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Clinical and microstructural aberrations of enamel of deciduous and permanent teeth in patients with autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy

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ABSTRACT

Objective: Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED) causes multiple endocrine deficiencies, oral candidiasis and different forms of ectodermal dystrophy including enamel hypoplasia, documented in permanent teeth. Our purpose was to examine dental aberrations associated with APECED, including possible manifestations in primary teeth.
Design: We studied clinically, radiographically, and by scanning electron microscopy (SEM) teeth of children belonging to two APECED families with different mutations in the *AIRE* gene.
Results: In addition to enamel defects in the permanent teeth we observed hypoplastic pits and hypomaturated patches in the deciduous teeth with underlying changes in the prismatic ultrastructure. The enamel of the permanent molars exhibited a layered arrangement with included whirl-like formations.
Conclusions: Our findings confirm that APECED causes enamel defects that are individually but chronologically distributed, and can alter enamel development early enough to affect deciduous teeth.

1. Introduction

Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED), also known as autoimmune polyglandular syndrome type I (APS 1), is a rare heritable disease (OMIM 240300), characterized by a broad clinical spectrum of signs that fall into three main categories: (1) multiple endocrine deficiencies including hypoparathyroidism (HPT), adrenocortical failure, gonadal failure, insulin-dependent diabetes mellitus, thyroid disease, gastric parietal-cell atrophy, and hepatitis, (2) chronic mucocutaneous candidiasis (CMC), and (3) different forms of ectodermal dystrophy including keratopathy, alopecia, vitiligo, tympanic membrane sclerosis, keratoconjunctivitis, and hypoplasia of the dental enamel.[1] APECED is due to homozygocity or, rarely, heterozygocity, for mutations in the *AIRE* (autoimmune regulator) gene.[2–4] This gene encodes a transcription factor with primarily intra-thymic expression and role in autoimmunity control. Consequently, *AIRE* mutations cause a multiorgan autoimmune reaction.[2,5,6]

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Abbreviations: AIRE gene, autoimmune regulator gene; APECED, autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy; APS 1, autoimmune polyglandular syndrome type I; BEI, back-scattered electron image; CMC, chronic mucocutaneous candidiasis; DEJ, dentine–enamel junction; HPT, hypoparathyroidism; PTG, panoramic tomogram; SEI, secondary electron image; SEM, scanning electron microscopy.

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The most consistent three components of the disease are CMC, HPT and primary adrenocortocal failure (Addison's disease),[1] two of which have classically been required for the diagnosis.[4] CMC is often the first manifestation of the disease, and any additional signs can occur at any time later.[1] APECED usually has an early onset—in the largest available patient material the median age at onset was 3.3 yrs.[4]

Most APECED patients display hypoplasia of dental enamel on their permanent teeth.[7–9] The development of the aberrations appears to be time-dependent, following the chronological sequences of enamel deposition. However, the timing and thus the pattern of enamel involvement differs from patient to patient.[7] Furthermore, the enamel defect can be present as generalized enamel hypoplasia varying from only pitted enamel surface to compete lack of enamel, or locally as horizontal lines of pits or horizontal hypoplastic patches which are clearly demarcated from normal enamel and suggestive of a transient autoimmune episode.[7,8] In the hypoplastic enamel, abnormal

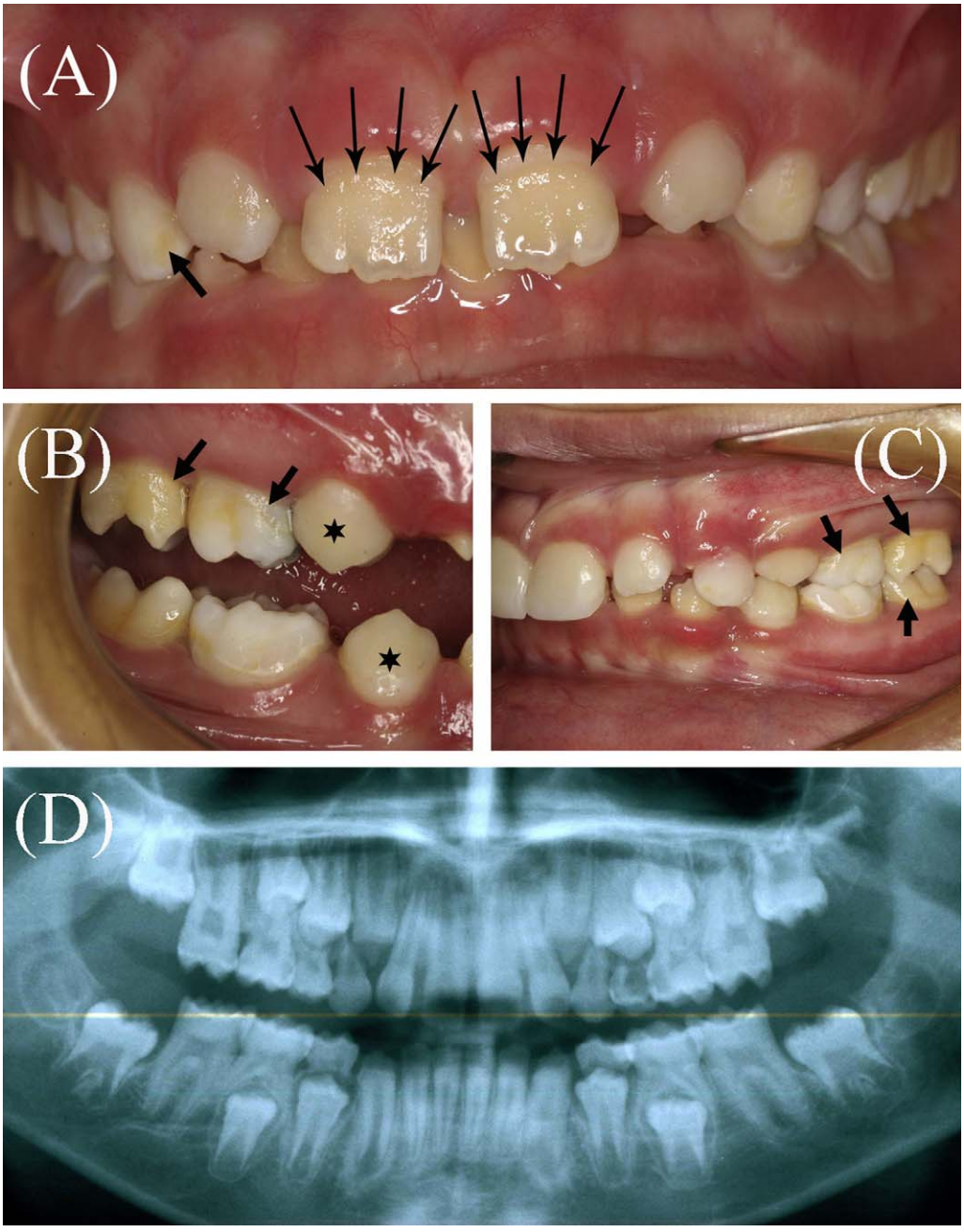

Fig. 1 – Mixed dentition of the female APECED patient A from family 1. (A) At the age of 9, the upper central permanent incisors display extremely thin enamel except for a cervical rim of a normal-appearing thickness (thin arrows). The upper lateral permanent incisors show minor hypoplastic grooves at incisal edges. The deciduous teeth are chalky with yellowish patches (single arrow). (B and C) Clinical photographs taken later illustrate hypoplastic enamel on all permanent first molars, as well as tiny hypoplastic pits on the upper deciduous second molars (arrows). First premolars appear normal (asterisks). Upper central incisors have been restored with composites. (D) The panoramic tomogram from the age of 8 shows thin enamel and sharp cusps on the permanent first molars.