



Johanson-Blizzard Syndrome: Dental Findings and Management

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ABSTRACT

Aim: Oral rehabilitation of a child with Johanson-Blizzard syndrome (JBS).

Background: JBS is an extremely rare inherited disorder characterized by unusually small nose that appears 'beak shaped' due to the absence (aplasia) or underdevelopment (hypoplasia) of the nostrils (nasal alae), abnormally small, malformed primary (deciduous) teeth and misshapen or absent secondary (permanent) teeth, hearing disorder, hypothyroidism, dwarfism, malabsorption and mental retardation. It is sometimes described as a form of ectodermal dysplasia.

Case report: Oral findings in JBS are very obscure in the literature. The present report describes oral findings in an 8 years old boy with JBS and his oral rehabilitation.

Conclusion: JBS has an emotional consequence for the affected individuals at early ages. Oral rehabilitation in this case had a very positive impact on the child's mind.

Clinical significance: Early identification and treatment of this disease is of great importance to rehabilitate the patient on functional, esthetic and psychological front.

Keywords: Ectodermal dysplasia, Autosomal recessive disorder, Nasal alar hypoplasia.

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INTRODUCTION

Johanson-Blizzard syndrome (JBS) is an autosomal recessive disorder associated with multiple anomalies including aplasia of the alae nasi, deafness, hypothyroidism, absent permanent teeth, scalp defects and pancreatic exocrine insufficiency. Ten or more cases of this syndrome have been reported but only one deals with the oral findings. Details of the oral findings in JBS are therefore obscure.¹

This report describes the oral findings and dental rehabilitation of an 8-year-old boy with this syndrome.

CASE REPORT

The propositus, an 8 years old boy, diagnosed with JBS was referred from Medical Department to the Dental Department for the treatment of decayed teeth. He was the second child of a 23 years old mother and 25 years old father, both healthy and nonconsanguineous. The elder sibling was healthy. The family history was unremarkable. The pregnancy was uncomplicated. This child was diagnosed as having JBS based on the characteristic features like aplastic alae nasi, scanty hair, posterior open fontanelle, slanting eyes, mild amount of sensorineural defect (deafness), slightly depressed T₄ levels.

On first examination, the height measured 116 cm and body weight 15 kg. His speech and ability to understand were good. As for the facial area, the right and left eyelids slanted and extended toward the midline (Fig. 1). The radix



Fig. 1: Slanting eyes, depressed nasal bridge, hypoplastic alae nasi

nasi was flat (Fig. 1). The parieto-occipital region of the skull was elevated and in the center of the vertex a scalp defect was apparent (Fig. 2).

Intraorally, soft tissue examination showed atrophy of papillae on dorsum of tongue (Fig. 3) and angular cheilitis (Fig. 4). Hard tissue examination gave a typical picture of rampant caries with majority of the deciduous teeth been affected by caries. All the first permanent molars were erupted and were caries free, with normal, sound



Fig. 2: Posterior open fontanelle



Fig. 3: Atrophy of papillae of tongue



Fig. 4: Angular cheilitis

morphology. The other permanent teeth erupted were maxillary central incisors (11, 21). The right and left maxillary and mandibular deciduous molars and canines were present. All the mandibular deciduous molars (74, 75, 84, 85) showed pulpal involvement; whereas the maxillary right first molar (54) showed deep caries approaching the pulp. The mandibular canines were conical in shape. The right deciduous mandibular canine had extensive mesial surface caries. Maxillary deciduous lateral incisors were also carious and the right lateral incisor was associated with preshedding mobility. At this dental age (7-8 years), permanent mandibular incisors were unerupted. As for the mandibular deciduous incisors (71, 72, 81, 82), only carious root stumps of those remained which were mobile. This clinical finding gave an assumption for congenitally missing permanent mandibular incisors.

Radiographic (OPG) examination (Fig. 5) confirmed the missing mandibular permanent central and lateral incisors (31, 32, 41, 42). The maxillary arch was normal in size while the mandibular arch was contracted. The molar relation recorded bilaterally was Angle's class I type.

Treatment began with pulp therapy of mandibular deciduous molars (74, 75, 84, 85) and maxillary right first deciduous molar (54). Stainless steel crowns were then placed over them as a postoperative restoration. Other carious teeth were restored with glass ionomer cement. Extraction of mandibular deciduous incisor root stumps were done under local anesthesia. As rehabilitative care for this patient was concerned, a modified partial denture/a functional space maintainer was fabricated for the mandibular anterior region (Fig. 6). This partial denture was fixed in the mandibular arch by soldering the wire component to the crowns on deciduous second molars and stabilizing the anterior acrylic part with adjacent teeth with the help of composite placed lingually (Fig. 7).

DISCUSSION

As far as we are aware, JBS is a rare disease with few detailed reports on oral findings in the literature.¹ Cases



Fig. 5: OPG showing missing permanent lower anteriors and pulpally involved deciduous posterior molars



Fig. 6: Anterior space maintainer soldered to the crowns on posterior teeth



Fig. 7: Anterior space maintainer



Fig. 8: Scalp defect



Fig. 9: Beak-shaped nose

reported by Johanson and Blizzard in 1971 as well as a case of 6 years old female reported in Gunma University, Japan demonstrated classical features of aplastic alae nasi, ectodermal defects of scalp, slanting eyes, deafness, absent permanent tooth buds.^{2,3} The appearance of our patient bears a striking similarity with those reported by other authors. This gives us a clear picture that our patient also manifested the classical features of JBS.³ Since, the manifestations of this case did not include mental development/retardation and speech defect, it represented a milder form of this complex of abnormalities.

Superficially the patients resemble individuals who have oculodentodigital dysplasia. Characteristics of that syndrome are hypoplasia of nasal alae, deafness, normal IQ. Other features not manifested by our patients are syndactyly, enamel hypoplasia.²

The ectodermal scalp defects can be best described as 'aplasia cutis' (Fig. 8). This is usually an isolated, congenital absence of skin, most often on the scalp and occasionally is associated with more generalized mesodermal and ectodermal hypoplasia.²

An obvious feature of this case was the hypoplastic alae nasi, (Fig. 9), the morphogenetic explanation of which is far from been straightforward. This malformation does not

correspond to any of the known facial furrows seen during embryonic life. The anomaly has therefore been described as a failure of formation of the entire mesenchymal wall around the olfactory pit.³

The etiology of this syndrome probably seems to be genetic in origin (OMIM # 243800) having an autosomal recessive mode of inheritance (UBR1 gene).⁴ This gene is probably extremely rare within the general population. Since, there has been no case of nasal defect, deafness, slanting eyes among the relatives of our patient, a dominant mode of inheritance is unlikely.³

Treatment of JBS which is a type of ectodermal dysplasia requires knowledge of growth and development, behavior management, fabrication of prosthesis, motivation of patient and parent with long-term follow-up. In our patient, full mouth rehabilitation was done. Absence of tooth buds is one of the major factors of JBS and is almost always present. Our patient had missing permanent mandibular incisors. Therefore, we planned prosthetic treatment using removable partial denture. It was modified by fixing the wire component to the crowns on mandibular second deciduous molars and stabilizing the acrylic portion anteriorly by placing composite lingually (Fig. 7). It had an objective of improving esthetics and act as a provisional type of treatment care. This partial denture was of great value from psychological point of view. After the prosthetic treatment, the patient was very happy with his appearance.

Periodic recall checkup is an essential step in treating these patients. Replacement/modification of prosthesis is required due to continued growth and development of jaws and eruption of permanent teeth.

CONCLUSION

JBS has an emotional consequence for the affected individuals at early ages. So, it is important to identify and

treat such individuals at an early stage. Oral rehabilitation in this case had a very positive impact on the child's mind.

CLINICAL SIGNIFICANCE

- Early identification and treatment of this syndrome
- Rehabilitating patients on functional, esthetic and psychological front
- Planning periodic recall to modify/replace prosthesis.

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