Case Report - Ginny and Valerie

S-Subjective Phase

Chief Complaint
My teeth are sensitive, smaller, and more yellow than others'.

History of the Chief Complaint
Ginny has always noticed that her teeth were sensitive to certain foods. Valerie reports the same complaint. Ginny reports that she has always known that her teeth were not like other kids. She says that her mother doesn't know why.

Medical History
Ginny is an 8 yr old female, born at 40 weeks gestation to a G1P1 30 year-old mother and 32 year- old father. The pregnancy had no complications. Birth weight was 8 lbs 11 oz, and length was 22 inches. All developmental milestones were reached at normal times, with the first tooth erupting at 8 mos. All immunizations are up to date, and she is currently taking no medications. Ginny has a history of allergy to red dye.
Valerie is a 6 yr old female born at 39 weeks gestation to a G2P2 32 year-old mother and 34 year old father. The pregnancy was complicated because the mother was diagnosed with breast cancer and started chemotherapy. Birth weight was 7 lbs 15 oz, and length was 21 inches. All developmental milestones were reached at normal times, with the first tooth erupting at 9.5 mos. All immunizations are up to date and Valerie is currently taking no medications. She has no known allergies.

Dental History
Ginny has had no caries and no restorations. She has always had sensitive teeth. Her first dental visit was at 1 year and she continues to have regular dental recalls every 6 months. She is currently taking a fluoride supplement and drinks well water. She drinks juice and pop between meals only on the weekends and her favorite snacks are gum, chips and crackers. She brushes her teeth twice per day.
Valerie has had no caries and currently has some composite bonded restorations to restore her occlusion on the mandibular anterior teeth. She has always had sensitive teeth. Her fist dental visit was at 1 year and she continues to have dental recall visits every 6 months. She is currently taking a fluoride supplement and drinks well water. She never drinks juice or pop in between meals. Her favorite snacks are apples, pretzels, and chips. She brushes her teeth twice per day with Crest for sensitive teeth or Aquafresh.

Family History
There are three female children, age 8, 6 and 3 years. The girls' mother reports that their little sister's teeth look similar to the older girls. The children's father and his mother have similar teeth. Everyone on the mother's side of the family reportedly has normal appearing teeth.

O-Objective Phase

Clinical Findings

Ginny: Oral examination showed an age-appropriate dentition. The teeth were yellow with spacing. There was generalized attrition, chipping of the enamel. There was a Class I skeletal, molar and canine occlusal relationship. The rest of the oral findings were within normal limits. Radiographic examination showed that the teeth all had large pulps and thin enamel.

Valerie: Oral examination showed an age-appropriate dentition. The teeth were yellow and spaced. There were composite restorations on teeth 7-C to 8-C. There was end to end occlusion in the anterior with Class I skeletal and molar occlusion. The rest of the oral findings were within normal limits. Radiographic examination showed that the teeth all had large pulps and thin enamel.

A-Assessment

Differential Diagnosis

1. X-linked Amelogenesis imperfecta
    a. The enamel is abnormal throughout the dentition and runs in the family. It is thin (hypoplastic) and structurally weak, as evidenced by the enamel attrition and chipping. This suggests hypoplastic amelogenesis imperfecta.
    b. The pedigree in this family supports a trait with X-linked inheritance, but an autosomal dominant pattern cannot be ruled out. Vertical bands of thicker and thinner enamel are suggestive of Lyonization—due to the pattern of X-chromosome inactivation, but these are very subtle.

2. Autosomal Dominant Amelogenesis imperfecta
    a. Inherited hypoplastic enamel
    b. The pattern of inheritance in this family could be autosomal dominant. Vertical bands of thicker and thinner enamel in affected females are not obvious, so it might not be X-linked.

3. Enamel Hypoplasia
    a. Enamel hypoplasia is characterized by localized defects of enamel, usually affecting teeth that develop at the same time. It is usually caused by an environmental insult such as fever or illness.
    b. This is considered in this case, because some of the family members may have been exposed to similar environmental insults. It does not fit well because all of the teeth are uniformly affected in this family.

4. Tetracycline Staining
    a. This can cause yellow appearance to the teeth. This is

excluded in this family because there is no history of exposure to tetracycline in the patients' history, and structural problems, besides staining, are evident.

Diagnosis
X-linked Amelogenesis Imperfecta. This diagnosis was confirmed by mutational analyses, which identified a defective translation initiation codon in *AMELX*.

Discussion
This is a genetic disease with a 100% risk of transmission from the affected father to his female offspring, with no risk to male offspring. All of the affected daughters offspring (male and female) will have a 50:50 chance of inheriting the trait. Affected male offspring of the daughters will have a more severe dental phenotype.

P-Treatment Plan

Management
Dental recall every 6 months with fluoride treatment
Fluoride supplements
Orthodontic consult

Treatment outcomes
Because of esthetic concerns and sensitivity, these teeth are usually restored using full crowns, once all permanent teeth have erupted and the pulp chamber size is reduced by the accumulation of secondary dentin. When rapid occlusal wear is a problem, the teeth may be temporarily restored with stainless steel and or composite crowns.

Restore teeth with full coverage composite and/or stainless steel crowns during mixed dentition, if necessary. Restore permanent teeth with full crown coverage at around age 18, when risk of exposing pulp chambers is reduced.

References
Kim JW, Simmer JP, Hu YY, Lin BP, Boyd C, Wright JT, Yamada CJ, Rayes SK, Feigal RJ, Hu JC. (2004) Amelogenin p.M1T and p.W4S mutations underlying hypoplastic X-linked amelogenesis imperfecta. J Dent Res. 83:378-83.
Coffield KD, Phillips C, Brady M, Roberts MW, Strauss RP, Wright JT. (2005) The psychosocial impact of developmental dental defects in people with hereditary amelogenesis imperfecta. J Am Dent Assoc. 136:620-30.
Lindunger A, Smedberg JI. (2005) A retrospective study of the prosthodontic management of patients with amelogenesis imperfecta. Int J Prosthodont. 18:189-94.
Hillson S, Bond S (1997). Relationship of enamel hypoplasia to the pattern of tooth crown growth: a discussion. Am J Phys Anthropol 104:89-103.
Reid DJ, Dean MC (2000). Brief communication: the timing of linear

hypoplasias on human anterior teeth. Am J Phys Anthropol 113:135-139.