogen peroxide or carbamide peroxide-based gels, is a non-invasive approach for addressing discoloration in hypomineralized enamel. These agents work by releasing oxygen radicals that break down discoloration within the enamel and dentin (Joiner et al., 2013). The procedure is most effective for generalized or mild discolorations but may be less suitable for cases involving severe structural defects or enamel loss.

Whitening products are available in various concentrations. Home-use products typically contain 3% to 6% hydrogen peroxide or equivalent concentrations of carbamide peroxide, while in-office procedures can involve concentrations as high as

35%, applied under controlled conditions to minimize risks such as enamel damage or sensitivity.

In the UK, tooth whitening for patients under 18 years old is restricted due to regulatory guidelines established by the EU Council Directive 2011/84/EU, which limits the use of products containing over 0.1% hydrogen peroxide to dental professionals for patients aged 18 or older. This presents challenges in managing discoloration for children and adolescents. However, exceptions can be made if the discoloration causes significant functional or psychosocial distress and is deemed a clinical necessity (Dental Professionals Council, UK). For younger patients, lower concentrations of hydrogen peroxide are typically preferred to reduce the risk of enamel damage and sensitivity, given their thinner and more porous enamel structure (McEvoy & Ziada, 2020).

When whitening is considered for younger individuals, strict monitoring during and after the procedure is crucial to identify and address potential side effects, such as increased tooth sensitivity or gum irritation. Desensitizing agents, including fluoride gels or potassium nitrate, are often recommended to enhance comfort following treatment (Joiner, 2013). Despite its benefits, tooth whitening is generally less effective for teeth with severe structural defects, such as post-eruptive breakdown or areas of hypoplasia, where discoloration is intrinsic and associated with enamel loss. For these cases, alternative treatments like microabrasion or composite masking may be required (Roberts-Harry & Norman, 1997).
Tooth whitening remains an essential tool in the aesthetic management of enamel discoloration, but its use requires careful consideration of age, enamel structure, and the severity of the underlying defect to ensure safe and effective outcomes.

Veneers (Porcelain / Composite)

Veneers are typically indicated for anterior teeth with significant discoloration or structural defects that cannot be adequately managed with composite resin. These thin shells of porcelain or composite bond to the tooth surface to mask imperfections and improve appearance (Joiner et al., 2013). While effective, veneers require enamel removal, making them more invasive. Their longevity is also influenced by enamel quality, with hypomineralized teeth posing a higher risk of bonding failure.

Full-Coverage Crowns (Porcelain / All Ceramic etc.)

Full-coverage crowns are necessary for teeth with severe structural loss as they can they provide both an aesthetic and functional component to a weak posterior tooth. Porcelain-fused-to-metal crowns or all-ceramic crowns are commonly used for anterior teeth, while metal-based crowns are preferred for molars due to their strength. Crown placement involves significant tooth reduction and multiple appointments, making it less suitable for very young or uncooperative patients.

Orthodontic Interventions

Severe AI can result in malocclusions, such as AOB, requiring orthodontic correction. Orthodontic treatment is often combined with restorative interventions to optimize both aesthetics and function (Silva et al., 2021).

1.3.4 Treatment Risks and Considerations

While treatment modalities for DEDs aim to restore function and aesthetics, each carries inherent risks, particularly in teeth with compromised enamel. Bonding failures are a significant concern in hypomineralized enamel, as the porous structure and reduced mineral content compromise adhesion, leading to higher risks of restoration debonding or reduced longevity (Bekes et al., 2021; Ghanim et al., 2013).

Tooth whitening, while non-invasive, carries the risk of exacerbating sensitivity, particularly in hypomineralized teeth with porous enamel. Excessive or improper use of whitening products can lead to gingival irritation, enamel damage, or uneven results, especially in cases of severe discoloration. This is particularly significant in children, where enamel thickness is already reduced in many DED cases.

Post-treatment sensitivity is another potential risk, especially for procedures involving enamel removal, such as microabraision, veneers or crowns. Increased dentin exposure during veneer or crown prep during these treatments may result in temporary or persistent sensitivity, which can be distressing for children (Silva et al., 2021). Restoration longevity also varies significantly; while composite resins and GIC restorations offer minimally invasive solutions, they may fail under high occlusal forces, particularly in molars, necessitating more durable options such as SSCs or full-coverage crowns. However, even these options often require replacement as the child grows due to late establishment of the gingival margin, adding to the long-term treatment burden (Wright et al., 2015).

Treatments like veneers and crowns, while effective in improving aesthetics, may necessitate multiple appointments, which can be logistically and emotionally challenging for children and their caregivers, particularly in younger or anxious patients (Albadri et al., 2014). Additionally, achieving aesthetic harmony in severe cases of hypomineralization is challenging, as matching the shade and translucency of natural teeth is technically demanding, sometimes resulting in suboptimal aesthetic outcomes (Arrow, 2017).

These risks underscore the importance of tailoring treatment plans to individual needs, considering not only the clinical condition but also the psychological and practical considerations of children and their families.

1.3.5 Challenges in Treating Children and Young Persons (CYPs)

The management of DEDs in CYP is often complicated by the need for sedation or anaesthesia to facilitate treatment. Due to their limited tolerance for dental procedures and heightened anxiety, CYP may require aids such as local anaesthesia (LA), inhalation sedation (IS), intravenous (IV) sedation, or even general anaesthesia (GA). Each of these approaches presents unique challenges.

Local anaesthesia, while effective, can be distressing for children due to needle phobia, with studies estimating that up to 20-30% of children experience significant anxiety or fear during dental injections (Cohen et al., 2013). Sedation options, such as nitrous oxide inhalation, are generally well-tolerated, with success rates of 70-90% for managing mild to moderate anxiety (Wilson et al., 2014). However, these methods are technique-sensitive, require specialized equipment, and may fail in highly anxious or uncooperative children, necessitating escalation to IV sedation or GA.
IV sedation is indicated for more complex procedures but carries risks such as respiratory depression, which occurs in approximately 0.1-0.3% of cases (AAPD, 2019). For extensive treatments or uncooperative children, general anaesthesia may be necessary. While GA ensures complete cooperation, it involves significant risks, including respiratory complications (1-5% of cases) and longer recovery times (Wilson et al., 2014). Parents are often reluctant to consent to GA due to its invasive nature, perceived risks, and the logistical and financial burden of hospital-based procedures.

The reliance on these aids not only adds complexity to treatment but also increases the likelihood of failure due to poor cooperation, sedation intolerance, or technical difficulties. Failed attempts may necessitate additional appointments, heightening distress for the child and burdening families with logistical and financial challenges. These factors underscore the importance of minimizing invasiveness wherever possible and tailoring treatment approaches to the needs and capabilities of the child.

1.4 DEDs Impact on Quality of Life

DEDs, including enamel hypoplasia, hypomineralization, and AI, have profound effects on children's OHRQoL. Beyond clinical symptoms, these conditions influence emotional well-being, functional abilities, and social participation, creating challenges for both affected children and their families. Accurate assessment of these impacts requires validated tools that measure patient-reported outcomes and provide critical insights into the psychosocial, functional, and economic burdens of DEDs. The following section outlines these tools and explores the multifaceted consequences of DEDs on OHRQoL.

1.4.1 Tools for Measuring Oral Health-Related Quality of Life (OHRQoL)

To quantify the psychosocial and functional impacts of DEDs, several validated tools have been developed, focusing on both the child's perspective and family burden. These tools play a vital role in evaluating the broader implications of DEDs, ensuring that interventions prioritize patient-centred outcomes.

Child Perceptions Questionnaire (CPQ)

The CPQ evaluates pain, emotional distress, and social limitations associated with oral health conditions. It is a widely recognized tool for assessing OHRQoL in children with visible enamel defects, providing a child-centred approach to understanding the impacts of DEDs (Jälevik & Klingberg, 2002).

Early Childhood Oral Health Impact Scale (ECOHIS)

Designed for preschool-aged children, the ECOHIS measures the effect of oral health problems on children and their families. It evaluates functional difficulties, dental pain, and the stress experienced by caregivers due to frequent dental visits (Pahel et al., 2007).

Child Oral Health Impact Profile (COHIP)

C-OHIP is tailored for school-aged children, focusing on the aesthetic, emotional, and functional consequences of oral health issues. It is particularly relevant for children with enamel defects that affect their self-esteem, daily functioning, and social interactions (Broder et al., 2007).

Patient-Reported Outcome Measures (PROMs)

PROMs, such as the AI PROM, provide comprehensive insights into patient experiences, including pain, treatment satisfaction, and quality of life. These tools emphasize a patient-centred approach and complement clinical evaluations by incorporating subjective experiences into care planning (Wright et al., 2015; Hasmun et al., 2020).

1.4.2 Impacts on QoL

Emotional and Social Impacts

Children with DEDs often face psychological and social hardships due to the visible nature of their enamel defects:

- **Aesthetic Concerns**: Children with DEDs often face visible enamel defects, including discoloration, pits, or thin enamel, which lead to embarrassment, low self-esteem, and social withdrawal (Marshman et al., 2009; Andrade et al., 2019). Adolescents are particularly vulnerable, as peer relationships and self-image become increasingly important during this developmental phase.

- **Bullying and Stigma**: CYP with MIH and AI report higher rates of bullying and teasing due to the appearance of their teeth (Dias et al., 2020). This social exclusion exacerbates emotional distress, anxiety, and reduced confidence.

- **Psychological Distress**: Fear of dental treatment, pain from hypersensitivity, and chronic embarrassment contribute to emotional struggles such as anxiety and depression (Silva et al., 2021). These cumulative effects may lead to avoidance of social interactions and reluctance to smile or speak publicly (Hasmun et al., 2020).

Functional Impacts

- **Sensitivity and Pain**: Hypomineralized enamel, as seen in MIH, is prone to fractures and post-eruptive breakdown (PEB), resulting in dental hypersensitivity to thermal or mechanical stimuli. This discomfort disrupts eating habits, sleep, and daily activities (Bekes et al., 2021).

- **Oral Hygiene and Caries Risk**: Defective enamel increases plaque retention, caries susceptibility, and periodontal issues. In AI, the rough surface texture or exposed dentin further complicates oral hygiene maintenance (Arrow, 2017; Wright et al., 2015).

- **Treatment Burden**: Frequent dental visits and repeated interventions, such as fillings, extractions, and crowns, contribute to disruptions in school and family routines, further amplifying stress and financial burdens for caregivers (Albadri et al., 2014).

Socioeconomic and Family Impacts

- **Economic Burden:** The cumulative costs of restorative and cosmetic treatments, combined with time off work, place disproportionate pressure on low-income families, exacerbating existing healthcare disparities (Folayan et al., 2018; Barros et al., 2022).

- **Parental Stress**: Parents experience emotional strain witnessing their child's discomfort, compounded by financial and logistical challenges (Coffield et al., 2005).

- **Disrupted Routines**: School absences, work disruptions, and ongoing appointments create long-term strain on family dynamics (Andrade et al., 2019).

Educational and Long-Term Impacts

- **Academic Disruption**: CYP with severe enamel defects miss school more frequently due to pain, dental appointments, or social embarrassment, impacting their academic performance and participation (Bekes et al., 2021).

- **Long-Term Health Risks**: Untreated DEDs can lead to chronic pain, early tooth loss, and increased risk of caries and periodontal diseases, emphasizing the need for early intervention and long-term care plans (Silva et al., 2021).

DEDs significantly impair children's quality of life, with wide-ranging functional, emotional, and socioeconomic consequences. Tools such as the CPQ, ECOHIS, and PROMs provide essential insights into patient-reported outcomes, enabling clinicians to better understand the lived experiences of affected children and families. Addressing these impacts requires a holistic, multidisciplinary approach that combines preventive care, restorative interventions, and psychosocial support. Recognizing the broader implications of DEDs ensures that treatment strategies prioritize both oral health and overall well-being.

1.5 Significance of this Study

The impact of DEDs extends beyond the clinical setting, influencing the oral health, daily functionality, and overall QoL of affected CYP. This study is significant in advancing understanding, addressing current gaps in the literature, and guiding future research, clinical practice, and public health initiatives.

1.5.1 Existing Knowledge on DEDs

DEDs are well-recognized for their impacts on dental health. Extensive research has identified their clinical manifestations, including enamel fragility, hypersensitivity, and susceptibility to caries. Studies have also explored treatment approaches ranging from minimally invasive techniques to complex restorative interventions. Tools such as PROMs have provided valuable insights into both functional and psychosocial impacts, though their application remains underutilized.

1.5.2 Gaps in Understanding AI, MIH, and Hypoplasia

Despite advancements, critical gaps persist in understanding the broader implications of DEDs. Research often emphasizes clinical outcomes, overlooking the nuanced psychosocial and emotional burdens experienced by CYPs. There is limited exploration of age- and gender-specific experiences, long-term health consequences, and socioeconomic impacts on families. Furthermore, the literature has limited information regarding the role of standardized assessment tools like PROMs in guiding patient-centred interventions. These gaps highlight the need for holistic, multidisciplinary approaches to understanding and managing DEDs.

1.5.3 Addressing Psychosocial and Functional Implications

This study advocates for a holistic, patient-centred approach that integrates psychological, social, and economic support into dental care pathways to comprehensively address both aesthetic and functional concerns associated with DEDs. By promoting the use of standardized assessment tools, such as validated PROMs like the CPQ and the ECOHIS, clinicians can better quantify the OHRQoL impacts and use this information to guide evidence-based clinical decision-making.

Furthermore, multidisciplinary collaboration is essential, with coordinated efforts between paediatric dentists, psychologists, educators, and policymakers ensuring timely intervention and ongoing support for affected CYP. By addressing these critical gaps, this study aims to inform evidence-based guidelines, advance public health initiatives, and promote equitable, accessible care for CYP with DEDs. Ultimately, this research underscores the need for a paradigm shift towards holistic care models that prioritize not only clinical outcomes but also the lived experiences of patients and their families, ensuring both a comprehensive and compassionate management scheme.

Chapter Two: Scoping Review

The effects of developmental enamel defects on quality of life in children and young persons: A Scoping Review

2.1 Aim and Objectives

2.1.1 Aim of the Review

The aim of this review was to explore the available literature on the effects of DEDs on the quality of life (QoL) of CYPs.

This review of published peer-reviewed studies aimed to identify evidence regarding the effect of DED on the QoL of affected individuals.

2.1.2 Objectives of the Review

To better capture the scope of the review, the following specific objectives were explored:

- Examining the nature, range, and extent of empirical research investigating the relationship between DED and OHRQoL.
- Gaining insight into the unmet dental needs associated with DED.
- Identifying gaps within the literature that could guide future research directions.
- Providing a critical overview of the current empirical literature to highlight areas for future studies focusing on CYPs with DED.

2.2 Methodology

2.2.1 Review Question

The review question was developed in collaboration with research supervisors (SP and FR): **The effects of developmental enamel defects on quality of life in children and young persons: A Scoping Review**

2.2.2 Protocol and Registration

The protocol was developed in accordance with the methodology of Joanna Briggs Institute (JBI) methodology for scoping reviews (Peters et al., 2021) and the Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR): Checklist and Explanation (Tricco et al., 2018). The final version of the protocol was registered with the Open Science Framework (OSF) to ensure transparency and replicability. It delineated the inclusion criteria using the Population, Concept, and Context (PCC) framework.

2.2.3 Eligibility Criteria

For the following scoping review, the eligibility criteria were based on the Population, Concept and Context (PCC) framework, recommended by JBI as a guide to identify the main concepts necessary to answer the review question.

Population: Individuals under the age of 18 regardless of gender and race with DED.

Concept: Covered all available research related to the assessment of the effect DED have on the QoL specifically in CYPs (<18 years of age) affected with such conditions.

Context: Considered all healthcare settings, with no specific cultural factors, location, or gender specified. Furthermore, reviews including systematic reviews, meta-analysis, meta-synthesis, narrative reviews, mixed-methods reviews, qualitative reviews, and rapid reviews must be identified and evaluated.

Inclusion Criteria

- Publications considered included cohort studies, cross-sectional studies, randomised controlled trials (RCTs), observational studies, and narrative review articles.
- Studies focused on QoL associated with DED in CYPs (< 18 years of age).
- Studies published in English only.
- No date limitation.
- Publications containing information on both children and adults were included only if results were presented separately for CYPs (<18 years of age).

Exclusion Criteria

- Non-peer-reviewed literature
- Reviews and Editorials
- Adult only studies
- In-vitro studies
- Animal-based studies
- Grey literature

These criteria were designed to ensure that the included studies provide robust, age-specific, and clinically relevant data to address the review question effectively.

2.2.4 Study Selection and Screening

Following the application of these eligibility criteria, the study selection process was conducted in a systematic manner to identify the most relevant literature.

All identified citations were collated and uploaded into EndNote X9.3.3 (Clarivate Analytics, PA, USA) for reference management, where duplicates were identified and removed. Titles and abstracts were then screened by two independent reviewers to assess their relevance against the inclusion criteria.

A pilot screening was conducted to ensure consistency between reviewers. Full-text articles of potentially relevant studies were retrieved and assessed in detail against the inclusion criteria. This process was facilitated using the JBI System for the Unified Management, Assessment and Review of Information (JBI SUMARI) (JBI, Adelaide, Australia). Reasons for exclusion of sources at the full-text stage were documented and are reported in the scoping review results.

This multistage process ensured the inclusion of only those studies that directly addressed the review question and met the predefined eligibility criteria.

2.2.5 Search Strategy

The search strategy aimed to identify both published and unpublished studies relevant to the review question. A preliminary search was conducted in MEDLINE and the Cochrane Database of Systematic Reviews (CDSR) to confirm the availability of sufficient studies to warrant the review. Search terms were drawn from keywords in titles, abstracts, and subject headings such as MeSH (Medical Subject Headings) and Emtree (Embase Subject Headings) and tailored for each database.

Relevant studies were identified from the following sources:

- Electronic databases: MEDLINE and EMBASE.
- Reference lists of included studies, which were manually screened to locate additional relevant studies.

The search was conducted without date restrictions to ensure that all relevant articles, regardless of publication year, were considered.

Regarding grey literature, no studies from grey literature sources (e.g., theses, reports, conference proceedings) were included in this review. Only peer-reviewed articles were selected to ensure high-quality and standardized evidence. The exclusion of grey literature was a methodological decision to maintain consistency and reduce the risk of bias in the data selection process.

2.2.6 Data Extraction

Data extraction was performed independently by two reviewers (SP and JT) using a customized tool developed by the authors. This tool, designed in Microsoft Excel™, was refined during the initial stages of data extraction to ensure all relevant variables were captured consistently.

The final version of the tool captured the following variables:

- Study metadata:
  - Author(s)
  - Year of publication
  - Country of origin
  - Study title.
- Study characteristics:
  - Study design: For example: the term 'longitudinal study' was used to describe studies that examined changes over time. It was essential to distinguish between longitudinal intervention studies and longitudinal observational studies. Intervention studies included treatments applied and tracked over time, while observational studies solely observed changes in the absence of such interventions. This clarification was made to ensure a more accurate comparison of different study types in terms of the impact of AI-related treatments versus natural progression.
  - Sample size
  - Participant demographics (e.g., age range).
- Methodological details: Tools and measures used to assess outcomes, including QoL impacts.
- Key findings: Primary results related to enamel defects and their impact on children's QoL.

This structured approach allowed for systematic validation and coding of data, ensuring a high level of reliability and reproducibility in the extraction process.

2.3 Results

2.3.1 Preliminary Search and Screening Outcomes

Initial screening resulted in identifying a total of **241 studies** based on relevant keywords, publication period, and age group. In Ovid Embase database, 115 titles were identified, while Ovid Medline yielded 126 titles. EndNote X9.3.3 automatically removed **77 duplicate** records which were obtained from the databases.

The remaining **164 titles** underwent abstract and methodology screening to assess their relevance against the inclusion criteria. Studies were included only if they explicitly addressed the impact of DED on (OHRQoL).

A total of **136 titles were excluded** for reasons such as:

- Irrelevant subject matter or outcomes.
- Lack of focus on age-specific populations.
- Absence of DED-specific findings.

Following the full-text review of **28 eligible studies**, a total of **3 studies were excluded** for reasons such as:

- One did not distinguish between DED and dental decay, leading to overlapping results.
- One focused on treatment outcomes rather than OHRQoL.
- The third emphasized co-morbidities rather than direct patient-reported outcomes.

Ultimately, **25 studies** were deemed eligible for inclusion in this review as shown in Fig. 13 (PRISMA Flow Diagram).

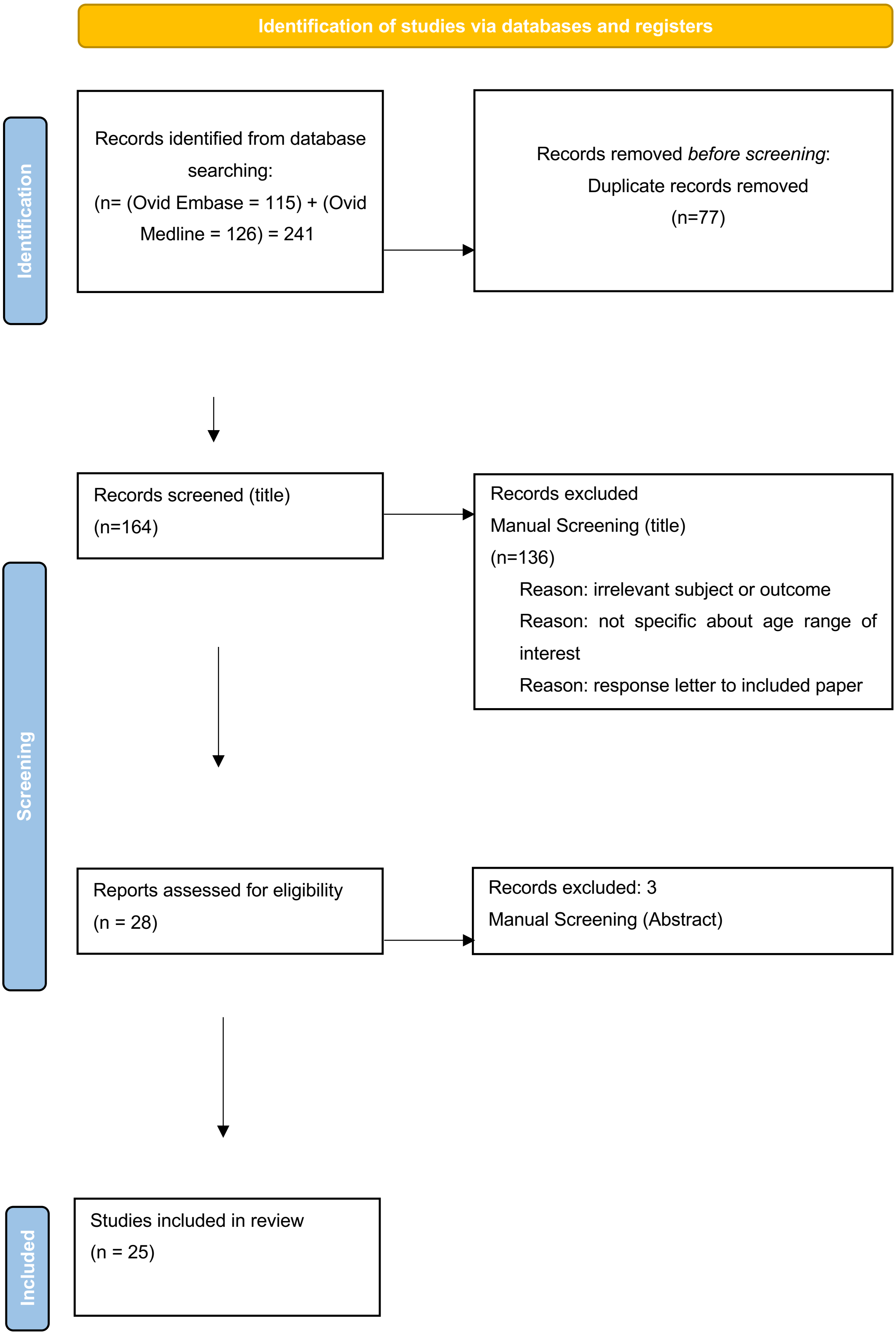


Figure 13- PRISMA FLOW DIAGRAM

2.3.2 Theme Identification Process

The themes presented in the following tables were identified through a systematic process of data extraction and qualitative synthesis. After the inclusion and exclusion criteria were applied, the full-text articles of the selected studies were analysed. Key findings related to the impacts of DEDs on OHRQoL were extracted using a structured data extraction tool designed for this review.

To identify recurring themes, the following steps were undertaken:

A. Coding Key Findings:
   Extracted data points, such as psychosocial, functional, and socioeconomic impacts, were categorized and coded using thematic analysis principles. Common phrases, patterns, and outcomes described across studies were highlighted and grouped under preliminary themes.

B. Refining Themes:
   The initial themes were iteratively reviewed and refined to ensure clarity and alignment with the review objectives. This process involved discussions among the research team to achieve consensus on theme definitions and categorizations.

C. Tabular Representation:
   Finalized themes were organized into tables, summarizing the primary impacts explored in the included studies. Each table corresponds to a specific impact area, providing a comprehensive overview of the evidence.

This systematic approach ensured that the themes accurately reflect the breadth and depth of the included studies, offering insights into the multifaceted consequences of DEDs on the lives of children and young persons.

Psychosocial Impact

Table 2 showing studies included in the scoping review that focused on exploring the psychological impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
UK	Community Dentistry and Oral Epidemiology	2009	Marshman et al.	To explore the impact of DED on young people through their experiences and its meaning to them.	DED	21 (10–15)	Interviews	TFI MDDE	Social withdrawal, self-esteem, and identity issues.	Emotional/ Behavioural
UK	Journal of Dentistry	2020	Hasmun et al.	To identify clinical and psychosocial predictors of OHRQoL in children with MIH after treatment.	MIH	103 (7–16)	L	COHIP-SF19 SPPC	Social and emotional well-being improvements post-treatment.	Emotional/ Behavioural
UK	British Dental Journal	2021	Lyne et al.	To identify key patient concerns using PROMs.	AI	60 (5–17)	CSS	AI PROMs	Social difficulties related to AI.	Long-Term Health
Brazil	Journal of Dentistry for Children	2019	Andrade et al.	To evaluate the DED impact on QoL of 5-year-old children.	DED	566 (5)	CSS	PedsQL, DMFT MDDE	Social struggles due to enamel hypoplasia and overall QoL.	Functional, Socioeconomic
Germany	International Journal of Environmental Health	2022	Elhennawy et al.	To assess the association between MIH and QoL.	MIH	317 (7–14)	CSS	COHIP-G19 EAPD MIH index	Social interactions and emotional well-being impacted by MIH.	Functional, Emotional/Behavioural
Mexico	Journal of Dentistry	2019	Gutierrez et al.	To evaluate MIH's social and self-perception impact in Mexican children.	MIH	411 (8–10)	CSS	CPQ, EAPD	Social anxiety and self-image challenges linked to MIH.	Functional, Emotional/Behavioural

Key:

AI:
Amelogenesis Imperfecta
COHIP-SF19:
Child Oral Health Impact Profile, Short Form
DED:
Developmental Enamel Defects
DMFT:
Decayed, Missing, and Filled Teeth Index
EAPD MIH Index:
European Academy of Paediatric Dentistry MIH Index
MDDE:
Modified Developmental Defects of Enamel Index
MIH:
Molar-Incisor Hypomineralization
OHRQoL:
Oral Health-Related Quality of Life
PedsQL:
Pediatric Quality of Life Inventory
PROMs:
Patient-Reported Outcome Measures
SPPC:
Self-Perception Profile for Children
TFI:
Thylstrup-Fejerskov Index

Functional Impact

Table 3 showing studies included in the scoping review that focused on exploring the functional impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
Brazil	BMC Oral Health	2021	Dias et al.	To evaluate parental and child perceptions of MIH's impact on OHRQoL.	MIH	253 (6–12)	CSS	P-CPQ, CPQ 8–10, CPQ 11–14	Chewing difficulties, sensitivity, and functional limitations.	Emotional/ Behavioural
Mexico	Journal of Dentistry	2019	Gutierrez et al.	To evaluate the impact of MIH on the OHRQoL in Mexican schoolchildren.	MIH	411 (8–10)	CSS	EAPD MIH index, CPQ	Functional limitations from pain and chewing issues.	Emotional/ Behavioural
Germany	Clinical Oral Investigations	2022	Altner et al.	To explore whether treatment of severe caries or MIH leads to better QoL improvements.	MIH, Caries	210 (7–11)	CSS	CPQ G8–10	Functional improvements post-treatment for pain and sensitivity.	Emotional/ Behavioural, Educational
Turkey	Journal of Pediatric Dentistry	2022	Tugcu et al.	To assess OHRQoL changes in children with MIH post Glass Hybrid treatment.	MIH	55 (11–14)	CSS	CPQ 11–14	Relief of hypersensitivity and improved chewing and eating ability.	Emotional/ Behavioural, Long-Term Health
Brazil	International Journal of Paediatric Dentistry	2022	Barros et al.	To assess MIH's role in functional impairment in relation to socioeconomic conditions.	MIH	1181 (8–9)	CSS	CPQ B8–10	Functional and caries-related limitations in daily life.	Socioeconomic, Long-Term Health

Key:

CPQ:
Child Perceptions Questionnaire
CPQ 8–10:
CPQ for children aged 8–10
CPQ 11–14:
CPQ for children aged 11–14
CPQ B8–10:
CPQ Brazilian version for ages 8–10
CPQ G8-10:
CPQ General version for ages 8–10
CSS:
Cross-Sectional Study
DED:
Developmental Enamel Defects
EAPD MIH Index:
European Academy of Paediatric Dentistry MIH Index
MIH:
Molar-Incisor Hypomineralization
OHRQoL:
Oral Health-Related Quality of Life
P-CPQ:
Parental-Caregiver Perceptions Questionnaire

Socioeconomic Impact

Table 4 showing studies included in the scoping review that focused on exploring the socioeconomic impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
Nigeria	BMC Oral Health	2018	Folayan et al.	To assess whether socioeconomic factors influenced QoL in children with MIH.	MIH, Enamel Hypoplasia	853 (6–16)	CSS	C-OIDP, EAPD MIH index	Socioeconomic disparities worsened QoL for MIH-affected children.	Functional, Emotional/Behavioural
Brazil	International Journal of Paediatric Dentistry	2022	Barros et al.	To model MIH's impact on caries experience and QoL through socioeconomic factors.	MIH, Dental Caries	1181 (8–9)	CSS	CPQ B8–10	Indirect QoL impacts via socioeconomic mediators.	Functional, Long-Term Health
Belgium	International Journal of Paediatric Dentistry	2022	Vanhee et al.	To assess dental anxiety and QoL among children with MIH.	MIH	280 (8–9)	CSS	CFSS-DS, C-OIDP	Economic challenges influenced treatment access and anxiety levels.	Emotional/Behavioural, Long-Term Health
Brazil	Journal of Dentistry for Children	2019	Andrade et al.	To evaluate the DED impact on QoL of 5-year-old children.	DED	566 (5)	CSS	PedsQL, DMFT, MDDE	Socioeconomic influences on QoL for hypoplasia-affected children.	Functional, Psychosocial

Key:

C-OIDP:
Child Oral Impacts on Daily Performance.
CFSS-DS: Children's Fear Survey Schedule-Dental Subscale.
CPQ B8–10: Child Perceptions Questionnaire Brazilian version for ages 8–10
CSS:
Cross-Sectional Study.

DED:
Developmental Enamel Defects.
DMFT:
Decayed, Missing, and Filled Teeth Index
EAPD MIH Index: European Academy of Paediatric Dentistry Molar-Incisor Hypomineralization Index.
MDDE:
Modified Developmental Defects of Enamel Index

MIH:
Molar-Incisor Hypomineralization
OHRQoL:
Oral Health-Related Quality of Life
QOL:
Quality of Life

Emotional and Behavioural Impact

Table 5 showing studies included in the scoping review that focused on exploring the Emotional and Behavioural impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
Belgium	European Archives of Paediatric Dentistry	2022	Jalevik et al.	To summarize evidence of MIH's links to dental fear and QoL.	MIH	6–18	Systematic Review	PRISMA	MIH-induced fear and anxiety affected QoL.	Long-Term Health
UK	Dentistry Journal	2018	Hasmun et al.	To evaluate minimally invasive treatments' emotional effects on children's OHRQoL.	MIH	93 (7–16)	L	COHIP-SF19	Reduced anxiety and distress post-aesthetic intervention.	Functional
Germany	Clinical Oral Investigations	2022	Michaelis et al.	To compare caries and MIH's emotional impact on children's QoL.	MIH, Caries	258 (7–10)	CSS	CPQ G8–10	Emotional distress from visible defects and associated pain.	Functional, Psychosocial
Germany	International Journal of Paediatric Dentistry	2019	Portella et al.	To evaluate MIH-related distress and emotional consequences on early mixed dentition.	MIH	728 (8)	CSS	CPQ 8–10, EAPD MIH Questionnaire	Emotional struggles in younger children linked to MIH sensitivity.	Functional, Psychosocial

Key:

COHIP-SF19: Child Oral Health Impact Profile (Short Form, 19 items).
CPQ 8–10: Child Perceptions Questionnaire for children aged 8–10.
CPQ G8–10: Child Perceptions Questionnaire (General Version) for children aged 8–10.

CSS: Cross-Sectional Study.
EAPD MIH Questionnaire: European Academy of Paediatric Dentistry Questionnaire for MIH.
L: Longitudinal Study.

MIH: Molar-Incisor Hypomineralization.
PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses.

Educational Impact

Table 6 showing studies included in the scoping review that focused on exploring the educational impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
Australia	Community Dental Health	2017	Arrow et al.	To evaluate the impact of DED on school participation and absenteeism.	DED	111 CPQ, 91 CE (13)	Cohort	CPQ, MCDAS	Pain and sensitivity caused school absenteeism.	Functional
Germany	Clinical Oral Investigations	2022	Bekes et al.	To assess whether MIH treatment led to better school engagement and fewer absences.	MIH	258 (7–10)	CSS	CPQ G8–10	Improved focus and reduced absenteeism due to pain alleviation.	Functional
Brazil	International Journal of Paediatric Dentistry	2021	Bekes et al.	To examine sealing MIH molars and its impact on child participation in school life.	MIH	38 (6–10)	L	SCASS, CPQ G8–10	Functional and academic performance improved post-treatment.	Emotional/ Behavioural, Functional

Key:

CPQ:
Child Perceptions Questionnaire
CPQ G8–10: Child Perceptions Questionnaire (General Version) for children aged 8–10.
CSS:
Cross-Sectional Study.

DED:
Developmental Enamel Defects.
L:
Longitudinal Study
MCDAS: Modified Child Dental Anxiety Scale.:
Longitudinal Study.

MIH:
Molar-Incisor Hypomineralization.
SCASS:
School Child Assessment Scale.

Long-Term Health Impact

Table 7 showing studies included in the scoping review that focused on exploring the Long-Term Health Impact DED had on CYPs.

Country	Journal	Year	Authors	Study Aims	Enamel Defect Explored	No. of Patients (Age Range)	Study Design	Scales Used	Primary OHRQoL Impact Explored	Secondary Impacts Explored
Brazil	Brazilian Oral Research	2011	Vargas-Ferreira et al.	To assess the long-term impacts of DEDon QoL in Brazilian children.	DED	944 (11–14)	CSS	MDDE, DMFT, CPQ B11–14	Chronic pain and untreated defects led to long-term oral health risks.	Functional
Colombia	Colombian Dental Journal	2018	Velandia et al.	To assess MIH's long-term impacts on children's oral health and QoL.	MIH	88 (7–10)	Interviews	Translated CPQ 8–10	Untreated MIH created risks of future complications and reduced QoL.	Functional, Socioeconomic
Brazil	International Journal of Paediatric Dentistry	2022	Barros et al.	To analyse socioeconomic and caries-related health risks associated with MIH in schoolchildren.	MIH	1181 (8–9)	CSS	CPQ B8–10	Chronic oral conditions linked to worse long-term OHRQoL outcomes.	Functional, Socioeconomic

Key:

CPQ 8–10: Child Perceptions Questionnaire for children aged 8–10.
CPQ B8–10: Child Perceptions Questionnaire (Brazilian version) for ages 8–10.
CPQ B11–14: Child Perceptions Questionnaire (Brazilian version) for children aged 11–14.
CSS: Cross-Sectional Study
DED: Developmental Enamel Defects
DMFT: Decayed, Missing, and Filled Teeth Index.
MDDE: Modified Developmental Defects of Enamel Index.
MIH: Molar-Incisor Hypomineralization

2.3.3 Overview of Included Studies

The 25 studies analysed covered diverse geographies, methodologies, DED, and QoL impacts:

- **Geographic Distribution:**
  Most studies originated from Europe (40%), followed by Latin America (32%), and Africa (20%).

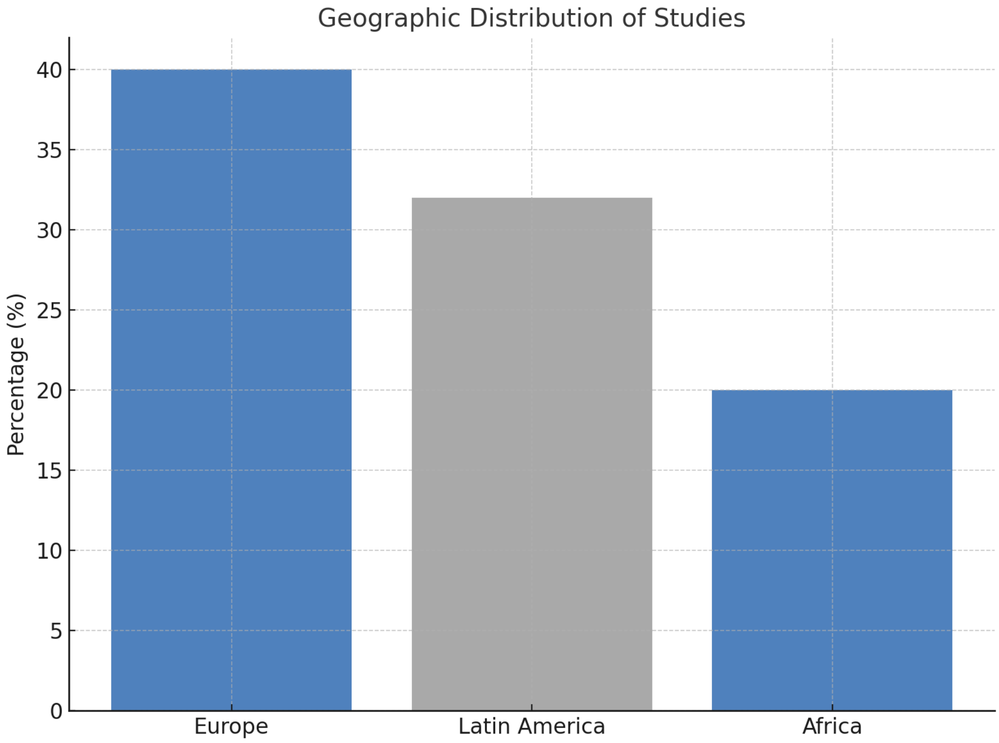


Figure 14 Bar chart displaying study percentages from Europe, Latin America, and Africa

- **Enamel Defect Types:**

  The distribution of papers discussing different enamel defects reveals varying research focus across categories. Most studies (60.0%, 15/25) addressed **MIH**, reflecting its prominence in paediatric dental research and its significant impact on oral health and QoL. A smaller proportion (20.0%, 5/25) examined **Chronological Enamel Hypoplasia**, which is often linked to disruptions in enamel formation during specific developmental periods. Only 12.0% (3/25) of papers specifically discussed **AI**, likely due to its rarity and genetic aetiology. Notably, 28.0% (7/25) of papers were categorized as addressing **Non-Specific Defects**, which include studies that used general or ambiguous terms such as "diffuse opacities," "generalized hypoplasia," or "DEDs" collectively without further classification. These non-specific defects encompass a wide range of enamel abnormalities that are often grouped together due to a lack of detailed diagnostic differentiation.

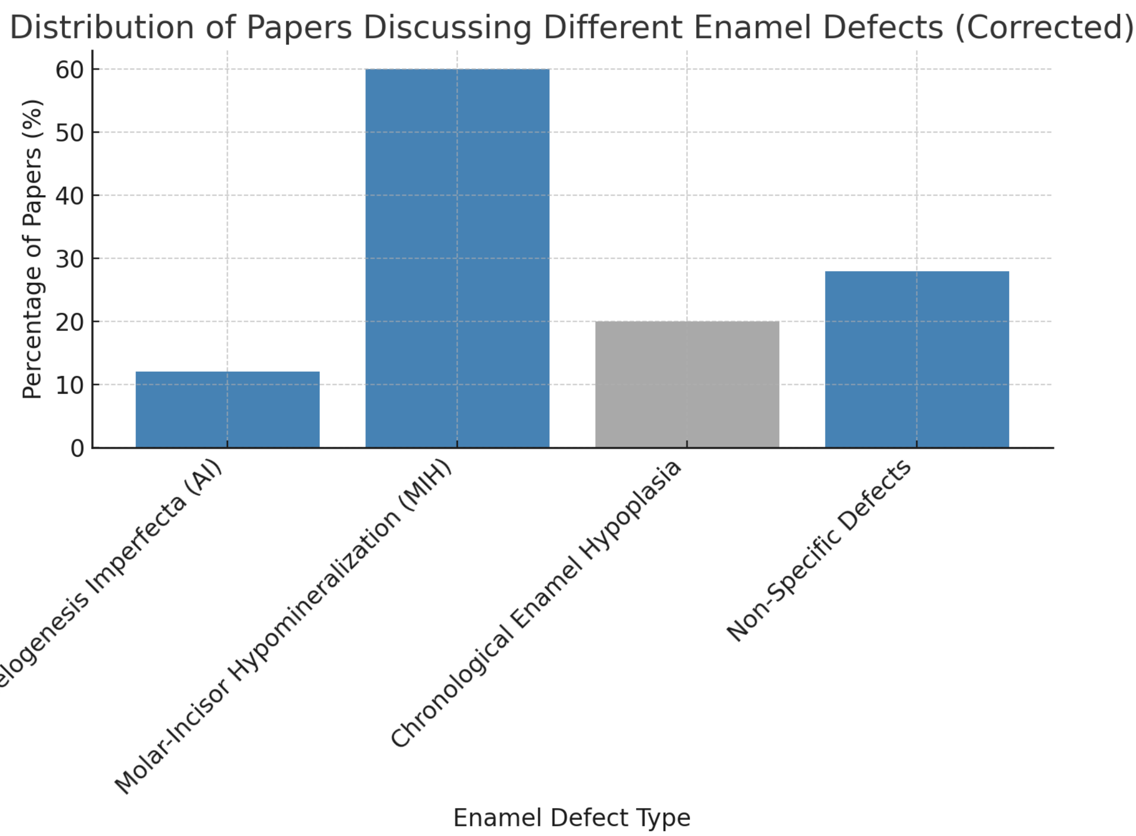


Figure 15 Bar chart displaying percentages of AI, MIH, Chronological Enamel Hypoplasia, and Non-Specific Defects.

- **QoL Impacts:**

  The psychological impact of enamel defects was the most frequently researched area (65% of studies), followed by functional limitations (50%) and socioeconomic challenges (40%). Educational impacts and long-term health outcomes were less commonly explored, appearing in only 20% and 10% of studies, respectively. This imbalance underscores the need for greater focus on educational and long-term health dimensions, as these can indirectly influence children's overall QoL and future well-being.

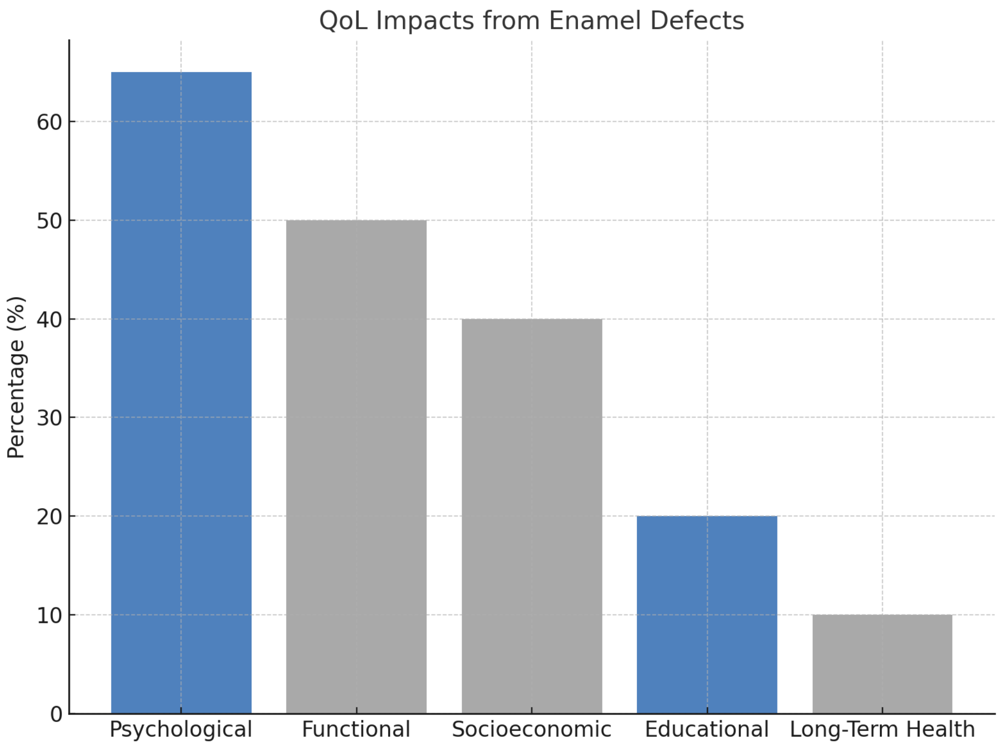


Figure 16 Bar chart displaying the percentages of various impact areas, such as psychological, functional, and socioeconomic.

- **Measurement Tools and Thematic Synthesis:**
  The included studies utilized diverse tools to measure OHRQoL, including the:

  - Child Perceptions Questionnaire (CPQ),
  - Child Oral Health Impact Profile (C-OHIP)
  - Early Childhood Oral Health Impact Scale (ECOHIS).

  Due to inconsistencies in scoring systems and scales, it was not possible to combine studies and therefore thematic synthesis was applied, grouping findings into overarching domains:

  - **Psychosocial impacts**
  - **Functional limitations**
  - **Socioeconomic challenges**
  - **Educational impact**
  - **Long-term health impacts**

  Where studies reported numerical scores (e.g., CPQ subdomains), data were analysed descriptively. Similar domains across tools (e.g., emotional well-being, functional limitations) were grouped for comparative analysis. However, direct statistical comparisons were not feasible due to methodological differences across studies.

2.3.4 Impact of Developmental Enamel Defects on Quality of Life

The studies revealed that DEDs impact children's QoL across several key domains: psychosocial well-being, functional limitations, socioeconomic burden, and broader emotional and educational impacts. Each category below highlights the specific effects of DEDs and provides referenced studies to support each finding.

Psychosocial Impact

- Self-Esteem:
  Children with visible enamel defects often experienced embarrassment and lowered self-esteem. Andrade et al. (2019) reported that 78% of children with DEDs felt uncomfortable smiling in public, while Boukhobza et al. (2022) observed that children with severe MIH reported higher levels of social anxiety due to concerns about appearance.

- Social Interactions:
  Many children with DEDs, especially those with MIH, faced social exclusion and bullying. For instance, Dias et al. (2020) found that 65% of children with severe MIH reported experiences of teasing or bullying, leading to social withdrawal and reduced participation in activities.

- Emotional Distress:
  DEDs often lead to emotional strain, including feelings of frustration, sadness, and anger. Dias et al. (2020) reported that 45% of children with severe enamel defects felt "upset" or "frustrated" due to their appearance and associated pain.

- Behavioural Avoidance:
  Many children with DEDs avoid social activities and behaviours like smiling, speaking in class, or participating in sports due to concerns about their appearance or fear of discomfort. Marshman et al. (2009) found that 30% of children with DEDs avoided smiling or speaking publicly to avoid drawing attention to their teeth.

The emotional and behavioural impacts of DEDs suggest that psychosocial interventions and support may play a vital role in improving QoL for affected children.

Functional Limitations

- Pain and Sensitivity:
  Functional limitations such as pain and temperature sensitivity affect children's eating habits and nutritional intake. Bekes et al. (2021) reported that 85% of children with MIH experienced pain when eating hot or cold foods, leading to dietary restrictions. Silva et al. (2021) found that 70% of children with enamel hypoplasia reported discomfort while eating.

- Oral Hygiene and Caries Risk:
  DEDs compromise oral hygiene, increasing susceptibility to caries. Arrow (2017) found that children with MIH had a 2.5 times higher prevalence of dental caries compared to peers without DEDs. Structural weaknesses in enamel make maintaining oral hygiene difficult, leading to further complications and the need for frequent interventions (Elhennawy et al., 2022).

Addressing pain management and ensuring children have access to dental care are essential in mitigating the functional impacts of DEDs.

Socioeconomic Challenges

- Financial Burden:
  Families often bear significant costs for the treatment and management of DEDs, especially for recurring interventions and aesthetic treatments. Bekes et al. (2021) reported that families spent between $300–$500 on initial MIH treatments, with additional costs over time, placing a financial strain on families from lower-income backgrounds.

- Time Lost from Work and School:

The frequent need for dental appointments and management of DED symptoms leads to school absences for children and missed workdays for parents. Andrade et al. (2019) noted that 35% of children with severe enamel defects missed 3–5 school days per year, and Silva et al. (2021) observed that 20% of parents missed workdays to care for their child's dental needs.

The socioeconomic impact of DEDs emphasizes the need for affordable treatment options and workplace support for affected families.

Educational Impact

- Academic Performance:
  Frequent absences due to dental issues, combined with the psychological impact of DEDs, can impair academic performance. Bekes et al. (2021) noted that children who missed school for dental-related issues tended to have lower grades in core subjects.

- Reduced Participation in School Activities:
  Pain, social anxiety, and discomfort related to DEDs discourage participation in extracurricular activities, limiting social development opportunities. This reduced engagement can affect peer relationships and social skills, potentially impacting long-term educational and personal outcomes.

Schools and educators may need to be made aware of the potential impact of DEDs on students' participation and academic performance and advised to offer supportive measures where needed.

Long-Term Health Impact

- Increased Risk of Future Oral Health Problems:
  DEDs often lead to further complications, such as a higher risk of caries, periodontal disease, and early tooth loss. Arrow (2017) noted that children with

MIH had a 50% higher risk of developing caries, necessitating ongoing dental interventions.

- Potential for Chronic Pain:
  In cases where DEDs are not effectively managed, children may develop chronic pain, affecting QoL over the long term. Chronic dental pain can disrupt sleep, mental health, and dietary habits, leading to a cycle of health challenges.

Addressing DEDs early and providing ongoing management may help reduce the risk of chronic pain and further oral health issues.

Key Findings by Enamel Defect Type

The included studies explored a range of DEDs, which were categorized into four major groups: AI, MIH, Chronological Enamel Hypoplasia, and Non-Specific Defects. These studies demonstrate the variable impacts of these conditions on OHRQoL.

The major types of DEDs and their recorded QoL impacts are summarized below:

Molar-Incisor Hypomineralization (MIH)

Prevalence: MIH was the focus of 60% of the included studies (15/25).

Key Findings:

- Functional impacts were significant, with hypersensitivity and pain due to post-eruptive breakdown (PEB) reported in 70% of children with MIH (Dias et al., 2020; Tugcu et al., 2022).
- Psychosocial effects included bullying and anxiety in 65% of affected children, especially adolescents, due to the visibility of affected teeth (Vanhee et al., 2022; Gutierrez et al., 2019).
- Aesthetic concerns were prominent, with 78% of children feeling embarrassed about their appearance (Michaelis et al., 2022; Hasmun et al., 2018).
- Studies like Boukhobza et al. (2022) emphasized that effective treatment improved both social and emotional well-being.

Chronological Enamel Hypoplasia

Prevalence: Addressed in 20% of the studies (5/25).

Key Findings:

- Chronological hypoplasia was primarily linked to systemic disturbances during enamel formation, such as malnutrition or childhood illness (Arrow, 2017; Silva et al., 2021).
- Functional impacts included increased susceptibility to caries and structural fragility, reported in 70% of children with hypoplasia (Vargas-Ferreira et al., 2011; Silva et al., 2021).
- Moderate psychosocial impacts were observed in younger children due to aesthetic concerns and functional limitations (Arrow, 2017; Andrade et al., 2019).

Amelogenesis Imperfecta (AI)

Prevalence: Covered in 12% of the studies (3/25).

Key Findings:

- AI caused psychosocial challenges, including social withdrawal and low self-esteem, especially in 65% of adolescents, due to the visibility of defects (Gutierrez et al., 2019; Barros et al., 2022).
- Functional limitations, such as enamel fractures and hypersensitivity, were reported in 80% of affected children, requiring frequent restorative interventions (Silva et al., 2021; Hasmun et al., 2018).
- Aesthetic concerns were severe, with 70% of children reporting negative self-image (Gutierrez et al., 2019; Dias et al., 2020).

Non-Specific Defects

Prevalence: Discussed in 28% of the studies (7/25).

Key Findings:

- These studies used generalized terms such as "diffuse opacities," "hypoplasia," or "DEDs without clear classification (Silva et al., 2021; Velandia et al., 2018).

- Aesthetic impacts were predominant, with 75% of adolescents experiencing self-esteem issues due to visible enamel abnormalities (Michaelis et al., 2022; Dias et al., 2020).
- Minimal functional effects were reported, with most studies focusing on aesthetic management rather than functional or psychosocial challenges (Vargas-Ferreira et al., 2011; Velandia et al., 2018).

2.4 Discussion

2.4.1 Study Comparability and Limitations

Direct statistical comparisons of studies were not possible due to variability in study designs, outcome measures, and reporting formats. However, thematic synthesis provided a framework to qualitatively compare findings. For example, despite using different tools (e.g., CPQ versus C-OHIP), studies consistently reported psychosocial impacts like reduced self-esteem and social anxiety among children with enamel defects.

Additionally, demographic, and contextual factors, such as socioeconomic status, were accounted for in studies like Folayan et al. (2018) and Barros et al. (2022), which highlighted disparities in access to care. These findings were included to provide a holistic understanding of QoL impacts rather than isolated comparisons.

In addition to the variations in study designs and outcome measures, another notable observation was the absence of studies using the ECHOHIS (Early Childhood Oral Health Impact Scale) in the scoping review. This tool, designed to assess the OHRQoL in very young children, was not widely represented in the studies included. The absence of ECHOHIS could be attributed to several factors.

First, it is primarily used for younger children, specifically preschool-aged children, which may limit its applicability to studies involving older children and adolescents. Furthermore, despite its utility in measuring functional and psychosocial impacts in early childhood, the ECHOHIS is less frequently used in studies of developmental enamel defects like AI, possibly due to a focus on other well-established tools such as the CPQ and C-OHIP, which are more widely used across different age groups.

Additionally, another possible explanation is that AI predominantly affects permanent teeth, while primary teeth are often less severely impacted. As a result, the issues associated with AI may not become evident until permanent teeth begin to erupt, reducing the likelihood of ECHOHIS being employed in AI-focused studies.

Future research could benefit from incorporating the ECHOHIS to capture a broader range of psychosocial impacts in younger populations, particularly given its sensitivity to the emotional distress and social anxiety associated with visible dental defects in early childhood. Further exploration into the broader use of ECHOHIS in AI-related studies could provide valuable insights into the condition's impact on younger children, an area that remains underexplored in the current literature.

2.4.2 Variability in Outcomes by Defect Type

The type and severity of DEDs significantly influenced their impact on children's QoL.

Molar-Incisor Hypomineralization (MIH)

MIH consistently led to the most severe impacts across psychosocial, functional, and aesthetic domains, making it the primary focus of 60% of the included studies. Pain and sensitivity from PEB were frequently reported, impairing basic functions such as eating and speaking (Dias et al., 2020; Tugcu et al., 2022). Adolescents with visible discoloration were particularly affected by social anxiety and bullying, as highlighted by studies like Gutierrez et al. (2019) and Marshman et al. (2009). While targeted interventions such as minimally invasive restorative techniques improved QoL, gaps remain in understanding the long-term outcomes, particularly in underserved populations (Folayan et al., 2018).

Chronological Enamel Hypoplasia

Enamel hypoplasia predominantly affected functional domains, with increased caries risk and structural fragility being common (Vargas-Ferreira et al., 2011; Silva et al., 2021). Younger children were particularly impacted by eating difficulties, highlighting the need for early dietary and pain management strategies (Andrade et al., 2019). Psychosocial effects were less frequently studied but remained a concern, particularly in visible cases.

Amelogenesis Imperfecta (AI)

AI presented a unique combination of psychosocial and functional challenges. The hereditary nature of the condition and its visibility led to significant self-esteem issues, especially among adolescents (Gutierrez et al., 2019). Functional challenges such as enamel fractures and hypersensitivity necessitated frequent and costly interventions, creating disparities in care based on socioeconomic status (Hasmun et al., 2020).

D) Non-specific defects

Non-specific defects, although less severe in terms of functional impact, had notable aesthetic implications. These defects were primarily associated with self-esteem issues in adolescents, as visible abnormalities often led to social anxiety (Michaelis et al., 2022; Dias et al., 2020). The lack of standardized classification and measurement tools for these defects limits direct comparisons, underscoring the need for consistent methodologies in future research.

This categorization underscores the need for tailored interventions that address the specific impacts of each defect type. While MIH requires priority in research and treatment due to its severity, greater focus on hypoplasia and non-specific defects is necessary to achieve a holistic understanding of DEDs.

2.4.3 Demographic and Contextual Influences

Age and gender significantly influence how enamel defects impact OHRQoL.

Age-Specific Differences

The impact of enamel defects varies significantly across age groups. Younger children, particularly pre-schoolers, are more affected by functional limitations such as pain, sensitivity, and difficulty eating. Studies such as Andrade et al. (2019) emphasize that these limitations often manifest through parental reports rather than self-reported impacts by children. Conversely, older children and adolescents are more likely to report psychosocial challenges such as self-esteem issues, social withdrawal, and embarrassment, particularly due to the visibility of defects like MIH and AI (Dias et al., 2020).

Adolescents also face unique social pressures that amplify the psychological burden of enamel defects. For example, Marshman et al. (2009) found that teens with visible enamel defects were more likely to experience bullying and social anxiety, leading to avoidance of social and academic activities. This highlights the need for age-appropriate interventions, such as pain management for younger children and counselling or aesthetic treatments for adolescents.

Table 8 showing the primary impact DED had in specific age groups.

Age Group	Primary Impact	Examples
Pre-Schoolers	Functional limitations	Eating difficulties (Andrade et al., 2019)
School-Aged (6–12)	Pain and social anxiety	Social withdrawal (Dias et al., 2020)
Adolescents (13–18)	Psychosocial challenges	Bullying, self-esteem issues (Marshman et al., 2009)

Gender-Specific Differences in the Impact of Enamel Defects

Although gender differences in QoL impacts are subtle, girls tend to report heightened concerns over the aesthetic effects of enamel defects compared to boys. Hasmun et al. (2020) found that 60% of girls with visible enamel defects expressed dissatisfaction with their appearance, compared to only 30% of boys. This aesthetic dissatisfaction often leads to social discomfort, withdrawal from social settings, and even avoidance of public speaking or smiling in photographs.

Cultural factors may amplify these differences. For instance, in societies with strong emphasis on physical appearance, girls are more likely to internalize aesthetic challenges, further impacting their self-esteem (Gutierrez et al., 2019). On the other hand, boys, while less concerned about aesthetics, may face functional challenges such as pain or sensitivity during physical activities like sports.
This highlights the need for possible gender-sensitive approaches in treatment planning. For example:

- **Girls:** Focusing on aesthetic improvements (e.g., cosmetic treatments like bonding) and counseling may boost self-esteem.

- **Boys:** May require more focus on addressing functional challenges through pain management and dietary guidance.

These demographic variables underscore the importance of tailoring interventions to specific age groups and considering gender-specific needs when designing treatment strategies and psychosocial support systems.

Cultural and Socioeconomic Influences

Cultural norms and societal attitudes play a pivotal role in shaping the perception and psychosocial impact of enamel defects.

- **Regional Differences:**
  In societies where aesthetics holds significant cultural value, children with visible enamel defects such as MIH or AI are at greater risk of stigma, social anxiety, and self-esteem issues. For instance, Dias et al. (2020) found that in Brazil, a country with a strong emphasis on appearance, children with MIH frequently reported embarrassment and avoidance of social activities.

- **Cultural Beliefs on Dental Health:**
  In Nigeria, Folayan et al. (2018) reported that cultural attitudes towards oral health often prioritized functional concerns over aesthetic issues, particularly in low-income families. This resulted in delayed or limited access to treatment for aesthetic concerns, which further compounded the psychosocial impact for affected children.

- **Implications for Research:**
  These findings highlight the need to consider cultural context when evaluating the psychosocial impacts of enamel defects. Future research should aim to explore cultural attitudes in a wider range of populations to understand how they influence treatment-seeking behaviors, stigma, and coping mechanisms.

2.5 Implications for Practice

2.5.1 Addressing Functional and Aesthetic Impacts

Given the multifaceted impact of enamel defects, a multidisciplinary approach is essential for effective management. Collaboration among paediatric dentists, psychologists, nutritionists, educators, and policymakers can address the broad range of challenges faced by children and their families:

Management strategies for enamel defects can be categorized by their impact on functionality and aesthetics, with treatments progressing from least invasive to more advanced interventions.

Role in Addressing Functional Impacts:

Minimally Invasive Treatments:

- Application of fluoride varnishes and remineralization agents to strengthen enamel and reduce hypersensitivity (Dias et al., 2020).
- Use of glass ionomer sealants to protect molars affected by MIH, which has been shown to reduce hypersensitivity by 40% (Hasmun et al., 2020).

Intermediate Interventions:

- Preformed metal crowns (PMCs) are effective in protecting structurally compromised teeth and preventing further breakdown, especially in cases of MIH and severe hypoplasia (Elhennawy et al., 2022).
- Composite resin restorations for localized enamel defects to restore both function and aesthetics, particularly in anterior teeth (Michaelis et al., 2022).

Advanced Functional Restorations:

- Placement of full-coverage custom-made crowns or overlays for severely compromised teeth, ensuring long-term durability and functional restoration (Vargas-Ferreira et al., 2011).

- Endodontic treatments in cases where structural defects lead to pulpal involvement, often necessary in severe cases of MIH or hypoplasia (Silva et al., 2021).

Role in Addressing Aesthetic Impacts:

Minimally Invasive Aesthetic Interventions:

- Microabrasion to remove superficial discoloration, particularly in cases of mild hypomineralization or diffuse opacities (Gutierrez et al., 2019).
- Use of RI techniques, which can improve aesthetics by masking discoloration and stabilizing enamel (Dias et al., 2020).

Intermediate Aesthetic Treatments:

- Composite bonding to improve the appearance of discolored or misshapen anterior teeth, providing a cost-effective and conservative option for adolescents (Michaelis et al., 2022).
- In-office bleaching for cases of generalized discoloration, which has shown to improve self-esteem in over 70% of adolescents with visible enamel defects (Lyne et al., 2021).

Advanced Aesthetic Restorations:

- Veneers or ceramic crowns for older adolescents and young adults to achieve optimal aesthetic outcomes in cases of severe discoloration or structural anomalies (Hasmun et al., 2020).
- Orthodontic treatments to address malocclusions that may arise due to enamel defects, improving both aesthetics and function (Barros et al., 2022).

2.5.2 Role in Collaborating with Other Sectors

Psychologists

Psychologists can play a vital role by implementing school-based or clinic-based counselling programs to boost self-esteem and reduce social anxiety. For instance, Dias et al. (2020) found that 65% of children with MIH reported social withdrawal due to teasing or bullying, highlighting the need for accessible psychological support. School counsellors and teachers should also be trained to recognize signs of emotional distress in children with visible enamel defects and provide appropriate interventions. Peer-support groups can further help children share experiences and coping strategies, fostering social integration, and reducing stigma. Additionally, psychologists can support parents by addressing the emotional toll of their child's condition, helping caregivers manage feelings of guilt or frustration, and equipping them with strategies to bolster their child's self-esteem.

Nutritionists

Collaboration with nutritionists is essential to address the dietary challenges posed by enamel defects. Tailored dietary guidance can help reduce enamel sensitivity and prevent further deterioration, as emphasized by Silva et al. (2021), who reported that 45% of children with enamel hypoplasia experienced eating difficulties. Dentists and nutritionists can conduct educational workshops for parents to equip them with strategies for managing their child's dietary needs, ensuring balanced nutrition while avoiding foods that exacerbate sensitivity. Addressing these challenges early can prevent further oral health complications and improve children's quality of life.

Educators:

Educators play a critical role in mitigating the academic and social impacts of enamel defects. Teachers should be trained to recognize the challenges these conditions pose, such as absenteeism, difficulty concentrating, and social withdrawal. For instance, Andrade et al. (2019) noted that children with enamel defects often avoided oral presentations or other social activities in class due to self-consciousness. Educators can adapt classroom activities to support these students, offering alternatives like written assignments in place of public speaking. School administrators can collaborate with dental professionals to ensure children have access to academic and emotional support, including flexibility for missed school days due to dental appointments or psychosocial challenges. Raising awareness among educators can foster a more inclusive and supportive environment for children with enamel defects (Arrow, 2017).

Policymakers:

Policymakers must address the systemic challenges associated with enamel defects by implementing public health initiatives to improve access to care. Subsidized treatment programs can reduce the financial burden on families, particularly those from low-income backgrounds (Folayan et al., 2018). Integrating oral health services into school health programs can facilitate early-stage screening, fluoride applications, and referrals for specialized care. Community awareness campaigns are also critical to reducing stigma and encouraging families to seek care for enamel defects. By addressing these systemic barriers, policymakers can improve health equity and ensure timely interventions for children in need.

2.6 Gaps in the Literature

Despite significant advancements in understanding the impacts of developmental enamel defects (DEDs) on children's quality of life, several critical gaps remain. These gaps highlight the need for more robust and comprehensive research to address methodological limitations, demographic variations, and underexplored outcomes

2.6.1 Methodological Gaps

Lack of Longitudinal Studies

The lack of longitudinal studies is a major limitation in the current literature. Most reviewed studies utilized cross-sectional designs, capturing data at a single point in time. This approach limits the ability to understand how DEDs influence oral health-related quality of life (OHRQoL) across different developmental stages or to assess the long-term outcomes of interventions. For example, studies by Arrow (2017) and Dias et al. (2020) focused on immediate impacts, leaving the long-term trajectory of these impacts unclear.

Variability of Assessment Tools

One of the key challenges in enamel defect research is the variability in assessment tools, which complicates cross-study comparisons and the synthesis of findings. Current tools such as the CPQ, ECOHIS, and Visual Analog Scale (VAS) vary in scope and application. For example, CPQ emphasizes psychosocial impacts, while ECOHIS is more focused on functional outcomes (Bekes et al., 2021; Dias et al., 2020).

To address this issue, a standardized framework combining the strengths of existing tools is needed. Such a framework could include:

- **Functional Domains:** Pain, sensitivity, and dietary impacts (e.g., from ECOHIS).
- **Psychosocial Domains:** Self-esteem, social interactions, and emotional well-being (e.g., from CPQ).
- **Cultural Adaptability:** Items tailored to reflect cultural differences in the perception and impact of enamel defects.

Developing and validating such a tool would enhance the comparability of future studies and improve clinicians' ability to monitor outcomes consistently across diverse populations.

2.6.2 Demographic Gaps

There is limited data comparing the impact of DEDs across different age groups. Studies like Bekes et al. (2021) and Boukhobza et al. (2022) focused on specific age groups without contrasting age-related differences in impact. Adolescents may face unique social and psychological challenges due to DEDs, highlighting the need for age-specific interventions. Gender differences are similarly underexplored, with preliminary findings suggesting that girls may be more affected by the aesthetic implications of enamel defects, while boys are more likely to report functional challenges (Hasmun et al., 2020).

DEDs impose significant socioeconomic and psychological burdens, yet these dimensions remain underexplored in the literature. While some studies, such as Folayan et al. (2018) and de Barros et al. (2022), have examined the role of socioeconomic status in shaping the impact of DEDs on OHRQoL, they often overlook how cultural perceptions influence treatment-seeking behaviours and coping mechanisms. Socioeconomic disparities, particularly in low- and middle-income populations, exacerbate access to care and financial barriers, leaving families with limited options for timely intervention. Cultural differences, including societal norms regarding aesthetics and oral health, further shape the experiences of affected children and their families, underscoring the need for cross-cultural studies.

In addition to these systemic factors, the psychological toll on families is under-researched. Silva et al. (2021) highlighted the socioeconomic burden on caregivers, but broader family dynamics, including caregiver stress, parental mental health, and relationships within the household, require further investigation. Caregivers of children with visible or functionally debilitating DEDs often report feelings of guilt, stress, and helplessness. For example, untreated enamel defects causing pain or social stigma can strain family routines and finances, deepening the psychosocial burden.

2.6.3 Outcome Specific Gaps

Several key outcomes of DEDs remain underexplored. One significant gap is the limited research on the long-term health impacts of untreated enamel defects. While studies like Vargas-Ferreira et al. (2011) have highlighted increased risks of caries and periodontal disease, there is little evidence on how these conditions affect systemic health or oral function over time.

Emotional and behavioral outcomes are also inadequately addressed. While studies by Dias et al. (2020) and Marshman et al. (2009) have reported behavioral avoidance and social anxiety among children with DEDs, few have examined deeper emotional impacts, such as depression or chronic anxiety. Comprehensive assessments of mental health outcomes are needed to fully understand the psychological toll of these conditions.

Finally, the academic consequences of DEDs have received insufficient attention. Although Andrade et al. (2019) noted school absenteeism and reduced participation in classroom activities, there is little evidence on how these disruptions affect academic performance or long-term educational outcomes. Future research should investigate these impacts in greater detail and explore strategies for mitigating them within educational settings.

2.7 Scoping Review Summary

This scoping review analyzed 25 studies to explore the impact of developmental enamel defects (DEDs) on children's oral health-related quality of life (OHRQoL). The findings highlight significant challenges posed by these defects, spanning psychosocial, functional, socioeconomic, educational, emotional, and behavioral domains. Among the studies, molar-incisor hypomineralization (MIH) was the most frequently explored defect (60%), followed by chronological enamel hypoplasia (20%), amelogenesis imperfecta (12%), and non-specific defects (28%). Psychosocial impacts, including bullying, social withdrawal, and low self-esteem, were the most frequently reported (65%), followed by functional impacts such as pain and sensitivity (50%). Despite these insights, research into educational, emotional, and long-term health impacts remains sparse, with fewer than 20% of studies addressing these areas.

The variability in tools used to measure OHRQoL, such as the CPQ, ECOHIS, and COHIP, presents a barrier to synthesizing findings across studies. Standardizing assessment tools to incorporate functional, emotional, and cultural dimensions would improve comparability and guide interventions. Additionally, significant gaps persist, including the lack of longitudinal studies, limited focus on age-specific and gender-specific impacts, and inadequate exploration of socioeconomic and cultural disparities. These gaps hinder a full understanding of how DEDs affect children's overall quality of life.

Addressing these limitations requires a multidisciplinary approach, involving pediatric dentists, psychologists, educators, policymakers, and families to develop tailored interventions. Schools have a vital role in providing support, while policies to reduce financial and access barriers can alleviate socioeconomic strain. Standardized methodologies and tools, coupled with greater focus on underserved populations, will advance research and clinical care.

Building on these findings, the next chapter focuses on a retrospective analysis of patient-reported outcomes using the AI Patient-Reported Outcome Measure (PROM).

This study aims to quantify the impact of treatments and demographic factors, providing actionable insights to enhance patient-centered care and address the identified knowledge gaps.

Chapter Three: Service Evaluation of Patient-Reported Outcomes Using the AI PROM

Chapter Three provides an overview of the retrospective analysis of AI PROM data collected from children and young people with AI. This chapter presents a service evaluation of their experiences through patient-reported outcomes before and during treatment, focusing on understanding the challenges they face and the impact of dental interventions. Key aspects include the aims and objectives of the AI PROM, its development and validation, and a summary of data collected in previous studies. These findings provide the foundation for this retrospective analysis, which seeks to identify longitudinal changes and subgroup variations in patient-reported outcomes to enhance clinical understanding and improve patient-centred care.

3.1 Background Information on the AI PROM

3.1.1 AI PROM Aims and Objectives

The purpose of the AI PROM was to explore the challenges that children and young people with AI face before, during, and after dental treatment.

The objectives included identifying the following: the impact of AI on the lives of children and young people living with such dental condition (such as school attendance, self-confidence, and peer relationships) and whether they were satisfied or happy with their teeth in terms of aesthetics and function.

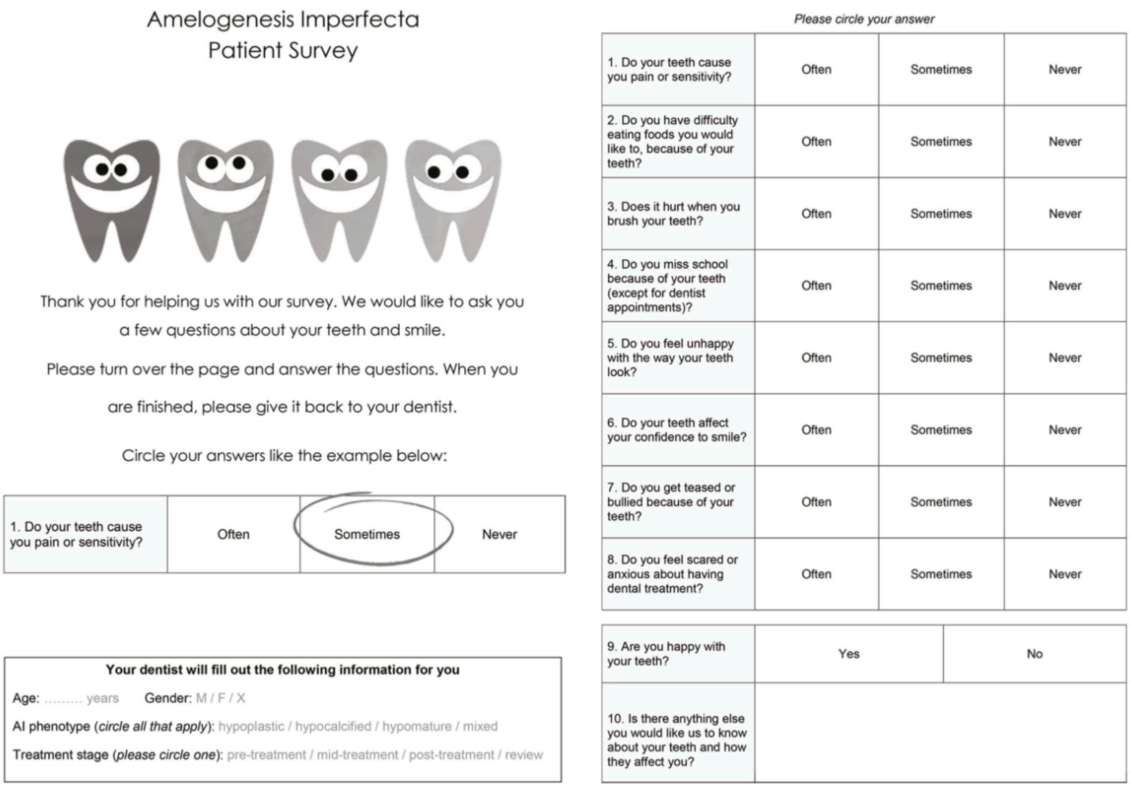
3.1.2 Development of the AI PROM

In 2019, a patient-reported outcome measure (PROM) for children and young adults with AI was developed at Eastman Dental Hospital (EDH) with input from both clinicians and patients. The PROM was peer reviewed by the national AI Clinical Excellence Network (AICEN) and piloted with ten children and young people, aged 9-17 years with AI, attending dental clinics at EDH and Sheffield Dental Hospital (Lyne et al., 2019).

The development process included rigorous testing of the PROM's psychometric properties, including assessments of its reliability and validity to ensure it effectively captured the impact of AI on OHRQoL. Cognitive testing was carried out with participants to confirm the clarity and relevance of the questions, and feedback was incorporated to refine the tool.

The final version of the PROM consists of two pages and is presented in a paper format rather than an electronic version to improve accessibility across diverse clinical settings (Figure 17). Visual aids, such as smiley face Likert scales, were incorporated to ensure it was child-friendly and intuitive. The questionnaire was designed to be completed within 5–10 minutes, reducing patient burden while maintaining comprehensiveness.

To accommodate children under ten years of age, the PROM was deemed suitable for completion with parental assistance. Furthermore, to ensure accessibility for individuals with colour vision deficiencies, the final questionnaire was presented entirely in black and white. A readability assessment using **Readable (2020)** confirmed the questionnaire was appropriate for children with a reading age of 9.5 years.



Amelogenesis Imperfecta
Patient Survey

Thank you for helping us with our survey. We would like to ask you a few questions about your teeth and smile.

Please turn over the page and answer the questions. When you are finished, please give it back to your dentist.

Circle your answers like the example below:

1. Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never

Your dentist will fill out the following information for you

Age: .... years Gender: M / F / X

AI phenotype (*circle all that apply*): hypoplastic / hypocalcified / hypomature / mixed

Treatment stage (*please circle one*): pre-treatment / mid-treatment / post-treatment / review

Please circle your answer

1. Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never
2. Do you have difficulty eating foods you would like to, because of your teeth?	Often	Sometimes	Never
3. Does it hurt when you brush your teeth?	Often	Sometimes	Never
4. Do you miss school because of your teeth (except for dentist appointments)?	Often	Sometimes	Never
5. Do you feel unhappy with the way your teeth look?	Often	Sometimes	Never
6. Do your teeth affect your confidence to smile?	Often	Sometimes	Never
7. Do you get teased or bullied because of your teeth?	Often	Sometimes	Never
8. Do you feel scared or anxious about having dental treatment?	Often	Sometimes	Never

9. Are you happy with your teeth?	Yes	No
10. Is there anything else you would like us to know about your teeth and how they affect you?		

Figure 17 Final Version of the AI PROM

3.1.3 Previous Data Collection Summary

The final PROM questionnaire was distributed to all patients with AI who attended six specialist units that agreed to participate in the study between January and March 2020. Two units were unable to access their completed questionnaires due to clinic closures during the Covid-19 Pandemic, resulting in their exclusion from the final analysis. Therefore, data from four out of six eligible units were analysed using descriptive statistics in Microsoft Excel™ 2010, representing 67% of the originally intended sample.

The PROM was administered to patients at three time points: before treatment (pre-treatment), during treatment (mid-treatment), and after treatment (post-treatment), allowing for an evaluation of patient experiences across the treatment pathway.

A total of 60 CYPs (aged 5-17 years) completed the PROM, of these:

- **20 CYP (33%)** were evaluated in the pre-treatment stage.

- **18 CYP (30%)** were evaluated in the mid-treatment stage.
- **22 CYP (37%)** were evaluated in the post-treatment stage.

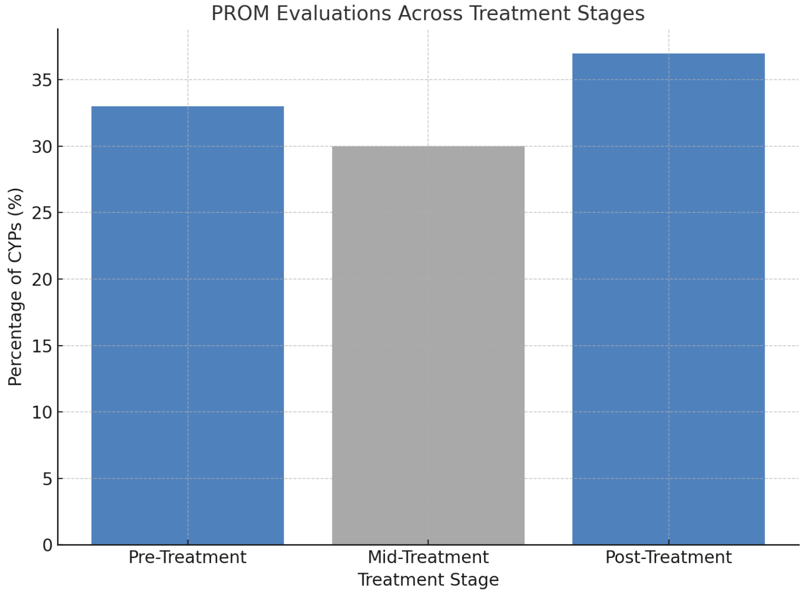


Figure 18 PROM evaluations across treatment stages, showing the distribution of CYPs in pre-treatment, mid-treatment, and post-treatment stages.

The following overall findings were observed across the entire cohort, combining data from all treatment stages:

- **72% (43 out of 60 CYP)** reported that they 'often' or 'sometimes' experienced pain or sensitivity with their teeth.
- **76% (46 out of 60 CYP)** reported that they 'often' or 'sometimes' felt unhappy with the way their teeth looked.

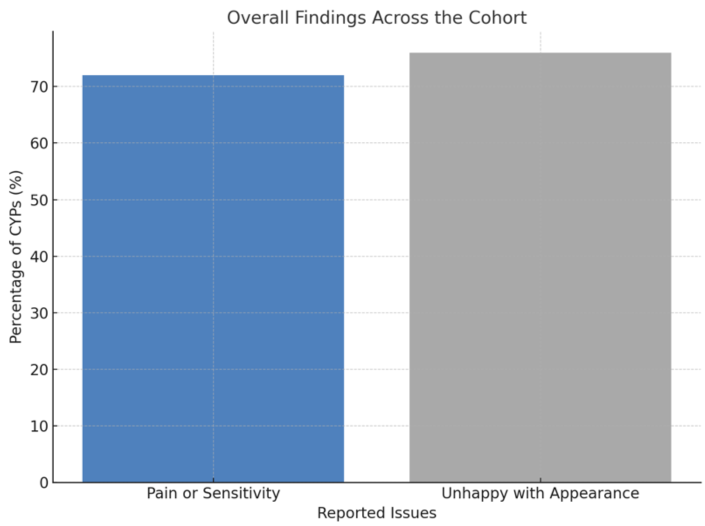


Figure 19 Bar chart showing the overall findings across the cohort.

Post-treatment evaluations showed a significant improvement in satisfaction, with 81% (18 out of 22 CYP) indicating that they were happy with their teeth. This contrasted with 41% (7 out of 18 CYP) at the mid-treatment stage and just 33% (7 out of 20 CYP) at the pre-treatment stage.

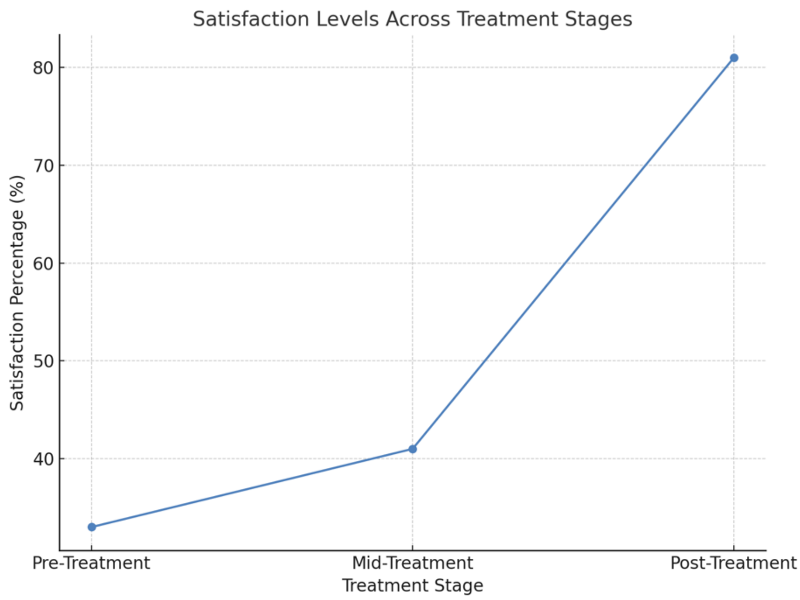


Figure 20 Line chart showing the change in satisfaction levels across treatment stages.

This dataset provides valuable insights into the evolving patient experiences and perceptions at different stages of treatment and highlights the positive impact of completed dental interventions.

The AI PROM was found to serve as an effective tool for children and young people to express their concerns, emphasising the need for personalised patient care. It revealed that patients with all types of AI experience a wide range of issues, as reflected in their responses. This reinforces the importance of providing holistic and individualised care to meet the diverse needs of these patients.

3.2 Aims and Objectives

3.2.1 Aims

The aim of this study was to explore the experiences of CYP with AI through PROMs. Building on the original study by Lyne et al. (2019), this research expanded the scope to address key questions and provide deeper insights into the challenges and outcomes of CYP with AI.

3.2.2 Objectives

The objectives of the study were to:

1. **Evaluate longitudinal changes** in PROM responses, specifically comparing pre-treatment and mid-treatment stages, to understand how symptoms, functional limitations, and psychosocial impacts evolve over the treatment pathway.

2. **Examine the relationship between specific treatments** (e.g., tooth whitening, anterior restorations) and changes in PROM responses, to assess the impact of these interventions on patient experiences and QoL.

3. **Explore subgroup differences** by analysing PROM responses based on age and AI type (Hypomature, Hypocalcified, Hypoplastic), providing an understanding of variations within the cohort.

4. **Explore the following aspects**:
   The changes in patient-reported symptoms, such as pain and sensitivity, over time during treatment.
   The influence of aesthetic and functional treatments, such as whitening and restorations, on psychosocial factors like confidence and satisfaction with teeth.
   The impact of age or AI type on patient-reported outcomes and the nature of any observed differences.
   The insights provided by the extended two-year timeframe of this study compared to the original three-month validation study.

By addressing these aims and objectives, this study aims to provide a comprehensive understanding of the lived experiences of CYP with AI, contributing valuable knowledge to support patient-centred care and PROM-based assessments.

3.3 Methodology

3.3.1 Approvals and Ethical Considerations

This project was carried out as a service evaluation to assess patient-reported outcomes using the AI PROM in routine clinical care. A service evaluation, as defined by NHS guidelines, involves a systematic review of services against established standards to improve patient care. This method was chosen as it allows the retrospective analysis of routinely collected data without the formulation of new hypotheses or experimental interventions, ensuring relevance to real-world clinical settings.

The service evaluation was registered with the clinical governance lead in the department, and the protocol and results were presented at departmental clinical governance meetings for peer review. Patient confidentiality was maintained throughout the study in full compliance with UCL's data management policies and the NHS Trust's data protection guidelines.

3.3.2 Study Design

This service evaluation analysed AI PROM responses collected from CYP diagnosed with AI. Data were collected over a two-year period (January 1, 2022, to January 1, 2024) to evaluate longitudinal changes in PROM responses between pre-treatment and mid-treatment stages. Unlike previous data collection efforts that included both a mid-treatment and post-treatment evaluation, for this analysis, mid-treatment was chosen to collectively represent anyone who has had undergone treatment for AI (whether completed or not) due to the understanding that AI patients require lifelong management and treatment is almost never definitive. The analysis aimed to explore physical symptoms, functional limitations, aesthetic concerns, social impacts, and overall patient satisfaction with treatment.

Key characteristics of the study design include:

- **Retrospective Nature**: Data were retrieved from existing electronic patient records (EPIC) at EDH.

- **Observational Focus**: The intervention in this study was the administration of the AI PROM, which was used to gather patient-reported outcomes. No new treatments were provided as part of the study; rather, this section evaluates outcomes based on treatments that had already been administered to patients.

- **Descriptive and Comparative Analysis**: PROM responses were summarized descriptively, and subgroup analyses were conducted to explore trends by treatment type, AI subtype, and age.

This type of study allows for a detailed examination of real-world patient experiences and treatment outcomes without the introduction of experimental variables, ensuring relevance to clinical practice.

3.3.3 Eligibility Criteria

3.3.3.1 Inclusion Criteria

- CYPs with a confirmed clinical diagnosis of AI (Diagnoses were made clinically by paediatric dentists at EDH based on clinical examination and detailed patient history)
- CYP attending the Anomalies clinic at EDH between Jan 1st 2022 – Jan 1st 2024.
- CYP who had at least 2 completed AI PROMs
    1. A Pre-Treatment PROM (First PROM completed between the ages of 6 and 18 years)
    2. A Mid-Treatment PROM (If multiple were available – the most recent AI PROM was selected)

3.3.3.2 Exclusion Criteria

- CYPs with no confirmed diagnosis of AI.
- CYPs with incomplete or missing PROM responses.
- CYPs with only 1 completed PROM

3.3.4 Instruments and Data Collection/Analysis

Instruments

The AI PROM (Figure 17) is a PROM developed for CYPs with AI (Lyne et al., 2019). It is accessible for individuals with a reading age of 9.5 years and is designed to be completed with parental assistance for younger children. The questionnaire was routinely administered to all AI patients attending EDH as part of standard clinical care and was scanned and uploaded to the EPIC database.

Data Collection

The researcher accessed the EPIC database to identify eligible patients and extract relevant data. The process involved:

A. Identification of Eligible Patients:
   - A database search was conducted using the electronic healthcare database at Eastman Dental Hospital (EDH), known as EPIC. The search focused on identifying patients diagnosed with amelogenesis imperfecta (AI) who attended the Anomalies Clinic during the inclusion period.
   - Patient ID numbers were recorded and managed in accordance with NHS's data protection policies.

B. Review of Patient Records:
   - The researcher accessed patient notes to confirm AI diagnosis, treatment details, and the availability of at least two completed AI PROMs (pre-treatment and mid-treatment.
   - After further revision, patient ID numbers that were recorded in step 1 that did not meet the inclusion criteria were removed.

C. Manual Data Extraction and Data Entry:
   - Systematically transferring PROM responses into a pre-designed Microsoft Excel™ spreadsheet.

     This spreadsheet was structured to include:
     - Dates of PROM completion

- Demographic details (Age at first PROM, gender)
- Clinical details (Type of AI, treatments received)
- AI PROM responses (Extracted for both pre-treatment and mid-treatment stages)

This meticulous process ensured that all relevant information was organized for the audit analysis while maintaining confidentiality and compliance with ethical guidelines. To minimize data entry errors, the researcher implemented a quality control process. Data was entered in batches of five records, followed by cross-checking each batch against the original source to ensure accuracy. Any discrepancies were promptly identified and corrected. By using Excel™, the researcher was able to streamline data organization and facilitate descriptive and comparative analysis with high reliability.

Data Analysis

The audit data was analysed using descriptive statistics in Microsoft Excel™ 2010. Longitudinal changes in PROM responses were examined to identify trends in pain, sensitivity, confidence, and social factors.

Subgroup analyses were conducted to explore differences by:

- **Treatment type**: e.g., teeth whitening, anterior restorations.
- **AI subtype**: Hypomature, Hypocalcified, Hypoplastic.
- **Age group**: <13 years vs. ≥13 years.

3.4 Results of AI PROM Service Evaluation

3.4.1 Demographic Overview

A total of 257 AI patients were identified on the patient record database (EPIC) when searching within the inclusion period (January 1, 2022, to January 1, 2024). After applying the inclusion criteria, which required patients to have at least two completed AI PROMs (pre-treatment and mid-treatment), 68 patients were found to meet all criteria. These 68 patients' AI PROM responses were included in this study. No unusable or spoiled questionnaires were identified during the data extraction process, as all PROMs reviewed were completed appropriately and uploaded to the EPIC database as part of routine clinical care.

The demographic analysis revealed that most participants in the study were female (58.8%), with males comprising 41.2% of the sample. The participants' ages ranged from seven to 19 years, with the first AI PROM completed at an average age of 12 years and the most recent PROM at an average age of 13 years. On average, there was a 10-month interval between the two assessments. The most common AI type observed was hypomature (72%), followed by hypoplastic (13%) and hypocalcified (9%). Combined types, such as hypomature and hypoplastic or hypocalcified and hypoplastic, were less frequently reported, respectively.

Table 9 provides an overview of the key demographic characteristics of the study cohort, including age, gender distribution, and the breakdown of AI types.

Demographic	Value	Percentage
Total Participants	68	100%
Mean Age (1st PROM)	12 years	-
Mean Age (Most Recent PROM)	13 years	-
Gender - Male	28	41.2%
Gender - Female	40	58.8%
AI Type – Hypomature (HM)	*49*	72%
AI Type – Hypocalcified (HC)	*6*	9%
AI Type – Hypoplastic (HP)	9	13%
AI Type – Hypomature & Hypoplastic (HM-HC)	3	4.5%
AI Type – Hypocalcified & Hypoplastic (HC/HP)	1	1.5%

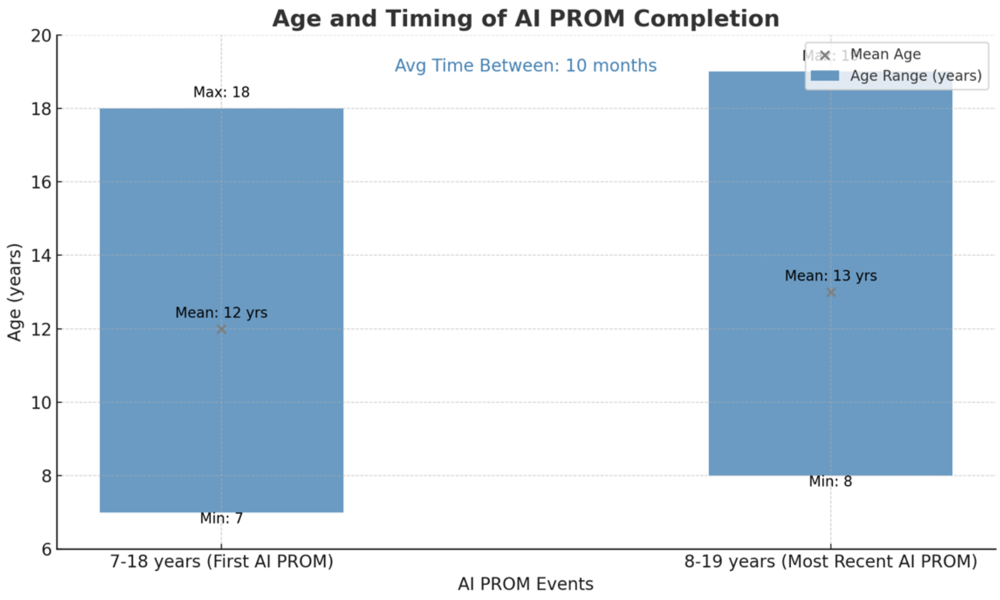


Figure 21 Range chart summarizing the age ranges and timing for the AI PROM completions.

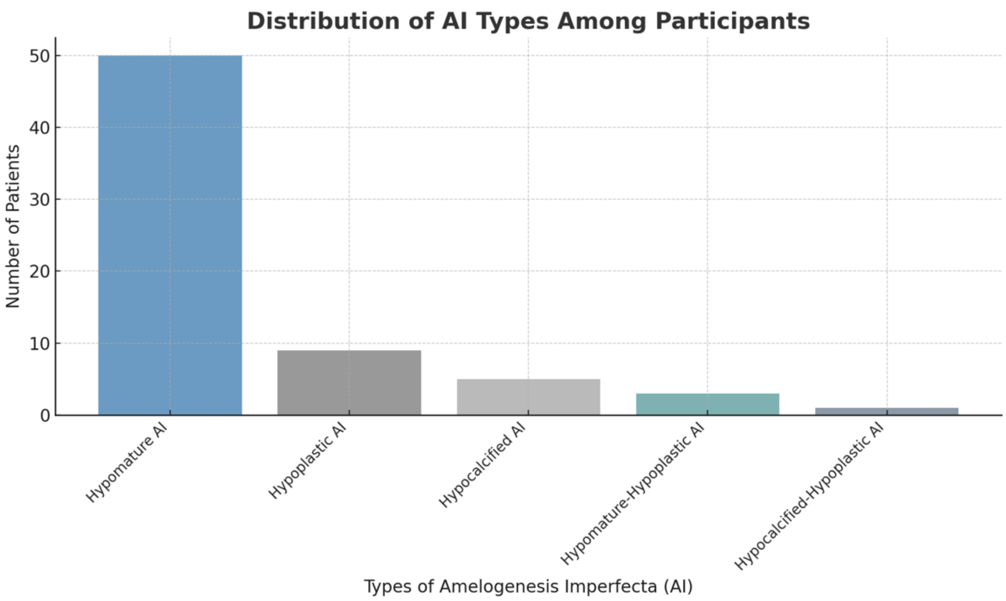


Figure 22 Bar chart representing the distribution of AI types among participants.

3.4.2 Treatments Provided

The treatments most frequently administered to CYP with AI included tooth whitening (26%) and fissure sealants (FS) (18%). These interventions underscore the dual focus on cosmetic and preventive care within this population. Less commonly provided treatments included ICON (4%), a resin infiltration system used for treating early caries and white spot lesions, and composite restorations, representing the smallest proportions among the available interventions. This distribution reflects a treatment approach tailored to managing both functional and cosmetic aspects of AI.

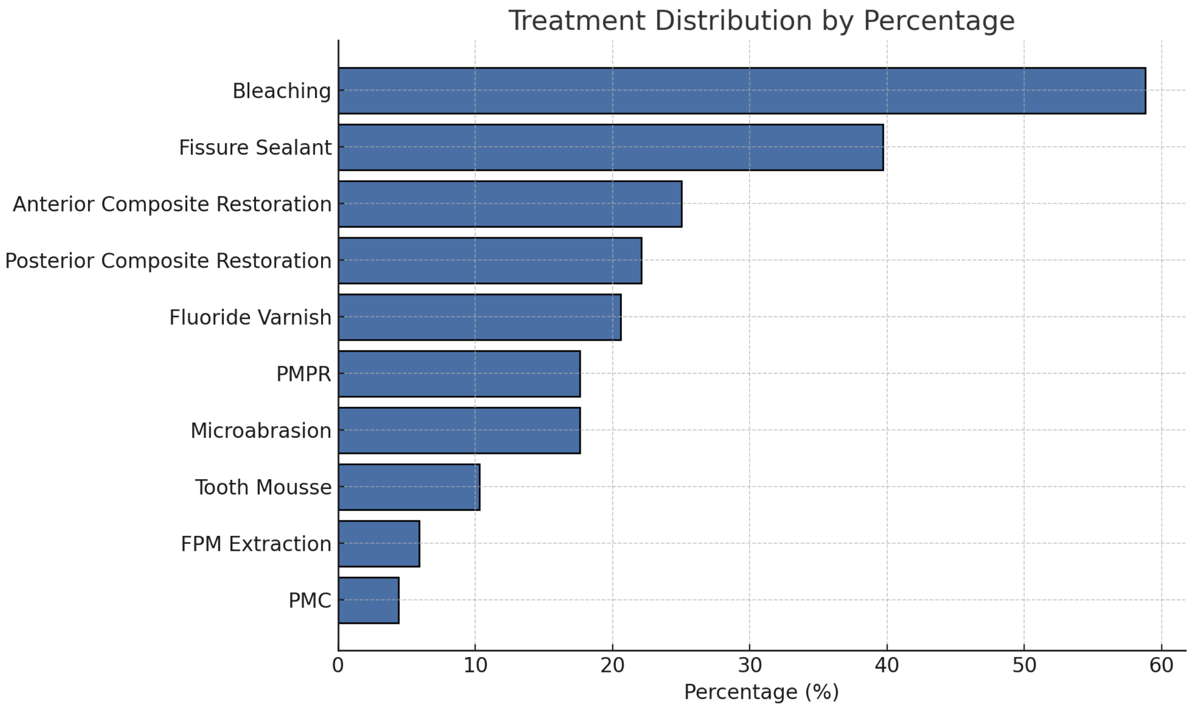


Figure 23 Horizontal bar chart displaying the distribution of treatments provided to patients in the cohort.

Among the study population, 25% (17 patients) received anterior composite restorations. The most common treatment involved six teeth (UR3-UL3), accounting for 41% of the recipients of anterior composite restorations (7/17 patients). By contrast, treatments involving one, two, four, 10, or 12 teeth were less common, each representing a smaller proportion of the recipients. This highlights the predominance of midline aesthetic treatments involving six teeth among patients receiving anterior composite restorations.

A total of 22% (15 patients) received posterior composite restorations. Of these, 73% (11/15 patients) involved single-tooth restorations, highlighting their primary use in addressing localized issues. Multi-tooth restorations, involving two or six teeth, were less common, reflecting their more selective application.

In the cohort, 17% (12/68) underwent microabrasion. The majority (58%) received treatment for two teeth, making it the most common application. Single-tooth and four-tooth treatments were less frequent, accounting for 25% and 16% of microabrasion recipients, respectively.

Among the patients who received teeth whitening treatments (n=40), the majority (20 patients, 50%) underwent two cycles, making it the most common regimen. hirteen patients (32%) received one cycle of treatment, while six patients (15%) underwent three cycles. Extended regimens, such as six cycles, were rare, with only one patient (2.5%) receiving this level of treatment. This distribution underscores the tendency toward shorter whitening regimens for AI patients.

Among the four patients who received PMC treatments, the distribution was evenly divided. Two patients (50%) had two PMCs placed, while the remaining two patients (50%) had four PMCs placed. This accounted for 3% of the total study population (n=68) in each category, highlighting the selective use of PMCs in specific clinical scenarios.

For dental extractions, the majority (75%) of patients had a single tooth extracted, accounting for 4% of the total study population (n=68). [The remaining patient (25%) underwent multiple extractions, having four teeth removed, which represented 1.5% of the total study group. This indicates a preference for conservative extraction approaches when managing AI cases.]

Among the treatments provided to CYP with AI, tooth whitening emerged as the most frequently delivered intervention, representing 26% of the total treatments. This was followed by fissure sealants (FS) at 18% and anterior composite restorations at 11%. Posterior composites (7%), microabrasion (7%), and ICON resin infiltration (4%) accounted for smaller portions of the treatments. Preventive measures, such as fluoride varnish (8%) and tooth mousse (5%), were also utilized, alongside more specialized treatments like PMCs (3%) and dental extractions (3%). This distribution highlights a multidisciplinary approach to managing both aesthetic concerns and oral health stability for children with AI.

3.4.3 AI PROM Responses

The AI PROM study aimed to evaluate patients' responses to various questions regarding their oral health and the impact of AI treatments over time. This study focuses on the comparison of pre-treatment and mid-treatment responses across several dimensions to assess changes in patient experiences. The analysis was conducted in a structured manner to ensure comprehensive insights into treatment outcomes.

First, an overall comparison of pre-treatment and mid-treatment responses was performed across all patients. This general overview provided a broad understanding of how AI treatments influenced patient perceptions and experiences over time.

Subsequently, AI-specific comparisons were conducted to explore how treatment impacts varied across the different AI phenotypes such as Hypomature (HM), Hypocalcified (HC), and Hypoplastic (HP) AI types. As each phenotype has distinct enamel characteristics, such as variations in thickness, hardness, and colour, it would be useful to explore the different clinical challenges and treatment needs of each subtype.

In addition to examining AI phenotypes, responses were analysed based on age groups to capture potential variations in treatment impact across developmental stages. The two age groups examined were patients under age 13 and those aged 13 and above.

These comparisons aimed to provide a nuanced understanding of how AI treatment outcomes are influenced by age and phenotype, ensuring a comprehensive evaluation of the PROM responses.

Question 1: Do your teeth cause you pain or sensitivity?

Pre-Treatment vs. Mid-Treatment Comparison:

After treatment, there was a slight increase in frequent pain or sensitivity and a decrease in the number of patients who reported no pain.

Table 10 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q1.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	33 (48%)	29 (42%)
Sometimes	31 (45%)	33 (48%)
Often	4 (7%)	6 (10%)

AI-Specific Comparisons:

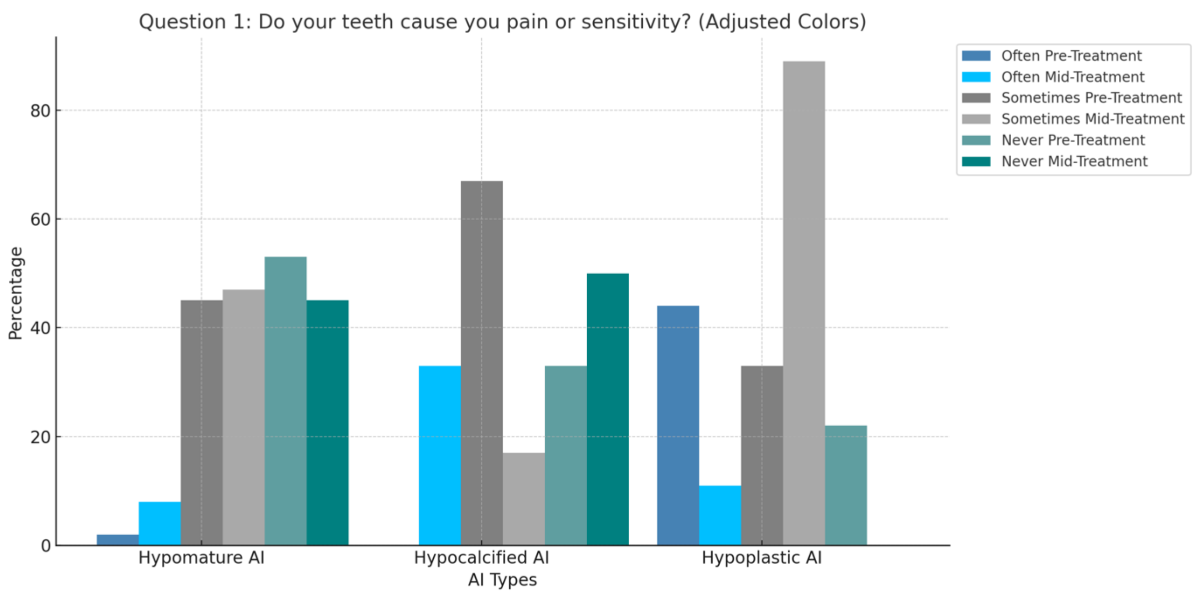


Figure 24 Grouped bar chart comparing reported pain and sensitivity levels by AI type at pre and mid treatment stages.

For **Hypomature AI**, there was minimal change in sensitivity post-treatment. The proportion of patients reporting no pain ("Never") decreased slightly from 53% (26/49) pre-treatment to 45% (22/49). Meanwhile, those experiencing frequent pain ("Often") increased from 2% (1/49) to 8% (4/49), suggesting a slight worsening in sensitivity for some patients.

In **Hypocalcified AI**, treatment showed mixed results. The proportion of patients reporting no pain improved from 33% (2/6) pre-treatment to 50% (3/6), but frequent pain increased from 0% to 33% (2/6), indicating both improvement and worsening within this group.

For **Hypoplastic AI**, sensitivity outcomes were the least favourable. No patients reported being pain-free post-treatment, with the "Never" category dropping from 22% (2/9) to 0%. Most patients shifted to the "Sometimes" category, which rose from 33% (3/9) to 89% (8/9), though frequent pain ("Often") decreased from 44% (4/9) to 11% (1/9).

Age-Based Comparisons:

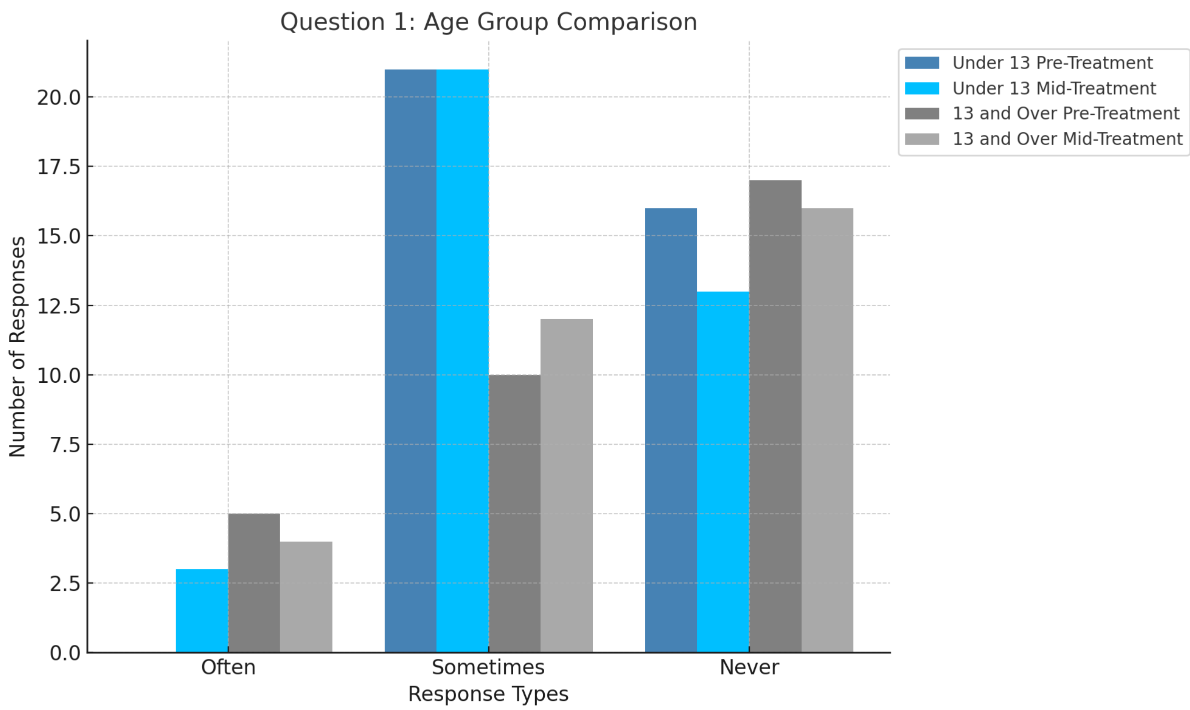


Figure 25 Grouped bar chart comparing reported pain and sensitivity levels by age group at pre- and mid-treatment stages.

Post-treatment, the proportion of patients reporting no pain ("Never") decreased in both age groups. Among those **under 13**, "Never" responses dropped from 73% to 68%, with a small increase in frequent pain ("Often") from 0% to 5%. In the **13 and over** group, the decline in the "Never" category was more pronounced, from 61% to

53%, and frequent pain responses remained slightly higher, increasing from 11% to 14%. These results suggest that older patients were more likely to experience persistent pain or sensitivity post-treatment.

Question 2: Do you have difficulty eating foods you would like to, because of your teeth?

Pre-Treatment vs. Mid-Treatment Comparison:

The number of participants reporting no difficulty increased, while those reporting "Sometimes" decreased slightly.

Table 11 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q2.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	45 (66%)	48 (70%)
Sometimes	22 (32%)	17 (25%)
Often	1 (2%)	3 (5%)

AI-Specific Comparisons:

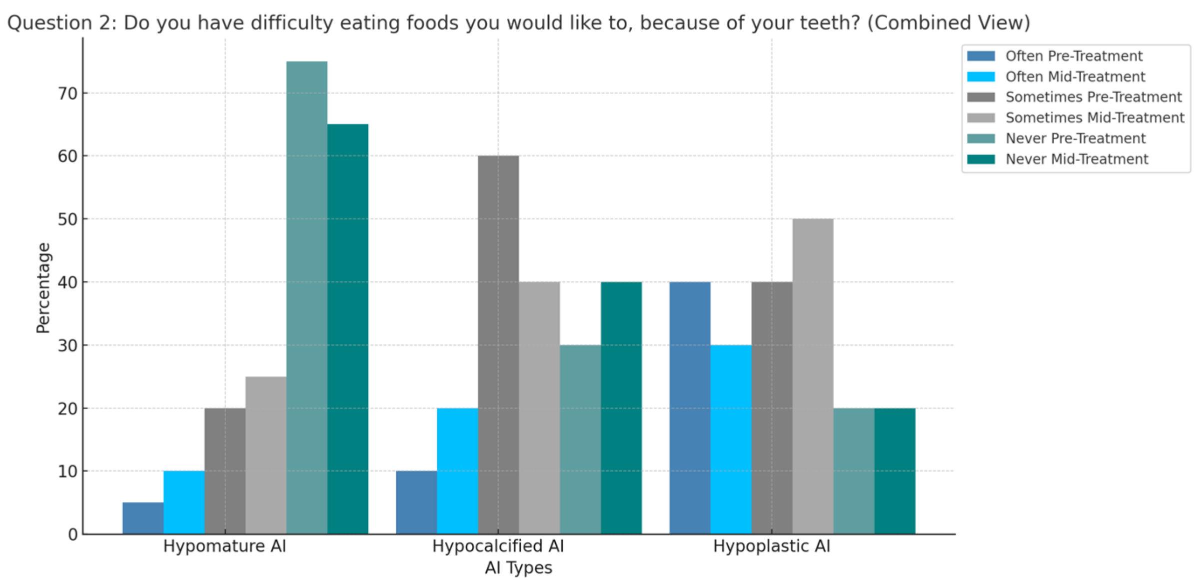


Figure 26 Grouped bar chart comparing reported difficulty in eating by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, there was a moderate improvement. The proportion of patients reporting no difficulty ("Never") increased from 59% (29/49) pre-treatment to 67% (33/49), while those experiencing occasional difficulty ("Sometimes") decreased from 37% (18/49) to 29% (14/49).

Patients with **Hypocalcified AI** also showed improvement. Those reporting no difficulty rose from 33% (2/6) to 67% (4/6), while "Sometimes" responses dropped from 67% (4/6) to 33% (2/6).

For **Hypoplastic AI**, moderate improvement was observed. The proportion reporting no difficulty increased from 33% (3/9) to 56% (5/9), while "Sometimes" responses decreased from 56% (5/9) to 33% (3/9). Frequent difficulty ("Often") remained consistent at 11% (1/9).

Age-Based Comparisons:

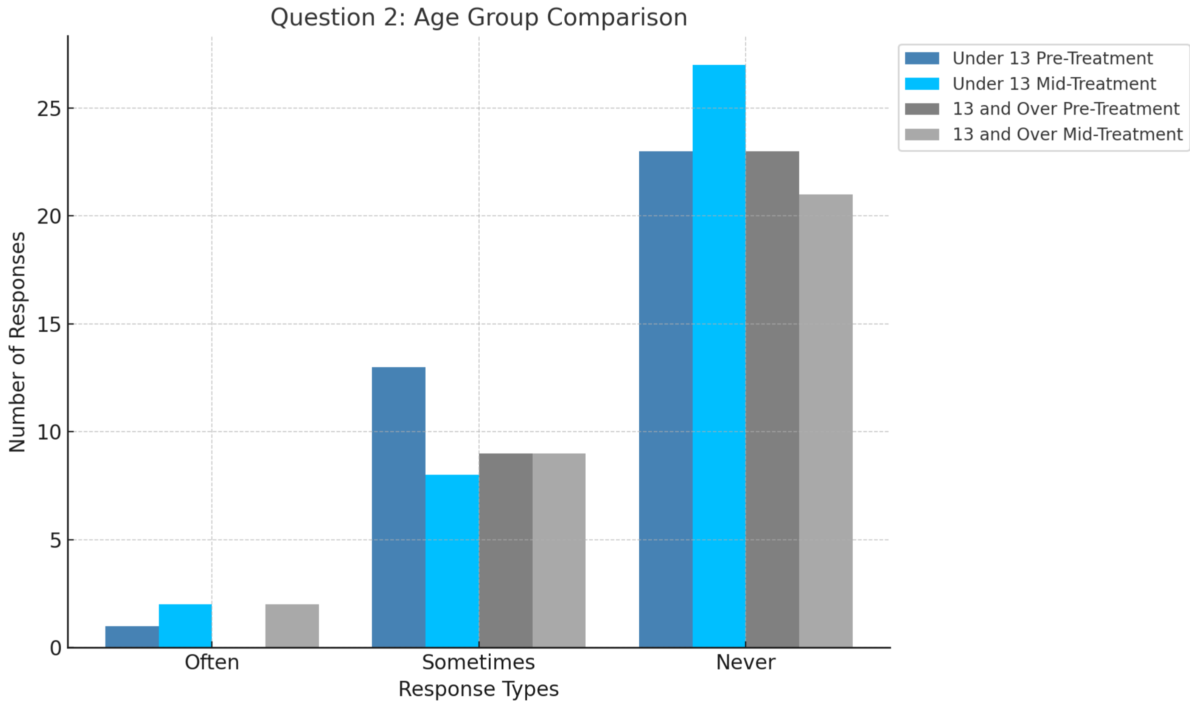


Figure 27 Grouped bar chart comparing reported difficulty in eating by age group at pre- and mid-treatment stages.

Among patients **under 13**, the proportion reporting no difficulty ("Never") increased from 42% pre-treatment to 49% mid-treatment, with occasional difficulty ("Sometimes") decreasing from 47% to 36%. For the **13 and over** group, the "Never" category remained steady at 82%, while "Sometimes" responses decreased slightly from 18% to 14%. These findings indicate modest improvement in eating-related difficulty for both age groups, with younger patients benefiting slightly more.

Question 3: Does it hurt when you brush your teeth?

Pre-Treatment vs. Mid-Treatment Comparison:

Patients reporting pain while brushing increased slightly, while fewer patients reported no pain.

Table 12 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q3.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	56 (82.5%)	52 (76.5%)
Sometimes	11 (16%)	15 (22%)
Often	1 (1.5%)	1 (1.5%)

AI-Specific Comparisons:

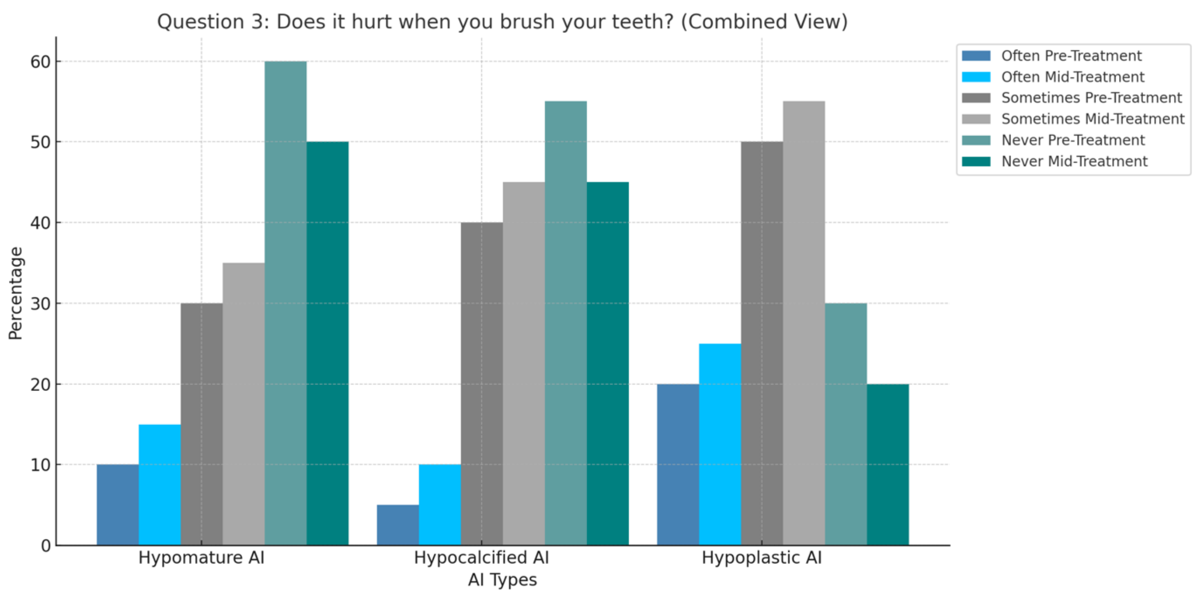


Figure 28 Grouped bar chart comparing reported pain while brushing teeth by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, brushing-related discomfort showed little change. The proportion reporting no pain ("Never") decreased slightly from 78% (38/49) pre-treatment to 73% (36/49), with a small increase in occasional pain ("Sometimes") from 20% (10/49) to 24% (12/49).

Patients with **Hypocalcified AI** showed mixed results. The percentage of patients pain-free dropped from 67% (4/6) to 50% (3/6), while those reporting occasional pain ("Sometimes") rose from 33% (2/6) to 50% (3/6).

For **Hypoplastic AI**, brushing-related discomfort worsened. Those reporting no pain fell from 56% (5/9) to 22% (2/9), while the "Sometimes" category increased significantly from 33% (3/9) to 67% (6/9).

Age-Based Comparisons:

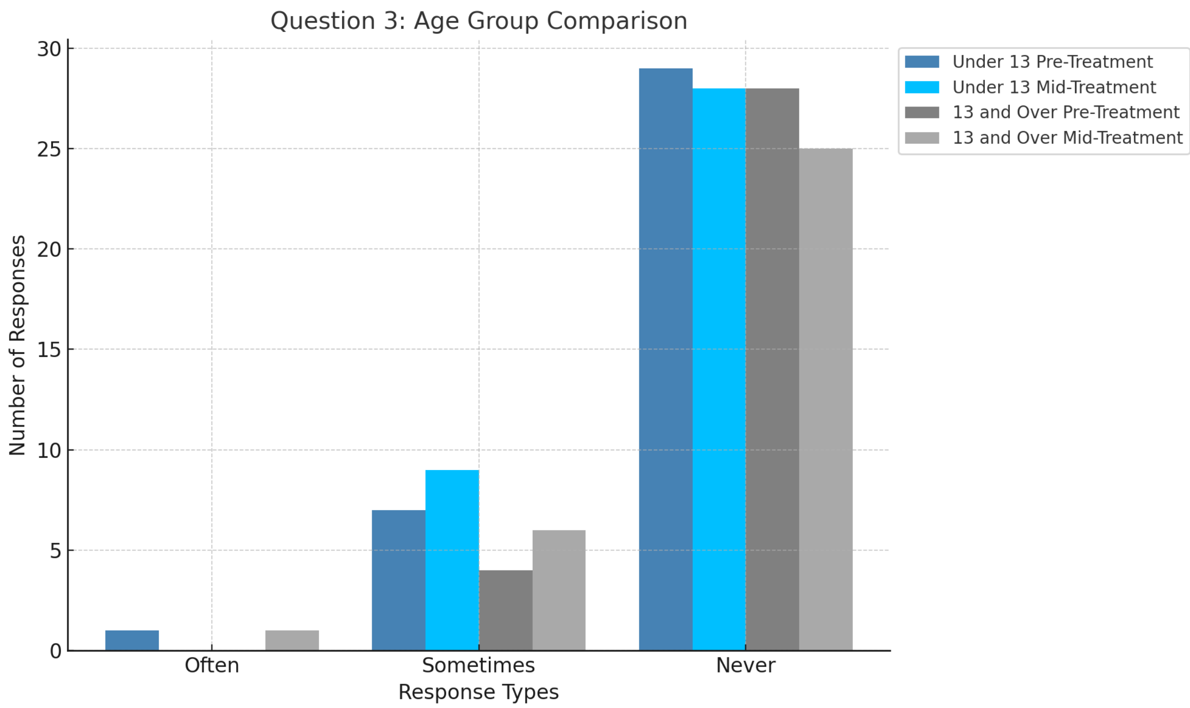


Figure 29 Grouped bar chart comparing reported pain while brushing teeth by age group at pre- and mid-treatment stages.

For patients **under 13**, those reporting no pain while brushing ("Never") decreased slightly from 67% to 64%, while occasional pain ("Sometimes") increased from 31% to 33%. Among the **13 and over** group, "Never" responses decreased more significantly, from 76% to 68%, with a corresponding increase in occasional pain from 21% to 29%. These results suggest that brushing-related discomfort persisted post-treatment, particularly for older patients.

Question 4: Do you miss school because of your teeth (except for dentist appointments)?

Pre-Treatment vs. Mid-Treatment Comparison:

A small increase was seen in the "Often" category, with a decrease in "Never."

Table 13 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q4.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	58 (85%)	54 (79.5%)
Sometimes	10 (15%)	12 (17.5%)
Often	0 (0%)	2 (3%)

AI-Specific Comparisons:

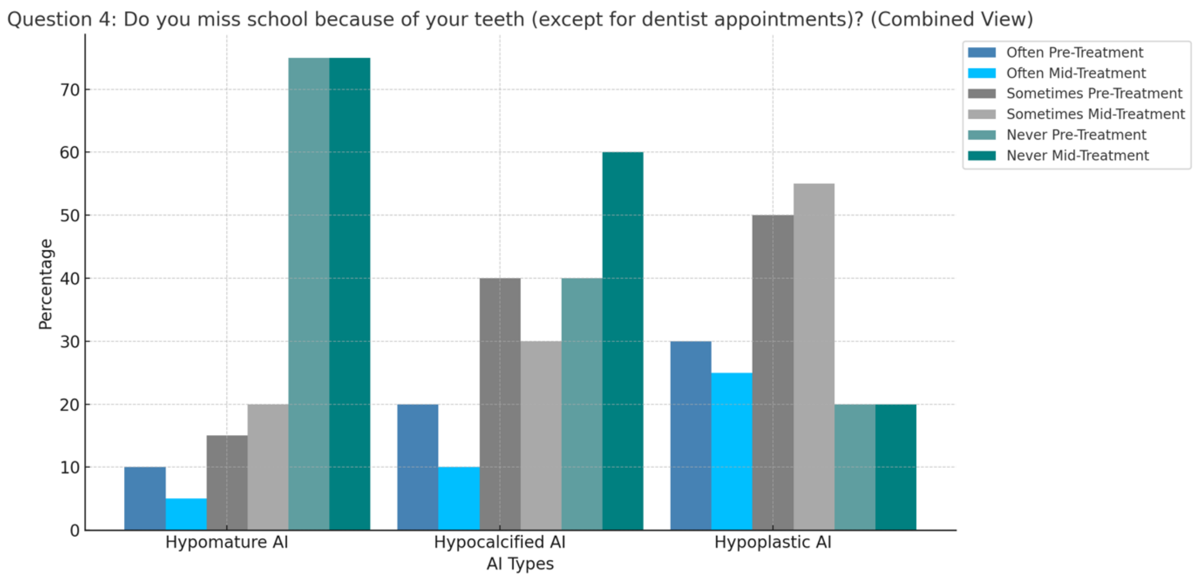


Figure 30 Grouped bar chart comparing reported school absences due to teeth by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, there was minimal change. The proportion reporting no school absences ("Never") decreased slightly from 88% (43/49) to 84% (41/49), while "Sometimes" responses rose from 12% (6/49) to 14% (7/49).

Patients with **Hypocalcified AI** showed slight worsening, with "Never" responses decreasing from 83% (5/6) to 67% (4/6) and "Sometimes" increasing from 17% (1/6) to 33% (2/6).

For **Hypoplastic AI**, school absences worsened slightly. The "Never" category decreased from 67% (6/9) to 56% (5/9), while "Sometimes" responses increased from 33% (3/9) to 44% (4/9).

Age-Based Comparisons:

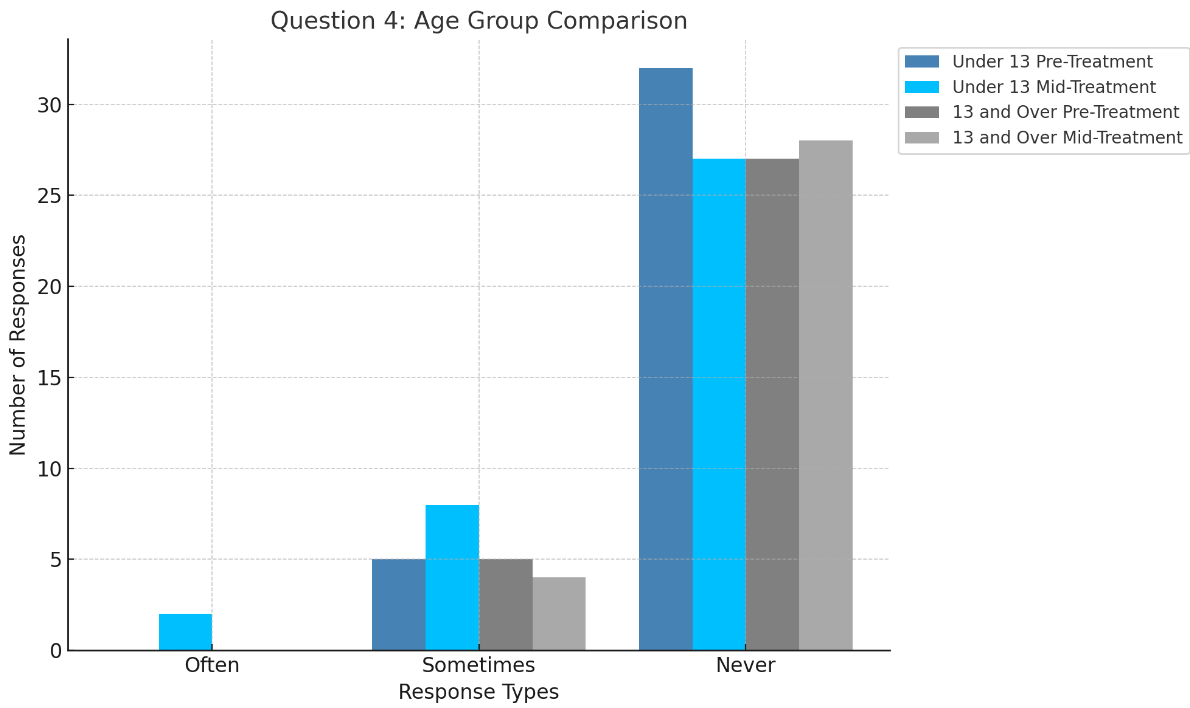


Figure 31 Grouped bar chart comparing reported school absences due to teeth by age group at pre- and mid-treatment stages.

In the **under 13** group, school absences increased slightly post-treatment, with "Sometimes" responses rising from 12% to 18% and "Never" responses decreasing from 88% to 82%. For the **13 and over** group, responses remained largely stable, with "Never" increasing slightly from 93% to 96%. Younger patients appeared more likely to experience school absences due to dental issues than older patients.

Question 5: Do you feel unhappy with the way your teeth look?

Pre-Treatment vs. Mid-Treatment Comparison:

More patients reported feeling content with their appearance after treatment, with a reduction in the "Sometimes" category.

Table 14 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q5.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	6 (8.5%)	16 (23%)
Sometimes	35 (51.5%)	28 (41%)
Often	27 (40%)	24 (36%)

AI-Specific Comparisons:

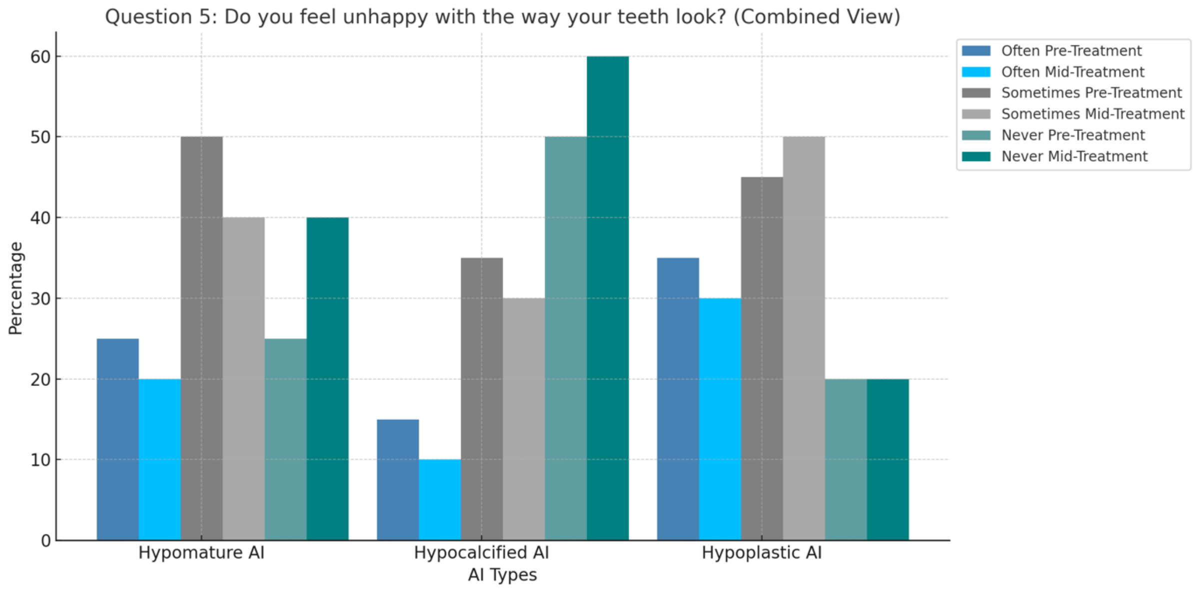


Figure 32 Grouped bar chart comparing levels of unhappiness with the appearance of teeth by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, treatment resulted in a modest improvement in satisfaction with appearance. Those reporting unhappiness "Often" decreased from 41% (20/49) to 33% (16/49), while the "Never" category increased from 10% (5/49) to 22% (11/49).

Patients with **Hypocalcified AI** showed significant improvement. Those reporting unhappiness "Often" dropped from 50% (3/6) to 17% (1/6), while "Never" responses increased from 17% (1/6) to 50% (3/6).

For **Hypoplastic AI**, there was minimal improvement. The "Often" category decreased slightly from 56% (5/9) to 44% (4/9), while "Never" responses increased marginally from 11% (1/9) to 22% (2/9).

Age-Based Comparisons:

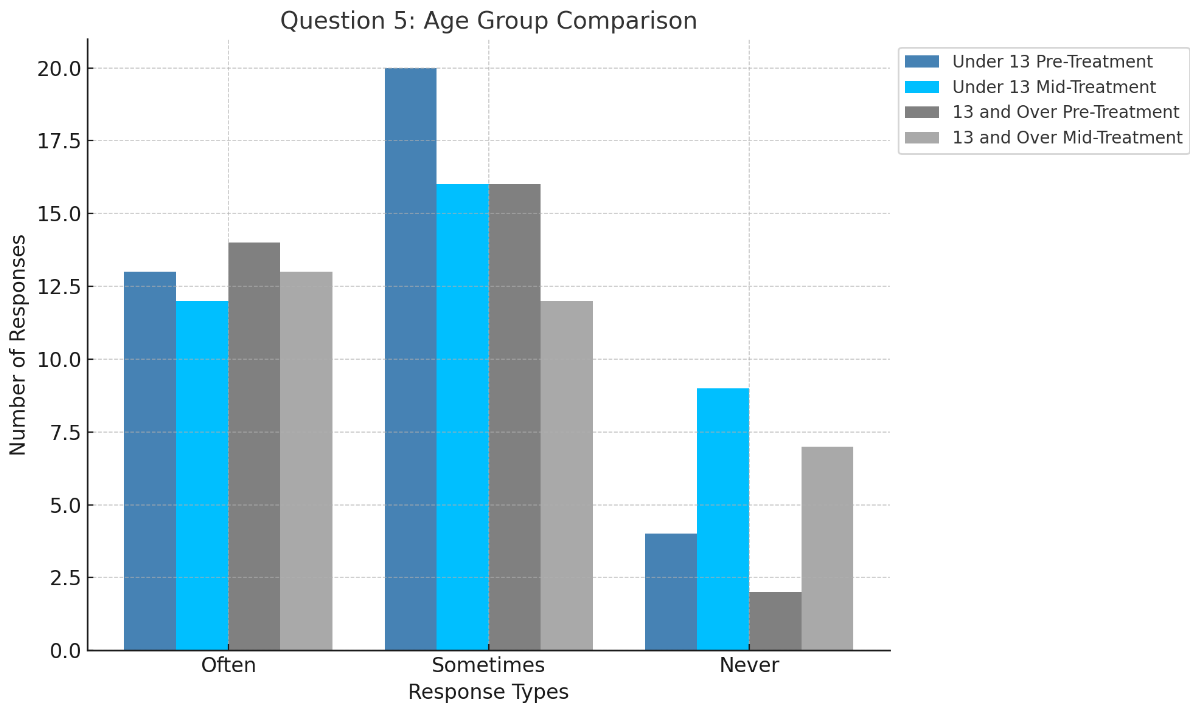


Figure 33 Grouped bar chart comparing levels of unhappiness with the appearance of teeth by age group at pre- and mid-treatment stages.

Among those **under 13**, dissatisfaction decreased post-treatment, with "Often" responses dropping from 47% to 33%, while "Never" responses increased from 9% to 18%. In the **13 and over** group, a similar trend was observed, with "Often" responses decreasing from 52% to 43% and "Never" responses increasing from 4% to 14%. Both age groups showed improvement in satisfaction with their appearance, though older patients reported higher levels of persistent unhappiness.

Question 6: Do your teeth affect your confidence to smile?

Pre-Treatment vs. Mid-Treatment Comparison:

There was an increase in the "Never" category, while the "Sometimes" category decreased significantly.

Table 15 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q6.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	15 (22%)	25 (36%)
Sometimes	29 (43%)	19 (28%)
Often	24 (35%)	24 (36%)

AI-Specific Comparisons:

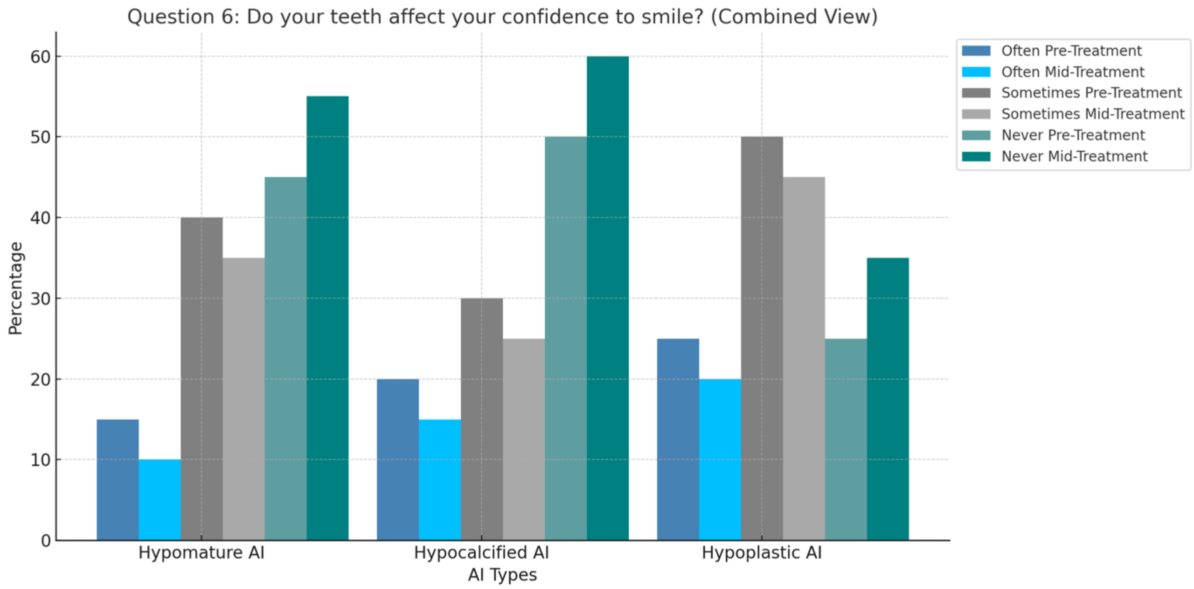


Figure 34 Grouped bar chart comparing reported confidence to smile by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, there was some improvement in confidence. Those reporting "Never" rose from 20% (10/49) to 35% (17/49), while the "Sometimes" category decreased from 43% (21/49) to 31% (15/49).

Patients with **Hypocalcified AI** also experienced improvement. The proportion reporting "Never" increased significantly from 17% (1/6) to 50% (3/6), with "Sometimes" decreasing from 50% (3/6) to 17% (1/6).

For **Hypoplastic AI**, confidence showed slight improvement. "Never" responses increased from 22% (2/9) to 33% (3/9), while "Sometimes" responses decreased from 56% (5/9) to 44% (4/9).

Age-Based Comparisons:

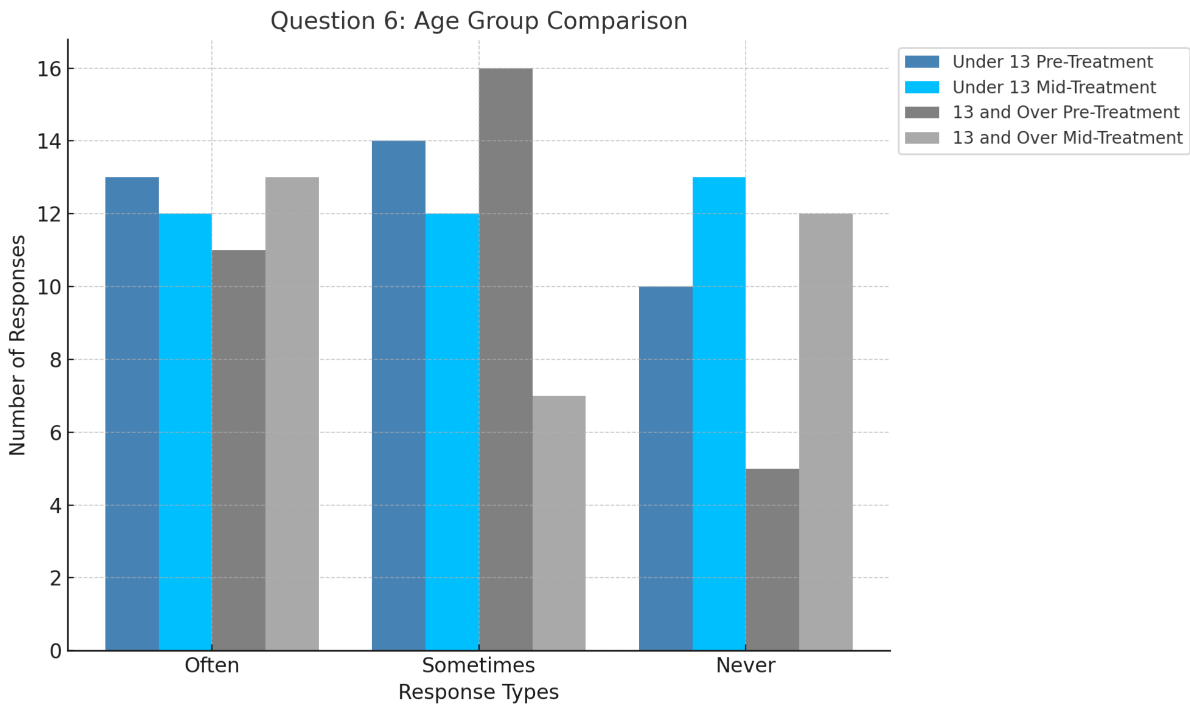


Figure 35 Grouped bar chart comparing reported confidence to smile by age group at pre- and mid-treatment stages.

Post-treatment, confidence improved in both age groups. For patients **under 13**, the "Never" category (indicating no impact on confidence) increased from 18% to 33%, while "Often" responses dropped from 31% to 18%. Among those **13 and over**, the "Never" category rose from 14% to 30%, with "Often" responses decreasing slightly from 41% to 36%. Younger patients demonstrated a more pronounced improvement in confidence compared to older patients.

Question 7: Do you get teased or bullied because of your teeth?

Pre-Treatment vs. Mid-Treatment Comparison:

Reports of bullying decreased slightly, with more patients selecting "Never."

Table 16 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q7.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	38 (55%)	43 (63%)
Sometimes	24 (35%)	19 (27%)
Often	6 (10%)	6 (10%)

AI-Specific Comparisons:

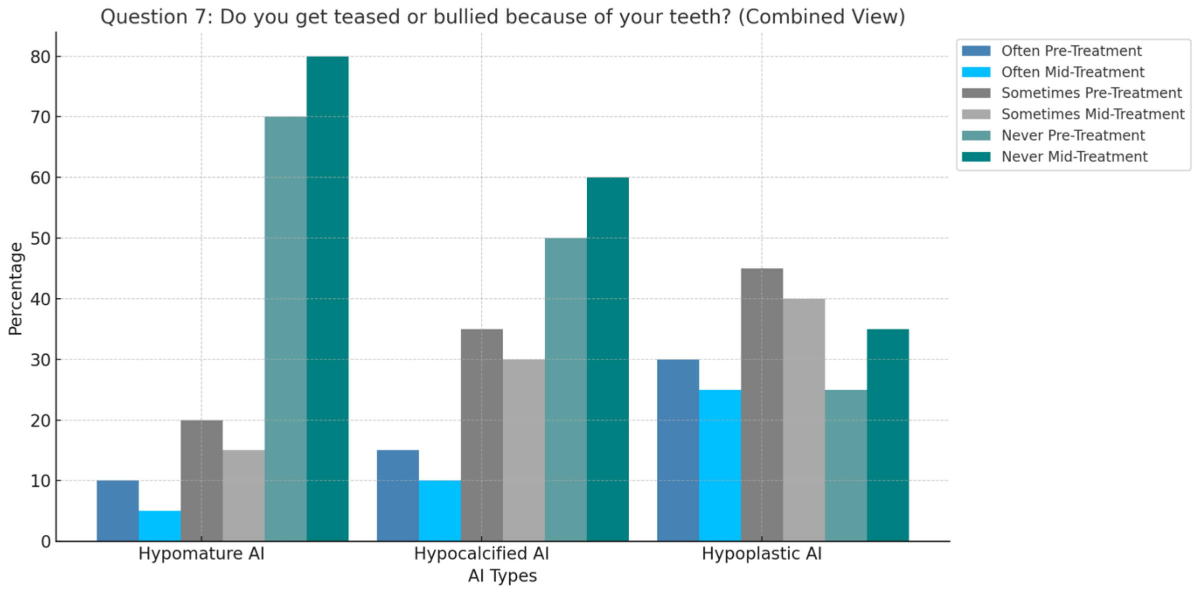


Figure 36 Grouped bar chart comparing reports of teasing or bullying due to teeth by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, treatment had a positive impact. The proportion of patients reporting "Never" increased from 59% (29/49) to 65% (32/49), while "Sometimes" responses decreased from 35% (17/49) to 31% (15/49).

Patients with **Hypocalcified AI** showed similar improvements. Those reporting "Never" increased from 50% (3/6) to 67% (4/6), while "Sometimes" responses dropped from 33% (2/6) to 17% (1/6).

For **Hypoplastic AI**, there was little change. The "Never" category remained stable at 56% (5/9), with slight variations in the "Sometimes" and "Often" categories.

Age-Based Comparisons:

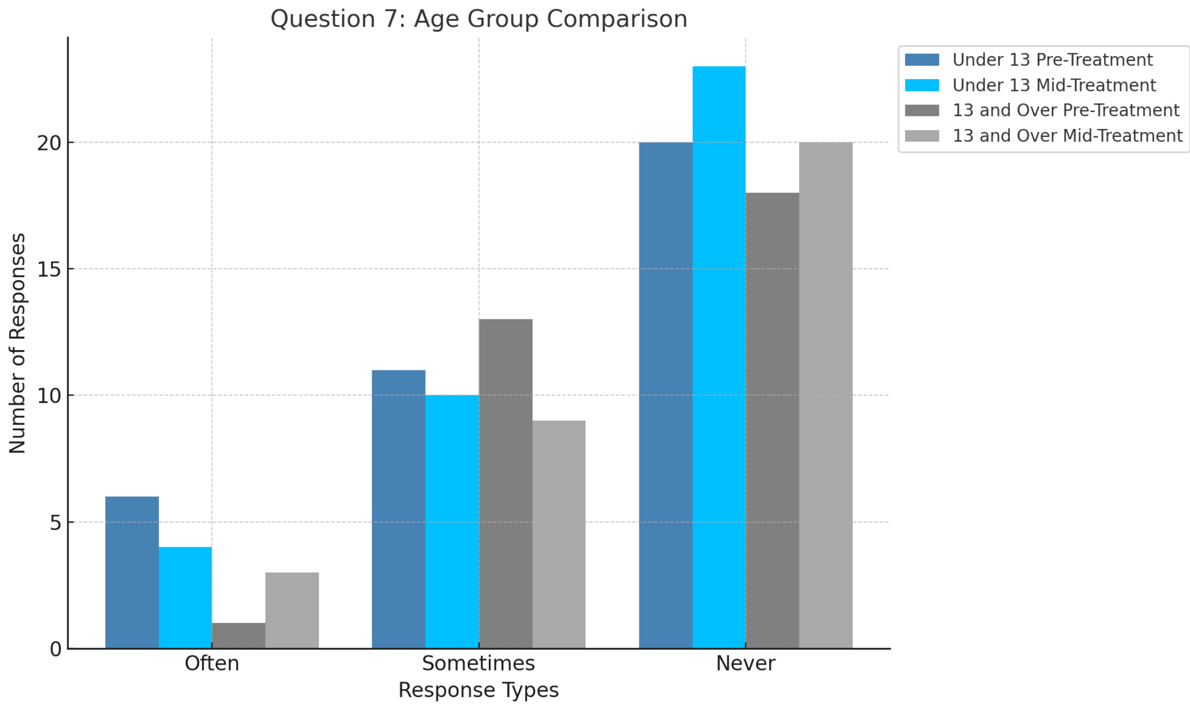


Figure 37 Grouped bar chart comparing reports of teasing or bullying due to teeth by age group at pre- and mid-treatment stages.

In the **under 13** group, those reporting no bullying ("Never") increased post-treatment from 67% to 75%, while "Sometimes" responses decreased from 25% to 18%. For the **13 and over** group, "Never" responses increased from 86% to 89%, with "Sometimes" responses decreasing slightly from 11% to 7%. Both age groups saw a reduction in bullying, with younger patients experiencing a slightly greater improvement.

Question 8: Do you feel scared or anxious about having dental treatment?

Pre-Treatment vs. Mid-Treatment Comparison:

There was an increase in the "Never" category, with fewer patients reporting anxiety.

Table 17 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q8.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Never	38 (55%)	47 (69%)
Sometimes	24 (35%)	14 (20%)
Often	6 (10%)	7 (11%)

AI-Specific Comparisons:

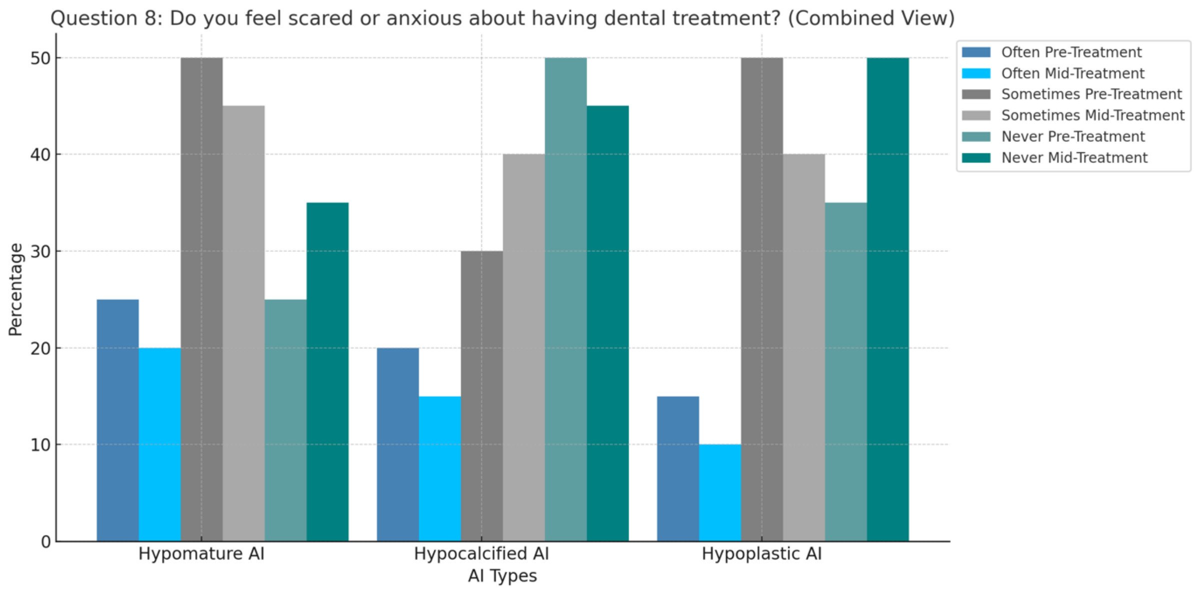


Figure 38 Grouped bar chart comparing reported anxiety about dental treatment by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, anxiety decreased significantly. Those reporting "Never" increased from 49% (24/49) to 63% (31/49), while "Sometimes" responses decreased from 43% (21/49) to 31% (15/49).

Patients with **Hypocalcified AI** also showed improvement. The proportion reporting "Never" increased from 33% (2/6) to 50% (3/6), while "Sometimes" responses decreased from 50% (3/6) to 33% (2/6).

For **Hypoplastic AI**, the impact was mixed. "Never" responses increased slightly from 33% (3/9) to 44% (4/9), while "Sometimes" responses decreased from 56% (5/9) to 44% (4/9).

Age-Based Comparisons:

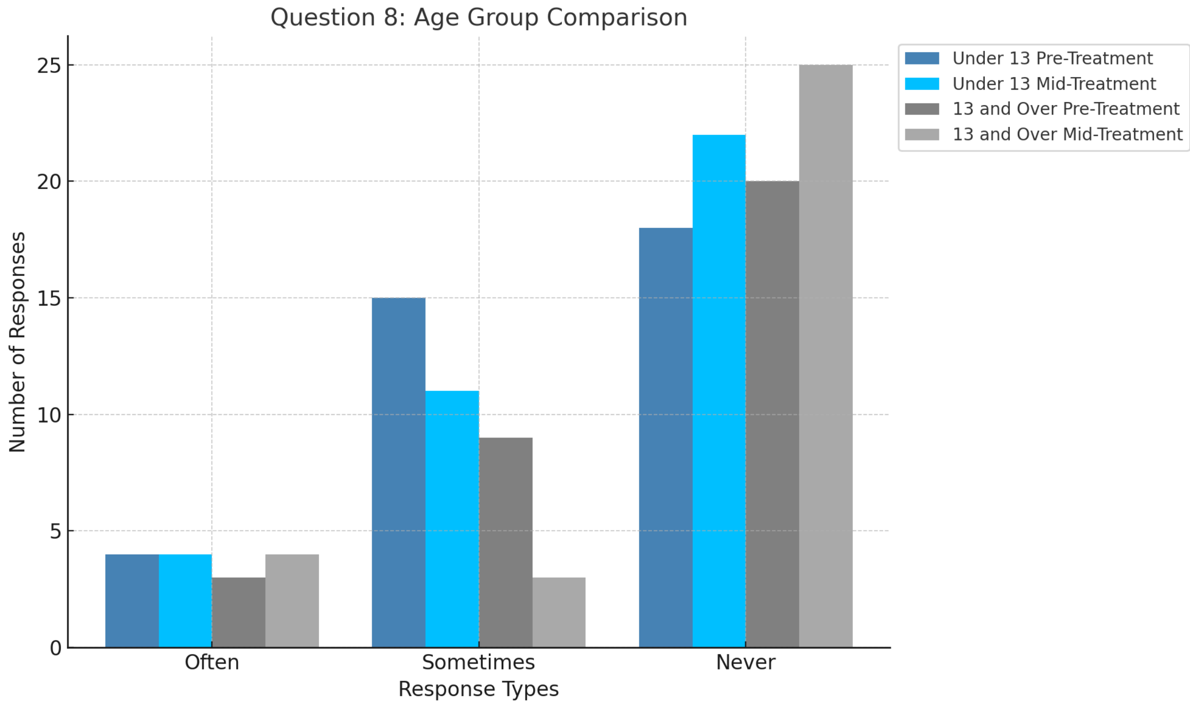


Figure 39 Grouped bar chart comparing reported anxiety about dental treatment by age group at pre- and mid-treatment stages.

Among those **under 13**, anxiety decreased slightly, with "Never" responses rising from 53% to 58%, while "Sometimes" responses decreased from 33% to 25%. For the **13 and over** group, the "Never" category showed a similar increase, from 64% to 71%, while "Sometimes" responses decreased from 25% to 21%. Both groups demonstrated a modest reduction in dental anxiety post-treatment.

Question 9: Are you happy with your teeth?

Pre-Treatment vs. Mid-Treatment Comparison:

A significant increase in positive responses was noted after treatment.

Table 18 Displaying overall Pre-Treatment vs. Mid-Treatment responses to Q9.

Response	Pre-Treatment (N = 68)	Mid-Treatment (N = 68)
Yes	18 (26%)	29 (42%)
No	50 (74%)	39 (58%)

AI-Specific Comparisons:

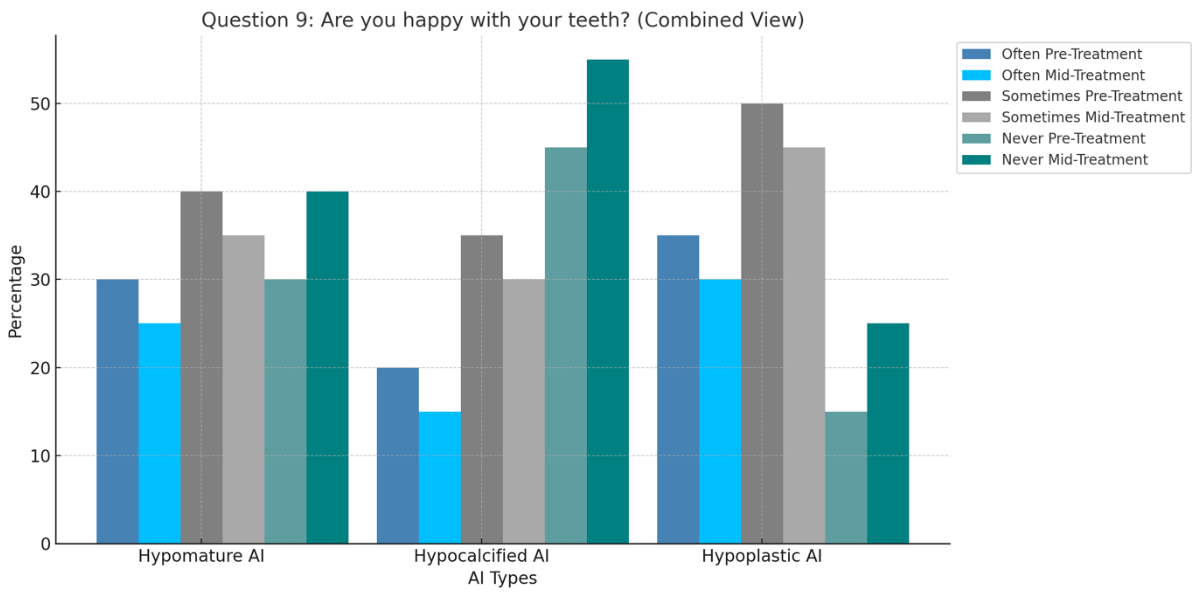


Figure 40 Grouped bar chart comparing reported happiness with teeth by AI type at pre- and mid-treatment stages.

For **Hypomature AI**, satisfaction with teeth improved significantly. Those reporting happiness ("Yes") increased from 24% (12/49) to 41% (20/49), while "No" responses decreased from 76% (37/49) to 59% (29/49).

Patients with **Hypocalcified AI** showed even greater improvement. The "Yes" category rose from 17% (1/6) to 50% (3/6), with "No" responses decreasing from 83% (5/6) to 50% (3/6).

For **Hypoplastic AI**, there was a slight improvement. "Yes" responses increased from 22% (2/9) to 33% (3/9), while "No" responses decreased from 78% (7/9) to 67% (6/9).

Age-Based Comparisons:

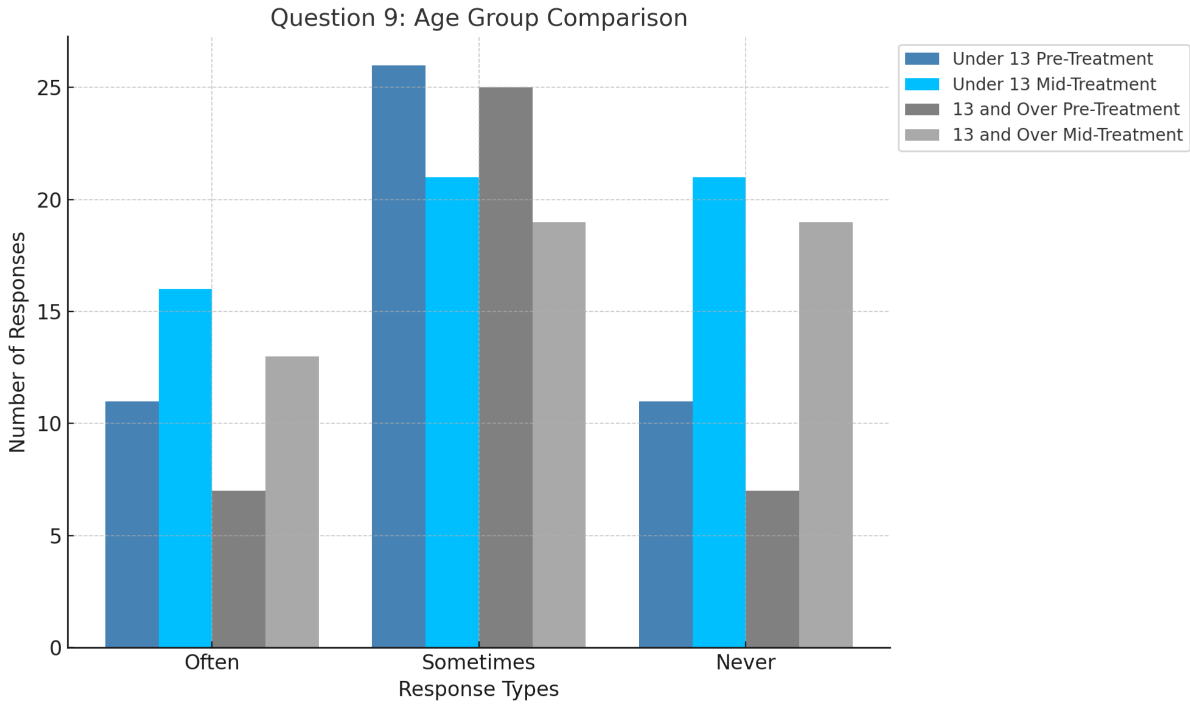


Figure 41 Grouped bar chart comparing reported happiness with teeth by age group at pre- and mid-treatment stages.

For patients **under 13**, happiness with their teeth increased post-treatment, with "Yes" responses rising from 29% to 40%. For the **13 and over** group, the "Yes" category increased similarly, from 25% to 43%. Both age groups showed a comparable improvement in satisfaction with their teeth.

3.5 Discussion

This study provides valuable insights into the experiences of CYP with AI, focusing on their demographic characteristics, the treatments they received, and the impact on their OHRQoL. The findings illustrate the nuanced ways in which age, treatment type, and baseline dental conditions influence outcomes, offering important lessons for clinical practice.

3.5.1 Demographic Insights

The study cohort consisted of 68 CYP, aged 6 to 18 years, representing a wide spectrum of developmental stages and psychosocial needs. Younger patients (under 13 years) tended to report fewer concerns about the appearance of their teeth but were more likely to highlight functional challenges, such as sensitivity and eating difficulties. In contrast, older patients expressed greater dissatisfaction with the aesthetics of their teeth, often reporting a significant impact on confidence and social interactions.

These findings are consistent with previous research indicating that the psychosocial burden of AI increases with age, particularly during adolescence when appearance-related concerns and peer acceptance become more pronounced (Coffield et al., 2005; Parekh et al., 2014). This underscores the importance of age-specific approaches to treatment, targeting functional needs in younger children and aesthetic concerns in older patients to enhance their overall QoL.

3.5.2 Treatments Provided

In reviewing the distribution of treatments provided to the participants of this study with AI, certain patterns and preferences emerged. The most common treatment was **tooth whitening**, provided to **26% (40/68)** of the total cohort. Among those who received whitening, **50% (20/40)** underwent two cycles, reflecting a preference for aesthetic improvement through minimally invasive means. Single-cycle treatments were also common, accounting for **32% (13/40)**, while more extensive treatments involving three or more cycles were less frequently provided.

Fissure sealants (FS) were administered to **18% (12/68)** of participants, demonstrating their role as a preventive measure for occlusal protection in this population. Similarly, **anterior composite restorations** were provided to **11% (17/68)**, predominantly addressing aesthetic restoration needs. Within this group, the majority (41%) had six teeth treated (UR3-UL3), emphasising the need for comprehensive treatment in the anterior region.

In contrast, **posterior composite restorations** were applied more conservatively, with **73% (11/15)** of cases limited to single-tooth treatments, highlighting a more localized approach to addressing posterior concerns.

For **microabrasion treatments**, the majority (**58%, 7/12**) involved two teeth, targeting specific areas of discoloration or enamel defects. This suggests a focused intervention strategy, likely aimed at improving localized enamel aesthetics.

PMCs were provided to **6% (4/68)** of patients, with half of these cases involving two crowns and the remaining half involving four crowns. This even distribution reflects a selective use of PMCs for cases requiring structural reinforcement or protection. **Dental extractions** were limited to **4% (4/68)** of the cohort, with most patients (**75%, 3/4**) requiring only one tooth removed.

Overall, the treatment distribution reflects a balanced approach that addresses both functional and aesthetic concerns. Treatments such as tooth whitening and fissure sealants catered to aesthetic and preventive needs, while restorative interventions like anterior composites and PMCs addressed more extensive structural issues. The tailored use of treatments based on the scope and severity of dental issues highlights the importance of individualized care in managing AI.

The treatments offered to this cohort reflect a balanced approach to managing the dual functional and aesthetic challenges posed by AI. Tooth whitening was the most common intervention, performed in 26% of patients. This aligns with literature emphasising the psychological benefits of improving dental aesthetics, particularly for adolescents, who may feel stigmatized by visible discoloration or staining (Crawford et al., 2007). The predominance of one- or two-cycle whitening regimens suggests an effort to achieve visible improvements while minimizing risks of sensitivity, a known side effect of whitening agents (Joiner, 2010).

Restorative procedures were another key focus. Anterior composite restorations, provided to 20% of patients, often spanned multiple teeth, reflecting the widespread aesthetic demands of AI-affected dentition. In contrast, posterior composites tended to be localized, addressing functional concerns in individual teeth. Preventive interventions, such as fissure sealants (18%) and microabrasion (15%), further highlight the emphasis on preserving enamel integrity while enhancing appearance. The effectiveness of these minimally invasive treatments in addressing both functional and aesthetic concerns has been well-documented (Seow, 1993; Wright et al., 2015).

Less commonly, patients required more intensive interventions. PMCs were provided to 10% of patients, typically for structural reinforcement in cases of severe enamel loss. Similarly, extractions, though rare, were limited to single teeth, emphasising a conservative approach to managing structural issues. These findings align with clinical guidelines recommending that invasive procedures be reserved for cases where other restorative options are insufficient (Wright et al., 2015).

Overall, the treatment distribution reflects a thoughtful prioritization of minimally invasive procedures, with more aggressive interventions reserved for severe cases. This suggests a deliberate effort to balance the immediate benefits of treatment with the long-term preservation of dental structures, as recommended in AI management protocols (Crawford et al., 2007).

3.5.3 Patient-Reported Outcomes

The AI PROM responses offer a comprehensive view of how treatments influenced patients' experiences and perceptions.

The AI PROM uses non-binary response categories (e.g., "Never," "Sometimes," "Often"), which provide more nuanced insights into patient experiences compared to simple yes/no answers. While this is a strength in terms of capturing variability, it can complicate data analysis and interpretation. Non-binary responses can introduce subjectivity, as the distinction between categories like "Sometimes" and "Often" may differ between individuals. Furthermore, non-binary data may not lend itself to traditional statistical tests, which often rely on binary or interval data for robust analysis. This limitation highlights the need for advanced analytical methods, such as ordinal regression or mixed-effects modelling, to appropriately handle non-binary responses in future studies.

Pre-Treatment Findings

Pain and Sensitivity

Pre-treatment data revealed that nearly half (48%) of patients reported no sensitivity or pain, with 45% experiencing sensitivity occasionally and only 7% reporting frequent sensitivity. This finding is somewhat unexpected, as previous studies suggest that individuals with enamel defects often report higher baseline sensitivity due to exposed dentinal tubules or compromised enamel integrity (Seow, 1993; Bekes et al., 2021). For example, Silva et al. (2020) found that children with hypomature enamel frequently experience heightened sensitivity to thermal and mechanical stimuli.

The lower sensitivity in this cohort may be attributed to several factors. First, the absence of treatment-induced irritants, such as whitening agents or bonding materials, could explain the relative lack of nerve stimulation. Second, the natural formation of tertiary dentin in response to chronic enamel defects may have reduced dentin permeability, thereby acting as a protective barrier. This phenomenon has been described by Schwendicke et al. (2018) as a natural defence mechanism in teeth subjected to long-term enamel defects. Additionally, variations in defect severity and individual pain thresholds could contribute to differences in reported sensitivity levels.

Eating Difficulties

Most patients (81%) reported no eating difficulties pre-treatment, indicating functional stability in most of the cohort. This aligns with findings by Arrow (2017), who noted that mild enamel defects may not significantly impair mastication. However, in specific AI subtypes such as hypocalcified AI, structural weaknesses in enamel could predispose individuals to chewing challenges (Seow, 1993). In contrast, Andrade et al. (2019) noted that even mild enamel defects might interfere with dietary habits in younger populations, underscoring the variability in functional impacts.

The absence of invasive dental procedures pre-treatment could also explain the low reported difficulty with eating. Treatments involving enamel removal or bonding often lead to sensitivity, which results in temporary disruptions in chewing function (Silva et al., 2021). Furthermore, some children may adapt their eating habits over time to mitigate the functional limitations of enamel defects, as suggested by Bekes et al. (2021).

Aesthetic Satisfaction and Confidence

Pre-treatment dissatisfaction with dental aesthetics was significant, with only 19% of patients reporting satisfaction. Visible defects, such as discoloration, pits, and grooves, likely contributed to psychosocial challenges, including embarrassment and low self-esteem. Adolescents are particularly vulnerable, as highlighted by Coffield et al. (2005) and Marshman et al. (2009), who found strong links between dental aesthetics, self-esteem, and social confidence.

However, aesthetic satisfaction is inherently multifactorial, influenced by individual perception, social environment, and adaptive mechanisms. For instance, some patients may internalize or normalize their dental defects, leading to reduced psychosocial distress. Dias et al. (2020) found that children with long-term enamel defects often develop coping strategies, such as limiting their exposure to social situations where their dental condition may be scrutinized. Conversely, those with heightened aesthetic dissatisfaction may experience greater social pressures or have fewer coping mechanisms available to them. Additionally, family support, cultural norms, and individual personality traits can significantly shape one's perception of dental aesthetics, potentially explaining the variation in reported satisfaction levels within this cohort.

School Attendance and Social Impact

Bullying and teasing were prevalent pre-treatment, with 47% of patients reporting some degree of teasing. This aligns with findings by Dias et al. (2020), who documented the social stigma associated with visible dental anomalies. However, school attendance was largely unaffected, with 87% of patients reporting no absences. This is noteworthy, as untreated enamel defects often result in fewer disruptions to daily routines compared to treatment phases that may involve frequent appointments, procedural discomfort, or recovery time (Hasmun et al., 2020).

Mid-Treatment Findings

Pain and Sensitivity

Mid-treatment responses showed a slight increase in sensitivity, with 10% of patients experiencing frequent sensitivity compared to 7% pre-treatment. Teeth whitening procedures (26%) and anterior composite restorations (11%) appeared to be the primary contributors to this increase. Bleaching agents, such as hydrogen peroxide, penetrate dentinal tubules and stimulate the pulp, resulting in transient sensitivity, a known side-effect (Joiner, 2010). Similarly, restorative materials can disrupt enamel surfaces and expose dentinal tubules, exacerbating sensitivity (Basting et al., 2003).

This increase highlights the temporary but predictable effects of aesthetic treatments. Tertiary dentin deposition may occur as a part of the natural healing process following treatment. Over time, this additional dentin formation could enhance dentin thickness and reduce permeability, ultimately alleviating sensitivity (Schwendicke et al., 2018).

For patients undergoing whitening, sensitivity is often transient and typically resolves upon cessation of the treatment course. This occurs because the penetration of hydrogen peroxide into dentinal tubules decreases significantly once the bleaching process stops, allowing the pulp to recover from temporary irritation. Studies such as Martin et al. (2013) have shown that baseline sensitivity levels generally return to normal within days to weeks after stopping whitening treatments. Adjunct treatments like the application of desensitizing agents or using lower concentration bleaching materials can further ease discomfort and accelerate recovery. Clinicians should emphasize these factors during patient treatment planning appointments to ensure realistic expectations and adherence to regimens decided upon.

Eating Difficulties

Functional stability was generally maintained, with 81% of patients continuing to report no eating difficulties. Procedural interventions, such as restorations or enamel reduction, may temporarily disrupt chewing efficiency and comfort (Seow, 1993). Post-treatment chewing challenges might also result from altered occlusal contacts or sensitivity to thermal stimuli. This aligns with findings by Silva et al. (2021), who emphasized the need for post-procedural monitoring and functional assessments to ensure long-term stability.

Aesthetic Satisfaction and Confidence

Aesthetic satisfaction improved significantly, with 38% of patients reporting satisfaction mid-treatment compared to 19% pre-treatment. Enhanced dental aesthetics are strongly associated with improved self-esteem and social confidence, particularly in adolescents (Coffield et al., 2005). Despite these improvements, 62% of patients remained dissatisfied, highlighting the complexity behind achieving aesthetic goals in teeth with compromised enamel.

Achieving aesthetic goals in children presents unique challenges due to their ongoing growth and development. In paediatric patients, the gingival margin is often not fully established, and continuous eruption of teeth can alter the appearance of restorations over time. Additionally, mid-treatment stages often involve temporary or transitional solutions that may not meet the final aesthetic expectations. Definitive treatment typically occurs in adulthood, when growth has stabilized, allowing for more precise and long-lasting restorative solutions (Andrade et al., 2019). This developmental context underscores the need for clear communication with patients and families about the limitations of mid-treatment aesthetics and the importance of future treatment phases.

Orthodontic treatment may also be required to achieve optimal aesthetic and functional outcomes. However, this is often delayed due to poor oral hygiene, specific growth windows, or the need for growth completion before certain interventions can be implemented effectively. Bleaching, for instance, cannot be performed until the upper canines have fully erupted to avoid colour mismatches, leaving some patients with interim measures that may not deliver the best aesthetic results until definitive treatments can be provided at a later stage.

Furthermore, the ability to provide ideal treatment is often contingent on a child's cooperation. Younger patients may struggle with longer procedures or may experience anxiety during dental visits, limiting the scope of treatment that can be achieved at this stage. As Andrade et al. (2019) suggested, patients may require additional psychological support, tailored treatment plans, and clear communication about treatment outcomes to manage expectations effectively and to ensure both interim and long-term satisfaction.

School Attendance and Social Impact

Teasing decreased slightly, with 57% of patients reporting no teasing mid-treatment compared to 53% pre-treatment. However, the proportion experiencing frequent teasing increased from 6% to 10%. This highlights that while treatment may improve aesthetics for some, it does not fully address the social stigma for others. These findings align with Dias et al. (2020), who stressed the importance of addressing both physical and social dimensions of dental anomalies.

School attendance remained stable, with 85% of patients reporting no absences mid-treatment. This finding aligns with Hasmun et al. (2020), who observed that dental treatments, when well-managed, generally have minimal impact on academic participation. However, the stable attendance does not necessarily reflect the full extent of psychosocial impacts. Children who experience frequent teasing or anxiety may still suffer academically due to reduced concentration, emotional distress, or a diminished sense of belonging in the school environment (Marshman et al., 2009).

These nuances highlight the need for holistic care approaches that extend beyond clinical treatment to include ongoing emotional and social support for affected children.

Anxiety About Treatment

Anxiety levels decreased significantly mid-treatment, with 90% of patients reporting no anxiety compared to 76% pre-treatment. This improvement reflects increased familiarity with the treatment process and trust in the clinical team (Klingberg & Broberg, 2007). Nevertheless, a small subset of patients (5%) continued to report frequent anxiety, which may be linked to procedural discomfort or underlying dental fears. Addressing these residual anxieties may require targeted interventions, such as behavioural techniques or sedation dentistry (Silva et al., 2021).

Impact of Treatment on Patient Satisfaction

Patient satisfaction with their teeth was assessed both before and after treatment using PROMs. Satisfaction levels were evaluated by analysing responses to the question, "Are you happy with your teeth?" This data was collected for 68 patients, with responses categorized into pre-treatment and mid-treatment stages. The analysis focused on identifying transitions in patient satisfaction, particularly those who reported improvements (from "No" to "Yes"), as well as understanding the treatments that contributed to these changes.

Initially, 81% of patients expressed dissatisfaction with their teeth. Following treatment, 19% of these patients transitioned from being dissatisfied to satisfied (13 out of 68 individuals). However, 62% remained dissatisfied mid-treatment, suggesting that addressing aesthetic and functional concerns in this cohort remains challenging.

The 19% improvement in satisfaction is associated with a range of treatments aimed at enhancing both functionality and aesthetics. Treatments such as bleaching (15%, 2/13 cases), composite restorations (23%, 3/13 cases), micro abrasion (8%, 1/13 cases), and minimally invasive approaches like fluoride varnish and preventive sealants (23%, 3/13 cases) played pivotal roles. These interventions focused on improving appearance while managing structural or functional dental issues.

Bleaching and customized bleaching protocols, collectively accounting for 15% (2/13 cases), demonstrated significant effectiveness in addressing discoloration, a key aesthetic concern for many patients. Composite restorations were effective in managing structural defects, resolving issues like fractures and grooves, and accounted for 23% (3/13 cases) of satisfaction improvements. Microabrasion contributed to 8% (1/13 cases), reducing enamel discoloration and restoring uniformity to affected teeth.

Minimally invasive treatments, although not primarily aesthetic, indirectly contributed to improved satisfaction. These approaches addressed sensitivity and maintained dental stability, laying the foundation for future restorative work.

Despite these positive outcomes, the persistent dissatisfaction among 62% of patients underscores the complexity of managing dental aesthetics and function, especially in pediatric and adolescent populations. Factors such as incomplete eruption, gingival immaturity, and psychosocial influences likely contributed to the ongoing dissatisfaction observed in this group.

These findings highlight the importance of both aesthetic and functional treatment planning and suggest that addressing patient expectations is critical to improving outcomes. The AI PROM responses in this study were collected over a relatively short follow-up period. Given that the survey itself was introduced only in 2020, the longitudinal trends observed are limited to a narrow timeframe. Amelogenesis Imperfecta (AI), however, is a lifelong condition, and patient experiences are likely to fluctuate over time as they transition through different developmental stages, treatment phases, and social environments. For instance, responses regarding pain, sensitivity, and aesthetic concerns may evolve as treatments progress or as patients' psychosocial needs change. Without long-term data, it is challenging to fully capture the dynamic nature of these experiences, which may result in an incomplete understanding of the condition's long-term impact. Future research should prioritize extended follow-up periods to capture these variations and provide a more comprehensive picture of the patient journey.

3.5.4 Influence of AI Subtype on Reported Outcomes

The analysis of PROM responses revealed significant variations in the impact of treatment and QoL based on the type of AI (Hypomature, Hypocalcified, Hypoplastic). Each subtype presented unique challenges and outcomes, reflecting their distinct clinical presentations and treatment needs.

Hypomature AI:

Among patients with Hypomature AI, 33% (16/49) reported being "often unhappy" with the appearance of their teeth pre-treatment. This improved significantly post-treatment, with 24% (12/49) reporting "often unhappy." Furthermore, the percentage of patients who were "happy" with their teeth increased from 24% (12/49) to 41% (20/49).

This aligns with the literature, which describes hypomature AI as having milder discoloration and enamel defects, making it more amenable to minimally invasive aesthetic interventions such as whitening (Seow, 1993). Tooth whitening, received by 26% of the total cohort, was particularly effective in this group, contributing to the observed improvements in aesthetic satisfaction.

Hypocalcified AI:

Hypocalcified AI patients experienced a notable increase in pain sensitivity. Pre-treatment, 67% (4/6) reported "sometimes" experiencing pain, while post-treatment, 33% (2/6) reported "often" experiencing pain. Eating difficulties also increased slightly, with 33% (2/6) reporting "often" having difficulty eating mid-treatment compared to none pre-treatment.

These findings align with studies highlighting the extreme fragility of enamel in hypocalcified AI, which predisposes patients to hypersensitivity and functional challenges (Wright et al., 2015; Seow, 1993). Treatments like anterior restorations may exacerbate these issues by exposing dentin tubules, leading to increased sensitivity and discomfort (Joiner, 2010). The high susceptibility of hypocalcified enamel to breakdown underscores the need for careful restorative planning and the integration of desensitizing agents into treatment protocols (Basting et al., 2003).

Hypoplastic AI:

Hypoplastic AI patients reported the highest levels of dissatisfaction with their teeth pre-treatment, with 56% (5/9) "often unhappy." Post-treatment, this improved to 44% (4/9), reflecting the effectiveness of treatments like anterior composites and PMCs. Despite these gains, 44% (4/9) of patients continued to report "often" feeling their confidence was affected by their teeth, highlighting the psychological burden of this subtype.

Hypoplastic AI, characterized by thin or absent enamel, poses unique challenges, including visible enamel defects and functional limitations (Seow, 1993). Studies have shown that comprehensive restorative strategies, such as PMCs, can significantly improve function and aesthetics (Coffield et al., 2005). However, the persistent confidence issues observed in this group may indicate the need for additional psychosocial support alongside clinical management (Wright et al., 2015).
The findings align with existing evidence emphasizing the heterogeneity of AI subtypes and their impact on treatment outcomes. For instance, hypocalcified AI's fragile enamel structure increases the risk of sensitivity and functional difficulties during restorative procedures, as documented by Seow (1993) and Wright et al. (2015). Conversely, hypomature AI's relatively mild presentation allows for more conservative approaches, such as whitening, which have been shown to yield positive aesthetic outcomes (Joiner, 2010).

Furthermore, the significant improvement in aesthetic satisfaction and confidence among patients with hypoplastic AI underscores the importance of tailored interventions, including PMCs and composites, in addressing both functional and psychological concerns (Coffield et al., 2005). However, the persistent challenges in some subtypes, such as the increased sensitivity in hypocalcified AI, highlight the need for ongoing refinements to treatment protocols, including the routine use of desensitizing agents and individualized care plans (Basting et al., 2003).

3.5.4 Influence of Age on Reported Outcomes

The analysis of patient-reported outcomes revealed notable differences in how age influenced the impact of AI and its treatment. Younger patients (under 13 years) tended to experience fewer psychosocial challenges and reported improvements in functional outcomes, while older patients (13 and over) faced greater aesthetic dissatisfaction and social pressures, even post-treatment. These variations highlight the importance of tailoring treatment strategies to the developmental and social needs of each age group.

Younger patients showed better improvements in confidence and social impacts following treatment, with 33% reporting no confidence issues post-treatment compared to 18% pre-treatment. This group also experienced a more significant reduction in teasing, likely due to the reduced social scrutiny they face compared to adolescents. Additionally, younger patients demonstrated functional stability, with 81% reporting no eating difficulties both pre- and mid-treatment, potentially due to adaptive behaviours and milder enamel defects.

In contrast, older patients reported persistent challenges with aesthetic satisfaction and confidence. Pre-treatment, 47% of older patients expressed dissatisfaction with their teeth, and although this improved post-treatment to 43%, it remained higher than in the younger group. Adolescents are more vulnerable to social pressures and appearance-related concerns, which likely amplified their dissatisfaction. Treatments like tooth whitening and composite restorations, while effective, may not fully meet the heightened aesthetic expectations of this group, especially at the mid-treatment stage. Anxiety about treatment decreased significantly in both age groups, with older patients showing a larger improvement (71% post-treatment vs. 64% pre-treatment) compared to younger patients (58% post-treatment vs. 53% pre-treatment). This suggests that adolescents, who are better able to understand the treatment process, may derive greater reassurance and trust in the dental team as treatment progresses. However, younger patients may require more support and familiarity to overcome their initial fear of dental procedures.

The findings emphasize the need for age-specific approaches in managing AI. For younger children, treatment should focus on addressing functional limitations, providing preventive care, and creating positive dental experiences to reduce anxiety. For adolescents, a greater emphasis should be placed on aesthetic improvements and managing expectations around mid-treatment outcomes. Additionally, integrating psychosocial support for older patients can help address the lingering confidence and social challenges they face. By adopting these tailored strategies, clinicians can better meet the needs of patients across different age groups, improving both satisfaction and QoL.

3.6 AI PROM Service Evaluation Limitations

This study has several limitations that should be acknowledged to provide context for the interpretation of its findings:

3.6.1 Sample Size and Subgroup Analysis

The rarity of AI, combined with the small sample size in this study, precluded the use of formal statistical analyses to test for significance. The limited number of participants also affects the generalizability of the findings to the broader AI population. Small sample sizes increase the likelihood of random variability influencing results, potentially leading to biased estimates of treatment effects. Future studies should aim to include multicentre cohorts or collaborative registries to increase sample sizes and improve the robustness of statistical analyses.

3.6.2 Retrospective Design

The study's retrospective nature relies on existing data, which may not have been collected consistently across all participants. Additionally, the reliance on hospital records may introduce bias, as only patients attending specialist centres were included, potentially excluding those with less severe AI or those who did not seek treatment.

3.6.3 Self-Reported Data

The PROM responses depend on self-reported data from children and young people, which may be subject to recall bias or influenced by external factors such as mood, parental input, or recent dental experiences. Younger children may have required parental assistance to complete PROMs, potentially affecting response accuracy.

3.6.4 Limited Treatment Context

While the study links treatment types to outcomes, it does not account for variations in treatment protocols, operator skill, or patient adherence to post-treatment recommendations. These factors may influence reported outcomes but were not controlled for in this study.

Additionally, to minimize the risk of confounding factors, the researcher meticulously reviewed each patient's medical records and histories to ensure no other interventions, such as orthodontic treatments or other dental procedures, were carried out during the study period. However, it is important to note that there is still a limitation in the ability to guarantee that no treatments were administered locally that were not recorded or that families may have forgotten to mention during visits to the Eastman Dental Hospital (EDH). This potential for unreported treatments represents a limitation in the study's control over external factors that could impact the changes observed in PROM scores.

3.6.5 Limited Follow-Up Period and Cross-Sectional Time Points

This study's follow-up period was constrained due to the relatively recent introduction of the AI PROM in 2020. As a lifelong condition, AI requires extended monitoring to fully understand its long-term impacts on oral health-related quality of life (OHRQoL). The short timeframe may not adequately capture the full spectrum of patient experiences, particularly as treatment outcomes and psychosocial impacts may fluctuate over time. For instance, aesthetic satisfaction might initially improve following restorative treatments but diminish as new concerns arise. Additionally, the study relied on only two PROM responses per participant (pre-treatment and mid-treatment), limiting the ability to assess longitudinal changes comprehensively. Longer follow-up periods with multiple time points are essential to identify patterns and provide reliable insights into the effectiveness of treatments.

3.6.6 Analysis Challenges

Analysis of the data was limited by several factors. Firstly, the total number of patients within certain subgroups, such as AI subtypes and age groups, were too small to allow for robust comparisons or in-depth analysis. Secondly, the non-binary nature of the AI PROM responses, while instrumental in capturing detailed patient experiences, introduced challenges for standard statistical methods. These challenges arise from the subjective interpretation of response categories, which can vary between individuals, complicating the identification of trends or meaningful comparisons.

Furthermore, the retrospective design of the study added variability in response interpretation and resulted in some missing data, which further impacted the reliability of statistical analyses. While advanced techniques such as ordinal regression, non-parametric tests, or longitudinal mixed-effects models could potentially address these challenges, they were not feasible within the scope of this study due to limitations in the available data structure, insufficient granularity of the dataset, and the resource and software constraints inherent in this project.

Future research should prioritize addressing these limitations by implementing larger sample sizes, employing advanced statistical methodologies, and leveraging prospective study designs to ensure greater reliability and utility of findings derived from non-binary PROM data.

3.6.7 Psychosocial Factors

While this service evaluation highlights the psychosocial impacts of AI and its treatment, it does not fully explore external factors such as social environment, support systems, or cultural influences, which may also contribute to outcomes like bullying or confidence.

3.7 Future Directions

Building on the findings and limitations of this study, the following recommendations are proposed for future research:

3.7.1 Larger/ Multicentre Studies

Future studies should aim to include larger and more diverse cohorts across multiple centres to improve generalizability and allow for more robust subgroup analyses, particularly for rarer AI subtypes.

3.7.2 Longitudinal Analysis

Incorporating additional time points, including post-treatment responses, would provide a more comprehensive view of how outcomes evolve over the entire treatment pathway. This would help to identify long-term benefits or challenges associated with AI treatments.

3.7.3 Standardized Treatment Protocols

Research should explore the impact of standardized treatment protocols on outcomes, allowing for more consistent comparisons. This could include specific guidelines for whitening, composite restorations, and preventive measures.

3.7.4 Integration of Objective Measures

Future studies could supplement PROM data with objective clinical assessments, such as enamel hardness, plaque scores, or aesthetic ratings by clinicians, to provide a more holistic understanding of treatment outcomes.

3.7.5 Focus on Psychosocial Support

Given the persistent issues with confidence, bullying, and aesthetic dissatisfaction, future interventions should integrate psychosocial support programs. Studies could assess the effectiveness of these programs in improving outcomes for children and young people with AI.

3.7.6 Exploration of Tailored Treatments

Research should investigate how tailored treatments based on AI subtype, age, or severity influence outcomes. For example, hypocalcified AI patients may require more intensive preventive measures due to their increased vulnerability to pain and functional challenges.

3.7.7 Technological Advancements

Investigating the role of emerging technologies, such as digital smile design or minimally invasive techniques, could provide innovative solutions to address both functional and aesthetic challenges in AI patients.

By addressing these areas, future research can build on the findings of this study, contributing to the development of more effective, patient-centred approaches to managing AI and improving the QoL for affected individuals.

Chapter Four: AI PROM – Clinician Feedback Survey

4.1 Aims and Objectives

4.1.1 Aims

AI is a rare inherited condition that affects the enamel, the hard outer layer of teeth. It can result in extreme tooth sensitivity, structural fragility, and aesthetic concerns, significantly impacting the OHRQoL of children and young people. Treating AI presents unique challenges due to its widespread effects on all teeth, necessitating lifelong, multidisciplinary care.

The AI PROM was developed in 2020 by Lyne et al. as a tool to capture patient-reported outcomes in individuals with AI. It is designed to provide clinicians with insights into the functional and aesthetic concerns of patients, aiming to guide treatment decisions and enhance patient-centred care. While it has gained widespread use in the UK, little is known about its effectiveness and utility from the perspective of clinicians who utilize it.

This study aims to evaluate the perceptions of paediatric dentists regarding the AI PROM. Specifically, it seeks to assess its utility in clinical decision-making, identify its limitations, and gather feedback to inform us about potential future improvements. By exploring clinicians' experiences, this research section aims to contribute to the refinement of the AI PROM, ultimately improving the quality of care for children with AI.

4.1.2 Objectives

This study was designed with the following key objectives:

A. To evaluate whether the AI PROM adequately addresses patient concerns, including functional, aesthetic, and psychological aspects of living with AI from the perspective of the treating clinicians.

B. To understand the perceived benefits of AI PROM, including its impact on treatment planning and patient engagement.
C. To identify challenges and barriers faced by clinicians when integrating AI PROM into their workflows.
D. To explore clinicians' recommendations for the timing of AI PROM implementation during different treatment phases, such as pre-treatment, mid-treatment, and at review.
E. To provide practical recommendations based on clinicians' insights for refining and enhancing the AI PROM to better meet patient and clinician needs.

4.2 Methodology

This mixed methods study employed a cross-sectional design using an online survey to collect data from paediatric dentists who regularly come across CYP with the genetic condition of interest (AI) and investigated their personal experiences with the AI PROM. The methodology was designed to capture both quantitative and qualitative insights into clinicians' perceptions.

4.2.1 Ethical Approval

Ethical approval for this study was obtained from University College London (UCL) under the Low-Risk Research Framework (Ethics ID: 26593/001). The ethical approval process adhered to rigorous guidelines to ensure the protection of participants' rights, privacy, and well-being. This process involved submitting a comprehensive application to the UCL Research Ethics Committee, detailing the study's aims, methods, and safeguards for participants. The project was classified as low risk because it involved no interventions or procedures beyond completing a brief online questionnaire.

Participants were provided with detailed information explaining the study's purpose, procedures, and voluntary nature of participation. They were informed that their responses would remain confidential, and that no personally identifiable data would be collected. Informed consent was obtained electronically via Qualtrics™ before participants could access the survey. Respondents had to confirm their understanding and agreement to terms such as confidentiality and the right to withdraw at any point before submission. This step ensured compliance with ethical standards while promoting informed participation.

Data security and anonymity were prioritised throughout the research. Responses were stored on UCL-encrypted servers compliant with General Data Protection Regulation (GDPR) standards. The Qualtrics™ platform was chosen for its robust security measures, including encryption and secure data storage, further ensuring participant confidentiality. Access to the data was limited to the research team, which included the student researcher and supervisors.

This study also accounted for potential risks and burdens to participants, which were deemed minimal, involving only approximately 10–15 minutes to complete the survey. The ethical approval process also established protocols for addressing any unexpected issues, ensuring that their well-being was safeguarded. By adhering to these principles, the study upheld the highest ethical standards while maintaining academic objectivity and reliability.

4.2.2 Survey Development

The survey was designed to gather qualitative and quantitative insights into clinicians' perceptions of the AI PROM. Hosted on Qualtrics™, the platform was chosen for its advanced features, robust security measures, and GDPR compliance. Qualtrics™ offered several advantages over alternative platforms, such as Google Forms, including encryption, flexible question formats, and logic branching, making it an ideal choice for academic research. Its user-friendly interface also ensured accessibility for participants, who could complete the survey conveniently on any device.

Training and Preparation

Formal training was undertaken by the lead researcher (JA) on how to use the Qualtrics™ software, which covered essential aspects such as survey creation, logic setup, and data security. Following this, a survey was designed that met the study's objectives while adhering to ethical and data management standards.
Setting up the survey involved creating an account through UCL's institutional license, which provided additional functionalities such as secure storage on encrypted servers.

The development process began with drafting preliminary survey questions informed by a thorough review of existing literature on methods used to collect useful clinician feedback. Initial drafts were discussed and refined collaboratively with the primary researchers' supervisors through multiple iterations. These discussions focused on ensuring the clarity, relevance, and alignment of the questions with the study's aims.

A pilot of the survey was conducted with staff members from the paediatric dental department within the Anomalies clinic, as well as with research supervisors. This test run was essential for assessing usability, identifying ambiguities, and confirming that the platform functioned as intended. Feedback from this pilot highlighted areas for improvement, including rewording certain questions for clarity and refining the survey logic to improve flow. These insights were instrumental in making final adjustments to the survey, ensuring it was robust and fit for purpose.

Consent Process

The finalised survey began with an introductory section that included a link to the Participant Information Leaflet (PIL) and an electronic consent form. The PIL provided essential information about the study's purpose, procedures, and confidentiality measures, ensuring participants were fully informed before proceeding. Consent was obtained electronically, with participants required to confirm their understanding and agreement to specific statements, including:

- Assurance that their responses would be handled securely and with strict confidentiality during analysis.
- Their right to withdraw at any time before survey submission without providing a reason.
- Acknowledgment that participation was voluntary and would not yield direct personal benefits.

These measures ensured compliance with ethical standards and established trust and transparency with participants, who could only access the survey after providing consent.

Sections and Corresponding Questions

The survey instrument used for this study is provided in Appendix 2, detailing the specific questions designed to gather clinician feedback on the AI PROM.

The main body of the survey was structured around thematic sections designed to align with the study's objectives and facilitate a comprehensive analysis. These themes were selected to capture key aspects of clinicians' experiences and perceptions of the AI PROM, providing a logical flow while ensuring coverage of all relevant areas.

The first section, Familiarity with AI PROM, aimed to assess participants' awareness of and engagement with the tool in their clinical practice. Questions such as "Are you familiar with the AI PROM?" and "Do you use the AI PROM in your unit?" provided foundational insights into how widely the AI PROM is known and used, setting the stage for subsequent analyses.

The second section, Demographics, collected contextual information about participants, including their clinical experience, educational background, and professional roles. This section sought to understand the diversity of respondents' professional profiles and how these factors might influence their perspectives on the AI PROM. Questions included "What is your clinical experience in years since qualifying?" and "What is your professional role (e.g., Consultant, Specialist, Trainee)?" along with inquiries about where participants obtained their primary dental degree and the number of AI patients they typically see in a month.

The AI PROM Content and Relevance sections focused on evaluating whether participants felt that the tool adequately addressed critical patient concerns related to functional, aesthetic, and psychological impacts of AI. Respondents were asked questions such as "Do you believe the AI PROM adequately covers how children feel about their teeth?" and "Do you believe the AI PROM adequately addresses AI symptoms, challenges, and treatment outcomes?" These questions were designed to gauge the effectiveness of the AI PROM in capturing the multidimensional challenges faced by AI patients and guiding clinicians in addressing these concerns.

The fourth section, Challenges in Using AI PROM, aimed to identify potential barriers to implementing the tool in clinical settings. Questions such as "Have you faced challenges using the AI PROM? (e.g., lack of time, patient reluctance, etc.)" and "Please elaborate on any specific challenges you've encountered" sought to uncover practical limitations clinicians might face, such as time constraints, patient motivation, and accessibility of the tool. Understanding these challenges was critical for identifying areas where improvements could enhance the tool's usability.

The fifth section, Perceived Benefits of AI PROM, explored the potential impacts of the tool on clinical decision-making, communication with patients, and overall patient satisfaction. Questions like "Have you observed any positive impacts on children after completing the AI PROM?" and "How useful do you find the AI PROM in making treatment decisions? (Rate 0–100)" were designed to assess whether the AI PROM successfully facilitated patient-centred care and improved treatment planning outcomes.

Finally, the Timing and Recommendations section sought feedback on the optimal timing for administering the AI PROM and suggestions for its refinement. Questions such as "When do you think the AI PROM should be administered? (Pre-treatment, Mid-treatment, at review)" and "Are there any areas you feel the AI PROM is lacking or should include? Please elaborate" encouraged respondents to reflect on their practical experiences and propose ways to enhance the tool for future use.

The sections were intentionally developed to address the study's key objectives and enable a detailed exploration of clinicians' experiences with the AI PROM. By including both closed-ended questions for structured data and open-ended questions to capture nuanced insights, the survey was designed to provide a balanced and in-depth understanding of its utility, limitations, and areas for improvement. The use of free-text comment boxes further encouraged participants to share detailed feedback, adding richness to the data collected.

To ensure accessibility and clarity, the questionnaire was also tested for readability using the Readable.io platform. This evaluation confirmed that the language and structure were appropriate for the intended audience, ensuring that the questions were easy to understand and engage with. These carefully chosen sections and questions, along with the readability assessment, formed the foundation for the subsequent analysis and discussion, ensuring that the survey results would contribute meaningfully to the refinement of the AI PROM.

4.2.3 Participant Recruitment and Data Collection

Participants for this study were recruited through the Amelogenesis Imperfecta Clinical Excellence Network (AI/DI CEN), an established professional network of paediatric dentists involved in managing children and young persons with Amelogenesis Imperfecta (AI). This targeted recruitment approach ensured that the survey reached clinicians with direct and relevant experience of the AI PROM. The recruitment process began with an email facilitated by the AI/DI CEN secretary. The email contained a summary of the study, its aims, and objectives, along with a link and QR code directing recipients to the online Qualtrics™ survey. Data collection was conducted between September 2024 and November 2024 using the Qualtrics™ platform.

The survey was distributed in two phases to ensure a comprehensive response rate. The first circulation of the survey was sent to colleagues on September 26th, 2024, and by October 22nd 2024, we had received a 28.2% response rate, corresponding to 22/78 completed surveys. After reviewing the initial responses, a second circulation was carried out on October 22nd, 2024. This second circulation resulted in an additional 2 responses, bringing the total response rate to 30.8%. This two-phase approach was aimed to increase participation and attempt to gain a more representative sample for analysis.

Upon accessing the survey, participants were presented with a comprehensive introductory page, which included a link to the Participant Information Leaflet (PIL). This document outlined the study's purpose, the voluntary nature of participation, and assurances of confidentiality (as Qualtrics™ does not store IP addresses). This was followed by an electronic consent form, requiring participants to confirm their understanding and agreement to key ethical statements, including their right to withdraw before submission and the anonymised handling of their responses.

Only after completing this consent process were participants able to access the main survey. This staged approach ensured that participants were fully informed and provided explicit consent, adhering to the rigorous ethical standards established in the approval process.

4.2.4 Data Analysis

The collected data consisted of both quantitative responses from closed-ended questions and qualitative feedback from open-ended questions and free-text comment boxes. A mixed-methods approach was employed to analyse the data, ensuring a comprehensive understanding of clinicians' perceptions and experiences.

Quantitative data were processed initially using Qualtrics™' built-in analytical tools. These tools facilitated the calculation of descriptive statistics, including percentages and averages, which helped identify key trends and patterns in the responses. For further exploration and visualisation, the data were exported to Microsoft Excel™. In Excel™, charts and graphs were created to illustrate the distribution of responses across themes, providing clear and impactful visual representations for inclusion in the final analysis.

Thematic content analysis was used to examine the qualitative data, focusing on identifying recurring patterns, insights, and suggestions provided by respondents. . The researcher read and familiarised themselves with the free text comments. The data were then sorted and coded into emerging themes. Further refinement of the process led to overarching categories/themes aligned with the survey's structure, such as familiarity with AI PROM, perceived benefits, challenges, and recommendations for improvement. This approach added depth to the findings, capturing clinicians' nuanced perspectives and highlighting areas for potential refinement of the AI PROM.

The combination of Qualtrics™ for initial processing, Excel™ for enhanced data visualisation, and thematic content analysis for qualitative insights ensured a robust and detailed exploration of the data. This methodology not only reinforced the reliability and validity of the findings but also provided a balanced understanding of the AI PROM's utility, challenges, and areas for enhancement. These results serve as a foundation for the subsequent discussion, aiming to inform improvements to the AI PROM and contribute meaningfully to patient-centred care.

4.3 Analysis of Qualtrics™ Survey Results

A total of 24 paediatric dentists engaged with the survey, providing insights into their experiences with the AI PROM. This represents a response rate of 30.8%, based on the 78 individuals listed on the AICEN mailing list. The findings are organised below under key themes derived from the survey objectives.

4.3.1 Consent and Engagement

A total of 24 individuals interacted with the survey, of whom 23 provided consent. This corresponds to a consent rate of 95.8%. Responses to subsequent questions varied depending on completion rates.

4.3.2 Demographics

The clinical experience of the respondents varied widely, with the largest proportion (42%) reporting more than 15 years of experience, representing a strong senior professional cohort. A third (33%) had 5-10 years of experience, indicating significant participation from mid-career clinicians. Early-career professionals with less than five years of experience accounted for 17% of the respondents, while a smaller proportion (8%) reported 10-15 years of experience. This diverse distribution highlights significant feedback from experienced professionals while also incorporating the perspectives of early-career clinicians.

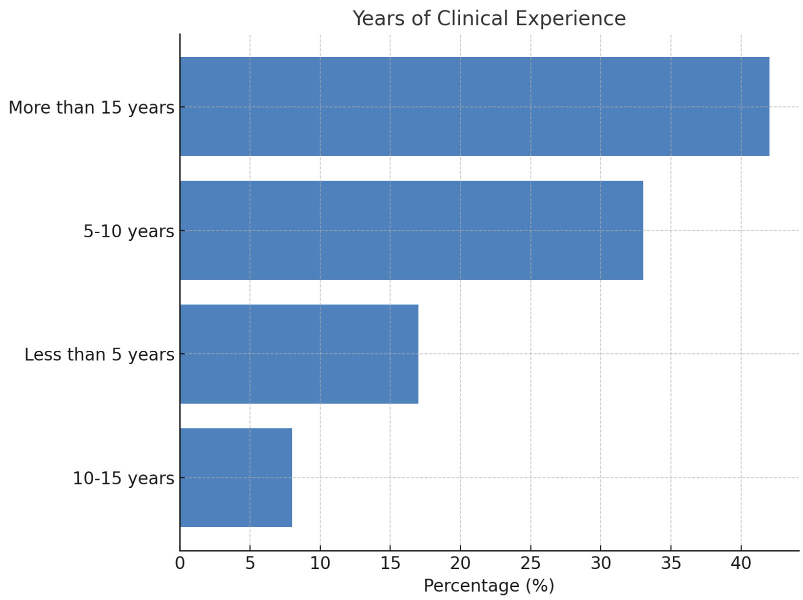


Figure 42 Bar chart illustrating the distribution of clinical experience amongst survey respondents.

Most participants (91%) obtained their primary dental degree (BDS) in the UK, with a smaller proportion (9%) completing their qualifications in Europe. This reflects the regional focus of the survey, emphasising insights from those trained within the UK system.

The professional roles of respondents were also diverse. The largest group (45%) identified as consultants, followed by HEE-Trainees (StR) at 36%. Smaller groups included post-graduate students and university academics or honorary NHS consultants, each contributing 9%. This distribution ensured representation from clinicians directly involved in patient care, treatment planning, and education, providing well-rounded feedback on the AI PROM.

Figure 43 Horizontal bar chart illustrating the distribution of professional roles amongst respondents.

In terms of clinical exposure, responses regarding the number of AI patients seen monthly were varied. Half of the respondents (50%) reported seeing 1-5 AI patients on average, while 25% saw 5-10 patients, and an equal proportion handled more than 15 cases per month. This range of workloads highlights the diversity in the sample, reflecting varied levels of familiarity with AI-related cases among participants.

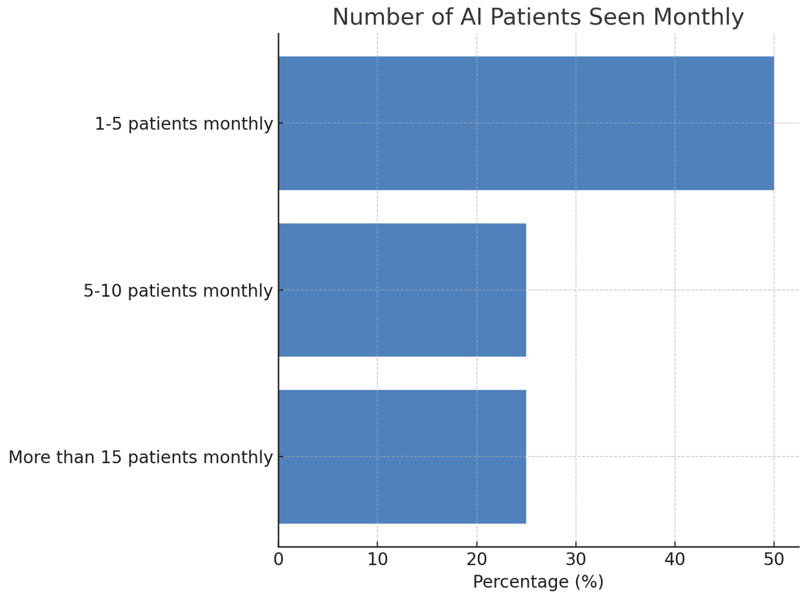


Figure 44 Horizontal bar chart illustrating the number of AI patients' clinicians see on average.

4.3.3 AI PROM Familiarity and Usage

Out of the 23 respondents who consented to participate in the survey, 22 provided an answer regarding their familiarity with the AI PROM. The majority, 81.8% (18 participants), reported being familiar with the AI PROM, while 18.2% (4 participants) indicated they were unfamiliar with it.

Out of the 18 respondents familiar with the AI PROM, 67% (12 participants) confirmed that they actively used the tool in their clinical unit, while 33% (6 participants) indicated that they did not. This suggests variability in the integration of the AI PROM into clinical workflows among those familiar with the tool.

Respondents were asked about the optimal timing for administering the AI PROM, and their feedback highlighted its relevance at multiple stages of the treatment process. All respondents (100%) agreed that the AI PROM should be administered both pre-treatment and at review stages. Pre-treatment was identified as critical for establishing a baseline understanding of patient concerns and guiding the development of personalized treatment plans. Similarly, review stages were recognised as essential for assessing treatment outcomes and ensuring that patient concerns were adequately addressed. One respondent remarked, "*Administering the AI PROM pre-treatment* *provides a baseline understanding of patient concerns, which informs treatment goals.*"

In addition to its pre- and post-treatment applications, a significant proportion (67%) recommended using the AI PROM mid-treatment. This was particularly emphasised in cases where the treatment course was very long or when there were significant changes, such as shifts in patient motivation or circumstances. One participant explained, *"I would include a mid-treatment PROM if the treatment course was very long, or if there was significant time elapsed such that a slight change of plan was warranted, or if there was a change in patient motivation etc."*

The distribution of responses reflects the perceived versatility of the AI PROM. While pre-treatment and review stages were universally favoured, mid-treatment usage was also recognised as valuable for tracking progress, making necessary adjustments, and evaluating long-term outcomes in more complex cases.

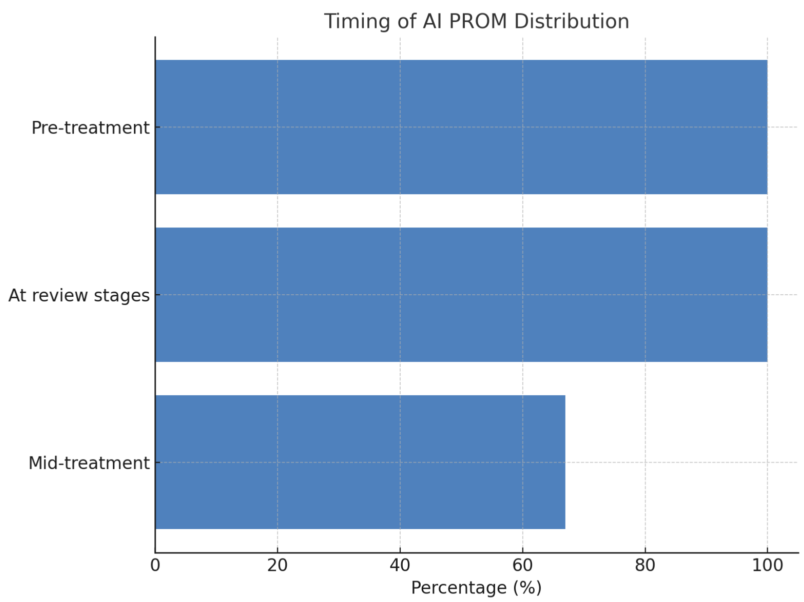


Figure 45 Horizontal bar chart illustrating respondents' preferences for the timing of the AI PROM distribution.

4.3.4 AI PROM Coverage

Clinicians were asked to evaluate whether the AI PROM adequately addressed key aspects of patient experiences, including functional and aesthetic concerns.
Responses indicated a generally positive assessment of the tool's coverage in certain areas. For example, 75% of respondents agreed that the PROM adequately addressed both how children feel about their teeth and AI symptoms, while the remaining 25% believed those aspects were somewhat addressed. Notably, no participants indicated that these areas were entirely neglected, reflecting a strong consensus on their inclusion in the PROM.

In contrast, opinions were more divided regarding the PROMs coverage of AI challenges. Half of the respondents agreed that this aspect was adequately addressed, while the other half felt it was somewhat addressed, highlighting potential areas for improvement.

The evaluation of how well the PROM assesses AI treatment outcomes revealed it to be the area of greatest dissatisfaction among clinicians. Only 33% felt that treatment outcomes were adequately covered, while 58% believed this aspect was somewhat addressed, and 8% indicated it was not adequately addressed. One clinician remarked, *"The survey could better capture long-term treatment outcomes to assess the lasting impact on the child's QoL."*.

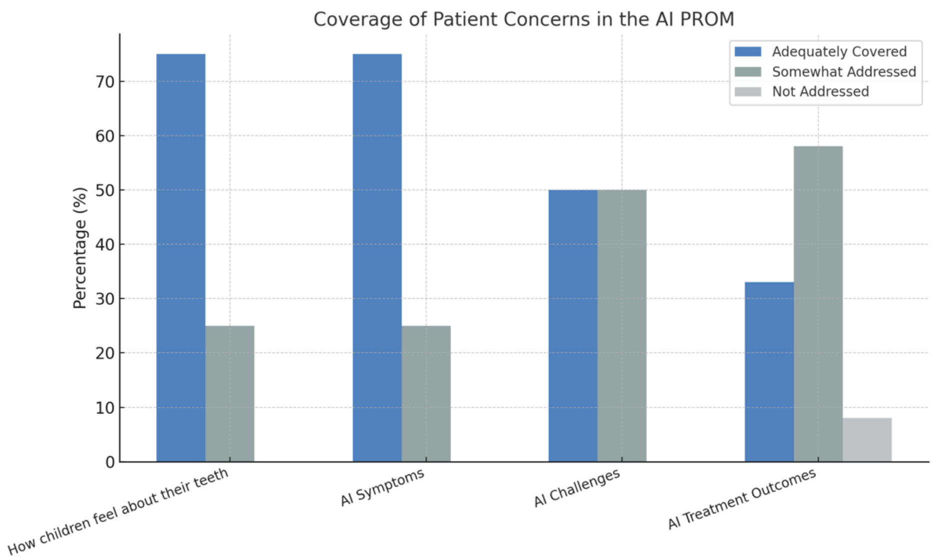


Figure 46 Grouped bar chart illustrating the coverage of patient concerns in the AI PROM based on respondent's feedback.

4.3.5 AI PROM Perceived Benefits

Respondents highlighted several benefits of the AI PROM, emphasising its value in improving patient experiences and guiding clinical practice. One of the key advantages noted was improved communication, with 64% of respondents indicating that the tool facilitated discussions about patient concerns, particularly regarding aesthetics and functionality. One clinician remarked, “The survey gave patients a platform to voice concerns they may have otherwise hesitated to share,” underscoring the PROM’s ability to encourage open dialogue. Additionally, 56% of participants observed enhanced patient satisfaction with the treatment process when the AI PROM was used, suggesting its role in fostering positive perceptions of care. The tool was also described as instrumental in tailoring treatments to the specific needs of children with AI, providing valuable guidance for treatment planning.

When asked whether they had encountered positive impacts on children after completing the AI PROM, 83% of respondents reported observing positive changes, while 17% indicated they had not noticed any specific benefits.

Respondents who reported positive impacts provided detailed insights into the specific benefits they observed. Improved communication emerged as a standout outcome, with all respondents (100%) agreeing that the AI PROM made patients feel more comfortable discussing their issues. One clinician commented, “The AI PROM provides children with an opportunity to express concerns they may not have felt comfortable raising otherwise.”

In addition to facilitating communication, the AI PROM was perceived by some respondents as contributing to a more structured and patient-centred consultation process. Twenty percent of respondents noted that children who completed the PROM prior to their visit appeared more engaged during consultations compared to those who had not. Similarly, 10% observed that these patients seemed more receptive to the advice provided during consultations. While these observations suggest potential benefits, it is important to note that direct causation between the use of the AI PROM and increased patient satisfaction cannot be definitively established from this feedback.

Other benefits were also reported, with 20% selecting "Other" in the survey. One respondent observed, “*Patients open-up about what challenges they are having. When we don’t use it, they often say everything is fine, but that is not always the case*.” Another clinician mentioned, “*Parents feel they understand issues better and value this tool as being helpful in that aspect*.”

4.3.6 AI PROM Challenges

Respondents identified several challenges associated with the use of the AI PROM in clinical settings. Time constraints emerged as the most significant barrier, with 50% of participants reporting a lack of time to incorporate the survey effectively into their workflow. Additionally, 33% noted a lack of motivation among patients, particularly children, to engage with the survey. While most clinicians (92%) felt that children were generally willing to complete the AI PROM, 8% observed that some children were reluctant, which can hinder its effectiveness in specific cases. Similarly, 25% of respondents indicated that a lack of parental motivation to encourage their children to participate further complicated the survey’s implementation. One clinician remarked that “*child and parent reluctance to open up about the barriers and challenges they face*” posed a significant challenge, while another mentioned “*child reluctance to complete the survey*” as a barrier.

Survey availability was cited as an issue by 8% of respondents, suggesting that logistical factors also play a role in limiting the tool’s accessibility. Additionally, 17% selected "Other" to describe fewer common obstacles not explicitly listed in the survey. These included instances of children or parents being hesitant to discuss the barriers and challenges they face, highlighting the nuanced nature of these challenges and the need for tailored strategies to address them.

Most respondents (92%) reported that they had not encountered any negative impacts on children after completing the AI PROM. However, one clinician (8%) noted that completing the survey was perceived by a patient as prolonging their appointment, suggesting that in some cases, the additional time required for the PROM may be viewed as a drawback.

4.3.7 AI PROM Usefulness in Clinical Practice

Respondents were asked to evaluate the AI PROM's usefulness in making treatment decisions and understanding the broader impact of AI on a child's life using a scale of 0–100, where 0 indicated "not at all useful" and 100 indicated "extremely useful." The average rating was 56 (rounded to the nearest whole number), with individual ratings ranging from 21 to 90.

In addition to its role in treatment planning, 83% of respondents agreed that the AI PROM enhanced their understanding of the impact of AI on a child's life, including psychological, functional, and aesthetic challenges. However, 17% indicated that it had not significantly contributed to their awareness. Moreover, one clinician remarked that the tool is particularly effective in highlighting aspects of a child's life that might not otherwise surface during routine consultations, emphasising its potential as a resource for improving patient-centred care.

4.3.8 AI PROM Qualitative Analysis

Clinicians provided several recommendations to improve the AI PROM, focusing on capturing more comprehensive treatment outcomes, such as psychological and functional changes, and introducing flexibility in timing to include mid-treatment assessments. Suggestions also emphasised refining the question design to maintain patient engagement and making the tool more accessible by integrating it into clinical workflows, such as through electronic health records or mobile platforms. These insights highlight the need to balance practicality and patient-centred care in future improvements to the AI PROM. The main themes identified are outlined in Table 10.

Table 19 Key themes for Enhancing the AI PROM Based on Clinician Feedback

Recommendation	**Percentage Mentioned (n)**	**Example Quote**
Expand treatment outcomes	44% (n=11)	"Include questions about emotional impacts and long-term QoL."
Streamline question content design	36% (n=9)	"Focus on severity rather than frequency and reduce the number of questions."
Flexible timing for assessments	36% (n=9)	"Introduce mid-treatment assessments to monitor progress and adapt treatment plans."
Improved accessibility	28% (n=7)	"A mobile app or integration into EHRs would make it easier to use in a busy clinic."

4.4 Discussion

The findings from the clinician feedback survey provide valuable insights into the perceived utility, challenges, and potential improvements of the AI PROM in clinical practice. Clinicians generally recognised the AI PROM as a meaningful tool for enhancing communication with patients, informing treatment decisions, and improving their understanding of the multidimensional impact of AI on children's lives. These findings align with broader evidence highlighting the importance of PROMs in paediatric dentistry, such as the COHIP and ECOHIS, which emphasise the need for patient-centred approaches to care (Broder et al., 2007; Marshman et al., 2015; John et al., 2020).

4.4.1 Demographics and Clinical Exposure

The demographics of the respondents revealed an interesting and diverse distribution of clinical exposure. While 42% of participants had over 15 years of clinical experience, this did not necessarily correlate with a higher number of AI patients seen monthly. Half of the respondents reported seeing only 1–5 AI patients per month, regardless of their level of specialisation or seniority.

Systemic barriers within healthcare may limit the flow of AI cases to specialised clinicians. These barriers include referral inefficiencies, geographic disparities, and the logistical challenges posed by managing rare conditions like AI. For instance, geographic disparities may result in patients being unable to access centralised services, while uneven referral pathways can lead to delays or patients being managed by non-specialist clinicians. These issues are particularly relevant in managing rare conditions such as AI, where the patient population is already limited, making streamlined and efficient pathways critical (Smith et al., 2018). Addressing these barriers could improve access to specialised care and ensure patients receive appropriate treatment. The hub-and-spoke model adopted in cleft lip and palate care nationally could serve as a template for designing the clinical pathway (reference).

Additionally, the professional roles of respondents were diverse, with 45% identifying as consultants and 36% as trainees, alongside smaller groups of postgraduate students and academics. This varied cohort ensures a range of perspectives, but it also reflects the wide scope of clinicians engaging with AI cases. Interestingly, the frequency of AI patient encounters did not significantly differ between senior consultants and trainees. One possible explanation for this is that junior grades, such as trainees and postgraduate students, often see patients under the supervision of consultants, which could influence the patient volume reported across different levels of experience. Centralised care systems or lengthy travel times may also disproportionately limit access to consultants, even though their expertise would benefit complex cases.

Literature in medical and dental fields supports these observations, emphasising that specialists in rare conditions often face challenges in accessing a sufficient patient population due to logistical constraints and inconsistent referral pathways (Smith et al., 2018).

4.4.2 AI PROM Perceived Benefits

Most respondents reported that the AI PROM facilitated better communication between children, their families, and clinicians by providing a structured platform to articulate concerns. This feedback mirrors research on OHRQoL tools like COHIP, which emphasise the role of PROMs in enhancing patient-clinician dialogue and addressing psychosocial impacts (John et al., 2020). The tool's ability to uncover issues that might otherwise remain unspoken reinforces its utility in promoting patient-centred care.

For example, some parents reported feeling more empowered to understand their child's condition and discuss concerns with the clinician. One clinician noted, "The AI PROM helps highlight issues children may hesitate to share, fostering a more open and collaborative treatment process." Such psychological benefits are vital in paediatric dentistry, where emotional challenges often accompany functional and aesthetic concerns.

Additionally, 83% of respondents indicated that the AI PROM improved their understanding of how AI affects a child's life. This finding highlights the AI PROM's ability to capture the emotional, functional, and social dimensions of oral health, consistent with the goals of PROMs to address broader quality-of-life factors (Marshman et al., 2015). However, the variability in its perceived usefulness for treatment decision-making (average score: 55.9/100) suggests that while the tool provides valuable insights, its practical application in guiding treatment plans could benefit from further refinement.

4.4.3 AI PROM Challenges

Despite its benefits, the AI PROM presents several challenges that may hinder its widespread adoption. The most reported barrier was time constraints (50% of respondents), which reflects the difficulty of integrating PROMs into busy clinical workflows. Additionally, issues related to patient or parental motivation were reported by 33% and 25% of respondents, respectively. These findings are consistent with broader literature on PROM implementation challenges in paediatric dentistry, where engaging younger patients and maintaining their attention can be particularly challenging (Foster Page et al., 2016).

Time constraints have been highlighted in other studies as a key barrier to adopting PROMs. For example, research demonstrates that integrating PROMs into electronic health records (EHRs) or mobile applications significantly reduces administrative burdens and increases clinician uptake (Brown et al., 2019). Incorporating the AI PROM into existing digital systems could similarly streamline its use and address time-related challenges.

Although 92% of respondents felt that children were generally willing to complete the AI PROM, occasional reluctance among younger patients was noted, emphasising the need for strategies to foster greater engagement. Child-friendly adaptations such as gamified designs or visual aids have proven effective in improving engagement in paediatric care settings (Foster Page et al., 2016). Implementing these features in the AI PROM could enhance its usability for younger patients.

Survey availability was another concern, with 8% of respondents indicating that the tool was not readily accessible in their clinical settings. Improving logistical access, such as offering the PROM in digital formats, could overcome this limitation. For instance, PROMs deployed via mobile apps or digital platforms have been shown to significantly increase availability and usage (Marshman et al., 2015).

4.4.4 Areas for Improvement

Respondents provided several constructive suggestions for enhancing the AI PROM to maximise its utility and ease of use. A key recommendation was to expand treatment outcomes, with 44% suggesting the inclusion of questions that address emotional and functional changes post-treatment. One respondent noted, "*Including questions about the emotional impact of AI would provide a more holistic view of patient experiences*." This reflects a broader need for PROMs and PREMs (Patient Reported Experience Measures) to capture the long-term impacts of treatment, consistent with findings from other tools like the ECOHIS (Marshman et al., 2015).

Another common suggestion was to introduce flexible timing for assessments, particularly mid-treatment evaluations. This was highlighted by 36% of respondents who believed that mid-treatment PROMs could help monitor progress and adapt treatment strategies in real-time. Mid-treatment assessments are particularly relevant for managing AI due to the condition's complex and often lengthy treatment pathways, which may involve multiple stages of care over an extended period. By incorporating these assessments, clinicians could monitor the evolving needs of patients, identify any shifts in patient motivation, and adapt treatment plans accordingly. For example, if a child's psychosocial concerns or functional challenges increase during treatment, mid-treatment PROMs could facilitate timely interventions, potentially enhancing both treatment adherence and outcomes. Such flexibility allows clinicians to provide care that is responsive to the dynamic nature of AI, supporting both short- and long-term patient needs.

Refining question design was also emphasised, with recommendations to reduce the number of questions and focus on severity rather than frequency to maintain patient engagement. Additionally, 28% suggested integrating the AI PROM into EHRs or mobile platforms to streamline its administration and improve accessibility. Streamlining the PROM reflects the need for brevity in busy clinical settings while balancing depth with practicality. Literature on PROMs in dentistry highlights the value of follow-up assessments to capture long-term outcomes and reduce survey fatigue by limiting the number of questions (Broder et al., 2007).

4.4.5 Study Limitations

This study has several limitations that should be considered when interpreting the findings. The sample size was small, with only 23 respondents completing the survey, reflecting the rarity of AI and the limited number of clinicians who manage this condition. While this may limit the generalisability of the findings, the response rate provides valuable context. With 78 clinicians on the AICEN mailing list, the survey achieved a response rate of approximately 29.5%, which is reasonable for surveys conducted in this professional demographic. However, the findings should still be interpreted with caution as they may not fully represent the perspectives of all AICEN members.

The study's geographic focus on the UK restricts its applicability to international contexts. Differences in healthcare systems, referral pathways, and clinical practices may mean that the experiences of clinicians outside the UK vary significantly.

Additionally, the survey captured only the clinician perspective, excluding patient and parental views, which are critical for a holistic evaluation of the AI PROM. Including patient perspectives in future research would provide richer insights into the tool's impact on quality of life. Literature on rare conditions emphasises the importance of multi-centre studies to overcome sample size limitations and incorporate diverse perspectives, such as those of patients and caregivers (Smith et al., 2018).

4.4.6 Study Implications

Broader Clinical Implications

The findings suggest that the AI PROM holds significant potential for advancing patient-centred care within paediatric dentistry. By capturing patient concerns related to functional, psychological, and aesthetic aspects of AI, the tool facilitates meaningful dialogue between clinicians and patients. This improved communication can inform individualised treatment planning and enhance patient satisfaction with care. However, to fully realise its potential, refinements are required to address the challenges identified in this study, such as time efficiency and patient engagement.

Similar adaptations in PROMs like the COHIP have demonstrated success in improving usability and adoption (Broder et al., 2007). For instance, the COHIP has been refined by reducing the number of questions to minimise survey fatigue, making it more practical for use in busy clinical settings. Additionally, integrating the COHIP into digital platforms, such as the REDCap (Research Electronic Data Capture) system, has enhanced its accessibility and streamlined data collection. REDCap allows clinicians to administer the COHIP electronically, automate scoring, and securely store patient data, enabling a more efficient workflow and reducing the manual effort involved in traditional paper-based PROMs. These adjustments highlight the importance of balancing comprehensiveness with practicality to ensure effective implementation and sustained usage in clinical workflows.

Policy Implications

Integrating the AI PROM into routine clinical practice requires systemic support. Providing structured training programs for clinicians and incorporating the tool into EHRs could streamline its use, reduce time burdens, and ensure consistency in implementation. Policy-level efforts to promote its availability in diverse clinical settings could also address accessibility barriers, as demonstrated in studies where digital tools significantly increased PROM usage (Brown et al., 2019).

Research Implications

This study highlights the need for continued research into the refinement and validation of the AI PROM. Future studies should focus on expanding its application to a broader range of clinical settings and comparing its performance with established OHRQoL measures such as the COHIP and ECOHIS. For instance, the COHIP has been extensively validated and shown to provide comprehensive insights into the psychosocial and functional impacts of oral health conditions in children (Marshman et al., 2015). Evaluating the AI PROM against such measures would provide a clearer understanding of its strengths and areas for development.

Additionally, validating the AI PROM would be particularly beneficial as it targets the AI population specifically, centralising its design and application to address the unique needs of this group. This contrasts with broader surveys like COHIP and ECOHIS, which, while valuable, often serve general dental populations and may not adequately capture the nuanced challenges faced by children with AI as they are not a condition-specific measure.

Moreover, longitudinal research is needed to examine the long-term impacts of the AI PROM on satisfaction with treatment outcomes and patient QoL. This would address a critical gap in the current literature, as most studies focus on short-term effectiveness. By establishing evidence for its sustained benefits, the AI PROM could emerge as a model for integrating patient-reported outcome measures into paediatric dental care globally.

4.5 AI PROM – Clinician Feedback Summary

This chapter presented the findings of a clinician feedback survey on the AI PROM, highlighting its perceived benefits, challenges, and areas for improvement. The AI PROM was valued for its role in enhancing communication and understanding patient concerns, though its application in treatment decision-making received mixed feedback. Key challenges include time constraints, patient engagement, and survey design, which clinicians believe could be addressed through refinements like mid-treatment assessments and a shift from frequency- to severity-focused questions.

The study not only underscores the AI PROM's potential to enhance patient-centred care but also contributes to the broader goal of refining PROMs to address rare conditions like AI more effectively. By targeting specific patient populations, such as children with AI, the tool offers a tailored approach that can inform treatment planning, improve communication, and enhance patient satisfaction. These insights align with global efforts to advance the role of PROMs in healthcare by addressing logistical barriers and integrating digital innovations. Future work should build on these findings to establish AI PROM as a model for condition-specific PROMs globally, ultimately contributing to a more personalised and inclusive approach to care.

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Appendix

AI PROM

Amelogenesis Imperfecta
Patient Survey

Thank you for helping us with our survey. We would like to ask you a few questions about your teeth and smile.

Please turn over the page and answer the questions. When you are finished, please give it back to your dentist.

Circle your answers like the example below:

1. Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never

Your dentist will fill out the following information for you

Age: .... years   Gender: M / F / X

AI phenotype (*circle all that apply*): hypoplastic / hypocalcified / hypomature / mixed

Treatment stage (*please circle one*): pre-treatment / mid-treatment / post-treatment / review

Please circle your answer

1. Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never
2. Do you have difficulty eating foods you would like to, because of your teeth?	Often	Sometimes	Never
3. Does it hurt when you brush your teeth?	Often	Sometimes	Never
4. Do you miss school because of your teeth (except for dentist appointments)?	Often	Sometimes	Never
5. Do you feel unhappy with the way your teeth look?	Often	Sometimes	Never
6. Do your teeth affect your confidence to smile?	Often	Sometimes	Never
7. Do you get teased or bullied because of your teeth?	Often	Sometimes	Never
8. Do you feel scared or anxious about having dental treatment?	Often	Sometimes	Never

9. Are you happy with your teeth?	Yes	No
10. Is there anything else you would like us to know about your teeth and how they affect you?		

Qualtrics Survey

Paediatric dentists' perceptions of the quality and impact of the Amelogenesis Imperfecta Patient Reported Outcome Measure (AI PROM)

Consent

Dear Colleague,

As part of my Doctorate (DDent), I am carrying out a research project titled: Paediatric dentists' perceptions of the quality and impact of the Amelogenesis Imperfecta Patient Reported Outcome Measure (AI PROM).

This study aims to evaluate dentists' views of the existing AI PROM for children with AI.

Your participation is completely voluntary, but we would very much appreciate if you could spare approximately 10 mins to help us with this study which forms part of my DDent.

Participation will involve completing a short online questionnaire on Qualtrics.

All data will be treated completely confidentially, and you will not be personally identified in any work arising from the research.

Please note, this research has gained ethical approval from University College London (Ethics ID number: 26593/001)

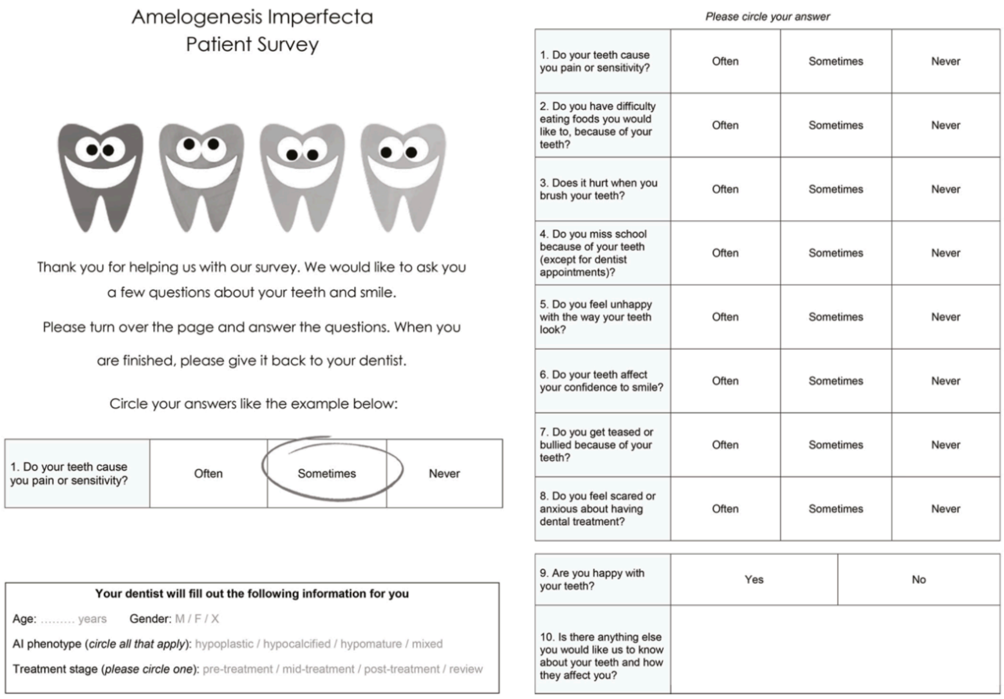
With very many thanks,
Jenan Altaher (DDent Student)
Prof Susan Parekh and Dr Fiona Ryan (Research Supervisors)

Please tick the statements below to indicate you consent to taking part in this study titled: Paediatric dentists' perceptions of the quality and impact of the Amelogenesis Imperfecta Patient Reported Outcome Measure (AI PROM).

You must TICK ALL THE BOXES (i.e. consent to all statements below) to complete the survey.

- I confirm that I have read and understood the Participant Information Leaflet/background information above for this study and also had an opportunity to consider the information and what I will need to do.
- I understand that the data I provide for this study will be stored confidentially and securely and that it will not be possible to identify me in any publications.
- I understand that there are no major benefits to me completing the questionnaire and taking part in this study, but it is hoped that future patients may benefit.
- I understand that I will not benefit financially from this study or from any possible outcome that may result in the future.
- I voluntarily agree to take part in this study and to complete the questionnaire.

Familiarity with the AI PROM



Amelogenesis Imperfecta
Patient Survey

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Treatment stage (*please circle one*): pre-treatment / mid-treatment / post-treatment / review

Please circle your answer

1. Do your teeth cause you pain or sensitivity?	Often	Sometimes	Never
2. Do you have difficulty eating foods you would like to, because of your teeth?	Often	Sometimes	Never
3. Does it hurt when you brush your teeth?	Often	Sometimes	Never
4. Do you miss school because of your teeth (except for dentist appointments)?	Often	Sometimes	Never
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6. Do your teeth affect your confidence to smile?	Often	Sometimes	Never
7. Do you get teased or bullied because of your teeth?	Often	Sometimes	Never
8. Do you feel scared or anxious about having dental treatment?	Often	Sometimes	Never

9. Are you happy with your teeth?	Yes	No
10. Is there anything else you would like us to know about your teeth and how they affect you?		

Are you familiar with the AI PROM?
The AI PROM (Lyne et. al, 2020) is used to help guide dentists in making clinical decisions based on the responses obtained to questions asked about functional and aesthetic concerns AI patients may experience.

- No
- Yes

Do you use the AI PROM in your unit?

- No
- Yes

Background Information

Clinical experience in years since qualifying:

- < 5 years
- 5 - 10 years
- 10 - 15 years
- > 15 years

Where did you get your primary degree (BDS)?

- UK
- Republic of Ireland
- Europe
- USA
- Asia
- Middle East
- Other

Are you a...

- Post-Graduate
- Specialist
- HEE Trainee (StR)
- University Academic / Honorary NHS Consultant
- Consultant

Within a 1 month period how many AI patients (including NP, treatment, review, etc.) would you see on average?

none

- 1 - 5
- 5 - 10
- > 10

AI PROM Content and Relevance

Do you believe the AI PROM adequately covers the following patient concerns:

	Yes	Somewhat	No
How children feel about their teeth	Yes	Somewhat	No
AI Symptoms	Yes	Somewhat	No
AI Challenges	Yes	Somewhat	No
AI Treatment outcomes	Yes	Somewhat	No

Applicability to the Clinical Setting

Please select all or none of the challenges that apply whilst using the AI PROM in your unit:

	Yes	No
Survey not readily available	Yes	No
Lack of time	Yes	No
Lack of patient motivation	Yes	No
Lack of parent motivation	Yes	No
Other reasons	Yes	No

As you have selected other reasons in the previous question, please elaborate below:

Patient Engagement and Experience

Do you find children are generally willing to participate in answering the AI PROM?

- No
- Yes

Have you ever encountered any positive impacts on children after completing the AI PROM?

- No
- Yes

Please select positive impacts encountered when using the AI PROM You may select more than one option

- Patient felt more comfortable to speak about issues after the survey was completed
- Patient appeared more satisfied with the advice received compared to children who had not completed the AI PROM at the start of the visit
- Patient appeared more satisfied with the treatment received compared to children who had not completed the AI PROM at the start of the visit
- Other

Have you ever encountered any negative impacts on children after completing the AI PROMS?

- No
- Yes

Please select negative impacts encountered after completing the AI PROM You may select more than one option

- Patient appeared more upset after completing the AI PROM
- Patient appeared more anxious / embarrassed after completing the AI PROM
- Patient felt completing the AI PROM prolonged the appointment
- Other

Outcome Measurement and Treatment Planning

How useful do you find the AI PROM in making treatment decisions for children with AI? Is it a decision-making guide?

0 = Useless

100 = Extremely Useful

	0 10	20	30	40	50	60	70	80	90	100
Usefulness scale										

Has using the AI PROM contributed to improving your understanding the impact of AI has on a child's life?

- No
- Yes

When do you think children should be asked to complete the AI PROM? You may select more than one option.

- Pre Treatment
- Mid Treatment
- At Review

Suggestions for Improvement

Are there any areas you feel the AI PROM is lacking or should include?

Thank You.