# The burden of dental care in Amelogenesis Imperfecta Paediatric Patients in UK NHS: A retrospective, multi-centred analysis

## Abstract

### Purpose

The burden of dental care in Amelogenesis Imperfecta (AI) has not been well described. This condition results in weak, discoloured, and often sensitive teeth. Specialist paediatric care is available for AI patients in the UK, but treatment protocols and care provided are inconsistent.

The aim of this study was therefore to analyse the provision of treatment and burden of care for children and families with AI across four Paediatric Dentistry centres in the UK.

### Methods

A retrospective evaluation of AI patient clinical records across four UK consultant-led Paediatric Dentistry centres was completed. Frequency and duration of care were recorded along with treatment and experience of inhalation sedation, local and general anaesthetic.

### Results

In total, 138 records were available for analysis. The average patient age at first referral was 7.7 years (range 1-16 years) and families travelled an average 21.8 miles per appointment (range 0.2- 286 miles). Patients attended on average 4.5 appointments per year for 5.8 years. In total, 65.2% had experience of local anaesthetic, 27.5% inhalation sedation and 31.9% general anaesthetic. Dental treatment including restorations and extractions were commonly required on multiple teeth per patient.

### Conclusion

AI carries a high burden of specialist dental care to patients and families. Specialist centres are required to provide longitudinal, comprehensive care.

**Key Words**
Amelogenesis Imperfecta, Burden of Care, Paediatric Dentistry

## Declarations

### Funding

Not applicable

### Conflicts of interest

Not applicable

### Ethics Approval

Local governance approval was gained in each setting. No further ethical approval was required as the study in each setting was carried out by those involved with direct clinical care of the patients.

### Consent to participate

Not applicable

### Consent for publication

All authors give consent for publication

### Availability of data and material

Data available from each unit locally

### Code availability

Not available

## Introduction

Amelogenesis Imperfecta (AI) is a heterogeneous group of generalised developmental enamel defects. It is inherited in a Mendelian manner with both dentitions affected (Aldred and Crawford, 1995). The burden of dental care for AI patients has not been well described or documented. This is despite the known adverse impact the condition has on patients and their families. AI disturbs the enamel structure with associated poor appearances and premature failure. Teeth can be discoloured, sensitive or vulnerable to breaking down (Crawford et al. 2007). Aesthetic concerns and sensitivity have been shown to often be the primary complaints (Koruyucu et al. 2014), and therefore reducing sensitivity and improving tooth colour is of high importance to this cohort of patients (Parekh et al. 2014). Patients describe fear of pain and chipping teeth (Pousette et al. 2016). AI patients have also been shown to have higher levels of distress, social avoidance and discomfort than those without AI (Coffield et al. 2005). A negative impact on quality of life has been found in adults with AI, including areas such as psychological discomfort and disability, social disability and physical disability (Hashem et al. 2013). AI can have an impact on whether a sufferer decides to have a family of their own (Pousette et al. 2016), with some concerned about passing the condition on to their children (Aldred et al. 2003 and Pousette et al. 2019). Understanding how care can be improved for these patients is therefore essential.

AI has a wide spectrum of severity with related clinical challenges. These challenges include: poor oral hygiene and calculus build-up, sensitivity, malocclusions such as open bites, eruption disturbance, periodontal problems and dental anomalies such as taurodontism, resorption and pulp stones (Poulsen et al. 2008). Treatment modalities for AI vary, with many different management strategies described. One systematic review has documented a wide range of treatment approaches but concluded that ultimately the evidence base for any interventions was weak (Dashash et al. 2013). Early intervention has been recommended to prevent further breakdown of the dentition and often requires a multi-disciplinary approach (Ayers et al. 2004). Prompt diagnosis along with treatment and preventative management at the correct stage is acknowledged as important if oral health is to be improved (Markovic et al. 2010 and Ortiz et al. 2019). This often means multiple visits and prolonged periods of treatment with multidisciplinary input, in specialist care centres.

The clinical burden of AI in the UK is poorly understood, which in part reflects absence of accurate prevalence data. This unknown burden limits service planning and delivery. The heterogeneous nature of AI means that individuals have different healthcare needs and access to specialist care can be difficult to achieve. In the UK, specialist Paediatric Dentistry services are mainly provided in a dental hospital setting. There are some limited areas offering specialist care in the local community services and this is a mixture of both consultant and specialist led. Across the UK there is a shortfall in the specialist paediatric dentistry workforce (BSPD 2015 and SDNAP 2017) with a number of geographical areas having no or very limited access to a Paediatric Dental Specialist (Mills, 2016). Seeking appropriate local care for paediatric patients with AI may be difficult. Access and provision of care should be equitable, affording the same opportunities for all patients, this is not the case for all AI patients in the UK. Understanding the treatment needs for these patients is important, and would help inform national treatment protocols and provide evidence for the need to expand specialist care services.

The aim of this study was to analyse the provision of treatment and burden of care for children and families with AI across four Paediatric Dentistry specialist care centres in the UK.

## Materials and methods

### Study design

A retrospective study of patient AI clinical records was carried out across four consultant-led Paediatric Dentistry centres in the UK between 2017 and 2019. Two of the care centres were hospital based: Leeds Dental Institute (LDI) and the Eastman Dental Hospital (EDH). The other two were based in the community dental service: North Yorkshire Community Dental Service (NYCDS) and Community Dental Service, Bradford District Care NHS Foundation Trust (BCDS). These centres all provide specialist care and combining the different settings was deemed to be beneficial to capture the diverse specialist workforce currently utilised in the UK.

In the EDH, NYCDS and BCDS all patients identified with AI were included in the study. In the LDI a randomly selected cohort from the AI database was included to match the same number of patients in the NYCDS. This was due to these two centres (NYCDS/ LDI) forming the original pilot study for this analysis and an aim initially to have the same number of patients to compare between them both.

### Ethical Approval

Local governance approval was gained in each setting. No further ethical approval was required as the study in each setting was carried out by those involved with direct clinical care of the patients.

### Patient selection

Patients were selected from the local AI/anomalies databases available in each centre. These included discharged and current patients. Please see Figure 1 for the selection process.

The inclusion criteria included:

- A diagnosis of AI documented in the patient notes
- Patient cared for primarily by the care centre analysing the data. In some cases, patients may have been treated in community and by the dental hospital specialist team if for example General Anaesthetic was not available in one setting.
- At least six months of records available for analysis as this would allow a new patient assessment and review period.

The exclusion criteria included:

- If a diagnosis of AI was unclear
- If records of the patient were not available
- Less than six months of records were available
- If it had been more than 20 years since paediatric dental treatment provision

## Data collation

A retrospective evaluation of AI patient computerised and paper records was completed in each setting and recorded on Excel spreadsheets for analysis. The data was then merged into one datasheet by the principal investigator (FL) for whole data analyses. Data analysis for each centre involved at least two of the authors involved in the primary care of these patients and familiar with the record keeping for these centres. Final analysis by the principal investigator allowed any further clarifications to be requested from these clinicians and standardised the data.

The data gathered included a wide range of information. This included:

- Patient demographics;
- Patient postcode (to calculate distance travelled);
- Type(s) of AI, as recorded in the notes;
- Details of appointments including average number of appointments attended each year and, for the discharged cohort, how many years of specialist care they received.

A detailed analysis of treatment was completed by working through each documented appointment for every patient. Experience of different treatment modalities was recorded in terms of local anaesthetic experience, inhalation sedation and general anaesthetic. The number of primary and permanent teeth undergoing individual treatments for each patient was also recorded.

## Statistical Analyses

Descriptive statistics were undertaken of the combined data including means and standard deviations. Results were entered onto a Microsoft Excel spreadsheet to aid with analysis.

## Results

### Demographics

The total number of patient records included in this study was 138 across all four clinical settings (Figure 1). The oldest record dated from 1994 and the most recent was 2019.This included 45.7% (n= 63) male and 54.3% (n= 75) female patients. The average age at first appointment at the paediatric specialist care settings (age at referral for BCDS) was 7.7 years (± 3.3). This ranged from 1 to 16 years old. The AI types as described in the notes, included many commonly used phenotypes (Table 1), and demonstrated the diverse nature of the condition.

### Appointments

More than half of the patients included in this study were currently under care: 58% (n= 80), with 42% (n=52) no longer under specialist care in any of the four settings. Families and patients were on average travelling 21.8 miles (± 24.9 miles) as a round trip from home to the specialist centre, with a range from 0.2 miles to 286 miles. The average varied considerably between centres, with BCDS the lowest with an average of 4.2 miles per round trip (± 2.9). The LDI patients travelled on average 22.9 miles (± 12.6), NYCDS patients 20.3 (± 24.1) and the EDH 67.6 miles (± 59.6 miles). The average number of appointments a year per patient was 4.5 (± 2.2). Including analysis of discharged and lost to follow-up patients (n=58), an average of 5.8 years specialist care was provided (± 4.5).

### Treatment

Anaesthesia modalities were varied and included local anaesthetic (LA), inhalation sedation (IHS) and general anaesthetic (GA). The majority (65.2%) of patients had experience of LA, with a further 27.5% and 31.9% having IHS and GA experience respectively. The repeat GA rate for those patients who had had GA at the time of analysis was 22.7%. The treatment modality experience can be seen in Table 2 and is the experience of both the current, discharged and lost to follow-up patients.

Patients required a number of different types of treatment and treatment modalities (Tables 3 and 4). These results show the average number of teeth per patient undergoing each treatment type. In the primary dentition, use of composite to restore teeth was low. Preformed metal crowns (PMCs) and extractions were the preferred treatment options with wide standard deviations of 2.9 and 3.4, respectively. In the permanent dentition, anterior composite restorations were the most common treatment provided, with both PMCs and composites used to restore posterior teeth. Other treatment modalities such as microabrasion or bleaching were used in a small number of cases.

Figure 2 demonstrates two patient journey examples of their longitudinal specialist care with many treatment modalities and appointments required.

## Discussion

This study provides evidence of the high burden of dental care for paediatric patients with AI. The most direct measure of this is the number of required dental appointments, with an average of 4.5 appointments a year, a number consistent with reports of similar cohorts in the literature (Pousette et al. 2016). One way of easing the burden of care is for ready access to specialist units. Our study demonstrated this was currently extremely variable with families travelling between 0.2 to 286 miles to access their care and reflects the documented inequality of geographical location of specialist units in the UK (Mills et al 2016). This disparity may also have been responsible for the relatively late presentation of many of the cases whose first contact with paediatric dentist was in the mixed dentition

The extensive variety of dental treatment recorded in our study was similar to those described previously in the literature, and likely reflected both the range of phenotype and the lack of good evidence base for specific interventions (Dashash et al. 2013). In the primary dentition the use of PMCs to restore posterior teeth has been well documented (Crawford et al. 2007). In the mixed and permanent dentition, there are many techniques and materials available with no consensus as to which is most appropriate. For example, cast adhesive copings are an option for ‘defective’ molars instead of PMCs, however more appointments are required, and the material cost is greater (Zagdwon et al. 2003). Both onlays and PMCs were used on permanent teeth in this study.

The most significant concern for many children with AI is the aesthetics (Rodd et al. 2011, Craig et al. 2015). In our study this was mostly addressed through the use of composite restorations in spite of known limitations. These restorations require commitment and long-term maintenance as they often need to be added to cervically given the gingival margin position changes during patient growth (Crawford et al. 2007). An investigation into long-term care of AI patients compared to a control group found the longevity of composite restorations was significantly lower in the AI cohort with a failure rate 2.5 times greater (Pousette et al. 2016).

Most patients in this study (73.9%) had experienced GA or LA for dental treatment. This figure can be examined alongside caries rates in England as a way of comparing treatment need with a non-AI patient group. Public Health England stated in their updated Child Oral Health Guidance, February 2018, almost a quarter of five-year olds had tooth decay (PHE, 2020). Although a crude comparison, it can be concluded from these figures that having AI does mean a higher treatment need than the general population where caries rates fall well below 73.9%. The well documented psychosocial impact of AI (Coffield et al. 2005 and Hashem et al. 2013), is likely further compounded by the multiple dental interventions recorded in this study. A history of extractions has been linked with dental anxiety in children (Tickle et al. 2009), and it is known that the dental setting is anxiety provoking as described by children themselves (Morgan et al. 2017). Inhalation sedation had been experienced by 27.5% of patients in this study, with dental anxiety a common reason for this modality to be chosen (Alexopoulos et al. 2007).

Of particular note regarding the added burden of care is the high number of patients who required GA (31.9%) for their dental treatment. This may reflect a proportion of the patients who are very young or anxious or it may be due to these patients requiring extensive dental treatment. Further significance can be seen with a repeat GA rate of 22.7%. This is far higher than rates quoted in the literature for caries e.g. 8.9% repeat GA rate has been found for child patients with 84% of the cases involving caries (Kakaounaki et al. 2011). This is further evidence of a high burden of care throughout childhood with further interventions required as new teeth erupt and previous restorations fail. Comprehensive treatment planning is an important aspect of paediatric dentistry specialist care which includes GA provision. In line with UK guidelines GA treatment planning should be carefully considered in order to avoid the need for a repeat GA whenever possible (Davies et al. 2008). Unfortunately, in a number of children with AI repeat GA may be unavoidable and that possibility should be incorporated into their overall planning and informed consent.

Given the rarity of the condition the authors chose a wide time period (6 months to twenty years) in order to capture as many patients as possible. Unfortunately, management of AI has not advanced significantly in this time period (Dashash et al. 2013). Whilst only including children with completed treatment may have provided a more consistent data set, unfortunately the study demonstrated that few children with AI ever have their treatment completed in the usual sense. Indeed, it can be argued that children with Amelogenesis Imperfecta should not be discharged but should remain under paediatric specialist care throughout childhood with transition to specialist adult services at the appropriate age.

This study is a comprehensive review of children requiring consultant-led paediatric dentistry treatment for AI. It included a large number of patients across 4 different sites in the north and south of England. Review of the notes was thorough in all clinical settings. The different elements examined allow a theme to build of what exactly these young patients are faced with and also provided evidence as to what specialist services are providing. This can be considered in terms of personnel required to treat these patients but also facilities needed e.g. access to sedation, lab facilities and theatres. These services also need to be accessible to families in order for treatment to be sought easily and lessen impact on other aspects of their life such as time off work and school. Some patients may have benefitted from an earlier assessment, before post-eruptive changes occur to their teeth, and so clearer access pathways for specialist care is needed.

The main limitation of this study was the number of descriptions that were used in the clinical records in an attempt to define and classify the AI type. These descriptions were extremely inconsistent between units, within units and even within the same patient records. For example, as illustrated in Table 1, the AI type for two patients was described as ‘part of a syndrome,’ a description which is insufficient as a diagnosis. This was in spite of the fact that patients were being seen in tertiary centres by clinicians familiar with managing AI. This inconsistency would have made any attempt to analyse provision of care by AI type redundant and the authors have therefore not attempted to do so. It also highlights the difficulty in classifying AI type by clinical phenotype. Accurate diagnosis is, however, critical in determining management and prognosis. The evolving area of genetic testing for AI will improve classification, inform development of treatment choices and contribute to service development (McDowall et al. 2018).

Further limitations to this study include the heterogenous nature of this data does make it difficult to analyse and interpret, as there are different elements of the results that could be viewed from numerous perspectives. Identification of the patients to include in the study was through access to databases in each unit and assumed this was an accurate method. However, there may be patients who had been missed from these databases and therefore not included in the study. Descriptions of treatment recorded and appearance of teeth including the diagnosis, was taken as the exact wording recorded in the notes by the treating clinicians. As such the severity and type of AI may not have been fully captured through this method.

Where it was unclear regarding the precise treatment, a consensus opinion was reached amongst the authors.

As presented in this paper, AI carries a high burden of care for both patients and care providers with further strategies to lessen the burden of care requiring careful consideration. The aim for paediatric dental patients with AI is to take them through to adulthood remaining motivated, with good oral hygiene practices and with future treatment options not compromised by previous dental work (Poulsen et al. 2008). More accessible care pathways for patients to receive appropriate timely care is important. This should be alongside continued workforce development for specialists in paediatric dentistry in appropriate, well-equipped care centres. Establishment in 2019 of a Clinical Excellence Network for Amelogenesis Imperfecta Paediatric patients in the UK has aided collaboration between specialist units. Development of national guidelines would also ensure equity of care across the nation and improve the service and treatment provided to these patients.

## Conclusion

- Amelogenesis Imperfecta carries a high burden of care with patients requiring consultant- led Paediatric Dentistry input frequently and for a significant period throughout their childhood.

- Access to local anaesthetic, inhalation sedation and general anaesthetic facilities is required to facilitate the delivery of comprehensive dental care.

- High treatment need is common, and this should be considered when planning care for these patients. Access to local specialist care can lessen the burden and national clinical guidelines would be of benefit.

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**Tables**

Table 1: AI types as recorded in the clinical notes

| **Types of AI** | |
|---|---|
| **AI phenotype as described in clinical notes** | **% of patients (Number of patients)** |
| Hypoplastic | **32.6% (45)** |
| Hypocalcified | **9.4% (13)** |
| Hypomaturation | **26% (36)** |
| Hypomineralised | **9.4% (13)** |
| Mixed | **12.3% (17)** |
| Unknown | **8.7% (12)** |
| As part of a syndrome | **1.4% (2)** |

Table 2: Anaesthesia modality experience in children with Amelogenesis Imperfecta

| **Anaesthesia modality experience** | |
|---|---|
| **Type of treatment** | **% of patients (Number of patients)** |
| No LA or GA | **26.1% (36)** |
| LA | **65.2% (90)** |
| Inhalation Sedation | **27.5% (38)** |
| GA | **31.9% (44)** |
| Repeat GA | **7.2% (10)** |

NB; a number of patients had treatment using various means and therefore the total does not add up to total number of patients

Table 3: Primary teeth treatment

| Primary teeth treatment | |
|---|---|
| **Type of treatment** | **Average no. of teeth/patient (SD)** |
| Posterior composite restoration | **0.1 (0.4)** |
| Anterior composite restoration (C-C) | **0.5 (1.5)** |
| Preformed metal crown | **1.7 (2.9)** |
| Exodontia | **1.9 (3.4)** |

Table 4: Permanent teeth treatment

| Permanent teeth treatment | |
|---|---|
| **Type of treatment** | **Average no. of teeth/patient (SD)** |
| Posterior composite restoration | **1.9 (3.1)** |
| Anterior composite restoration (3-3) | **4.1 (5.2)** |
| Preformed metal crown | **1.2 (2.3)** |
| Onlay | **0.3 (1.6)** |
| Exodontia | **0.7 (1.5)** |
| Microabrasion (Episodes/patient) | **0.1 (0.5)** |
| Bleaching | **0.2 (0.6)** |

## Figures

Figure 1: Number of patient records included in the study

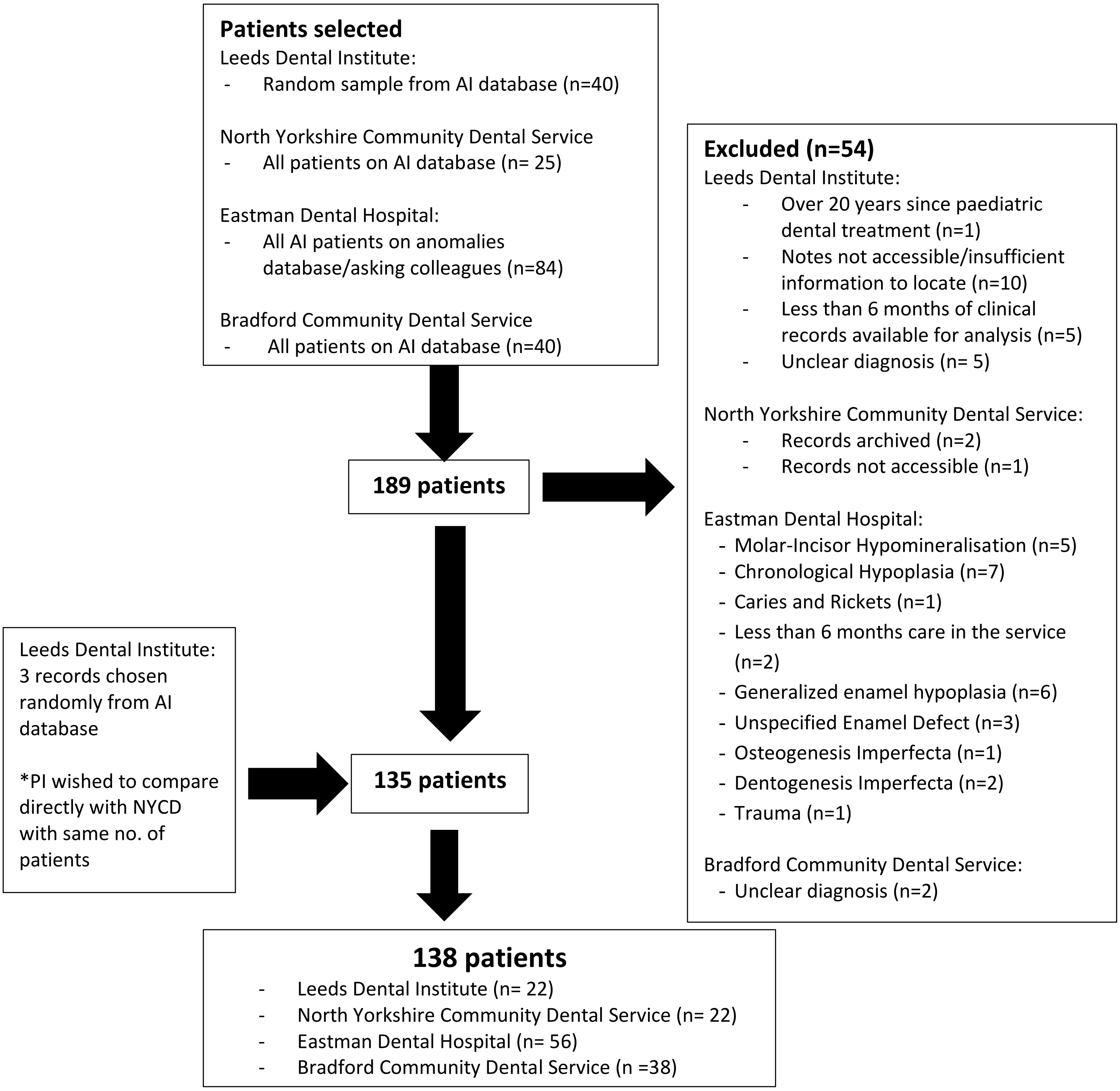
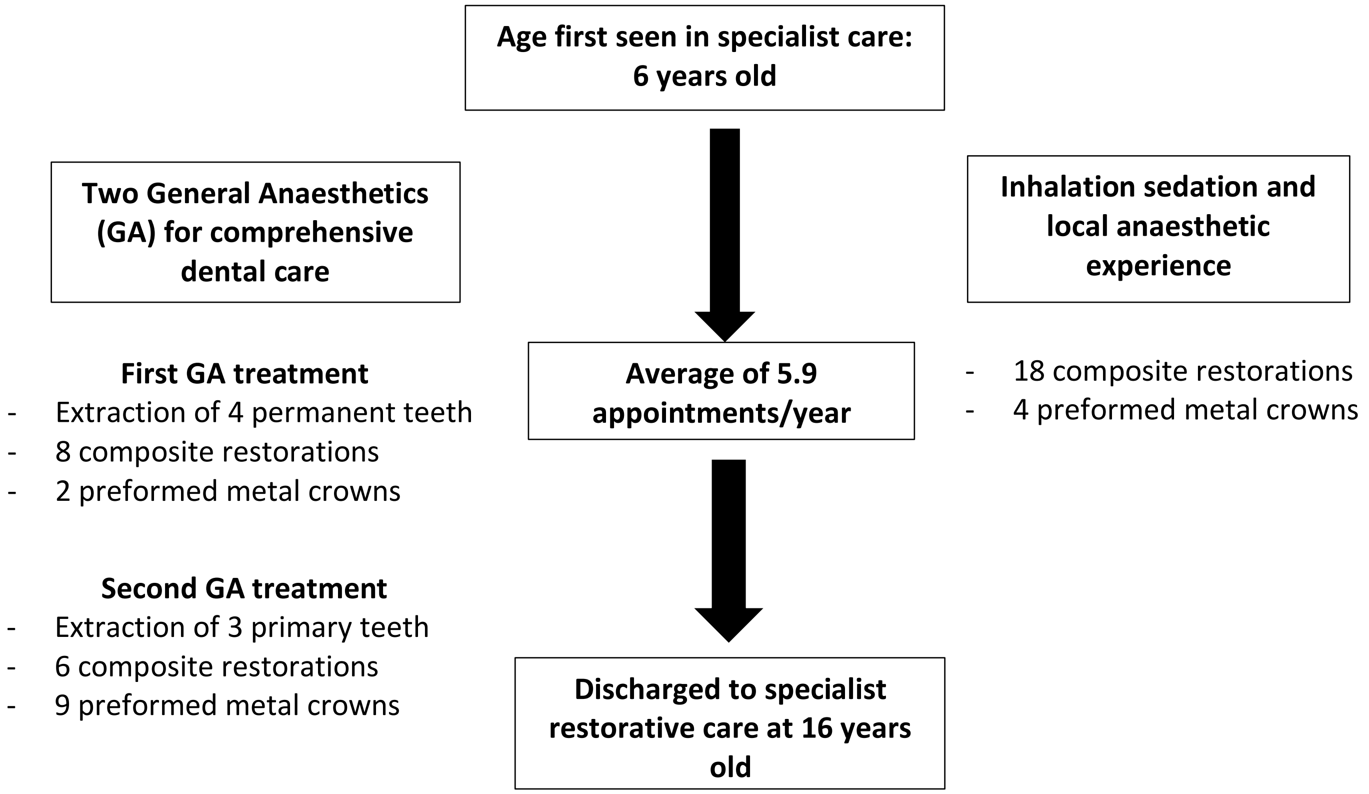


Figure 2: Example patient journeys

Patient A (NYCDS)



Patient B (EDI)