

AAO Foundation Award Final Report

Principal Investigator	Heleni Vastardis, DDS, DMSc
Co-Investigator	
Secondary Investigators	
Award Type	Biomedical Research Award
Project Title	Identification of a gene locus for peg-shaped maxillary lateral incisors in the human genome
Project Year	1997
Institution	Harvard School of Dental Medicine
Summary/Abstract	<p>Previously, we have demonstrated using human molecular genetics approaches that a missense mutation (Arg31Pro) in the homeodomain of <i>MSX1</i> causes agenesis of second premolars and third molars in one family. Tooth agenesis presents with a wide range of phenotypic expressivity. It may display complete expression, such as absent teeth, or incomplete such as rudimentary third molars or peg-shaped maxillary lateral incisors. To test the hypothesis of genetic heterogeneity within the tooth agenesis phenotype, a large family with peg-shaped (conical) lateral incisors has been identified and clinically evaluated. Mapping assessment of this family to the <i>MSX1</i> locus provided evidence against linkage to <i>MSX1</i>. Other candidate gene linkage and mutational screening have been supportive to the hypothesis that defects in different genes contribute to the clinical variation of the disorder.</p>