Johanson-Blizzard Syndrome: A case report

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Abstract

Johanson-Blizzard syndrome is a rare genetic condition that results from a defect on the UBR1 gene located on chromosome 15. This mutation affects multiple organ systems such as the pancreas, central nervous system, nose, ears, thyroid, and oral cavity. This includes dental anomalies such as small, malformed primary teeth and the absence of permanent dentition. This case report consists of medical background and dental considerations regarding the treatment planning of an 8-year 2-month-old female patient with Johanson-Blizzard Syndrome.

Background

JBS was first discovered by Dr. Johanson and Dr. Blizzard in 1971. The defective gene was later identified by Dr. Zenker in 2006.

Etiology/Genetics: This congenital condition is autosomal recessive. The URB1 gene on chromosome 15 is affected. Mother and father carry the gene variant that causes JBS and their child will have a 25% chance of being born with JBS. The UBR1 protein (encoded by the gene) is important for normal pancreatic function. When UBR1 protein is deficient it leads to the physical features of JBS but the mechanism is not well understood at the present time. Diagnosis of JBS is confirmed with genetic testing which is typically recommended with the child's appearance at birth.

Prevalence: It is estimated that JBS happens every 1 in 250,000 births. The distribution is equally male and female.

Craniofacial features: First signs of the condition is often the "beak-shaped" nose present at birth. This is caused by aplastic alae nasi (missing the outer wall of cartilage in the nose). Other craniofacial features include microcephaly, posterior open fontanelle and slanting eyes.

Systemic Symptoms

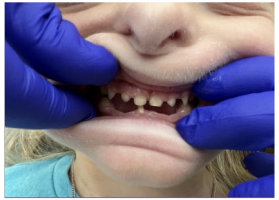
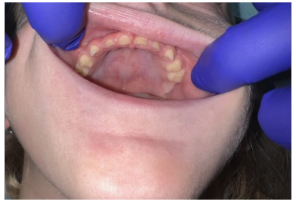
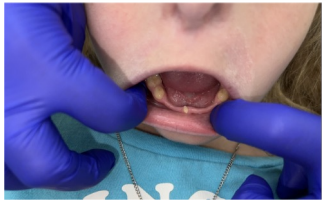
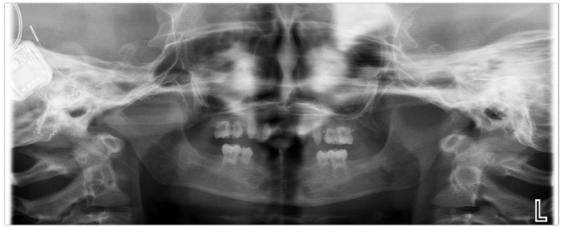
Exocrine pancreatic insufficiency: Insufficient amount of pancreatic enzymes to break down food and absorb nutrients. Treated with pancreatic enzyme replacement therapy.

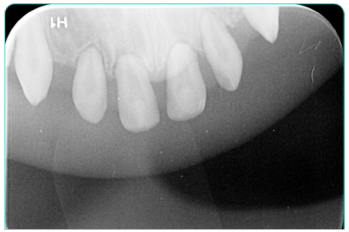
Hypothyroidism: Underactive thyroid resulting in low amounts of thyroxine (T4) and slightly increased levels of thyroid stimulating hormone. This is treated with synthetic thyroxine.

Sensorineural hearing loss: The middle ear is affected in JBS resulting in bilateral hearing loss ranging from severe to profound. Multiple case reports show treatment with cochlear implantation.

Additional symptoms include poor growth, developmental delay, and in 40% of cases imperforate anus.

Case Report



8-year 2-month-old female patient presenting to the University of Michigan school of Dentistry with her mother for new patient exam:

Health history: Johanson-Blizzard Syndrome, deaf (has CI and uses ASL), epilepsy, stage III kidney disease (stable)
Medications: Levothyroxine, Pertzye, Keppra
Social history: She lives at home with mother and father
Education history: She attends first grade. She is on track and learning at an appropriate level.
CC: Mother is aware from physicians that patient will likely be edentulous later in life and wanted to establish care and address best course of treatment
OH: Patient is using crest toothpaste and brushing 1x per day.
Diet: Patient eats meals (minimal snacking) that include meat, noodles, rice, and seafood. She drinks water and capris suns.
Plaque score: 10%
Caries risk: Low - due to no caries risk experience, generalized spacing, and fluoride toothpaste use.

Extraoral examination : Facial presentation consistent with JBS. Microcephaly and aplastic alae nasi. No extraoral swelling.
Intraoral examination: All maxillary primary teeth present as microdonts and non-mobile. Mandibular teeth present include 4 primary molars, and 1 central incisor that is conically shaped. Mandibular teeth are also non-mobile. Dentition has generalized spacing. Soft tissue is WNL. Central area of alveolar ridge is severely deficient in width.
Treatment completed: 1 OCC, 1 Panorex, New patient exam, prophy, fluoride varnish
Behavior: Does well with TSD, patient is very friendly. Patient responds in ASL and mom was translating.
Treatment plan: Continue with 6 month recall in graduate pediatric clinic at the School of Dentistry. Assess primary tooth mobility and tooth loss at future appointments. When patient has functional concerns with dentition recommend working with prosthodontics program to approach fabrication of removable appliances.

Dental Concerns

Common dental presentations of JBS include abnormally small and malformed primary teeth. The permanent teeth can range from hypodontia or complete absence of permanent tooth buds. Permanent dentition can also be misshapen.

Treatment considerations for JBS patients surrounds general anesthesia. Due to higher likelihood for malnutrition or hypoproteinemia, drug distribution can be complicated in these patients. It is also important to assess the thyroid status prior to proceeding with GA.

Conclusion

JBS is a rare genetic condition that can have implications in a patient's systemic health as well as dental health. There are many challenges surrounding definitive treatment for patients with JBS. Agenesis of permanent teeth and in this particular case a thin alveolar ridge and no possibility for abutment teeth make treatment planning complicated. At the present, the information and case studies are lacking in this topic. Many case studies currently show treatment with removable prosthesis. When prosthesis is fabricated frequent recall for adjustment is advised due to patient's growth and development.

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