(International Print/Online Journal)

SJIF IMPACT FACTOR: 5.565
PUBMED-National Library of Medicine ID-101739732

ISSN (Print): 2209-2870
ISSN (Online): 2209-2862

International Journal of Medical Science and Current Research (IJMSCR)
Available online at: www.ijmscr.com
Volume 5, Issue 1, Page No: 340-343
January-February 2022

IJMSCR

Oral Rehabilitation of a Pediatric Patient with Johanson Blizzard Syndrome – A Case Report

[1]Dr. Bisma Saher Khan, [2]Dr. Jyothsna V Setty, [3]Dr. Anand M, [4]Dr. Janani Rajesh, [5]Dr. Syeda Ayesha, [6]Dr. Ila Srinivasan

[1,4,5]Post Graduate Student, [2]Professor, [3]Reader, [6]Professor and Head,
[1,2,4,6]Department of Pedodontics and Preventive Dentistry, [3,5]Department of Prosthodontics,
M.R. Ambedkar Dental College and Hospital, Bengaluru

***Corresponding Author:**
Dr. Bisma Saher Khan
Post Graduate Student, Department of Pediatric and Preventive Dentistry
M.R. Ambedkar Dental College and Hospital, Bengaluru, Karnataka, India-560005

Type of Publication: Case Report
Conflicts of Interest: Nil

Abstract

Johanson-Blizzard syndrome (JBS) is an extremely rare genetic disorder that affects multiple organ systems of the body and presents with distinct features namely, pancreatic insufficiency, hypothyroidism, growth retardation, intestinal malabsorption, midline ectodermal scalp defects, and defects with permanent teeth. In this case report, we present the dental management of a 7 year old patient suffering from JBS who presented with features of poor weight gain, anasarca secondary to hypoproteinemia, severe anemia, sensorineural hearing loss and speech impairment secondary to the hearing loss, microcephaly, frontal upsweep of hair, sparse and coarse scalp hair with areas of patchy alopecia, prominent eyes with long eyelashes, small beak-shaped nose with alar hypoplasia, inferiorly set ears, thin and upturned upper lip. Intraoral presentation was that of hypodontia with only conical primary canines and grossly decayed primary 2nd molars being present. Extraction of the grossly decayed primary second molars was followed by full mouth rehabilitation using a canine supported acrylic interim partial denture to restore esthetics and masticatory function. This treatment improved the child's morale immensely as well. Given the young age and actively growing phase of our patient, replacement/ modification of prosthesis is required to accommodate for the continued growth and development of jaws. Recall and review is hence being conducted regularly every 2-3 months.

Keywords: Johanson Blizzard syndrome, Hypodontia, Interim denture.

Introduction

Johanson-Blizzard syndrome (JBS) is an extremely rare genetic disorder that affects multiple organ systems of the body which is estimated to be found in 1 in 250,000 live births. It was described by Johanson and Blizzard in 1971 and Park et al. in 1972 in 3 unrelated girls[1].The features noted by them included, nasal (alar) aplasia or hypoplasia, congenital deafness, exocrine pancreatic insufficiency, hypothyroidism, growth retardation, intestinal malabsorption, mental retardation, midline ectodermal scalp defects, and defects with permanent teeth.Treatment measures include oral administration of pancreatic enzymes; surgery for genitourinary and anorectal malformations; hearing aids for hearing impairment; specific education and early intervention programs for patients with intellectual disabilities and dental implantation during teenage years. Genetic counselling will also be of benefit to affected individuals and their families.

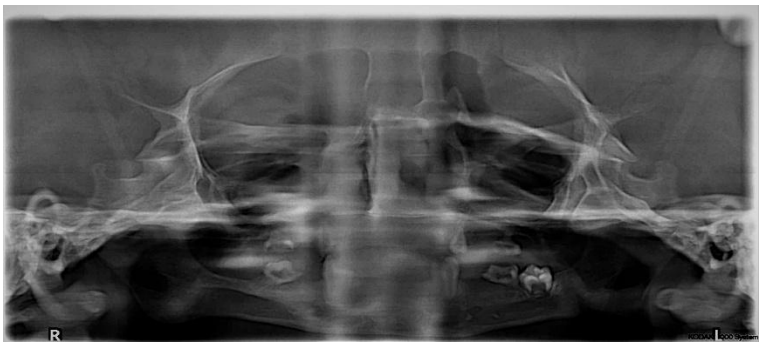
Case Report

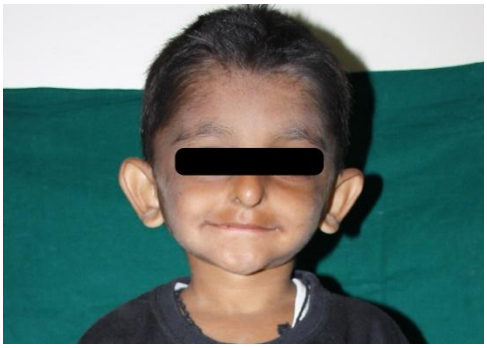
A 7-year-old male patient reported to the Department of Pedodontics and Preventive Dentistry with the

chief complaint of missing upper and lower front teeth since birth. A careful examination of his family history revealed that, he is the 4th child of a second-degree consanguineous marriage. His parents reported that their 1st child is a 10-year-old healthy female, the 2nd conception had resulted in a miscarriage at 8 months of intrauterine life and the 3rd was a male child who had been suspected/diagnosed of JBS and succumbed to the disease at 3 months of age on account of the overwhelming sepsis. The present patient who is the 4th child weighed 2.8 kg at birth. He was found to have an imperforate anus and hence a transverse colostomy was done on the 2nd day of life. His serum amylase and pancreatic lipase levels were found to be lower than normal limits. The patient was hence administered fat soluble vitamins and pancreatic enzyme replacement therapy. The child had the following features of poor weight gain, anasarca secondary to hypoproteinaemia, severe anaemia, sensorineural hearing loss and speech impairment secondary to the hearing loss. However no abnormalities were observed with the MRI of his brain and his thyroid profile. Karyotyping also revealed the normal 46XY. He had signs of global developmental delay and also presented with delay in skeletal maturation. He was started on regular physiotherapy along with early intervention programs to correct and treat his developmental delay. The child was advised to use bilateral hearing aids for the hearing impairment. The general physical examination of the patient revealed pallor and hypotonia, and a frail frame measuring 80 cms in height and weighing 8.1 kgs in weight. Extra oral examination showed some of the striking features of JBS namely, microcephaly, frontal upsweep of hair, sparse and coarse scalp hair with areas of patchy alopecia, prominent eyes with long eyelashes, small beak-shaped nose with alar hypoplasia, inferiorly set ears, thin and upturned upper lip. Intraoral presentation was that of hypodontia with only conical primary canines and grossly decayed primary 2nd molars being present. The orthopantomogram revealed the absence of all tooth buds except the left mandibular permanent first molar. A provisional diagnosis of Johanson Blizzard syndrome was arrived at considering these findings. Differential diagnoses considered were that of Shwachman-Diamond syndrome, Pearson Marrow-Pancreas syndrome.

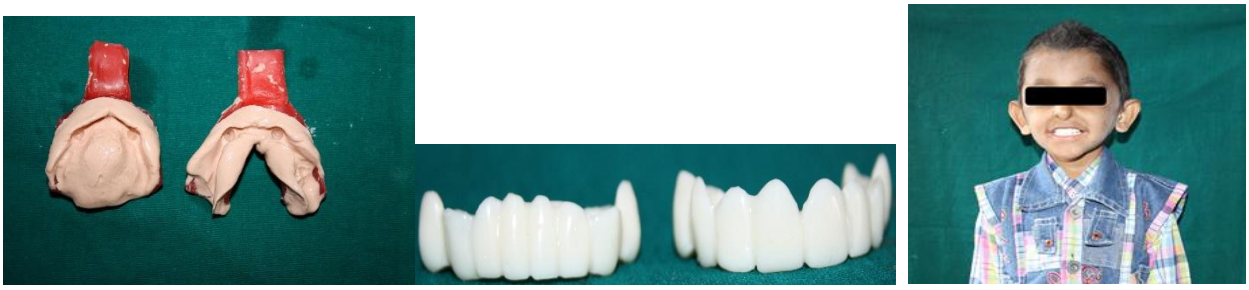
Management

Extraction of the grossly decayed deciduous second molars was done after obtaining of the treating physician's consent for the same. On follow up visits, eruption of the left mandibular first permanent molar was noticed. The treatment plan was devised with the goal of alleviating the patient's complaints regarding appearance, phonetics and nutrition. A canine supported acrylic interim partial denture was planned with the aim of improved esthetics, functional rehabilitation and better quality of life. Customized impression trays for both the arches were made using modeling wax. Bite registration was done and the casts were mounted on to an articulator. An interim partial denture extending from canine to canine was fabricated. Canines were not prepared to avoid the risk of pulpal exposure owing to their conical shape. The prosthesis was first cemented using temporary cement (non-eugenol zinc oxide based cement) in order to evaluate the compliance of the child patient. After a follow up period of two weeks, the denture was cemented using Type I Glass ionomer cement. Recall and review is being conducted regularly every 2-3 months.





Volume 5, Issue 1; January-February 2022; Page No 340-343



Discussion

Johanson Blizzard syndrome is a rare autosomal recessive disorder which affects many body systems with a broad range of congenital anomalies. A small beak-shaped nose and exocrine pancreatic insufficiency are considered to be pathognomonic of Johanson Blizzard syndrome, while other features may occur at differing frequencies in the affected patients.

Our patient had striking similarities with those reported in literature and hence the diagnosis of JBS was arrived at. However, as he did not present with any significant mental retardation, this can be categorized to be a milder form of the syndrome complex.

As literature states, JBS is a rare disease with few detailed reports on oral findings in the literature. Dental abnormalities may be treated with bonding agents, use of fixed or removable prosthesis along with the other supportive techniques.

In our patient, full mouth rehabilitation was done with an objective of improving esthetics and masticatory function. Along with this function it was of great psychological benefit for both the patient and the parents. After the prosthetic treatment, the patient was very happy with his appearance. Given the young age and actively growing phase of our patient, replacement/ modification of prosthesis is required to accommodate for the continued growth and development of jaws.

Conclusion

Johanson–Blizzard syndrome requires early identification and treatment. The treatment should focus not only on the specific symptoms that present in each individual, but also on the general well-being and health of the patient so as to lead a comfortable life. It 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.

References

1. Johanson A, Blizzard R. A syndrome of congenital aplasia of the alae nasi, deafness, hypothyroidism, dwarfism, absent permanent teeth, and malabsorption. J Pediatr. 1971;79(6):982-7.
2. Ono K, Ogawa T, Matsuda N. Oral findings in Johanson Blizzard syndrome. J Oral Med 1987;42(1):14-6.
3. Helin I, Jodal U. A syndrome of congenital hypoplasia of the alae nasi, situs inversus and severe hypoproteinemia in two siblings. J Pediatr 1981;99(6):932-4.
4. Alkhouri N, Kaplan B, Kay M. Johanson-Blizzard syndrome with mild phenotypic features confirmed by UBR1 gene testing. World J Gastroenterol 2008;14(44):6863-6.
5. Santhosh BP, Jethmalani P. Johanson-Blizzard syndrome: dental findings and management. J Contemp Dent Pract. 2013;14(3):544-7.
6. Asha GS, Shree KK, Revathi TN, Shilpa K. Kin with no skin: Johanson–Blizzard syndrome in siblings: A rare association of aplasia cutis congenita. Indian J Paediatr Dermatol. 2021;22(4):370.

Legends

Figure 1: OPG

Figure 2: Pre operative image of the patient

Volume 5, Issue 1; January-February 2022; Page No 340-343

Figure 3: Alginate impression made from customized trays

Figure 4: An interim partial denture extending from canine to canine fabricated using cold cure acrylic resin

Figure 5: Post operative image of patient with denture

Volume 5, Issue 1; January-February 2022; Page No 340-343