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CASE REPORT

Prosthodontic presurgical treatment of midline facial cleft in West Syndrome Infant

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Keywords: Midline facial cleft, Modified PNAM, West syndrome

ABSTRACT

Midline facial cleft is extremely rare congenital malformation. It may be associated with other syndromes, including West syndrome (WS), a severe form of epilepsy in infancy. This report presents modified Presurgical Nasoalveolar Molding (PNAM) treatment of midline facial cleft in WS patient. An infant girl born with midline facial cleft, characterized by flat nose due to complete absence of nasal cartilage, columella, and premaxilla. She was also diagnosed with WS, hydrocephalus, and cerebral palsy. PNAM treatment was performed, consisting of feeding plate and extraoral nasal hook. The feeding plate prevented tongue protrusion which could interfere the airway during multiple spasms and trained her to place the tongue in its normal position. The extraoral strapping was retracted across the upper lip and feeding plate was grinded selectively to approximate the lip and alveolar segment concurrently. An elastic band was paired on nasal hook and attached to the forehead to elevate nasal dome and maintain the airway during retraction. After 1 year follow-up, alveolar gap was reduced by 4,5mm, lip segment moved closer to the midline, and bodyweight increased sufficiently. Modified PNAM can help infant with WS breath, increase nutritional intake and mold the alveolar tissue properly. (IJP 2024;5(1):78-82)

INTRODUCTION

Craniofacial clefts are extremely rare congenital malformation compared with the orofacial cleft (cleft lip and/or cleft palate), with prevalence less than 1 per 100.000 births or only 0,4% to 0,7% among cleft population.[1] It is important to distinguish between facial cleft and ordinary cleft lip. The exact etiology of craniofacial cleft is still unclear, but presumably it appears to be as a result of the genetic predisposition, neural crest central disorganization, mutations during craniofacial embryogenesis, and the impact of environmental risk factors, with 4 major category of risk factors: radiation, infection, alcohol and drug use during pregnancy and maternal metabolic imbalance.[2-4]

According to the time of the embryological accident, clinical manifestations are variable. The malformation may concern brain, bone, and soft tissues either together or isolated.[5] In 1976, Tessier described the numeric classification for rare craniofacial clefts into 0 to 14, based on anatomical position of the cleft.[1]

Tessier number 0 is the most common type of rare craniofacial clefts.[4,5] It is a median craniofacial dysgraphia and also referred as midline facial cleft (MFC). MFC can be associated with other congenital defects and central nervous system (CNS) malformations. Patient with MFC presenting agenesis of the premaxilla, may be suspected of being associated with holoprosencephaly (HPE) sequence.[4] Infant with MFC also may be attributed and suffered from epileptic spasms during the neonatal period.[6] Prior to our report, there has no cases reported in the literature regarding MCF patient associated with West syndrome.

West syndrome (WS) is regarded as a subtype of Infantile Spasms Syndrome (ISs) and is the most frequently reported subtype of ISs. WS is a unique and severe form of epilepsy in infancy, characterized by a triad of (1) episodic spasm and occurring in the cluster, (2) characteristic almost continuous interictal epileptic activity with abnormal brain wave pattern on EEG so-called "hypsarrhythmia" and (3) delayed psychomotor development.[7-9] During spasms, acute respiratory compromise can occur when ictal epileptic activity directly affects autonomic control centre, involvement of respiratory centre of the brain stem.[9] Lower heart rate and loss of respiratory rate are commonly observed in WS patient, because of continuous

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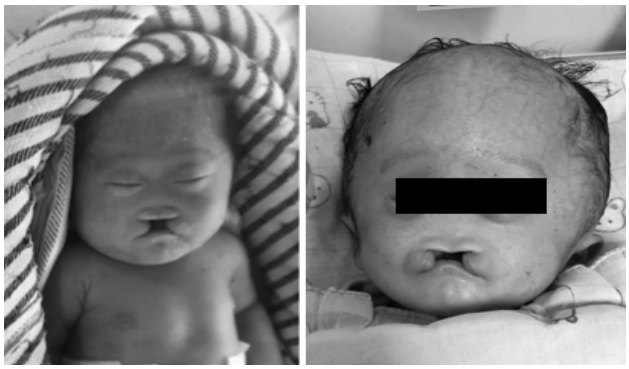

Figure 1. (left) Three days old newborn girl with midline facial cleft; (right) Congenital hydrocephalus clearly seen in this picture. Ventriculoperitoneal shunt was performed at the age of two months.

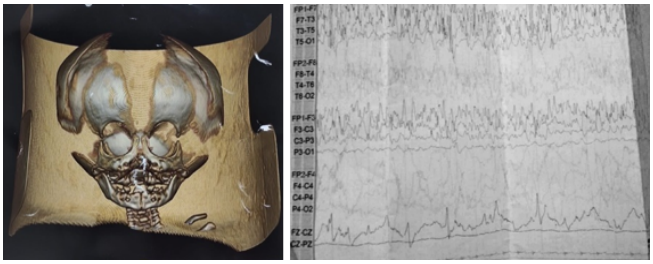

Figure 2. (left) Volume rendering technique (VRT) of head computed tomography (CT) scan demonstrating absence of the premaxilla bone; (right) EEG pattern shows the spike waves and polyspike waves at F7, T3, T5, F8 and T4 especially at left hemisphere. The EEG is abnormal with epileptiform waves.

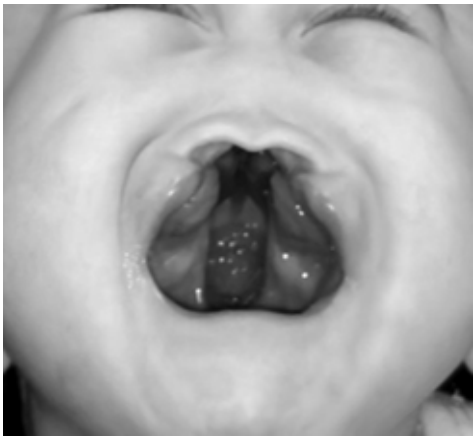

Figure 3. Frontal view with open mouth shows prolabium-premaxilla-columella complex agenesis in complete hard and soft palate cleft.

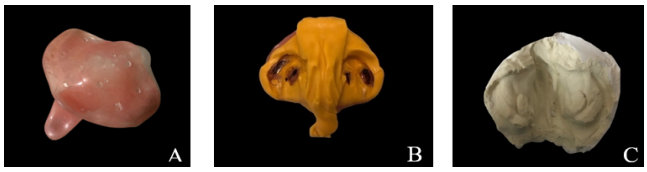

Figure 4. A. Individual tray from self-cure acrylic with holes for retention, B. Intraoral impression using putty elastomer material, C. Cast model.

adjustment of respiration during interictal spams activity. Both cardiac and respiratory dysfunction have been implicated as possible precipitating causes in sudden unexplained death in epilepsy patients (SUDEP).[9,10] We report here the first case of prosthodontic approach as presurgical treatment using presurgical nasoalveolar molding (PNAM) for infant with the aforementioned conditions.

Presurgical nasoalveolar molding (PNAM) has been widely recognized for a number of years in the early treatment of cleft neonates. The efficacy of PNAM before primary surgical repair has been described. They are the improvement of feeding, eliminate or reduce the risk of aspiration while feeding by providing a separation between oral and nasal cavity, normalizing of tongue position, and resulting presurgical reduction of cleft width.[3,11] The aim of this case report is to describe a modified PNAM treatment approach in MFC with WS infant who has respiratory compromise and feeding problem.

CASE REPORT

An infant girl born at term (40 weeks of gestation) by spontaneous vaginal delivery to a 22-year-old mother with a birth weight 2400grams dan length 41cm. The pregnancy was uncomplicated, but the mother experienced mild nausea and vomiting during the first trimester and light bleeding was reported in the 5th month of pregnancy. According to the parent, the family history was negative for birth defect or genetic disease, and the mother didn't smoke or consumed alcohol. The infant was referred to Dr. Hasan Sadikin general hospital (RSHS) due to her multiple abnormalities. She was treated for two months in the infectious pediatric care unit and then transferred to the non-infectious unit after her condition stable. The clinical diagnostics are then established including cerebral palsy type spastic quadriplegia, congenital hydrocephalus, West syndrome, and midline facial cleft with alobar holoprosencephaly. Figure 1 shows patient with congenital hydrocephalus before ventriculoperitoneal shunt (VP shunt) treatment. The VP Shunt had been performed at the age of 2 months.

Patient had neonatal spasms 5 hours after the birth for 2 minutes. The spasms displayed abnormal movements, intermittent jerks of head and recurrent spasms multiple times in a day followed by a high fever. Episodic spasm continued to recur in her early months of life, consisted of tonic activity of both arms like a forward hugging position with subsequent spread the other limbs. Spasms tend to begin soon after arousal from sleep within 3-5 seconds duration and occur in dozens of clusters, up to 100 spasm per day. Diagnostic work-up such as head computed tomography (CT) scan showed the absence of the premaxilla bone and electroencephalography (EEG) confirmed pattern of hypsarrhythmia, therefore she was diagnosed as having WS (Figure 2). The incidence of spasms slowly decreased over time as she got older, and by the time the patient was 6 months old, spasms became less frequent.

Patient got referred to Prosthodontic department RSHS for feeding treatment of cleft. Clinical features of her cleft were completely absence of premaxilla, columellar and nasal septal cartilage, therefore the nasal dome was flat without any support

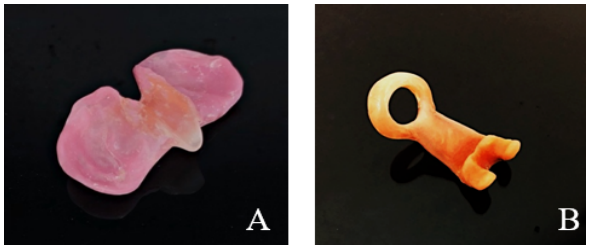

Figure 5. A. Hotz-type feeding plate, B. Extraoral nasal hook.

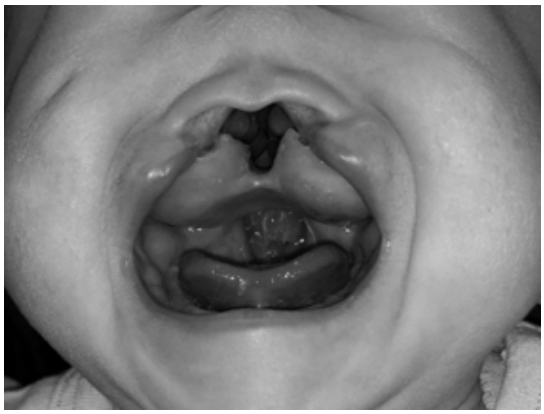

Figure 6. Feeding plate is placed inside the mouth with a good retention.

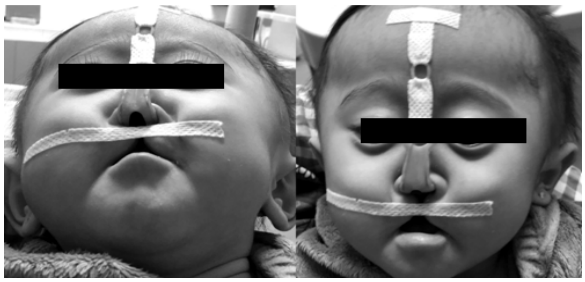

Figure 7. Extraoral nasal hook is performed to elevate the flattened nasal dome up during lip retraction.

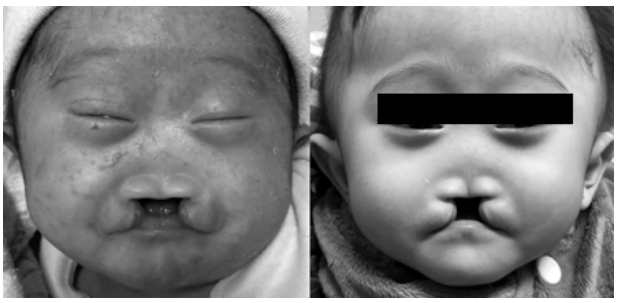

Figure 8. (left) photo before PNAM treatment, (right) photo after 10 months PNAM treatment. Note the gap of lip segment moves narrower.

as shown in Figure 3. The cleft defect extended to the soft palate. The patient's tongue protruded outward and upward, occupying the cleft most of the time. This activity caused respiratory problems for the patient since her palatum was low.

Intraoral impression was taken at 10 days postnatal using individual infant oral tray and putty impression material (Figure 4). The modified presurgical nasoalveolar molding (PNAM) was then delivered a week after. This PNAM consisted of two separate appliances, the first appliance was a Hotz-type feeding plate and the second appliance was an extraoral nasal hook, as shown in Figure 5. The feeding plate was inserted and the patient's ability to suck milk from the bottle was evaluated (Figure 6). It had taken about 20 minutes to drink 40ml of milk in the first trial of feeding with the appliance. Over time, the ability to drink milk increased and at the next control visit, she was able to drink 60ml of milk in 15 minutes. The weight gain was observed regularly and it was increased sufficiently.

During the onset of multiple spasms, the feeding plate helped pushing the protruded tongue back to its normal position and preventing it to enter the defect area which could interfere the airway. Selective grinding was performed gradually on the feeding plate to approximate the gap of alveolar segment. Strapping was placed across the upper lip to retract the lip closer to the midline. As a consequence of the retraction process, the nasal dome got flatter due to there was no bone to support it. Therefore, an extraoral nasal hook is applied to elevate the nasal passages and maintain the airway during the lip segment retraction. A plaster tape with elastic orthodontic band were paired on the nasal hook and attached vertically on the forehead. Figure 7 shows the procedures described above.

After 1-year follow-up, the 12,5mm alveolar gap reduced to 8mm and the gap between the lips that used to be wide had become narrower, a favorable shape for repair surgery preparation, as shown in Figure 8. Due to patient's unstable conditions, lip surgical repair was performed when she was 14 months old. The repair showed favorable esthetics result and the tissue healed under minimal tension (Figure 9).

DISCUSSION

MFC results from failure of the two medial nasal processes to fuse in the midline.[1,4] It is commonly associated with HPE as a result of failure of cleavage of the embryonic brain. In 1963, DeMyer classification previously used to describe HPE into five categories: (I) cyclopia, (II) ethmocephaly, (III) cebocephaly, (IV) midline facial cleft lip (premaxilla agenesis) dan (V) Facial dysmorphism. DeMyer groups I-III still strongly correlate brain defects, while DeMyer's group IV and V correlate normal or near normal mental development.[1,2] HPE then categorized based on brain morphology. There are three broad categories of HPE: alobar, semi lobar and lobar. Patient in this case is diagnosed with alobar HPE, presenting class IV DeMyer classification: midline facial cleft. In alobar HPE, the brain has not divided at all, usually associated with severe facial deformities and lack of psychomotor development.[12,13] Children with alobar HPE associated with severe facial

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anomalies (cyclopia, ethmocephaly and cebocephaly) have very low survival rate or rarely survive the immediate postnatal period. While those with less severe facial malformation, for example MFC, can survive for months or in a minority of cases, longer than one year.[14]

HPE patients have a strong correlation with epileptic spasm. A retrospective study from Butow and Zwahlen,[6] proposed data of 85 patients with cleft and HPE, 63.0% of them with prolabium-premaxilla-columella agenesis suffered from epilepsy presenting with intermittently compromised airways during epileptic seizures. Our patient also had a complex cleft with prolabium-premaxilla and columella agenesis (class IV DeMyer classification) and she had neonatal spasm and later diagnosed with WS.

WS is an episodic epileptic spasm that affects individuals during infancy and early childhood. The onset of spasms varies from the first week of life to 3 years of age, with a peak at 6 months of age and in about 80-90% of cases, the spasms manifest within the first year of life.[7,8] In the present case, spasms were present from as early as 5 hours after birth for 2 minutes and associated with violent jerking of the upper limbs. The spasm continue to occur multiple times in a day, followed by a tonic contraction lasting a few seconds (3-5 seconds) with involvement mainly of the muscles of the neck, trunk, and limb. The spasm may appear with episodes of cry or scream and having a peak at 5 months of age. This patient is not achieving physical and mental milestones, she can't even roll over from tummy to back at one year old. She is being subjected to physiotherapy and she also receives routine medication from neuropediatric department for her spasm.

There can be cardiac and respiratory involvement in WS patient. During spasms, acute changes in heart rate and/or respiration can occur, therefore, respiratory dysfunction could be another risk factor for morbidity and mortality in patients with WS.[9] A study by Jansen et al.[9] evaluate respiratory control in 10 patient with WS and 14 control subject. The results show that there is a clear difference in autonomic respiratory control in patients with WS compared to control subject, it is the loss of respiration rate in patients with WS. Case specificity in our patient is that WS is associated with tongue protrusion, moving upward and forward entered and covered the cleft area. Hotz-type feeding plate then was planned to prevent the tongue from entering the defect and interfering the growth of the palatal shelves. It also trained the patient to normalize and place the tongue in the right position. Respiratory issues occurred several times in this patient, including during intraoral impression for PNAM treatment. We took intraoral impression under strict paediatrician supervision and monitor oxygen saturation due to the saturation dropped drastically during the impression process.

The management of cleft in this patient is multicomplex. The greatest obstacle in the reconstruction of the rare craniofacial clefts relates to the extent of soft tissue hypoplasia.[13] In our case, as a presurgical repair treatment, prosthodontic approach is carried out by retracting the lip tissue with extraoral strapping, so that the tissue is moulded closer to the midline, maximizing the aesthetic result of the labioplasty. Unfortunately, in this case, the nasal is flat without any bone

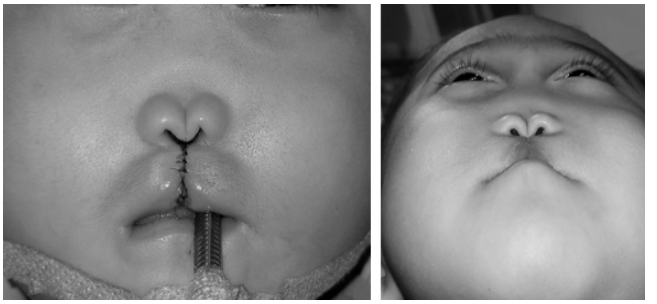

Figure 9. Labioplasty as primary surgical repair result.

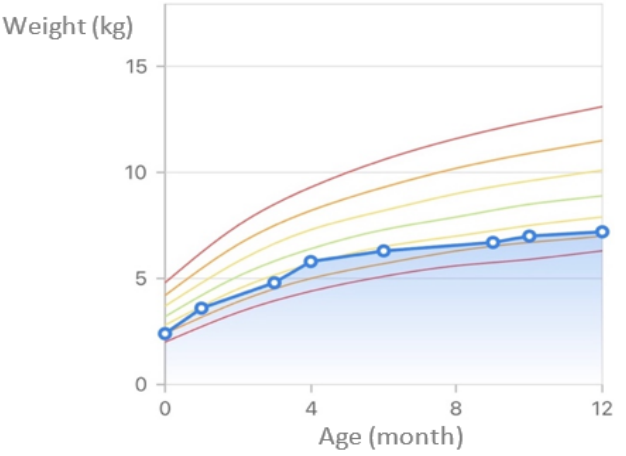

Figure 10. Patient's first year weight chart shows favourable weight gain.

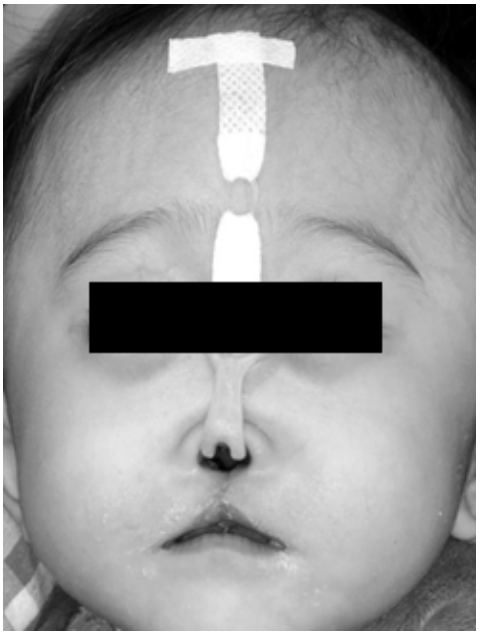

Figure 10. Post labioplasty, patient using extraoral nasal hook to maintain the airway, until the nasal reconstruction in the next surgical phase.

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necessary to be performed. A study by Monasterio et al.[15] compare the effect of nasoalveolar molding with nasal stent and nasoalveolar molding with extraoral nasal elevator in unilateral cleft lip and palate patient. The result showed both methods produced similar result. We used extraoral nasal elevator or nasal hook to simplify the procedure of elevating the nasal dome. Nasal hook was made with a wire lined with resin, paired with elastic band and fixed to the forehead. This simpler way of nasal traction helps to maintain the airway during the lip segment retraction. It is also easier for parent to understand and manage the appliance.

Patient's weight increased favourably, indicate the proper function of PNAM (Figure 10). She had not gain much weight in 5th month to 8th month of age due to her peak spasms and high fever that kept on recurring. However, her weight was regained after that. Not only for facilitate nutritional intake, the feeding plate is also useful for presurgical reduction of cleft width in the alveolus and palate. It provides guidance for the growth of each alveolar segment. In this case, we performed selective grinding of the feeding plate in every routine control visit, and we also performed build up at the under portion of feeding plate to facilitate the jaw development. The 12,5mm alveolar cleft width was reduced to 8mm in one year follow up.

Timing and sequence of cleft reconstruction in MFC with infantile spasm must encompass some criteria: successful weight gain, control of epileptic spasm, presence and the successful wearing of a feeding plate, and one-year survival.[6] It was suggested to perform primary repair surgery in MFC with HPE is not before 12 months of age and a minimum of 5 kg body weight.[6] There is a high risk of postoperative aspiration problem after surgical repair especially in cases with epileptic episodes. In our case, surgery was not performed until the spams completely disappear. The primary labioplasty in this patient was performed at 14 months of age due to patient's unstable condition. The second surgical phase, nasal repair and palatoplasty, is planned to be performed when the patient is 2 years old.

CONCLUSION

The presurgical treatment of MFC with WS has not been reported previously in the literature. Treating patient with these conditions, particularly those with agenesis of facial structures remains a very challenging task and multidisciplinary approach. The prosthodontic approach with PNAM has been expected as an effective treatment for patient with these conditions. It helps the patient to breath better, facilitate nutritional intake so the body weight gain sufficiently and mould the lip and alveolar tissue closer to the midline to provide a more aesthetic primary surgical result.

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