Case report

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

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. However, following the operation, she experienced respiratory complications that required her to be intubated. Fortunately, with improvement of her respiratory condition, she was successfully extubated and discharged home few days later. 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

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 [1-5].

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 [6-7].

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.

Case Presentation

Our case is a 3-year-old female, diagnosed with Marden Walker Syndrome. She has history of bilateral talipes equinivoras which was surgically treated. She underwent tenotomy and is wearing splints, boots and bars that help her in ambulating. 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 figure 1 and 2.

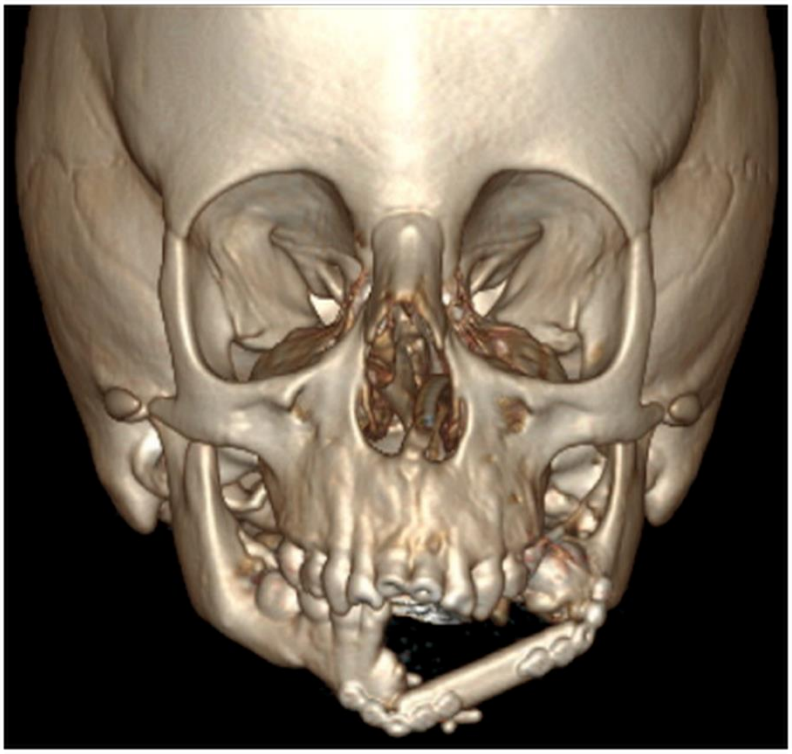

Figure 1 Figure 1. 3D image of post mandibular reconstruction revealing the left mandibular bone graft

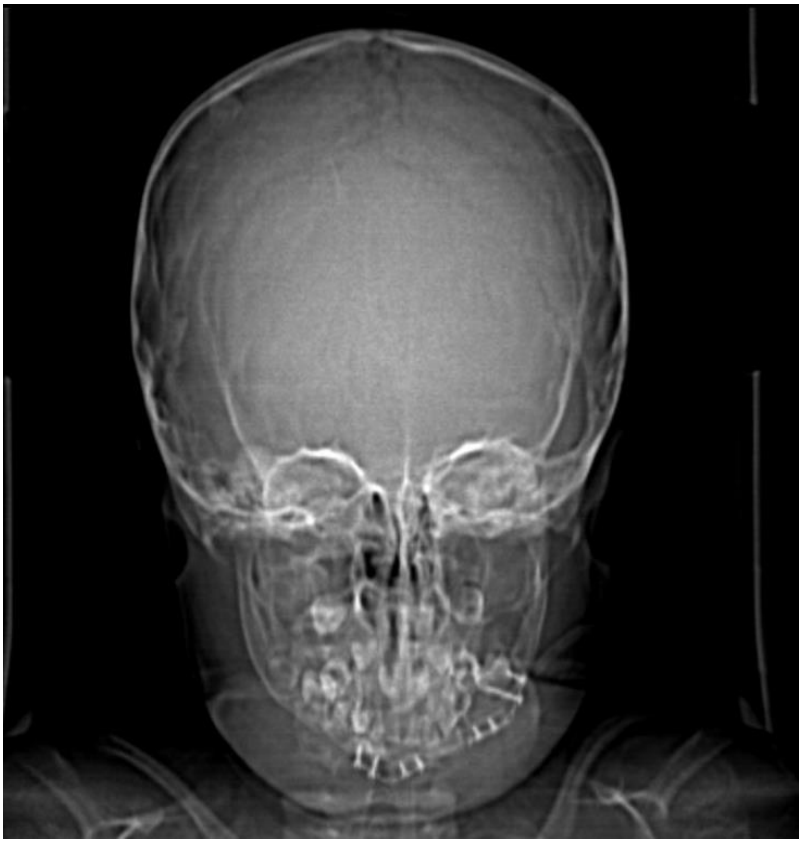

Figure 2 Status post mandibular reconstruction. Bone graft is in place. Surgical staples are seen

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).

Post operative arterial blood gas showed PH- 7.254, PCo2- 56.3 mmHg and HCO3- 22.5 mmol/L.

During her PICU stay, she underwent a series of interventions to address her respiratory distress. After a period of sedation and mechanical ventilation, she exhibited promising respiratory progress. Several days after operation, the patient transitioned from high-flow nasal cannula to room air, marking significant improvements. Despite her complex condition and recent surgery, her vital signs and general appearance were reassuring and she displayed no signs of acute distress.

On discharge, her parents were advised gradual advancement of her diet, from puree smooth 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.

Case Discussion and Literature Review

Marden Walker syndrome is a rare connective tissue disease that is inherited as an autosomal recessive pattern. In 1944, Ealing described an infant who may have had a description of Marden-Walker syndrome. It was then discovered in 1966 by Marden and Walker. Patients usually have multiple deformities, ranging from craniofacial abnormalities for instance micrognathia, blepharophimosis, masked faces and high arched palate to joint contracture, growth retardation and failure to thrive. Therefore, more often than not, they are required to undergo corrective surgical procedures to restore normal anatomy and reestablish the function of the respected organ [1][3-5].

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 [8-12].

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 [13].

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 [14].

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.

Summary and Conclusion

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. The patients who undergo 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 follow them up in the long-term.

Statement of Ethics

Written informed consent was obtained from the patient for publication of the details of their medical case and any accompanying images. Ethical approval is not required for this study in accordance with Dubai Health.

Conflicts of Interest Statement

The authors have no conflicts of interests to declare.

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.

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