CASE REPORT
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Oral Findings of a Rare Case of Johanson-Blizzard Syndrome: A Case Report

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Abstract:

Johanson-Blizzard Syndrome (JBS) is a rare congenital disorder characterized by an autosomal recessive inheritance pattern due to a mutation in the ubiquitin protein ligase E3 component n-recognin 1 (UBR1) gene. Notable features of this syndrome include a beak shaped nose, sensorineural hearing loss, hypothyroidism, pancreatic dysfunction, growth disorders, and dental abnormalities such as generalized microdotia and oligodontia. This study presented the case of a 10-year-old girl who visited the dental department with the chief complaint of absence of permanent teeth eruption. The diagnosis of JBS had been established at an early age through genetic testing, which had revealed a mutation in the UBR1 gene. Clinical observations indicated the presence of a beak shaped nose and hearing impairment in the patient. Clinical and radiographic examinations revealed that the patient's primary teeth were present in normal count, but exhibited microdontia. Additionally, only the first permanent molars were formed and visible in the oral cavity, while the buds of other permanent teeth were not observed on radiographic evaluation. Rare syndromes such as Johanson-Blizzard can have significant effects on dental structures, requiring specialized care and treatment planning.

Keywords: Johanson–Blizzard Syndrome; Oral Manifestations; Sensorineural Deafness

Introduction

Johanson-Blizzard syndrome is a rare genetic disorder first described in 1971 by two pediatricians, Johnson and Blizzard [1]. This syndrome affects males and females equally in an autosomal recessive pattern [2].

The genetic cause was not known until 2005. In 2005, the locus associated with Johanson-Blizzard syndrome was localized to chromosome 15q14–21.1, where mutations—primarily truncating types—were discovered in the UBR1 gene across 12 unrelated families. This gene encodes an E3 ubiquitin ligase that is part of the N-end rule pathway, a conserved proteolytic mechanism responsible for targeting proteins with destabilizing N-terminal residues, and functions redundantly with at least three other E3 ligases [3].

The syndrome usually affects structures that develop between the 4th and 8th weeks of gestation[4]. Pancreatic insufficiency is a consistent feature in these patients. Other Symptoms include hypoplasia or aplasia of ala nasi, hypothyroidism, sensorineural hearing loss, midline ectodermal scalp defects, genital malformation, short stature, microcephaly, cardiac abnormalities, and dental defects such as oligodontia [5-8]. People with this syndrome usually have moderate to severe mental retardation, but cases with normal intellect have also been reported [9]. Fewer than 100 cases of this disorder have been reported in the literature to date [10].

This report described oral manifestations of Johanson Blizzard syndrome in a 10-year-old girl.

Case

A 10-year-old female patient diagnosed with Johanson-Blizzard syndrome presented to the Department of Pediatric Dentistry at Shahid Beheshti Dental School, Tehran, Iran for routine dental evaluation. This syndrome had been diagnosed in the child at the age of 11 month. The patient was born at full term following an uncomplicated vaginal delivery to first-cousin parents—her mother was 27 years old and her father 30 at the time of birth. As their eldest child, she represented the family's first encounter with this rare genetic condition. Her 5-year-old sister was healthy with no signs of the syndrome.

At the time of examination, the patient weighed 30 kg and measured 135 cm in height. Her gait and motor development were within normal limits. Speech and comprehension were appropriate for her age, and her academic performance was reported to be good.

Her medical history was significant for congenital sensorineural hearing loss, for which she had undergone cochlear implantation. At the time of examination, she utilized both a hearing aid and cochlear implant with good functional outcomes. Pancreatic dysfunction had also been diagnosed. However, given the mild severity of the condition, only periodic monitoring was recommended and the patient did not receive active pancreatic treatment. No additional systemic abnormalities were documented.

Extra-oral examination demonstrated facial symmetry with normal ocular positioning. Characteristic features of JBS were observed, including a high anterior hairline, thin lips, and nasal alar hypoplasia. These findings were consistent with the established craniofacial phenotype associated with this syndrome (Figures 1 and 2).

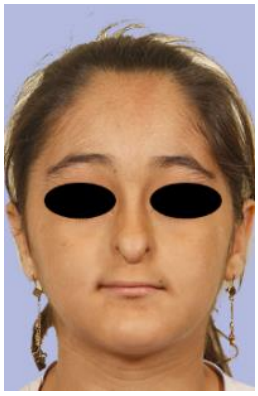

Figure 1: Full face frontal view of the patient

Figure 2: Profile view of the patient

Intra-oral soft tissue examination revealed papillary atrophy of the tongue and a frenal tag of maxillary labial frenum (Figures 3 and 4). Localized gingival erythema was noted in the region of the mandibular left primary first molar (Figure 3).

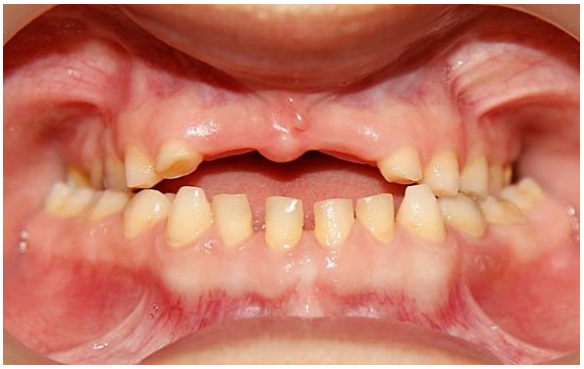

Figure 1: Frontal Intraoral view

Clinical examination of the dentition showed generalized micrognathia affecting both jaws. Congenital absence of the maxillary central incisors was observed. The permanent dentition was limited to the first molars in all quadrants, which exhibited microdontia. The remaining teeth were all decideous. Multiple carious lesions were identified affecting all permanent first molars and several primary molars, specifically the mandibular left primary molars, and maxillary bilateral primary molars (Figures 4 and 5).

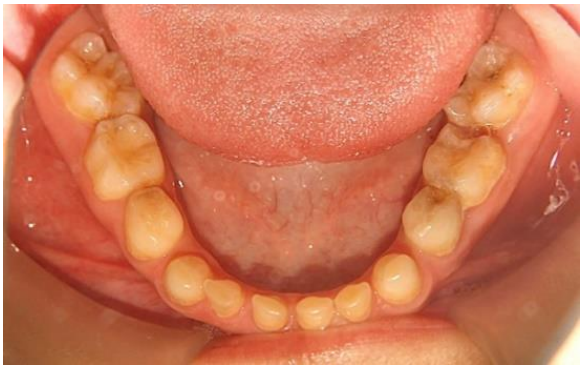

Figure 2: Occlusal view of the lower arch and tongue

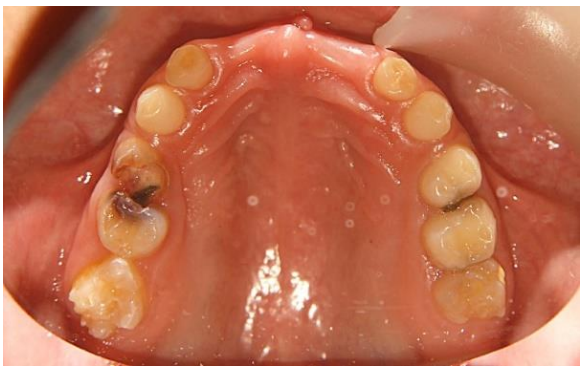

Figure 3: Occlusal view of the upper arch

Panoramic radiographic evaluation confirmed the absence of all permanent teeth except for the first molars. Advanced root resorption was evident in the retained primary teeth. The permanent first molars displayed characteristic microdont morphology. Maxillary and mandibular alveolar bone showed no structural abnormalities (Figure 6).

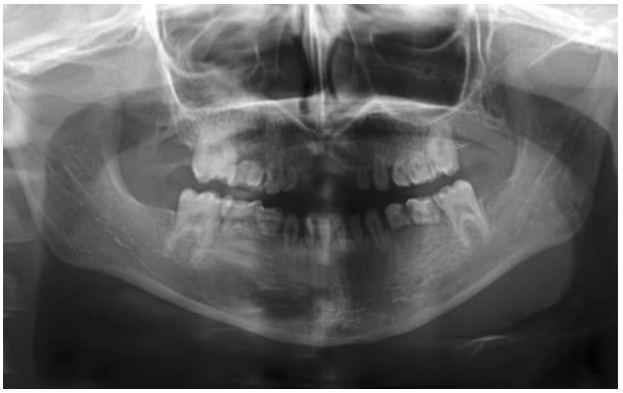

Figure 4: Panoramic radiograph of the patient

Treatment planning prioritized the management of active infections and preservation of viable tooth structure. Extraction of severely compromised primary teeth with extensive root resorption was performed. The carious permanent first molars were treated with direct composite resin restorations. Comprehensive oral hygiene instruction was provided, and a structured recall protocol was established for ongoing monitoring and planning for the future prosthodontic rehabilitation as she continued to grow up.

Discussion

Johanson-Blizzard syndrome is a genetic disorder with an autosomal recessive inheritance pattern caused by a mutation in the URB1 gene. This syndrome is very rare and very few cases have been reported to date. Due to the inheritance pattern, this condition is usually seen in children whose parents are consanguineous [1].

This syndrome has multiple symptoms and affects different areas of the body in different individuals. However, the

common feature in almost all patients with this syndrome is the pancreatic exocrine insufficiency [10, 11].

The diagnosis of Johanson-Blizzard syndrome is usually made at an early age by a pediatrician based on the characteristic features, laboratory, radiographic, and genetic tests.[4] Laboratory tests include Serum amylase, Lipase, Low level of trypsinogen, fat soluble vitamins, and Stool Examination, which help diagnose pancreatic disorders[12,13]. Oral and dental manifestations such as microdontia, missing of primary and permanent teeth, and oligodontia are common features of this syndrome.

In a study conducted by Santhosh et al., an 8-year-old boy with Johanson-Blizzard syndrome was reported with oral manifestations including the papillary atrophy of the tongue and missing permanent mandibular central and lateral incisors [2]. Barroso et al. also reported a case of 18- year-old girl with this syndrome that showed multiple missing of permanent teeth [11].

In the present case, the patient was the first child of consanguineous parents, which naturally increases the risk of developing genetic diseases with an autosomal recessive pattern. This child had been diagnosed at an early age due to the characteristic features of Johanson Blizzard syndrome, such as pancreatic secretion disorder and ala nasi hypoplasia. Other symptoms observed in this child included bilateral hearing impairment, high anterior hair line, and thin lips. Oral signs included papillary atrophy of the tongue, the absence of some permanent tooth buds, and microdontia of the primary teeth. The child had normal intellectual function, which could indicate a milder form of the condition. In this child, the infected and carious teeth with pulp invasion were extracted, but due to the parents' reluctance, prosthetic treatment to restore the dentition was not performed.

Conclusion

Early diagnosis of Johanson-Blizzard syndrome and careful recognition of its associated oral manifestations are essential for comparative long-term treatment planning and successful dental rehabilitation.

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Using AI: AI was not used at any stage of writing this article.

Conflict of Interest: The authors declare no conflict of interest.

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