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### Amelogenesis imperfecta - a systematic literature review of associated dental and oro-facial abnormalities and their impact on patients

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REVIEW ARTICLE

## Amelogenesis imperfecta – a systematic literature review of associated dental and oro-facial abnormalities and their impact on patients

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### Abstract

**Objective.** Amelogenesis imperfecta (AI) is a disease primarily affecting amelogenesis, but other aberrations have been reported. The purposes of this review were: (1) to identify other anomalies associated with AI, and (2) to describe the impact of the disease and its associated conditions on the oral health-related quality of life of patients, and the economic consequences. **Material and methods.** A literature search was conducted in the following databases: PubMed, EMBASE, Bibliotek.dk, The Cochrane Library, Web of Science, and OMIM, supplemented by a search for selected authors. Based on titles and abstracts, 137 papers were identified. **Results.** Most articles were case reports or case series with few cases. Aberrations were reported in the eruption process, in the morphology of the crown, in the pulp-dentine organ, and in the number of teeth. Gingival conditions and oral hygiene were usually reported to be poor, and calculus was a common finding. Open bite was the most commonly reported malocclusion. A negative impact on patients' oral health-related quality of life was described, but information was scarce. No information was found on the economic impact. **Conclusions.** A number of aberrations associated with AI have been reported, but not sufficiently systematic to allow for a secondary analysis and synthesis of the findings. The impact on patients in terms of reduced quality of life and economic burden needs to be studied.

**Key Words:** *Amelogenesis imperfecta, associated anomalies, systematic review*

### Introduction

Amelogenesis imperfecta (AI) is defined as a “disorder in which there is faulty development of the dental enamel owing to agenesis, hypoplasia, or hypocalcification of the enamel. It is marked by enamel that is very thin and friable and frequently stained in various shades of brown” [1]. Recent research has emphasized the close interaction during odontogenesis between the different components of the developing tooth germ [2]. This research indicates that defects in the amelogenesis may be associated with developmental defects or abnormalities in other components of the developing tooth, as well as in dental and skeletal development (e.g.

taurodontism, agenesis of teeth, delayed eruption, and frontal open bite).

From the patient's perspective, AI may have severe consequences in the form of high sensitivity of the teeth, low esthetic quality of the dentition, and poor mechanical properties of the dental tissues, all of which can result in reduced oral health-related quality of life. Furthermore, some forms of AI require extensive treatment and may therefore be an economic burden on patients.

The rarity of AI makes it difficult to obtain large study groups. Our knowledge of the disease is therefore based on case reports and case series with small numbers of patients. No primary systematic literature review has been identified focusing on

associated dental anomalies, dental and skeletal development, impact on the oral health-related quality of life of the patients, or the economic burden the patients have to carry.

### Purpose

The purposes of this review were: (1) to identify dental and oro-facial anomalies described as being associated with AI, and (2) to describe the impact of AI complications on patients' oral health-related quality of life, and also the economic consequences of this condition.

### Material and methods

The study was performed as a systematic review of original, primary articles reporting clinical or radiographic information on patients diagnosed as having AI.

#### Search strategy

The term "amelogenesis imperfecta" was searched for as a controlled search term where possible. The MeSH and Emtree terms were "exploded" and synonyms included where applicable. The controlled terms were OR combined with "amelogenesis imperfecta" as a free text term or key word in the databases where possible. Some databases were searched using free text search alone.

The following databases were searched by applying almost identical search strategies: PubMed, EMBASE, Bibliotek.dk (the Danish national book and journal catalogue), The Cochrane Library databases, Web of Science, and OMIM. All databases were searched as far back as they go. The final update of the search was on 15 May 2007. Separately, we searched for the following authors in combination with the text word "amelogenesis" in an attempt to avoid unintentional omission of relevant papers: Aldred MJ, Crawford PJ, Bäckman B, Seow WK, Sundell S, Winter GB, Witkop CJ, Witkop CJ Jr., Hoppenreijts TJ. The search strategy details can be obtained on request from the first author.

Papers describing for example hereditary hypoplastic enamel were not included, nor were articles where AI is described as part of a syndrome. Only papers written in English, German, Danish, Norwegian and Swedish were included. Abstracts and reviews were not included.

The search produced 2,225 citations, which were reduced to 642 references after screening by the first author for duplicates. Subsequent screening of the title and abstract identified 137 papers in which the term "amelogenesis imperfecta" was the diagnostic

term used by the authors, and thus relevant for this review [3–139].

#### Data extraction

Only findings actually described by the authors were used. We did not include additional findings that could be drawn from clinical photographs or radiographs in the article and that were not reported by the authors. A special form (a copy can be obtained from the first author on request) was designed for extraction of the following data:

1. Study design and material
2. Diagnosis of AI
  - a. Classification system
  - b. Diagnostic criteria
3. Eruption of teeth
4. Dental anomalies
  - a. Aberrations in crown
  - b. Aberrations in root
  - c. Aberrations in dentine and pulp
  - d. Aberrations in number and shape
5. Periodontal conditions
6. Occlusion, space conditions, and cephalometric findings
7. Impact on patient
  - a. Quality of life
  - b. Economic burden

The papers were distributed between six of the authors (SP, HG, DH, GH, HH, HL), whose data extraction was calibrated by comparing their scorings of five papers.

### Results

The studies varied considerably depending on study design. Only three population-based epidemiological studies designed to determine the prevalence of AI were identified. The original study by Witkop [140], based on a sample of 64,000 children from the State of Michigan, reported that "enamel defects loosely classified as amelogenesis imperfecta occurred once in 12,000 to 14,000" or 0.07 to 0.08 per 1,000. Later, two large Swedish epidemiological studies reported prevalence rates ranging from 0.25 per 1,000 [120] to 1.4 per 1,000 [16]. Large case series (> 10 cases) were relatively rare [6,15,27,29,33,40,47,64,68,69,82,92,98,103,108,112,113,116,119,136], and the majority of the papers were case reports, some with a description of family members, others presenting the clinical management of one or a few AI patients. Pedigrees describing patterns of inheritance of the condition were presented in 42 papers, some reporting more than one pattern, and three showing a pedigree without stating an inheritance pattern. Autosomal dominant inheritance was described in 18 papers

[6,17,24,27,37,38,45,47,58,59,64,74,79,104,106,119,125,138,141], autosomal recessive inheritance in 6 [17,28,46,67,92,119], and X-linked inheritance in 15 [10,17,19,23,36,37,48,55,57,62,63,88,116,119,138] out of the 137 papers.

The most frequently used classification system was that described by Witkop and Rao [142] and by Witkop [143], which was used in 41 (30%) of the 137 papers, while the classification system described by Sundell and co-workers [118,120] was used less frequently (10 papers). More than half of the papers used other classification systems, or did not report having used a classification system at all. The clinical criteria used for classification were specified in 48 (35%) of the 137 papers.

### Eruption

The eruption process of the permanent teeth was reported to be affected by AI. Some studies reported follicular cysts [66,99,113], others delayed eruption [8,26,28,31,46,53,90,93,106,113,125,127,135,139] and retention or impaction of teeth [9,26,28,46,58,59,66,80,81,87,105,114,124,126–128].

### Dental anomalies

The most frequently reported dental anomaly was taurodontism, followed by reduced crown size (Table I). Changes in the pulp-dentine organ-like enlarged pulp chambers and intrapulpal calcification and pulp stones were also relatively frequent findings. Crown resorption was reported in six papers and agenesis in nine.

Table I. Summary of dental findings in the crown, the root, and the pulp-dentine organ reported in AI patients.

	Feature	References
Crown	Resorption	[31,76,85,114,125,135]
	Small crowns	[20,26,36,46,51,53,73,83,90,100,106,114,133]
	Attrition	[8,19,21,28,35,45,106,111,139]
	Deviant morphology	[85,144]
Root	Resorption	[26,31,131]
	Taurodontism	[4,6,31,33–35,40,45,55,57,67,85,101,118,119,132,136,145]
	Short roots	[26,66,106,135]
Pulp-dentine	Deviant morphology	[66,85,99,135]
	Enlarged pulp chamber	[6,33,35,40,91,111,118,119,126,136]
	Pulp stone	[31,53,58,59,66,85,93,99,101,114,118,119,135]
Number and shape	Dens in dente	[8]
	Agenesis	[8,9,20,21,31,46,81,91,138]

Table II. Summary of reporting of periodontal conditions in AI patients.

Feature	References
Gingival enlargement	[9,22,46,53,93,117,124]
Gingivitis	[7,20,21,24,26,35,38,41,56,58,77,81,82,85,87,91,101,104,111,112,114,117,118,124,126,131,135]
Periodontitis	[21,51,82,91,101,114,126,127]
Calculus	[7,37,59,83,101,118,131]
Oral hygiene	[7,20,21,35,56,59,72,79,81,82,85–87,91,101,104,111,114,124,131,139]

### Periodontal conditions

Information on periodontal conditions was available in 40 of the papers, ranging from gingival enlargement and gingivitis to periodontitis, often combined with poor oral hygiene and calculus (Table II).

### Occlusion, space conditions, and cephalometric findings

Thirty-two of the papers reported malocclusion, 8 anomalous space conditions, and 12 findings from cephalometric analysis (Table III). Open bite was reported in 23 papers. Fewer reported exact measurements of overjet or overbite. Sagittal molar relationship according to the Angle classification was reported in seven of the papers [8,11,20,46,81,98,105], but in none of the large epidemiological studies [14,16,120]. A limited number of studies with emphasis on cephalometric findings were identified [15,25,68,98,103]. However, cephalometric information was also presented in some of the case reports, which added up to a total of 11 studies with cephalometric information (angular measures: [15,25,68,75,91,98,103,108], linear measures [15,25,68,75,91,98,103], and facial proportions [15,81,108]).

### Patient impact

Dental hypersensitivity is frequently reported by AI patients as a symptom that severely influences their daily lives and was accordingly reported in several

Table III. Summary of occlusal traits, space conditions, and cephalometric findings reported in AI patients.

	Feature	References
Occlusion	Open bite	[6,8,11,17,20,23–25,46,53,67,68,73,75,81,91,92,98,103,108,112,114,131]
	Overjet	[15,24,25,46,68,69]
	Overbite	[15,25,46,68,69,75,98,103,108]
	Cross bite	[46,75,105,114]
Space conditions	Other occlusal traits	[6,69,80,112]
	Crowding	[11,46,81]
	Excess of space	[20,28,34,73,92]

papers [8,11,18–21,23,25,30,41,48,53,73,77,82,83,94,101,103–105,107,108,111,112,117,124,126,138]. One paper reported improvement in quality of life and weight gain after treatment [20], but only one was based on formal assessment of oral health-related quality of life [29]. None of the papers reported information on the economic burden on AI patients due to their extensive treatment need.

## Discussion

The present literature review identified a number of supplementary findings in papers on AI, i.e. findings involving the hard dental tissues, the periodontal tissues, the occlusion, and development of the dentition and facial skeleton. A few studies also reported on how the disease affected the patient.

AI is known to be part of several syndromes (e.g. Tricho-dento-osseous syndrome). However, as many syndromes are associated with several different dental abnormalities, we decided not to include papers in which AI was described as part of a syndrome.

The review identified a number of problems relating to identification of the relevant literature in this field and to methodological issues. Thus, the concept of “amelogenesis imperfecta” was searched for in its broadest form. This implies that any subcategory that could occur in some of the databases, e.g. other types of hypoplasia or colour changes, was included in the search. Initial attempts were made to make the search more specific and precise, but the high degree of sensitivity aimed at in this review could only be achieved by making the search almost all-inclusive. This may partly be due to the many types of reporting utilized for this condition and its associated abnormalities. We therefore had to perform an extensive manual search of papers meeting the inclusion criteria for this particular review. This was a time-consuming process and was therefore confined to titles and abstracts, which could mean that some papers with clinical information were missed.

The many findings supplementary to AI identified by this review suggest that the basic defect in amelogenesis carries a risk of damage to a number of other dental tissues and orofacial structures. This requires that a broad range of findings supplementary to AI be considered when reporting AI cases. The large majority of reports identified for this review were case reports, which often emphasized restorative treatment. Case reports are useful for communicating information on rare conditions such as AI, but this review has shown that supplementary, related, findings should also be considered, and that it should be indicated whether the conditions were found or not. Guidelines for case reports should be considered in order to improve the quality of this type of publication.

Some of the types of AI were reported to result in severe sensitivity of the teeth, but other effects on the patient's life and quality of life were rarely well described. With the presently increasing interest in non-clinical outcomes, this may be an important focus in future studies.

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