RESEARCH REPORTS
Clinical

S.A. Frazier-Bowers[1]*, H.M. Hendricks[1], J.T. Wright[2], J. Lee[1], K. Long[2], C.F. Dibble[3], and S. Bencharit[3,4]

[1]Department of Orthodontics, School of Dentistry; [2]Department of Pediatric Dentistry, School of Dentistry; [3]Department of Pharmacology, School of Medicine; and [4]Department of Prosthodontics, School of Dentistry, University of North Carolina at Chapel Hill; *corresponding author, sylvia_frazier@dentistry.unc.edu

J Dent Res DOI: 10.1177/0022034513513588

Novel Mutations in *PTH1R* Associated with Primary Failure of Eruption and Osteoarthritis

APPENDIX

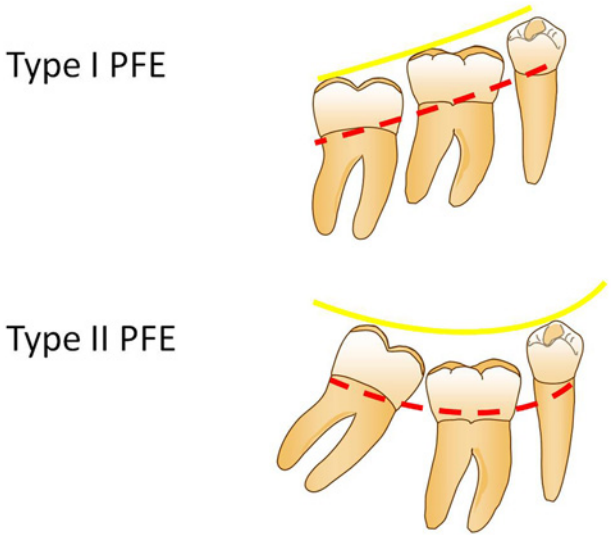

Figure. Diagrammatic representation of type I versus type II primary failure of eruption based on a previous study (Frazier-Bowers *et al.*, 2007). Illustrations provided by Dr. Sonny Long.

Appendix Table 1. Genotype-Phenotype Correlation of 2 Families Segregating PFE

	PFE Affection	Other Phenotype	Arthritis	Mutation
Family 1				
I:3	Affected	Edentulous: reported type I	Y	Unavailable
II:4	Affected: type I	Unilateral/supraosseous	Y	c.996_997insC
II:13	Unaffected	N/A	N	No mutation
III:1	Affected: type I	Bilateral/supraosseous	UNK	c.996_997insC
III:3	Affected: type I	Bilateral/supraosseous	UNK	c.996_997insC
III:9	Unaffected	N/A	N	No mutation
III:10	Unaffected	N/A	N	No mutation
III:11	Unaffected	N/A	N	No mutation
Family 2[a]				
I:1	Unaffected carrier	N/A	Y	c.572delA
II:2	Unaffected carrier	N/A	Y	c.572delA
II:4	Affected[b]	—	Y	Unavailable
III:1	Type I	Bilateral/supraosseous	UNK	c.572delA
III:2	Affected[b]	—	UNK	Unavailable

N/A, not applicable; PFE, primary failure of eruption; UNK, individuals of an age before the onset of symptoms/diagnosis of arthritis.
[a]Family 2 shows incomplete penetrance with 2 individuals carrying mutation but not affected with the eruption phenotype.
[b]Two individuals were not available for sequencing, but one reported symptoms of arthritis, and both reported eruption failure as diagnosed by their dentist.

Appendix Table 2. Phenotype-Genotype Correlation of Additional Families Without a Functional *PTH1R* Mutation

	PFE Status	Affection Pattern	Genetic Alteration/SNP
Family 3			
1	Unaffected	N/A	No alteration
2	Unaffected	N/A	No alteration
3	Unaffected	N/A	No alteration
4	Affected: type I	Bilateral/supraosseous	c. 1389 T>C, known variant
Family 4			
1	Unaffected	N/A	No alteration
2	Affected	Undetermined	c.638+97G>A, intronic
3	Affected: type II	Unilateral/supraosseous	c.638+97G>A, intronic
Family 5			
1	Unaffected	N/A	No alteration
2	Unaffected	N/A	No alteration
3	Affected: type I	Bilateral/supraosseous	c. 646 C>A, synonymous
4	Unaffected	N/A	c. 646 C>A, synonymous
5	Unaffected	N/A	No alteration
Family 6			
1	Affected: type II	Bilateral/supraosseuos	c. 1389 T>C, known variant
2	Affected: type II	Bilateral/supraosseous	c. 1389 T>C, known variant
3	Affected: type UNK	UNK	c. 1389 T>C, known variant

N/A, not applicable; PFE, primary failure of eruption; SNP, single-nucleotide polymorphism; UNK, individuals of an age before the onset of symptoms/specific phenotype unknown.
Summary of additional families with *PTH1R* SNPs: 4 of 8 showed nonfunctional alterations in *PTH1R*, not including the more commonly occurring SNP c. 1116 +58 T>C, which was found in several affected and unaffected individuals.