

Long-term followup of a rare dental eruption disorder

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Nonsyndromic primary failure of eruption (PFE) is a rare disorder of dental eruption with no obvious local mechanical interference or associated systemic or syndromic conditions. The purpose of the present study was to genetically and clinically characterize a family affected by PFE and describe the natural evolution of the disorder. Three generations of a family with 18 members, eleven of them clinically affected by PFE were evaluated periodically during 20 years. No treatment was carried out for open bite correction, except for tooth extractions for periodontal reasons. DNA sequence analysis of all exons and the exon/intron boundaries was carried out in index patient II.9, and seven probands were genotyped. PFE was observed in varied degrees of severity, with no difference between genders. Clinical presentation became more severe at adulthood. An incisal attrition facet was found in the left superior canine of proband II.9 at the age of 42, although it was in infraocclusion. Proband II.11 presented spontaneous reeruption of the left superior first molar and the right inferior first molar. Genetic analysis revealed a heterozygous mutation in intron 9 of the PTH1R gene (c.639-2A>G). The twenty-year follow-up permitted to conclude: (i) the onset of PFE may occur despite the occurrence of functional occlusion of involved teeth; (ii) PFE is closely related to ankylosis; (iii) affected teeth maintain the eruptive potential even in adulthood; and finally (iv) the earlier the onset of PFE the more severe is the open bite.

Keywords: Primary Failure of Eruption; PTH1R; mutation analysis; Tooth Eruption; Ankylosis.



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