Asian Journal of Pediatric Research

Volume 14, Issue 12, Page 62-67, 2024; Article no.AJPR.127786
ISSN: 2582-2950

Mandibular Reconstruction in a 3 Year Old Patient with Marden Walker Syndrome

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

Article Information

DOI: https://doi.org/10.9734/ajpr/2024/v14i12410

Open Peer Review History:
This journal follows the Advanced Open Peer Review policy. Identity of the Reviewers, Editor(s) and additional Reviewers, peer review comments, different versions of the manuscript, comments of the editors, etc are available here: https://www.sdiarticle5.com/review-history/127786

Case Report

Received: 12/10/2024
Accepted: 14/12/2024
Published: 17/12/2024

ABSTRACT

Introduction: Marden Walker Syndrome is a rare genetic disorder with distinctive craniofacial features, primarily characterized by micrognathia or a small jaw.
Patients' affected by this condition may undergo corrective surgical procedures in order to manage their deformities and improve their overall quality of life.
Case Presentation: A 3-year-old female, known case of Marden Walker Syndrome with severe micrognathia, was admitted for elective mandibular reconstruction surgery. The surgery involved bone grafting with microvascular anastomosis and subperiosteal implantation. Postoperatively, the patient experienced respiratory complications, requiring intubation and subsequent admission to the

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Cite as: *Amir, Roumman, Maryam Haitham Salman, Hana Iqbal, Masiha Tasneem Banu, and Farheen Khan. 2024. "Mandibular Reconstruction in a 3 Year Old Patient With Marden Walker Syndrome". Asian Journal of Pediatric Research 14 (12):62-67. https://doi.org/10.9734/ajpr/2024/v14i12410.*

Pediatric Intensive Care Unit (PICU) for 16 days. She was managed with mechanical ventilation, extubation trials and gradual weaning to room air. Following stabilization, she was successfully extubated and discharged home. The patient is currently receiving follow up with the pediatric plastic surgeons and otolaryngologists.

Conclusion: This case illustrates the challenges faced in managing rare genetic conditions and complex craniofacial anomalies in pediatric patients.

Multidisciplinary care including surgical expertise, respiratory support, and vigilant monitoring, is crucial to achieve successful outcomes in such cases.

Keywords: Marden walker syndrome; craniofacial anomalies; genetic disorder; micrognathia; mandibular reconstruction surgery.

1. INTRODUCTION

Marden Walker Syndrome is a rare genetic autosomal recessive disorder characterized by distinctive craniofacial features, notably micrognathia or a small jaw. In 2014, McMillin et al. identified a de novo heterozygous mutation in the PIEZO2 gene in a patient with Marden Walker Syndrome. These patients present with distinctive facial characteristics including an abnormal jaw structure, droopy eyelids, a flat nasal bridge, low set ears and a fixed facial expression. Additional symptoms may encompass microcephaly, heart irregularities, anomalies in the sexual and urinary systems, osteoporosis, pectus excavatum or carinatum, preauricular tag, microphthalmia, a short neck, a small mouth and a low hairline (INSERM US14. (n.d.), U.S. Department of Health and Human Services. (n.d.), Jancar 1985, Garavelli et al. 2000, Williams et al. 1993).

In 1966, Marden and Walker described an infant displaying blepharophimosis, micrognathia, immobile facial features, kyphoscoliosis, limb contractures, pigeon- breasted appearance, arachnodactyly and microcystic kidney disease, however, unfortunately, the infant passed away at 3 months of age. This case bore some similarities to the siblings with myotonic myopathy described by Aberfeld et al. in 1965 (Vincent & Hohman 2023, Baskaran 2017.

Managing and treating these patients necessitates multidisciplinary approach from medical and surgical specialities and usually require long term follow ups due to the complexity of this syndrome.

2. CASE PRESENTATION

A case of a 3-year-old female, diagnosed with Marden Walker Syndrome. She has a history of bilateral talipes equinivoras, which was surgically treated. She underwent tenotomy and is wearing splints, boots and bars to assist with ambulation. She presented with severe micrognathia and hypoplastic mandible with missing paramedian segment and hypoplastic lateral segments. She came for elective free fibula mandibular reconstruction surgery in order to reconstruct her jaw and restore her facial structure. She successfully underwent mandibular reconstruction with bone grafting, microvascular anastomosis and subperiosteal implantation, as seen in Fig. 1 and Fig. 2.

However, she faced postoperative complications mainly related to airway compromise and severe respiratory acidosis, which led to her admission in the Pediatric Intensive Care Unit (PICU) for 16 days.

On admission to the PICU, her arterial blood gas showed pH of 7.254, PCo2 of 56.3 mmHg and HCO3- 22.5 mmol/L, indicating respiratory distress requiring intensive monitoring.

During her PICU stay, she underwent a series of interventions to address her respiratory distress. She was intubated and mechanically ventilated initially to manage her respiratory distress. On Day 9 in PICU, a trial of extubation to high- flow nasal cannula (HFNC) was attempted, but she developed severe stridor, respiratory acidosis, and desaturation, which didn't improve with supportive measures. As a result, she was re-intubated and managed with mechanical ventilation using PC-AC mode with VG (volume guaranteed), with settings including tidal volume (VT), respiratory rate, and PEEP (positive end-expiratory pressure) to ensure adequate ventilation and prevent airway collape. After a period of sedation and mechanical ventilation, she exhibited promising respiratory progress. Several days postoperatively, the patient transitioned from high-flow nasal cannula (HFNC) to room air. By Day 16 in PICU, she showed significant improvement with no stridor, good respiratory effort, and tolerated room air well.

Despite her complex condition and recent surgery, her vital signs remained stable, and her general appearance was reassuring with no signs of acute distress.

On discharge, her parents were advised gradual advancement of her diet, from pureed smooth foods, progressing to soft solids and to avoid foods that require chewing. Future follow ups of the patient revealed the donor site to be completely healed with no complications. Moreover, the profile of her jaw was very superior and satisfactory. She had started eating solid foods, her speaking and opening of mouth had improved. The patient continued to have subsequent follow ups.

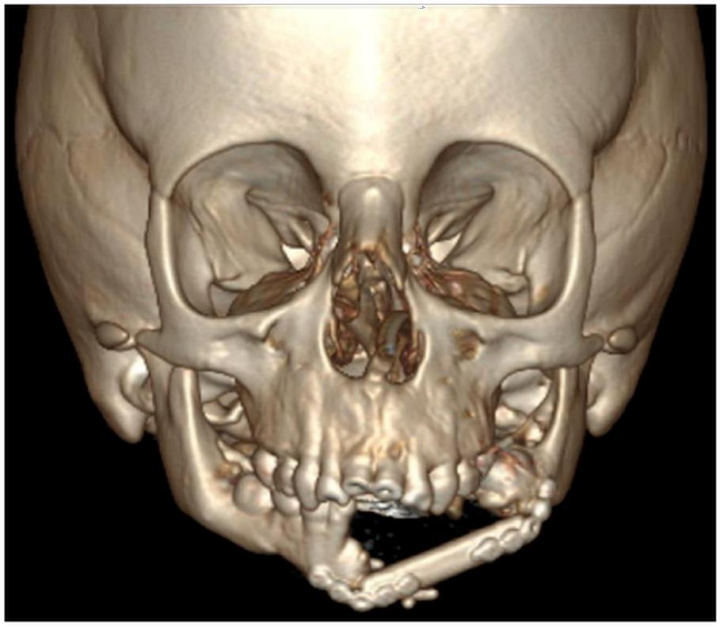

Fig. 1. 3D image of post mandibular reconstruction revealing the left mandibular bone graft

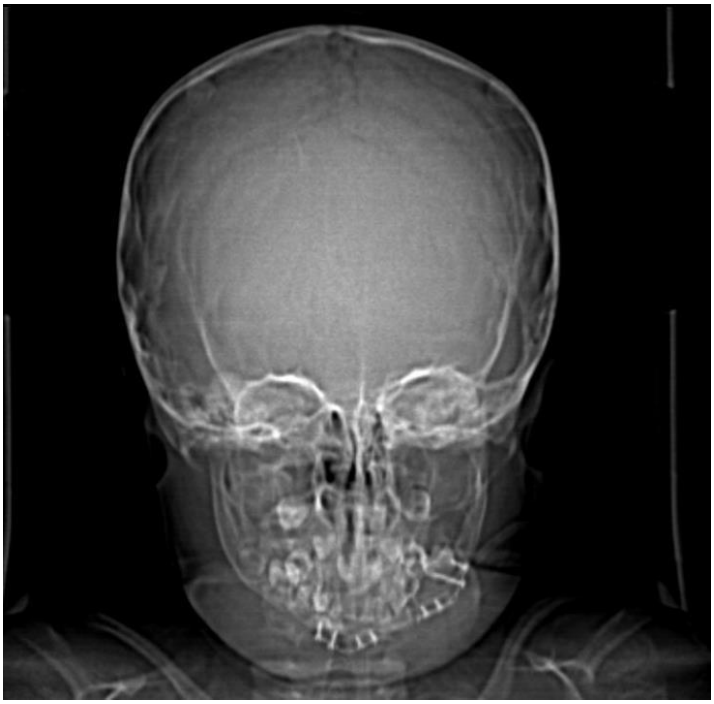

Fig. 2. Status post mandibular reconstruction. Bone graft is in place. Surgical staples are seen

3. CASE DISCUSSION AND LITERATURE REVIEW

Marden Walker syndrome is a rare connective tissue disease that is inherited as an autosomal recessive pattern**.** The first description of the syndrome was suggested by Ealing in 1944, but it was more formally recognized in 1966 by Marden and Walker, who characterized the disorder and its key features (King & Magenis 1978). Patients usually exhibit a range of deformities, including craniofacial abnormalities such as micrognathia, blepharophimosis, masked faces, and high-arched palate, as well as

joint contracture, growth retardation, and failure to thrive. These patients often require corrective surgical procedures to restore normal anatomy and reestablish the function of the affected organs (INSERM US14. (n.d.), Jancar 1985, Garavelli 2000, Williams et al. 1993). A review by Begum & Nayek (2002) further explores the clinical spectrum of Marden Walker syndrome, emphasizing the variability in its presentation and the importance of early intervention to manage the associated deformities (Begum & Nayek 2002).

Our patient presented with severe micrognathia and hypoplastic mandible with missing paramedian segment and hypoplastic lateral segments. She was scheduled electively for free fibula mandibular reconstruction surgery in order to reconstruct her jaw, restore her facial structure, and improve her overall health, quality of life and well-being.

The mandible has crucial functions including mastication, oral phase of swallowing, speech, dental occlusion and cosmesis of the face which is necessary for everyday living. Severe defects in the mandible, micrognathia as an example, can disturb the patients' quality of life in various ways.

Micrognathia also known as small jaw, is when the mandibular or maxillary skeleton does not grow to the normal required size, thus resulting in upper airway complications. Majority of children born with micrognathia are either asymptomatic or can be treated conservatively with prone positioning and nasopharyngeal airways. Few patients, however, may outgrow their micrognathia with time. On the contrary, children having micrognathia with severe upper airway obstruction, require mandibular reconstruction surgery (Kimple et al. 2014, Dodge 1965, McMillin et al. 2014, Mandell et al. 2004, Denny et al. 2001).

Mandibular reconstruction surgery is when an attempt is made to advance the tongue base anteriorly via its muscular attachments to the distracted mandible, thus pulling the tongue out of the hypopharynx and relieving upper airway obstruction (Denny & Kalantarian 2002).

Nevertheless, each surgery has its own complication and few of them include, penetration of the floor of the mouth with a pin or loosening of a pin after a fall, development of an abscess at the pin site, inadequate distraction requiring a second procedure and facial scarring requiring revision (McCarthy et al. 1992).

Post operatively, our patient suffered from severe stridor with respiratory acidosis and desaturation which did not improve even when supportive measures were taken. Therefore, the patient was re-intubated and kept in the PICU. Fortunately, our patient improved over the course of her stay, as later she maintained saturation on room air.

A possible reason for developing the respiratory distress in our patient, could be attributable to the surgical alteration of her facial and jaw structures that could have induced temporary airway compromise. This necessitated vigilant, specialized anesthesia techniques and postoperative care to address and subsequently mitigate the respiratory distress effectively.

4. CONCLUSION AND SUMMARY

In Marden Walker syndrome, it is crucial to understand that complications can vary from one individual to another and are often influenced by the severity of the craniofacial abnormalities, the surgical approach, and the overall health of the patient. Patients undergoing mandibular reconstruction should be closely monitored in the postoperative phase and receive ongoing multidisciplinary care, including orthodontic, speech, and occupational therapies, as well as psychological and medical support to address any complications and ensure long- term recovery.

This case report is significant for the scientific community as it contributes valuable insights into the management of a rare and complex genetic disorder. The detailed surgical approach, postoperative care strategies and challenges discussed here are not only beneficial for clinicians faced with similar cases but for the broader medical community including future generations of healthcare professionals.

Moreover, it underscores the importance of multidisciplinary approach to treatment, which is essential in addressing the multifaceted needs of patients with facial asymmetry and related conditions. This report offers practical guidance and helps raise awareness about the complexities involved in the treatment of Marden Walker Syndrome, ultimately contributing to better patient outcomes and fostering ground for further research into rare genetic disorders.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declare that generative AI technologies such as Large Language Models, etc have been used during writing or editing of this manuscript. This explanation will include the name, version, model, and source of the generative AI technology and as well as all input prompts provided to the generative AI technology.

Details of the AI usage are given below:

AI technology (OpenAI GPT-4, version 4.0) was used solely for text refinement, focusing on improving grammatical structure and punctuation, with no content generation or substantial modifications made.

DATA AVAILABILITY STATEMENT

All data generated during this case report study are included in this article. Further inquiries can be directed to the corresponding author.

ETHICAL APPROVAL

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

CONSENT

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Peer-review history:
The peer review history for this paper can be accessed here:
https://www.sdiarticle5.com/review-history/127786