Amelogenesis Imperfecta: Genetic Diagnosis and Treatment at UCSF Pediatric Dental Clinic

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Dana Point, CA
Inherited Enamel Defect in Primary Teeth
(3 y.o. female patient)
Studied Family for WDR72 Mutation

Katsura K. et al.
Studied Family for WDR72 Mutation
Diagnose by Collecting Saliva Samples of Subjects.
Mutational Analysis of WDR72 Results in Frameshift Mutation (p.G255fsX294)

A  Homozygous wild-type (V1):

B  Homozygous recessive (affected proband V3)

C  Heterozygous (father & mother)
Post Treatments with Posterior SSCs and Anterior Esthetic Composite Crowns
At 6 y.o., Congenital Enamel Defect of 1st Permanent Molars
GI Sealant Placement to Reduce Thermo-Sensitivity

Before GI placement

After GI placement
SSCs for 1st Permanent Molars Soon after Fully Erupted, ~ 7 y.o.
Pt’s at 8 years old

Pictures obtained on 3/25/15

Temporary composite veneers were done for partially erupted incisors using self-etch bonding agent Clearfil S3, helping to reduce thermal sensitivity and improved esthetics.
Team of participants

• Kei Katsura (genetic mutation work)
• Dr. Thuan Le
• Dr. Ling Zhan
• Dr. Rung Warotayanont
Family Pedigree and Patient’s Clinical Presentation

(A)

Family Pedigree
Autosomal Dominant or X-linked Dominant

![Pedigree Diagram](image)

![Patient's Clinical Presentation](image)
Mutational Analysis

g.10602C>G (p.Pro83Arg)
Clinical management for ENAM mutation, 14 y.o. female

- SSCs for posterior upper and lower premolars and molars
- Fiber-reinforced resin crowns for upper anterior teeth
- Ortho txt completed
- Planning Cerec crowns using CAD/CAM system for her anterior permanent teeth
Diagnostic wax up model used for provisional crowns fabrication
Provisional resin crowns were pre-fabricated
Pre-op photos in O.R.
Pre-op PA X-rays for anterior teeth
Crowns preps
Upper anterior crowns preps and placement of retraction cords for impressions, showing crown shoulder margins
Retraction cords placement
Taking impressions techniques
Upper PVS impression
Lower anterior crowns preps
Retraction cords placement, showing shoulder margins for impression

Aluminum chloride used for retraction cords
Lower PVS impression
Re-lining provisional crowns with flowable composite
Removing excess flowable composite, light curing, further re-shaping the contour of temp crowns.
Cementation of temp crowns
After CAD/CAM milling of final crowns, which are comprised of ~ 85% ceramic and ~15% composite, the final esthetic crowns will be cemented for both upper and lower anterior teeth.

If chipped or defect detected in the future, crowns can be restored with regular composite resins.
Completed die trimming for upper and lower anterior teeth, showing shoulder margins were captured.
Final provisional wax-ups for CAD/CAM milling procedures
Lab fabrication of CAD/CAM ceramic crowns (85% ceramic + 15% composite)
Crowns with adequate thickness were evaluate on casts with proper occlusion, and clearance.
Prepared teeth after removing temporary crowns

Patient was under minimum oral conscious sedation using 10 mg of Diazepam (2 tabs, 5 mg/tab), and 50% N2O/50% oxygen for anti-anxiety.
Crown cementing system (composite cement)
Mixing A + B = self-etched adhesive

Applied for 20 sec, then air dry gently, followed by light cure
Preparation of ceramic crowns for increased bond strength with composite cement

Etch ceramic crown with 9.6% hydrofluoric acid (HF)

Remove debris post etching

Silanate ceramic crowns to chemically modify surfaces
Composite cement was added to chemically-modified ceramic crowns.
Crown placement, remove cement excess, and light curing
Flowable composite was added at any opened margins’ areas, and removal of fine excess cement.
Immediate post-op photo after crowns cementation
Final X-rays
Team of participants

• Dr. Pam Den Besten
• Dr. Sam Huang
• Dr. Thuan Le
• Dr. Jessica Massie (genetic mutation work)
• Dr. Ram Vaderhobli