

AAO Foundation Award Final Report

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Secondary Investigators	H. Kapadia, D.D.S. and J. Dykes, D.D.S.
Award Type	Biomedical Research Award
Project Title	Genetic Studies on Human Primary Failure of Eruption
Project Year	2001
Institution	University of Texas Dental Branch at Houston
Summary/Abstract	<p>Please note that four abstracts have been published from this study which is still ongoing.</p> <p>R. MIDDLETON, O.J. DYKES, IV, <u>H. KAPADIA</u>, J.D. ENGLISH, and R.N. D'SOUZA, University of Texas Health Science Center at Houston - Dental Branch, USA</p> <p>Primary Failure of Eruption (PFE), a rare anomaly of tooth eruption, is poorly characterized and remains one of the most difficult to diagnose and treat. Objectives: The aim of this study was to characterize the clinical phenotype for PFE by: (1) developing inclusion and exclusion criteria for a database of individuals/families with PFE and (2) delineating by phenotypic and partial segregation analyses of the affected individuals whether PFE occurs as a sporadic or familial condition. Methods: Using patients identified with PFE, as outlined first by Proffit and Vig in 1981, we collected x-rays, models, photos, medical and dental histories, and arranged for or performed a thorough clinical evaluation. Results: Our inquiry from the dental/medical community yielded sixteen cases of PFE. Unlike previously published data, our results indicate that 37.5% of the cases had one or more family members affected, suggesting PFE can be an inheritable condition. This will allow for studies of candidate genes found in the dental follicle. Consistent with previous data, all of the patients presented with a posterior open bite, with a slightly higher percentage of unilateral (56.25%) than bilateral open bite (43.75%). Our results also showed posterior teeth were most commonly affected (100%), while anterior teeth were less often involved (25%), with every affected tooth being distal to the first involved tooth in that particular quadrant. Over 43% of the individuals were Angle Class III molar occlusion. In the affected area, 25% of the cases had ankylosed primary molars, and</p>

37.5% had congenitally missing permanent teeth. Conclusion: Results of our phenotypic analyses of 16 individuals affected with PFE provides new information about its clinical features. Such knowledge will aid in establishing phenotype-genotype correlations for PFE. Supported by NIH/NIDCR Grant DE13368 (RDS), DE14237 (HK), and AAOF grant (RDS and JE).

Seq #339 - Development and Growth of Teeth, Sutures, and Muscle

11:00 AM-12:15 PM, Saturday, March 9, 2002 San Diego Convention Center Exhibit Hall C

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General Session (March 6-9, 2002)

The abstract is published in:

1. #3781 - A phenotypic analysis of primary failure of eruption presented at 2002 IADR in San Diego
2. Hinman abstract - Genetic studies of Human primary failure of eruption presented to the Hinman Meeting in October 2002 in Memphis, TN
3. #2340 - Genetic Studies on PFE presented at IADR on June 24, 2003 in Goteborg, Sweden
4. #0415 - Genetic Studies on PFE presented at AADR on March 13, 2003 in San Antonio, TX