Lip And Palate Reconstruction On Median Cerebrofacial Malformation Patient

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Background: Median cerebrofacial malformations are developmental anomalies of the midline brain and facial structures. The clinical manifestations ranges from holoprosencephaly with agenesis of central facial structures, to those seen in median facial dysplasia. Patients who survive are usually born with severe functional limitation and die during infancy. Nowadays, with improved perinatal care, patient with severe midline craniofacial abnormalities can be expected to live longer. The purpose of this paper is to show how reconstruction of the lip and palate on patient with median cerebrofacial malformation is done.
Patient and Method: Reporting 2 cases of child with cleft median cerebrofacial malformations. First case was a 2 year-old child with complete median cleft lip and palate. Clinical examinations and CT scan revealed a holoprosencephaly. She was also diagnosed with laryngomalacia. The second case was a 4 month-old patient with median cleft lip and palate also microcephaly. We performed labioplasty in both patients and did the palatoplasty in the first patient.
Result: Both cases showed no sign of complication after the operation. They both discharged one day after the operation. The first patient reported dead 1 week after the surgery at home. The cause was unknown.
Discussion: Disturbed maxillary growth is postulated to be caused by the manipulation and suturing of the maxillary vomer, and scarring of the dentoalveoli post-surgery. Another contributing factor is the intrinsic tissue deficiency.
Summary: With proper perinatal care and holistic teamwork that expand the survival rate we expect those with the best prognosis deserve to appear as normal as possible.
Keywords: *Holoprosencephaly, median facial dysplasia, median cleft lip and palate*

Latar Belakang: Malformasi cerebrofacial median adalah kelainan dalam perkembangan garis tengah otak dan struktur wajah. Manifestasi klinis dapat berupa *holoprosencephaly* dengan agenesis struktur wajah sentral, hingga dysplasia wajah median. Pasien yang dapat bertahan hidup pada saat lahir akan memiliki keterbatasan fungsional dan akan meninggal dalam beberapa bulan. Dengan semakin berkembangnya perawatan perinatal, pasien memiliki ekspektasi hidup yang lebih tinggi. Tujuan penulisan adalah menunjukkan bagaimana rekonstruksi bibir dan palatum pada pasien dengan malformasi cerebral median.
Pasien dan Metode: Dilaporkan 2 kasus pasien anak dengan malformasi cerebrofacial median. Kasus pertama adalah anak 2 tahun dengan bibir sumbing dan langit-langit median. Pemeriksaan klinis dan CT scan menunjukkan holoprosencephaly. Pasien juga didiagnosis laringomalasia. Kasus kedua adalah pasien usia 4 bulan dengan bibir sumbing dan langit-langit median dengan microcephali. Kami lakukan labioplasty pada kedua pasien dan palatolasty pada pasien pertama.
Hasil: Kedua pasien tidak menunjukkan tanda-tanda komplikasi setelah operasi. Keduanya pulang satu hari setelah operasi. Namun, satu minggu setelah operasi, pasien pertama diketahui meninggal dirumah. Penyebab pasien meninggal tidak diketahui.
Ringkasan: Kami mengharapkan dengan perawatan perinatal secara holistik dapat meningkatkan angka harapan hidup. Pasien dengan prognosis baik dapat memiliki penampilan luar yang baik juga.
Kata Kunci: *Holoprosencephaly, median facial dysplasia, median cleft lip and palate*

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Median cerebrofacial malformation defines as developmental anomalies of the midline brain and facial structures. This malformation occurs at the developmental phase where the median part of the anterior neural plate is obstructed, and this event influences the shape of the midline brain and facial structures. Signs and symptoms are

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various from the critical type such as holoprosencephaly including agenesis of central facial formation to those present with median facial dysplasia. The majority of malformation on embryonic forebrain is holoprosencephaly (prosencephalon) that appears about one in every 250 embryos.[1]

The categorization of median cerebrofacial malformations are based on embryology of the deformity, location, and also signs and symptoms. Characteristic of the cleft that occur is also substantial sign to describe the defect. Median facial dysplasia was diagnosed if there are a sign of deficient median facial structures with less brain abnormalities. Unilateral or bilateral cleft lip and palate may occur in this type. The type of cleft lip and palate should be examined carefully to decide the reconstruction. Holoprosencephaly was a form of malfunction embryonic process of a brain that is related to midline facial hypoplasia. Patient with holoprosencephaly mostly die during intrauterine or in neonatal age caused by serious functional limitation. Those who survive usually have normally developed brain, perform in advance, and have a good prognosis.

All patients with median facial dysplasia who has gone through surgery present with type III malocclusion related to disorder of the midfacial development or various surgical pressure such as scarring which obstructs the facial development in every median facial dysplasia patients. Patients with common variety of cleft lip and palate patient have possibilities to defeat this pressure and develop naturally, nevertheless, median facial dysplasia patients that develop poorly are incapable to defeat the extra pressure of surgery.

The best equipment to evaluate the brain malformations is MRI. By the initiation of advanced CT scan and MRI techniques, specific abnormalities on the brain morphology can be detected. With a holistic approach by a team of medical expertise those patients who have potential for full life expectancies are candidates for surgical correction.

The decision for the best treatment plan would be achieved if parents were involved during the process, they have to be fully informed about the patient condition, the risk of any intervention, and the long term prognosis. Sometimes, despite how harsh the prognosis, some parents are willing to do everything for their child.[2-4]

PATIENT AND METHOD

We report 2 cases of child with cleft lip and palate accompanied by median cerebrofacial malformation.

Case 1

First case was a 2 year-old child with complete median cleft lip and palate. Clinical examinations and CT scan revealed a holoprosencephaly. She was also diagnosed with laryngomalacia. We performed Randall's technique to repair the lip. We cooperated with pediatricians to take care the patient holistically.

One and a half year after the cleft lip repair, we performed two-flap palate repair (Figure 2). A week before surgery, the team held a meeting with the parents to discuss about the patient condition, the complication and the risk of the operation. After discussing for several hours the conclusion was to still perform an operation to the patient with permission from the parents who longs for a better functional outcome for their daughter.

The distance of the uvula was 1.5 mm, distance of the nasal spine was 9 mm, and the mucoperiosteal flap width was 11 mm both left and right side (figure 3). The operation used two flap palatoplasty techniques and without difficulties intraoperatively (figure 3, 4).

Case 2

The second case was a 4 month-old patient with median cleft lip and palate and also microcephaly (figure 5). We performed labioplasty with Millard technique. The patient was wearing obturator while waiting for the optimum condition for surgery procedure. A day before the scheduled surgery, the patient

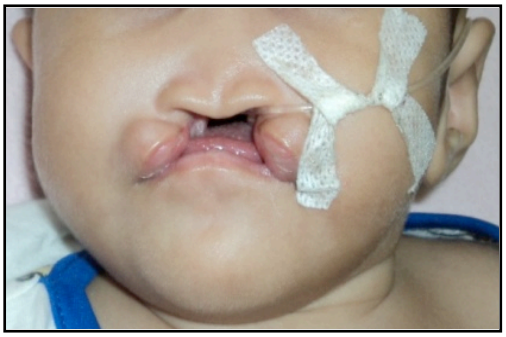

Figure 1. First patient, before Labioplasty

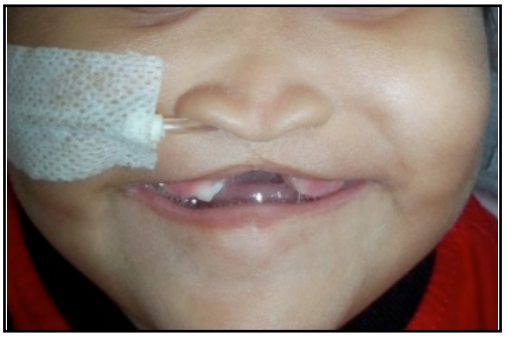

Figure 2. First patient, 1,5 years after labioplasty

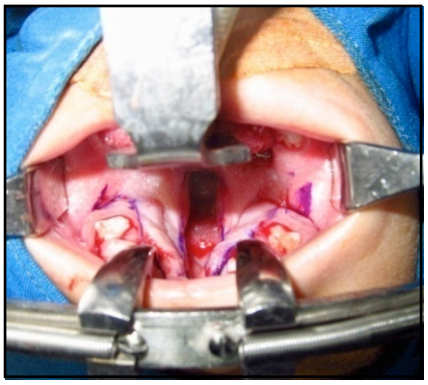

Figure 3. First patient, intraoperative

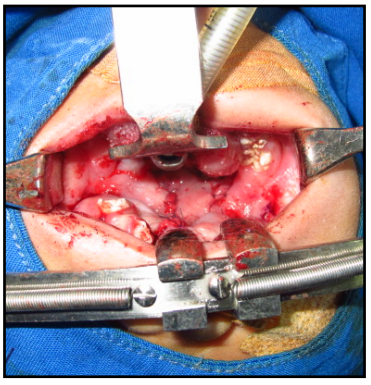

Figure 4. First patient, post operative Procedure

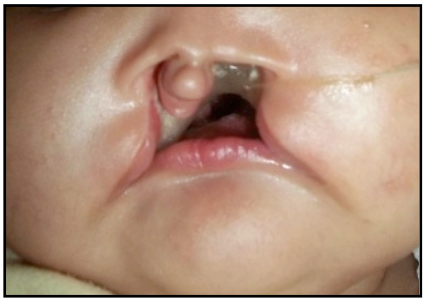

Figure 4. Second patient, before Procedure

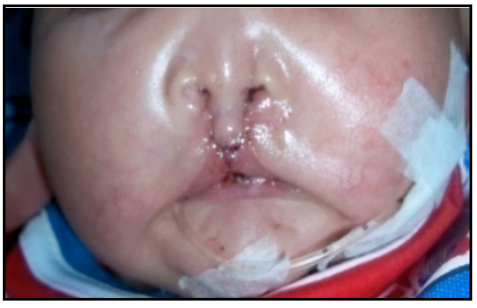

Figure 5. Second patient, one day after procedure

was having an airway management problem that has to be stabilized in intensive care unit, but require no ventilator and the condition was recovered the day after. The schedule was delayed until the patient is in ideal condition for operation. And the day after the operation was held. During the operation the airway problem also occurred but can be stabilized. And the operation was done in 2 hours.

RESULT

In the first case, the patient recovered and discharged the day after the operation. One week afterwards, the parents called to inform that the patient passed away while sleeping. The parents confirmed that there was no aspiration happened, and the patient was not acting unusually before. The parents declined the post mortem inspection.

In the second case, the patient was discharge from the hospital the day after (figure 6). And there were no significant complications reported until now. The patient will be scheduled for palate repair at 1 year old.

DISCUSSION

Holoprosencephaly is a genetically diverse malformations that occur 1 every 16.000. Mainly it was a random occurence, but there were 6 percent recurrence that has been recognized.[5] Sometimes the sign and symptom are delicate to detect such as mild microcephaly or hypotelorbitism, anosmia, hyposmia,or single central maxillary incisor.

Holoprosencephaly was classified into five classifications with differentiation of malformations of the face and the cerebral. Patient with cyclopia, ethmocephaly, or cerebrocephaly (groups I to III) usually die during infancy. This is the reason why surgical intervention in this stage of life was contraindicated. Patients with less facial defect and alobar or semilobar cerebral malformation may survive during infancy but suffer from severe neurological compromised and mental retardation. Those patients mostly require feeding tube, and gain minimal advantage from surgical intervention. However those kinds of patients have a promising extended life expectancy, therefore procedure of lip and palate repair is substantiate to perform. Further craniofacial interventions are not suitable for this patient.

Patients with lobar and normal cerebral malformation and facial cleft or midface hypoplasia (groups IVB, VA, VB) are good candidates for surgical intervention. Although their brains are anatomically normal, the risks of subtle improvement and neurogically insufficiency exist.[2]

Patient with holoprosencephaly usually associated with other problems such as impaired mobility, risk of aspiration, epilepsy, temperature instability, and endocrine dysfunction. Therefore treatment and management for holoprosencephly require approach from team of medical professional.

In our case we work along with the pediatrician, ENT, and from rehabilitation unit. Our patient was diagnose with holoprosencephaly with median cleft lip and palate. She also suffered from larygomalacia. She has a promising extended life, but requires further management to support the daily activities. Before the operation was held, the team has been discussed about the further management that might require. At the moment feeding tube was used to provide oral intake of the patient, further the patient will be require a swallowing exercise, and the parents was also has been explain to pay attention on the patient airway after the operating procedure. After a long discussion among the team and the parents, we decided to perform the operative procedure.

Since holoprosencephaly is an uncommon condition among living children, most of the parents were lacking in information and lead to difficulty in finding appropriate medical providers, services and resources. There were organizations called "Families for HoPE (www. Familiesforhope.org) which help parents whose children suffer from Holoprosencephaly to gather and share their feeling, experiences and other information.[6,7]

Indonesian people are still lacking information about rare disease, such as holoprosencephaly. This lack of information will lead to miscommunications that cause

misunderstanding between the parents ant the doctor. Parents need to be told about their children condition so they can be involved in the discussion to decide what management will be choose. In our case the parents was involved in a discussion that has been held, they were fully informed about condition of the patient, the risks of the procedure, the advantages and disadvantages of the procedure. They also have been informed about management that need to do after operation, and how their cooperation is a huge influence. The last decision was on their hand, whether they agree to allow their child being operated or not. And the parents agreed to let their child being operated with all consequences.

SUMMARY

We expect with the proper perinatal care and holistic teamwork, median cerebral malformations patient could have higher life expectancy. And patients with a higher life expectancy is a good candidate to perform surgical procedure. With that surgical procedure, the medical team hope that the patient can live as normal as possible.

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