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Title

Patient reported outcome measure for children and young people with Amelogenesis Imperfecta

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Abstract

Background. Amelogenesis imperfecta (AI) is a genetic enamel defect that can affect both the primary and permanent dentition. It has a range of clinical phenotypes, and children and young people often present with challenging oral health needs. Patient reported outcome measures (PROMs) can identify key patient concerns.

Methods. This was a multi-centre service evaluation across several specialist Paediatric Dentistry services in the UK. A PROM questionnaire was created with clinician and patient input, through peer review with the national AI Clinical Excellence Network, as well as piloting the PROM with 10 children and young people with AI. The final PROM questionnaire was distributed to all patients with AI attending each unit between January and March 2020.

Results. Sixty children and young people (ages 5 to 17 years) across four specialist units participated, with 72% reporting that they 'often' or 'sometimes' experienced pain or sensitivity, 76% reported that they 'often' or 'sometimes' felt unhappy with the way their teeth look. Of the patients who were post-treatment, 81% indicated that they were happy with their teeth, compared to just 41% of patients who were mid treatment, and 33% of patients who were pre-treatment.

Conclusion. Children and young people with AI experience a range of issues related to their function and psychosocial wellbeing. This simple PROM demonstrates the range of issues this group of patients face; and could be used to monitor an individual's progress to ensure that treatment is planned to address the patient's individual concerns and needs.

Introduction

Amelogenesis imperfecta

Amelogenesis imperfecta (AI) is an uncommon genetic condition characterised by defective enamel formation, with reported prevalences of between 1 in 200 to 1 in 8,000.¹ Several genes associated with AI have been identified, with varying inheritance patterns and clinical presentations, at present diagnosis is often based on the clinical phenotype.^{1,2} The phenotype classification¹ is shown in Figure 1, along with some typical clinical presentations. AI affects all teeth in the primary and permanent dentitions, although the clinical presentation can vary between dentitions and teeth, with the primary dentition usually more mildly affected.³

This enamel defect may have a significant impact on quality of life. Children with severe clinical presentations often report eating difficulties, pain, or worry about aesthetics. Many children also report being bullied or teased by their peers.⁴

Children with AI often require extensive dental treatment, attending more routine and emergency dental appointments over time than unaffected individuals.^{5,6,7} Treatment itself can be very challenging due to increased sensitivity, poor oral hygiene, compromised enamel bonding, decreased vertical dimension, delayed eruption and difficulties in providing orthodontic treatment. Children may also be more anxious about dental treatment, and delayed gingival maturation linked to delayed eruption may limit restorative options for a growing child.⁸

There are currently no U.K. guidelines for the management of AI in children and young people. Due to the wide-ranging clinical presentation in this group of patients, a personalised approach to care is needed. The American Academy of Pediatric Dentistry (AAPD) guidelines advise that *“clinicians treating children and adolescents with AI must address the clinical and emotional demands of these disorders.”*⁹ Alongside enhanced preventative care, patients may benefit from interventions to address their symptoms, function and aesthetics. Potential interventions at the different stages of development are

described in Table 1, although suitable options will depend on the individual's concerns and presentation. Treatment planning in patients with AI is complex, often requiring multi-disciplinary input, and so assessment and treatment planning in a specialist-led service may be preferable for children with AI. Adolescents and young adults with AI reported they often felt that dentists lacked knowledge about AI, or did not consider the patient's views and wishes.⁴

Patient reported outcome measures

Patient-reported outcome measures (PROMs) refer to health outcomes directly reported by the patients experiencing the condition, rather than by the clinicians treating them.¹⁰ Patients complete questionnaires related to their symptoms, condition, and quality of life. Some questions are specific to a condition, while others can be used for any diagnosis or treatment. PROM data can be used longitudinally, collected by the same patients before and after a particular procedure. The results can be used to monitor patient progress and as part of quality improvement for a health service. The key issues for patients may be different to the clinical outcomes measured by clinicians, therefore PROMs are typically developed by both health professionals with relevant expertise and patients with the condition.¹¹

The use of PROMs are encouraged within the National Health Service (NHS). For example, in 2009, NHS organisations providing four common elective procedures (hip replacement, knee replacement, varicose vein surgery, and hernia surgery), were required to provide PROM data, and the results were published nationally.¹⁰ In dentistry, commissioning guidelines encourage the use of PROMs at a local and national level,¹¹ but there is no requirement or national body to collect dental PROM data.

There are currently few published PROMs in dental services, and no existing PROMs for patients diagnosed with dental anomalies, such as AI.

Aims & objectives

The aim of this service evaluation was to understand the issues that children and young people with AI experience before, during, and after dental treatment using a PROM.

The objectives were to identify the following:

- What physical symptoms patients experienced (pain, sensitivity, difficulty eating, pain on toothbrushing)
- What impact their AI had on their lives (school attendance, self-confidence, relationships with peers)
- How they felt about having dental treatment
- Whether they were satisfied / happy with their teeth

Methods

Development and pilot of the questionnaire

The PROM questionnaire was developed in September 2019. It was piloted nationally by 10 children with AI, at two units (Eastman Dental Hospital and Sheffield Dental Hospital). The children were aged 9-17 years and included both boys and girls. Changes were made to the questionnaire following feedback from the pilot evaluation and are described in Table 2. The reading age (assessed using Readable© 2020)¹² of the final questionnaire was 9.5 years, and therefore parents were asked to help children under 10 to complete the questionnaire. To avoid confusion for individuals with colour vision deficiencies, the final questionnaire was black and white.

The PROM questionnaire was peer reviewed, pre- and post- pilot, by the National AI Clinical Excellence Network (AI CEN). The AI CEN consists of specialists and consultants in Paediatric Dentistry across the U.K. who have an interest in the management of children and young people with AI, and was set up in 2019 to improve care pathways and research for children with AI. The final questionnaire is shown in Figure 2.

Data collection

Originally, six units nationally participated in data collection. The PROM questionnaire was registered according to local clinical governance procedures at each Trust. No patient identifiable information was collected and patients and their accompanying adult gave verbal consent/assent to complete the PROM.

The plan was to include every child or young person with an AI diagnosis attending as an outpatient between 01/01/2020 and 31/03/2020. Due to the COVID-19 pandemic, most hospital units closed in March 2020, and therefore it was agreed to terminate data collection early, on the 16th of March 2020. Unfortunately, due to clinic closure and redeployment of staff during the pandemic, two units were unable to access their completed questionnaires, and these units were excluded from data analysis. Therefore, the final data came from four national specialist-led units (Eastman Dental Hospital, Guy's and St Thomas' Hospitals, Birmingham Dental Hospital, and Leeds Dental Institute). Descriptive statistics were produced using Microsoft Excel 2010.

Results

Sixty patients completed the questionnaire between 01/01/2020 and 16/03/2020, (n= 40 from the Eastman Dental Hospital, n= 6 from St Thomas' Hospital, n= 7 from the Birmingham Dental Hospital, and n= 7 from the Leeds Dental Institute). Clinicians collecting the data verbally reported that no patients or families refused to participate; however this information was not formally recorded.

The following sample characteristics were collected and described in Table 3: age, sex, phenotype, and stage of treatment.

The patient responses to the PROM questionnaire are shown in Figure 3. One patient did not answer Q3 and Q6, otherwise data was complete for all 60 patients. 72% of patients reported that they 'often' or 'sometimes' experienced pain or sensitivity from their teeth. 76% of patients reported that they 'often' or 'sometimes' felt unhappy with the way their

teeth look (Q5); and 29% reported that they 'often' felt their teeth affect their confidence to smile (Q6).

Q9, "Are you happy with your teeth?", was a yes/no question. Half of all patients answered Yes (50%, n=30) and the other half answered No (50%, n=30). Of the 16 patients who were 'post-treatment', 81% answered Yes. This dropped to 41% for patients 'mid-treatment' (n=29) and 33% for patients 'pre-treatment' (n=9). Irrespective of treatment stage, patient responses to Q9 "Are you happy with your teeth?" according to AI phenotype are shown in Figure 4.

19 out of the 60 patients made additional comments on their questionnaires; these are summarised in Table 4.

Discussion

The diagnosis and management of children and young people with AI can be complex and lifelong. As AI is a spectrum of enamel disorders, with varying severity, treatment plans are often individualised to the patient's concerns. This is the first study in the UK to develop and use a PROM to help identify patient needs and explore the concerns and issues that children and young people face as they move from the primary dentition to the permanent dentition.

The age range in this study was 5 to 17 years old, which is reflective of the specialist Paediatric Dentistry setting, but also indicates that AI can be diagnosed in the primary, mixed, and permanent dentitions. Clinically, AI typically affects the permanent dentition more severely than the primary dentition,¹ and this is reflected in the average age of the participants which was 12 years. There were more females than males in the sample (38 females and 20 males).

Patients with all AI phenotypes experienced a range of concerns, reflected in their PROM answers and comments. This reinforces the need for holistic and personalised patient care; to explore individual concerns and plan treatment to address these issues, rather than plan their treatment based on clinical phenotype.⁹

Despite patients experiencing a variety of concerns about their teeth, appearance and confidence relating to smiling (Q5 and Q6) was the most common concern, followed by pain. Fewer patients indicated that they experience bullying or teasing about their teeth, which highlights that individual perception of their teeth is not necessarily linked to comments or teasing from their peers. This finding is consistent with previous studies involving children with enamel defects.^{7,13,14} Aesthetic concerns, and the impact this has on a child's overall wellbeing, should not be underestimated and should be considered as part of a holistic treatment plan. One patient specifically mentioned how tooth whitening had improved their smile and confidence. Although the European Union directive states that tooth whitening (using concentrations of greater than 0.1% hydroxide peroxide) cannot be used in under 18s,¹⁵ the General Dental Council are supportive of this approach in this age group, when used to treat disease.¹⁶ Tooth whitening remains a valuable treatment option to have available in specialist units that manage these children, often used in conjunction with other procedures. However, there are barriers to its use and it may not be suitable for all presentations of AI, and particular attention should be paid to the patient's baseline level of sensitivity.^{17,18}

Over half of the sample was 'mid-treatment', which is reflective of the lengthy and evolving treatment plans for children and young people with AI, especially in this age group, as they move from the primary to permanent dentition. This finding is consistent with previous studies, showing that patients with AI attend more dental appointments than their unaffected individuals.^{5,6,7} For children and young people, frequent appointments will impact both their own school attendance and their parent/carer work and childcare arrangements; however, this impact was not measured and is therefore a limitation in this study. Excluding dental appointments, 15% of the sample indicated that they 'often' or 'sometimes' missed school because of their teeth.

Reassuringly, a majority of patients who were 'post-treatment' (including those attending for reviews) indicated they were happy with their teeth (81%, compared with 33% for the 'pre-treatment' patients). This suggests that current AI care, in specialist-led centres across the country, has a positive impact on the patient's satisfaction with their teeth. Given the

personalised approach to oral care, it is not possible to isolate specific procedures that are effective in this group.

This study was conducted across multiple centres in the secondary care / specialist setting; and may represent more severe AI phenotypes with a greater treatment need than those managed in primary care.

Anecdotally, no patients refused to complete the questionnaire; although this information was not formally recorded. Despite reassuring families that answers would remain anonymous, the questionnaire were completed on the dental clinic or in the waiting room; patients may have felt their answers were identifiable and this could have affected their responses.

This simple PROM questionnaire was developed with input from children with AI. It could be used by the dental team for individual patients, repeating the PROM at various stages in their growth and development, as part of treatment planning to address personal patient concerns, and highlight whether treatments have addressed those concerns.

Conclusion

This service evaluation which involved the development and use of an AI-specific PROM has highlighted the range of issues that children and young people with AI face; from sensitivity and pain, to problems with their appearance and bullying. The individual issues that patients faced were not linked to the clinical presentation of AI; and therefore clinicians could use this PROM at various stages of the child's development and throughout treatment in order to explore all the individual's issues and concerns. The results suggest that children who have completed treatment in this specialist-led setting have high levels of satisfaction with their teeth, and therefore timely diagnosis and referral from primary care is important.

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Table legends

Table 1. Potential treatment options available for children and young people with AI

Table 2. Feedback and changes made following the pilot

Table 3. Sample characteristics

Table 4. Additional comments from children and young people

Figure legends

Figure 1. Phenotype classification of AI

Figure 2. Final PROM

Figure 3. Bar chart showing PROM responses from 60 children and young people with AI

Figure 4. Histogram showing patient responses to the AI PROM question 9 'Are you happy with your teeth?' from 60 children and young people with AI.

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