Title

A UK-based Consensus on Clinical Decision Flowcharts for Managing Childhood Amelogenesis Imperfecta in the Permanent Dentition.

Declarations

The authors have no relevant financial or non-financial interests to disclose.

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Abstract

Purpose: Amelogenesis imperfecta (AI) is a rare genetic condition characterised by defective enamel formation, with variable presentations. Diagnosis traditionally involves family history, clinical presentation, with genetic testing being increasingly integrated into clinical practice. Children and young people (CYP) with AI face clinical and psychosocial challenges over extended periods. Given the absence of specific guidelines for AI management in CYP, this project aimed to develop flowcharts to guide dental practitioners in managing the three primary AI phenotypes: hypoplastic, hypocalcified, and hypomaturation. The flowcharts focus on the permanent dentition and provide a structured, yet flexible treatment approach, addressing key clinical issues such as hypersensitivity management, and aesthetic restoration.

Methods: The flowcharts were developed collaboratively by the UK Amelogenesis Imperfecta/Dentinogenesis Imperfecta National Clinical Excellence Network (AI/DI CEN). Expert opinion was sought over a series of meetings with specialists who treat children with amelogenesis imperfecta. Initial drafts of the flowcharts were discussed in two discussion forums until consensus was achieved.

Results: The flowcharts prioritise prevention, minimally invasive treatment, and shared decision-making, adapting interventions to phenotype type and severity as well as individual patient needs.

Conclusion: This guidance is presented to assist dental professionals in delivering comprehensive, empathetic, and effective care for CYP with AI.

Key words

Amelogenesis imperfecta, paediatric dentistry, phenotype-genotype correlation, psychosocial impact, preventive care, multidisciplinary treatment.

Introduction

Amelogenesis imperfecta (AI) is a rare genetic condition resulting in the formation of defective enamel. The prevalence varies depending on the population studied with values of 1:14,000 reported in the USA (Witkop, 1957) and 1:700 in Sweden (Bäckman & Holm, 1986). AI affects both the primary and permanent dentitions. The presentation can vary between the two dentitions with primary teeth appearing less affected.

The classification of AI is challenging reflecting inconsistent use of terminology and interpretation of post-eruptive changes to enamel. The Witkop (1988) classification identifies three major phenotypes of AI: hypoplastic, hypocalcified and hypomaturation. It is not an exhaustive list and significant variation does exist within each phenotype.

The potential for genetics to add value to clinical classification has long been recognised (Aldred et al, 2003). Traditionally, AI diagnosis is based on family history, pedigree plotting and clinical presentation. Genetic diagnosis is a growing field with increased availability of genetic testing in clinical practice including in the UK via the state-funded National Health Service (NHS) (PanelApp, 2023) with a need to upskill the dental workforce to use the tests appropriately (McDowall et al, 2018). There is an increasing understanding of phenotype-genotype associations (Dong et al., 2023; Wright et al., 2011), however currently most clinicians do not have access to information on the genetics of each presenting phenotype. Consequently, the Witkop classification is still broadly used among UK clinicians and taught as an approach to treatment planning. Given that clinicians are the target audience of this paper, it therefore considers the three AI phenotypes as identified in the Witkop classification. As genotype-phenotype associations are developed, there may be potential to provide treatments options by genotype, but currently this is not the situation in most cases.

Children and young people (CYP) with AI have higher burden of dental care than unaffected individuals (Lafferty et al., 2021). Treatment for AI can be challenging due to hypersensitivity, rapid tooth wear (Toupenay et al., 2018), and compromised enamel bonding (Faria-e-Silva et al., 2011). Further to these issues, delayed eruption and gingival maturation may limit restorative options in young patients (Patel et al., 2013). Orthodontic care is equally challenging, due to bonding difficulties and common presence of anterior open bite (Persson & Sundell, 1982; Arkutu et al., 2012). The psychosocial impact of AI is well documented, with higher levels of distress, social avoidance and poorer oral health-related quality of life than unaffected peers (Coffield et al., 2005; Parekh et al., 2014; Pousette-Lundgren et al., 2015).

Children with AI often require a combination of specialist dental input and care from a general dental practitioner (GDP), with the latter being instrumental in identifying CYP who require specialist management (Lafferty et al., 2022), in addition to provision of routine and preventative care. Currently there are no guidelines specific to managing AI in CYP. There is a paucity of research in this area and we do not aim to provide an exhaustive list of evidence or evidence-based guidelines. Instead, we felt as a group that given the lack of evidence, expert discussion and clinical guidance might help dentists managing this significant and complex condition.

Therefore, the aim of this project was to develop AI management flowcharts to inform clinical decision-making for the care of CYP in the permanent dentition with the three main AI phenotypes (hypoplastic, hypocalcified, hypomaturation) focusing primarily on the permanent dentition. The flowcharts were developed with input from the membership of the UK Amelogenesis Imperfecta/Dentinogenesis Imperfecta (AI/DI) National Clinical Excellence Network (CEN). This is a network of specialist paediatric and trainee paediatric dentists with an interest in AI and DI (Monteiro et al., 2024).

Materials and Methods

Scope: Develop flowcharts that guide dentists towards suitable treatment options for children in the permanent dentition with the three main types of AI.
Target Audience: Dentists with an interest in Paediatric Dentistry (both specialised and non-specialised).
Flowchart development
In April 2022, a panel of AI/DI CEN members (Paediatric Dentists), specialists in Orthodontics, Restorative Dentistry and Oral Medicine met to discuss developing flowcharts for management of children with the three main phenotypes of AI. The group established key treatment aims for the anterior and posterior dentitions of each phenotype, which are summarised in Table 2. Premolars were classed as posterior teeth, however, if they are in the aesthetic zone and their appearance compromised, the options for the anterior dentition should be considered.

Discussions of cases representing the main types of AI were held at the national AI/DI CEN study day in May 2022. This meeting was held in person and included postgraduate students, trainees as well as specialists/consultants in paediatric dentistry, restorative dentistry and orthodontics with expertise in treating children with AI. This meeting was the "brainstorming" phase of this project. Overarching goals and management themes were identified for each phenotype, focusing on the permanent dentition, with group members suggesting and discussing the main lines for development. Following this meeting, preliminary flowcharts were created.

In November 2023, an online meeting was held to discuss the flowcharts. This meeting was attended by AI/DI CEN members from across the globe, including paediatric dentistry specialists and consultants with experience in treating children with AI. The flowcharts were presented with cases (clinical photographs and radiographs) for each of the three AI phenotypes in small groups, to encourage discussion. Participants were asked to use the flowcharts to develop treatment plans and to discuss their use in breakout rooms. This was followed by presentation of findings/treatment plans and discussion with the wider group. The flowcharts underwent further refinement before being circulated among the AI/DI CEN membership for peer review and final ratification.

Results and Discussion

Before discussing the individual flowcharts, it is important to consider undertaking the following health histories and patient assessments to inform diagnosis and influence treatment planning.

- Radiographs provide crucial information for both diagnosis and treatment planning, such as the quantity of enamel or the presence of other anomalies. They also help establish a baseline for future monitoring.
- Pedigree charts to determine the family history of AI.
- Genetic testing, if available, may provide a definitive molecular diagnosis for AI with associated opportunities for genetic counselling.
- Photographs are vital in monitoring the dentition and aid treatment planning.
- Patient-reported outcome measures (PROMS), such as the AI specific tool (Lyne et al., 2021), can deliver insight into a patient's priorities, which may differ from the clinical aims of treatment.

Overarching Principles of the Flowcharts
The flowcharts can be used individually; however, it is important to consider the principles that underlie them.

1) Given the significant variation between phenotypes in the three main types of AI, the flowcharts are a guide highlighting key considerations and management options relevant to each type of phenotype and not a comprehensive list for each form.

a. However, the authors recognise that severe hypomaturation AI phenotypes may present with significant enamel loss and hypersensitivity, often requiring management options similar to those used for children with hypocalcified AI phenotypes. Furthermore, hypocalcified and hypomaturation AI types are increasingly considered to represent a spectrum of hypomineralisation severity which can complicate the selection of appropriate management pathways or diagnostic flowcharts for these cases.
b. Consequently, it is important to recognise that these flowcharts are broad in scope and intended as a general guide. Clinicians must assess the specific features of the presenting phenotype and modify the flowcharts accordingly to support individualised management.
c. It is important to recognise that in clinical practice, AI phenotypes do not always present in isolation. Mixed or overlapping features are common, which can complicate diagnosis and management. In such cases, clinicians must apply sound clinical judgement to identify the individual needs of each patient and adapt treatment plans accordingly. This may involve drawing on elements from more than one flowchart to develop a personalised management strategy that addresses the specific clinical presentation. Flexibility and thoughtful interpretation of the guidance are therefore essential to ensure effective and appropriate care.

2) In some instances, preventing further deterioration of the dentition (due to the AI) and monitoring alone are appropriate – see Table 3. UK national prevention guidelines are outlined by the Department of Health (2021). Under these guidelines children with AI are considered at high risk of caries development and should be under an enhanced preventive regimen.
3) The flowcharts aim to guide dentists towards suitable options under either local anaesthetic and/or sedation, not for treatment planning under general anaesthesia.
4) Although the flowcharts are divided into anterior and posterior dentitions, they can be used flexibly.
5) The flowcharts are designed for the permanent dentition and can be used from the early mixed dentition up to transition to adult dental services.
6) After an intervention, review and reassessment is recommended, by returning to the start of the flowchart.

General Considerations

Oral hygiene and sensitivity

Suboptimal oral hygiene is often a barrier to providing successful restorative care in CYP with AI. Hypersensitivity may prevent teeth from being brushed effectively and is a key treatment consideration across all three phenotypes. Accordingly, each flowchart recommends that sensitivity is addressed at the outset and throughout treatment. Significant calculus deposits are frequently observed in AI cases, more commonly in hypomineralised AI types, but particularly in hypocalcified AI. (Quandalle at al., 2020). The following can help manage hypersensitivity in children with enamel defects:

- Applying warm water to the toothbrush prior to brushing.
- Regular rinsing with fluoride mouthwash (Yates et al., 2004).
- Desensitising agents such as Tooth Mousse (10% CPP-ACP; GC Corp., Japan) (Pasini et al., 2018); and/or arginine containing toothpastes (Bekes et al., 2017).
- Local anaesthesia for scaling. For gingivae that are inflamed, hyperaemic and/or hyperplastic, consider using adjuncts such as chlorohexidine mouthwash for a limited time prior to scaling to help manage bleeding. This should be undertaken before commencing any restorative care.

- Provision of restorations in cases where hypersensitivity relates to reduced enamel thickness or post-eruptive breakdown.

Bonding
In hypomineralised AI, resin bonding is compromised due to the reduced mineral content and retention of organic matter. The use of a 5% sodium hypochlorite pre-treatment application has been suggested to enhance resin bonding in hypocalcified AI, but the evidence for this is currently weak (Venezie et al., 1994). This should be completed with rubber dam isolation.

Masking discolouration with composite placement
A non-bonded resin composite trial can be used to demonstrate what can be achieved and manage expectations. Restoration involving layers of resin composites with different translucencies (including an opaquer) may help mask the discolouration.

Interceptive extractions
Interceptive extractions may be necessary for teeth with poor prognosis. However, it is vital to be mindful that, unlike children with molar-hypomineralisation (MIH) or caries lesions, the unerupted teeth in AI will also be affected and may have an equally guarded prognosis. Consequently, interceptive extractions should only be considered on an individual basis, and, if necessary, in consultation with an orthodontist. There may be some benefit in retaining roots of teeth without apical pathology (root burial) to maintain bone and facilitate future tooth replacement options.

Anomalies
A range of dental anomalies are associated with AI and may add to the challenges of managing children with AI, including:

- Taurodontism
- Pulpal calcification
- Anterior open bite
- Failed/delayed eruption
- Tooth impaction
- Root dilaceration
- Idiopathic crown resorption

A multidisciplinary approach is recommended with orthodontic and restorative input often required. Orthodontic care is not within the scope of this paper. Dentists should be aware of the high prevalence of anterior open bite, delayed/failed eruption and difficulties in resin bonding which all may increase difficulty of orthodontic management. Liaison with local orthodontic teams may be available in these cases. Timing for transition to adult restorative care will be guided by provision and availability of local services.

Managing expectations
The psychological impact of AI should not be underestimated. An empathetic approach is paramount in building rapport with CYP and their families. Non-pharmacological and pharmacological behaviour management techniques, including general anaesthetic, may be necessary in managing these children. In some cases, professional psychological support may be required. If this is not currently locally available, then consideration should be given for development of a business case for provision of dental psychology support for affected individuals, to enhance the quality of the service provided. It is crucial that patients and parents have an appreciation of the intensive treatment burden from the outset (Lafferty et al., 2021). A frank discussion regarding time commitment, potential financial implications and maintenance regimens can help prevent treatment burnout. Organising treatment into manageable blocks with realistic goals can also help sustain adherence to treatment. Given the

genetic nature of AI, parents may have personally experienced the demands of treatment and acknowledging this can gain insight into their expectations.

The flowcharts were developed to aid dental care professionals in managing children with AI. Each flowchart is described in detail below with clinical cases for each type.

Hypoplastic AI

The flowchart for Hypoplastic AI is displayed in figure 1. Figure 2 shows a patient in the mixed dentition with reduced enamel quantity on all teeth (small yellow teeth), gingival enlargement and increased overbite. The main concerns are likely to be aesthetic (size/colour), whereas the main goal posteriorly is likely to be to maintain tooth tissue and vertical dimension. The flowchart can be started in posterior or anterior regions, according to the main clinical and patient's concerns. The following section will work through the flowchart for treatment planning.

Anterior dentition

Colour:

If appropriate, microabrasion or tooth whitening with 10% carbamide peroxide should be considered as a first line of treatment (consideration should be taken regarding the European Council Directive 2011/84/EU, 2011). The alternative option may be to mask more prominent discolorations with resin composite restorations, which may also help protect against hypersensitivity. Alternating the use of the bleaching product with desensitising agents in the trays or reducing the bleaching periods may be necessary to reduce hypersensitivity.

Crown Size and Shape:

Teeth with unfavourable crown-root ratios have an added layer of complexity and consequently liaising with restorative and/or orthodontic colleagues prior to intervention is necessary. In situations where the crown-root ratio is favourable, and the clinical crown height is sufficient, resin composite restorations may be indicated. Preparation for restorations should be avoided to preserve tooth structure. Direct options such as cellulose crown forms and the use of putty indices may be helpful for placement of more extensive restorations. Indirect restorations may be helpful in cases of more significant enamel loss. However, as gingival maturation is not complete in younger patients, indirect restorations prior to maturation may lead to aesthetic and sensitivity concerns. Gingivectomy can be considered in older patients, should the clinical crown height be inadequate for restoration. This must be carried out with care, being mindful of the biological width of the tooth.

Texture:

In phenotypes where pits and ridges are prominent, direct resin composite veneers can create smoother tooth surfaces, aiding aesthetics and oral hygiene.

Posterior dentition

The chief concern is the presence of vulnerable enamel. Fissure sealants are recommended, with flowable resin composite being considered in sites with markedly reduced enamel thickness. Direct resin composite restorations are advisable in the absence of pulpal involvement. For multi-surface and cuspal defects, indirect cuspal coverage restorations such as onlays or crowns should be considered. Resin composite and ceramic materials are increasingly being used for indirect restorations in AI cases aided by digital scanning which enables both printing or milling of such restorations (Lundgren et al., 2018). Preformed metal crowns (PMCs) are advised as an interim measure only. Prefabricated zirconia crowns are also now available in both the primary and permanent dentition and offer an alternative to resin composite restorations and PMCs, although preparation of teeth is required for Zirconia crowns.

Hypocalcified AI

The flowchart for Hypocalcified AI is displayed in figure 3. Figure 4 demonstrates minimal enamel on the posterior teeth. Starting with the posterior dentition on the flowchart, there are concerns regarding vulnerable enamel, and cuspal protection is needed. This leads to consideration of the option of PMCs as a cuspal coverage restoration, given that the patient is in the mixed dentition. Due to the reduced mineral density in hypocalcified AI, managing post-eruptive breakdown (PEB) and hypersensitivity are major considerations for both anterior and posterior teeth. The flowchart is divided into anterior and posterior dentitions.

Anterior dentition

Similar to hypoplastic AI, considerations should be made regarding PEB and colour. In the mixed dentition, the restorative choice for anterior teeth are direct resin composite restorations, which can easily be refined as the gingival margin matures. Cellulose crown forms may be useful, especially in cases with significant PEB. If the underlying tooth structure is discoloured, opaquers may help mask the discolouration. Indirect restorations are suitable for fully erupted teeth and/or extensive restorations or when bonding fails.

Colour:

Figure 4 highlights the yellow-brownish discolouration commonly related to hypocalcified AI. If there are no concerns regarding sensitivity and it is available, tooth whitening may be effective in improving aesthetics in this case. In cases where bleaching is not possible or of limited effect, restorations can be used to mask discolouration. Alternating the use of the bleaching product with desensitising agents in the trays or reducing the bleaching periods may be necessary to reduce hypersensitivity. It is worth noting that microabrasion is not recommended in hypocalcified AI types to prevent further inadvertent tooth tissue loss due to the 'softness' of the enamel.

Posterior dentition

If no PEB is present, caries preventive measures, such as resin sealants or flowable resin composite, should be provided. If PEB is limited, direct resin composite restorations can be used, as illustrated in figure 4. As in anterior teeth, indirect restorations are indicated for extensive defects. In the mixed dentition PMCs may be appropriate interim solutions in maintaining the occlusal vertical dimension. Prefabricated zirconia crowns are an alternative, although they require tooth preparation. In the permanent dentition they should be replaced with indirect cast restorations once patient co-operation allows. When deciding between onlays and full coverage crowns, the integrity of the cervical margin is key. If PEB is limited to the occlusal surfaces and cusp tips, cast (or milled) onlays are suitable in preserving intact tooth structure, assuming the margins are bonded to sound enamel. Full-coverage restorations are indicated if the cervical margin is compromised, and they provide further resistance to occlusal loading.

Hypomaturation AI

The flowchart for Hypomaturation AI is displayed in figure 5. Figure 6 shows disrupted enamel quality with yellow and white diffuse opacities and areas of enamel breakdown in the premolar regions. Both hypocalcified and hypomature AI are hypomineralised forms of enamel with opacities and marked enamel loss can be observed in both phenotypes The main issue in figure 6 is likely to be anterior aesthetics and managing the posterior breakdown. If PEB is present anteriorly in hypomature phenotypes, it should be managed accordingly (see anterior region of hypocalcified flowchart, which considers anterior PEB). Hypersensitivity can also be a challenge in hypomature AI phenotypes, even in cases of full thickness enamel (due to poor mineralisation) and should be managed appropriately if present (review above advice on managing hypersensitivity in children with enamel defects). The flowchart can be started in posterior or anterior regions, according to the main clinical and patient's concerns. The following section will work through the flowchart for treatment planning.

Anterior dentition

Colour:

If discolouration is predominantly yellow/brown, microabrasion can be effective in removing superficial 'staining' (figure 6). For more persistent discolouration, tooth whitening (if available) may be successful. Resin infiltration systems, such as ICON (DMG, Hamburg, Germany), can be used to treat more penetrating white opacities (the evidence for this is based on milder cases of hypomature enamel without marked enamel breakdown (Cagetti et al., 2017)). Evidence from *in vitro* studies, however, suggest that the resin may limit the uptake of hydrogen peroxide and consequently inhibit the effectiveness of any future bleaching (Rocha et al., 2020; Santos et al., 2020). Direct resin composite restorations are the final option.

Post-eruptive breakdown:

In cases with PEB, replacement of the lost tooth structure using resin composite is recommended.

Posterior dentition

The primary consideration in these cases is the management of vulnerable enamel, which may be plaque-retentive or at risk of fracture due to PEB. The material of choice for sealants may be resin or flowable composite. Concerns around PEB in hypomaturation AI, may be managed as per hypocalcified AI.

Conclusion

The aim of these flowcharts is to aid the dental management of CYP with AI in the permanent dentition. The flowcharts provide a structured approach to addressing key issues associated with each type of AI. If appropriate, conservative treatment options should be considered first, progressing to more invasive treatment if the phenotype severity necessitates it. It must be emphasised that the flowcharts are to be used as guidance only and the underpinning evidence base is weak. AI is a complex condition with multiple phenotypes of varying severities and consequently treatment should be tailored to individual cases. Finally, a shared decision-making approach with the child or young person and their parent is paramount.

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Tables & Figures

All tables created with Microsoft Word® (Microsoft Corporation, Washington, United States)
All figures created with Microsoft PowerPoint® (Microsoft Corporation, Washington, United States)

Table 1 The three most typical forms of AI and the key histological, clinical and radiograph features

	Hypoplastic	Hypocalcified	Hypomaturation
Enamel formation	Reduction in enamel quantity formed	Normal enamel thickness with qualitative failures of normal mineralisation due to presence of organic remnants from enamel developing cells	Normal enamel thickness with qualitative failures of normal mineralisation due to presence of organic remnants from enamel developing cells.
Clinical Appearance	• Thin enamel • Pitting and grooves • Occasionally hard and translucent enamel • Poor quality enamel may be present	• Prone to early, rapid PEB • Rapidly wear and stain • Chalky or opaque appearance	• Mottled appearance • Vulnerable to tooth wear, but not as severe as hypocalcified type
Radiographic Appearance	• Reduced thickness of enamel immediately before eruption • Variable radiographic contrast between enamel and dentine depending on the underlying genetic cause	• Normal thickness of enamel immediately before eruption • Enamel less radiopaque than dentine	• Normal thickness of enamel immediately before eruption • Enamel more radiopaque than dentine

Table 2 Key treatment aims for each type of AI.

	Hypoplastic	Hypocalcified	Hypomaturation
Anterior	Address discolouration prior to replacing missing enamel	• Improve oral hygiene • Prevent PEB • Improve appearance	Improve colour (ideally without restorations that require long term management)
Posterior	Protect the thinner enamel (sealing or restoring)	Protect from PEB and/or sensitivity	Prevent PEB and caries

N.B. Sensitivity is a symptom common to all AI types and consequently managing it is aim unanimous to all type.

Table 3 Factors to be considered for prior to treatment planning for AI cases.

Patient Level	Oral Level	Tooth Level
Age of patient	Developmental Stage	Type of AI
Medical History	Caries Risk	Severity of AI
Cooperative ability	Presence of other dental anomalies	Remaining tooth structure (quantity and quality)
Presence or Absence of Symptoms	Occlusion	Pulpal and Apical status
Current Child and Parental Priorities	Orthodontic need	
Motivation	Oral Hygiene	

Figure 1 Flowchart for hypoplastic AI

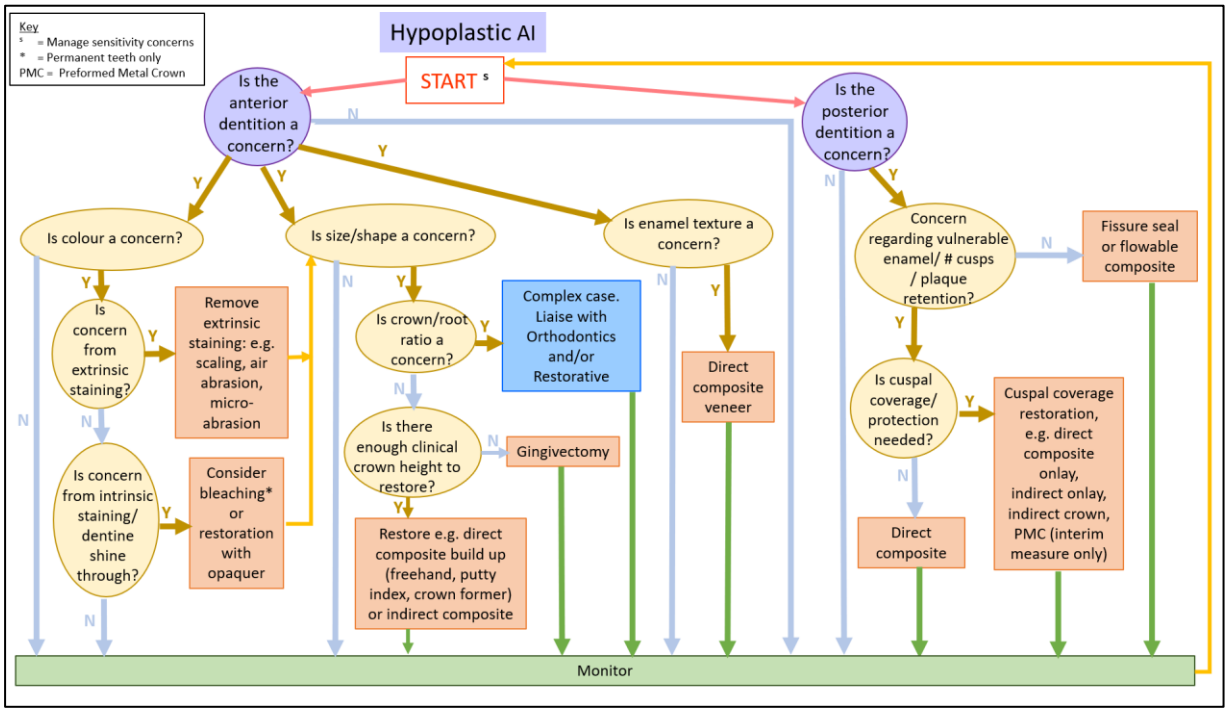
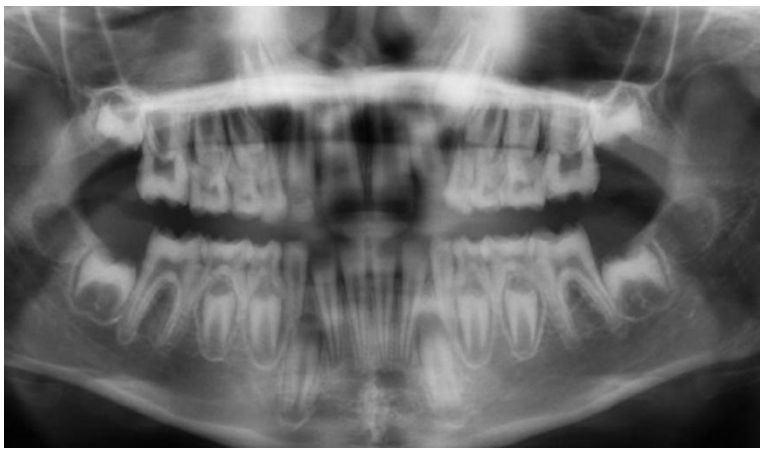


Figure 2 Intraoral photographs (2a, 2b, 2d) and orthopantomogram (2c) of a patient with hypoplastic AI.

(2a) Anterior view of hypoplastic AI

(2b) Maxillary occlusal view of hypoplastic AI



(2c) Orthopantogram of hypoplastic AI

(2d) Mandibular view of hypoplastic AI

Figure 3 Flowchart for hypocalcified AI

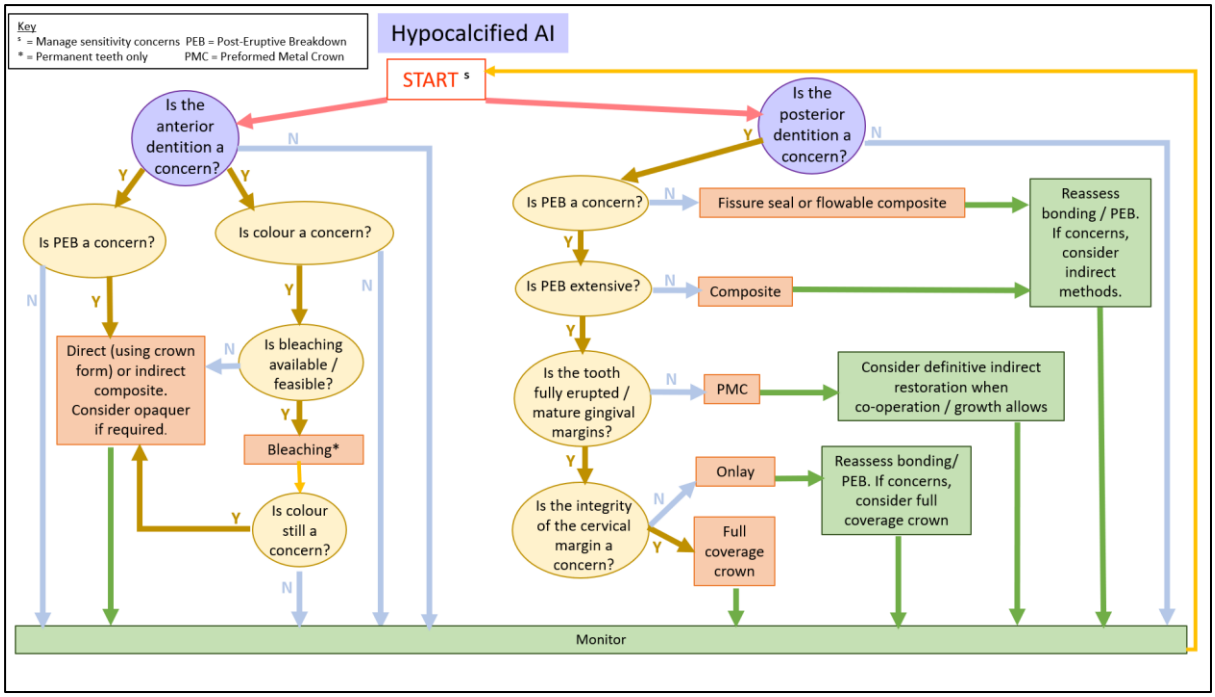
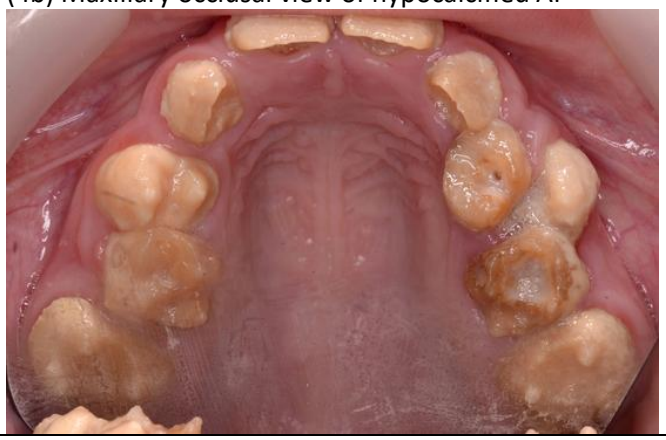


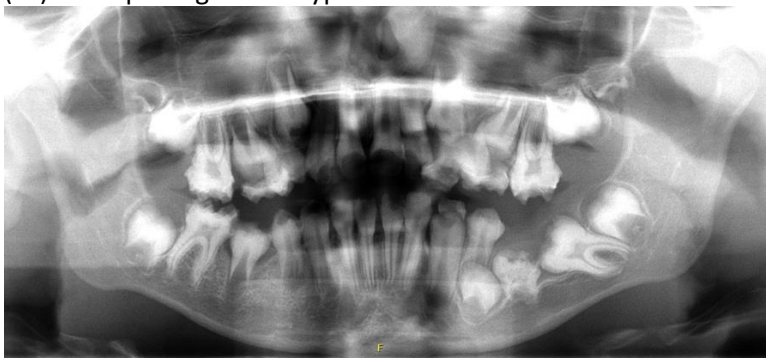
Figure 4 Intraoral photographs (4a, 4b, 4d) and orthopantogram (4c) of a patient with hypocalcified AI, demonstrating marked discolouration and failure of direct composite restorations.

(4a) Anterior view of hypocalcified AI

(4b) Maxillary occlusal view of hypocalcified AI



(4c) Orthopantogram of hypocalcified AI



(4d) Mandibular occlusal view of hypocalcified AI

Figure 5 Flowchart for hypomaturation AI.

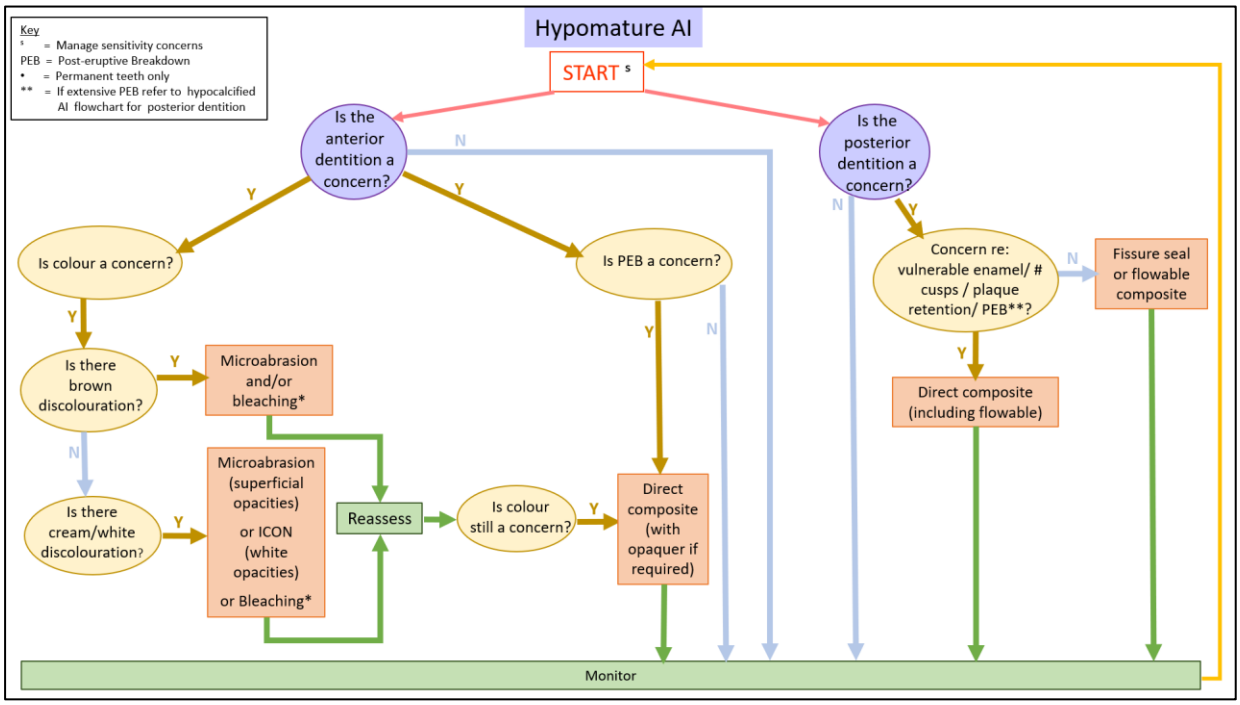
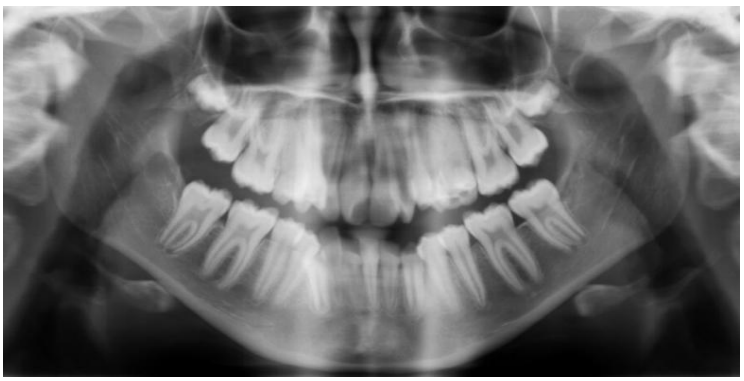


Figure 6 Intraoral photographs (6a, 6b and 6d) and orthopantomogram (6c) of a patient with hypomaturation AI illustrating white and brown/yellow opacities

(6a) Anterior view of hypomaturation AI

(6b) Maxillary occlusal view of hypomaturation AI



(6c) Orthopantogram of hypomaturation AI

(6d) Mandibular occlusal view of hypomaturation AI