Case Report: Steven

S - Subjective Phase

Chief Complaint

Referred by the oral surgeon after patient presented with toothache. Recommended further treatment for rampant caries.

History of Chief Complaint

"I had a bad toothache and my mom took me to the dentist. He took out my tooth and told my mom that I had a lot of cavities and something might be wrong with me".

Medical History

Patient is not taking any current medications. He did take erythromycin for a recent toothache and dental abscess. He has taken Nystatin for thrush associated with antibiotics in the past.

Allergy to penicillin. No other known allergies.

Prenatal and Developmental History:

Steven is the firstborn son of his two parents. He has two half-siblings on his father's side. No problems during mother's pregnancy. She received regular pre-natal care. He was delivered via C-section a week late and was bottle fed. When he was a baby he had surgery to re-cannulate the nasolacrimal duct and close a fistula below the lower eyelid. He has great vision but his mother reports he never made a lot of tears as a baby. He has reached all developmental milestones on times according to the mother. No other medical problems present.

Family History

Steven's sister has a similar history of multiple carious lesions in primary and permanent dentitions. She has no allergies, is not taking any medications, and has similar diet. She also had surgery to re-cannulate the nasolacrimal duct and close a fistula below the lower eyelid when she was a baby. She complains of cracking around the lips. Their father had all his teeth extracted at age 18 and wears complete dentures. The father's mother also wore dentures, as did her brother. Steven has two older half siblings from his father's previous marriage with no dental problems. Steven's mother has all of her own teeth and is in good dental health. There have been no stillbirths nor miscarriages.

Stevens' sister was also examined. She was missing her parotid ducts but had submandibular ducts/glands. Her mouth was dry but not as dry as Steven's. She also had rampant dental caries but had several teeth without caries as well.

Father was missing both parotid ducts/glands. He wore complete upper and lower dentures.

Dental History

Steven's first tooth erupted at 6 months and looked normal. Steven's mother reported that when he was around three years old he had his first toothache. Eventually most of his primary teeth decayed and fractured or were extracted by a dentist. His previous dentists said that Steven had had "baby bottle decay" although his mother denies giving him a bottle at bedtime or continuous sipping form a bottle or cup. He only drank water from a sippy cup. He has a history of dental abscesses, toothaches and missed school due to mouth pain. He chews on the opposite side of his mouth when he has a toothache. Steven's first permanent molars erupted at age 6 and also looked normal. Tooth # 14 was extracted because of severe dental caries. The tooth was nonrestorable.

Steven reports brushing his teeth after every meal. Mother reports low sugar intake, and Steven drinks milk or water with meals. Steven drinks from a fluoridated water source. He complains of having dry mouth and needing frequent sips of water to swallow food. Steven is not satisfied with the appearance of his teeth and wishes they looked normal and did not hurt so much.

O - Objective Phase

Clinical Findings

The oral mucosa was moist with very scant saliva present. There is no saliva pooling in the anterior floor of mouth. The openings to the parotid and submandibular glands could not be detected. The parotid and submandibular glands could not be palpated. He presents with rampant decay in his permanent dentition. Clinical exam shows Steven is missing #1, #6, #14, #16, #17, and #32. All anterior teeth have carious lesions, with extensive destruction of the lower anterior teeth. Large multi-surface carious lesions are present on #3, #19, and #30. There is incisal caries on all maxillary anterior teeth, cervical caries on #7 facial and #8 facial and generalized severe decalcification. Generalized inflamed free gingival margins. Bulge on gingivae lingual to #27. Panoramic radiograph shows that #27 is rotated; there is transposition of the # 6 between #4 and #5; developing tooth buds are present for #1 and #16, but not present for #17 and #32.

A - Assessment Phase

Differential Diagnosis

Increased Risk for Dental Caries, a multifactorial trait: Dental caries is a chronic, complex, multifactorial disease. For most patients, many etiologic host and environmental factors play a significant role in risk for caries. Dental morphology is under genetic control; therefore features such as occlusal surface topography, fissure depth, and fissure wall inclination, all have genetic influence. These can increase risk for caries. Tooth size and arch size are also under genetic regulation; dental crowding can increase risk of caries. The composition and development of

dental hard tissues are under genetic control. Several genes have been identified: amelogenin, ameloblastin, and enamelin are a few. Other genes are likely involved as well. There are several well known inherited defects of enamel and dentin development which could greatly increase risk for dental caries and need to be considered. Saliva composition is under genetic control. Gene expression of the many protein components of saliva that play a role in host defense against microbes, electrolyte balance in saliva, and proteins that facilitate tooth mineralization are variable and influence caries risk. Taste function and food preferences are also well described genetically determined traits that can play a role in caries risk. Therefore Steven's rampant caries could simply be due to a combination of diet and genetic factors.

S**jögren's Syndrome (SS):** A common autoimmune disease, that usually presents in middle aged or older patients. It is rare in children. Susceptibility to develop Sjogren's syndrome is familial –multiple genes likely play a role in risk, primary SS is not associated with any other autoimmune disease; secondary SS is associated with other autoimmune disease such as rheumatoid arthritis. The primary clinical features of SS are dry mouth and dry eyes. May present in some as a sudden increase is caries rates of oral burning complaints, rather than complaints of dry mouth. SS is associated with decreased salivary output and altered protein composition of saliva. Other oral complications of SS include oral mucosal sensitivity, oral discomfort, increase oral ulcers with trauma, dysphagia, dysgeusia, increased rate of dental caries, a higher prevalence of missing teeth, recurrent painful salivary gland enlargement and oral candidiasis. There have been cases of familial Sjogren's syndrome reported and rarely it can occur in children.

Aplasia of the lacrimal and major salivary glands (ALSG): is a rare, autosomal dominant disorder that is characterized by aplasia, atresia, or hypoplasia of the lacrimal and salivary glands. Affected patients may have aplasia or hypoplasia or minimal involvement of these glands, as there is variable expressivity, a common feature of autosomal dominant traits. It has been found to be linked to mutations in fibroblast growth factor 10 (FGF10) gene. Symptoms are associated with hypoplasia or aplasia of the major salivary glands include: xerostomia, oral inflammation, dental caries and dental erosion. Other clinical signs of this disorder include atresia of the nasolacrimal duct and absence of the lacrimal puncta.

Lacrimo-ariculo-dento-digital Syndrome (LADD): a rare, autosomal dominant disorder characterized by aplasia, atresia, or hypoplasia of the lacrimal andsalivary systems, cup-shaped ears, hearing loss, and dental and digital anomalies. There is significant phenotypic overlap between LADD syndrome and ALSG. Both the disorders have variable expressivity. These two condition are allelic; both are associated with mutations in FGF10.

Diagnosis

Aplasia of the lacrimal and major salivary glands (ALSG) confirmed by the clinical absence of the parotid and submandibular ducts. MRI can be performed to further confirm the absence of the glands but is generally not deemed necessary, because the ducts must be present for development of the salivary glands to occur. Currently genetic tests for this condition are unavailable.

Discussion

As an autosomal dominant genetic defect ALSG doesn't skip a generation. Steven's children will all have a 50% chance of being affected if his wife doesn't have a history of the disease. For rare disease such as this one, the genetic counselor would assume that any future mate for Steven would NOT carry the gene for the condition, based on the low frequency of this gene in the population. Knowing her status would not generally have significant impact on recurrence risk, because this is an autosomal dominant trait. As such, for every pregnancy, Steven has a 50% chance of passing the affected gene to his offspring. Only one copy of the gene is needed to have the disease. So, the mother's status would not affect this. If by rare chance she also had the disease, then they would have a 25 % chance of having a normal child (0.50 x 0.50 =0.25 represents the chance of having two normal copies of the gene).

The contrast between the features of Steven and the features in his sister demonstrate the variable expressivity of the trait.

P - Treatment Plan

Management

1. Refer to a medical geneticist for confirmation of diagnosis and genetics counseling. This is important because as dentists we are not qualified to provide genetics counseling. Further it is possible that there are other abnormalities signifying some other genetic disorder –need to be sure the diagnosis is correct and that appropriate counseling has been provided. We can help by providing clear description of our clinical findings and diagnoses, and that we have ruled out other causes for the rampant tooth decay.
2. Preventive therapy would include topical fluoride and chlorhexidine mouth rinse and stimulate the minor salivary gland secretions by chewing sugarless gum with xylitol. Restore the lesions and keep the teeth as long as possible, knowing that ultimately he will lose all of his teeth –prognosis is poor for Steven. Implants or dentures would be an option later. His sister –better prognosis because she has functioning major glands. All of the above should be included for her and consider prescribing a medication such as pilocarpine to further stimulate salivary flow. Could consider this also for Steven – not sure how much more saliva he would make but it might be worth a try.
3. Diet counseling- to minimize exposure to fermentable carbs.
4. Social work – they had limited resources to pay for their needed dental treatment. Help them get financial assistance, to fund the care; dentist also has option to do this for free as philanthropy. Another key learning issue - treat the family not just the child!
5. Salivary substitutes and mouth moisturizers – for comfort only. Humidifier in bedroom. Bland toothpaste. No alcohol in mouthwash if possible.

Treatment Outcomes

The sister had cracking at the corners of her mouth consistent with angular cheilitis (candidiasis) this was treated with Nystatin ointment. She had no evidence of intraoral candidiasis.

Two case reports are attached for additional references.

Resources

1. Leung, KCM, McMillan, AS Cheung, BPK, Leung, WK. Sjögren's syndrome sufferers have increased oral yeast levels despite regular dental care. *Oral Diseases.* 2008 Mar;14(2):163-73. http://www.ncbi.nlm.nih.gov/pubmed/18302677?ordinalpos=14&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum

2. Mathews, S.A., Kurien, B.T., and Scofield, R.H. *Oral Manifestations of Sjögren's Syndrome*. *J Dent Res* 2008; 87; 308, DOI: 10.1177/154405910808700411. http://jdr.sagepub.com/cgi/content/full/87/4/308

3. Chapman, D.B., Shashi, V., and Kirse, D.J. International Journal of Pediatric Ortohinolaryngology. *Case report: Aplasia of the lacrimal and major salivary glands (ALSG).* 2009 Jun;73(6):899-901. Epub 2009 Apr 18. http://www.ncbi.nlm.nih.gov/pubmed/19376597?ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum

4. Bretz WA, Corby PM, Schork NJ, Robinson MT, Coelho M, Costa S, Melo Filho MR, Weyant RJ, Hart TC. Longitudinal analysis of heritability for dental caries traits. *J Dent Res.* 2005 Nov;84(11):1047-51.