

Lip Repair Surgery for Bilateral Cleft Lip and Palate in a Patient Diagnosed with Trisomy 13 and Holoprosencephaly

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We report a case of lip repair surgery performed for bilateral cleft lip and palate in a patient diagnosed with trisomy 13 and holoprosencephaly. At the age of 2 years and 7 months, the surgery was performed using a modified De Hann design under general anesthesia. The operation was completed in 1 h and 21 min without large fluctuations in the child's general condition. The precise measurement of the intraoperative design was omitted, and the operation was completed using minimal skin sutures. It is possible to perform less-invasive and short surgical procedures after careful consideration during the preoperative planning. Considering the recent improvements in the life expectancy of patients with trisomy 13, we conclude that surgical treatments for non-life threatening malformations such as cleft lip and palate should be performed for such patients.

Key words: cleft lip and palate, trisomy 13, holoprosencephaly, surgery

INTRODUCTION

Trisomy 13 was identified by Patau in 1960 [1]. Several severe systemic abnormalities, such as congenital heart disease or central nervous system malformations, have been associated with trisomy 13 [2, 3], and most of the affected infants do not survive beyond the first year of life [4]. Therefore, children with trisomy 13 may not have surgeries for non-life threatening malformations, such as cleft lip and palate [2, 5]. Here we report a case of lip repair surgery performed for bilateral cleft lip and palate in a patient diagnosed with trisomy 13 and holoprosencephaly.

CASE REPORT

The patient was a 2.5 year-old girl. She was referred to our department by her attending pediatrician for examination of her cleft lip and palate. She was born at the gestational age of 35 weeks and 2 days. Both parents were Asian. Her mother was 40 years old, and had been admitted to the hospital because of intrauterine growth retardation and oligohydramnios. The baby was delivered by an emergency Caesarean; her birth weight was 1630 g, and Apgar scores were 8 and 9. At the examination after birth, she had microcephaly, orbital hypotelorism, blepharophimosis, bilateral cleft lip and palate, and laryngomalacia. The computer tomographic examination of the brain indicated an atrophy of the frontal lobe, incomplete cerebral inter-hemispheric fissure in the lower part of frontal lobe, and obscurity of the third ventricle and frontal horn of the lateral ventricle (Fig. 1a, b, c). These findings were compatible with holoprosencephaly. The chromosomal

analysis revealed free trisomy 13; therefore, she was diagnosed with trisomy 13 and holoprosencephaly. No congenital heart diseases were detected. Because of her severe psychomotor retardation, she received nasogastric tube feedings instead of oral intake. When she was 8 months old, she was discharged from the hospital to home health care. When she was 2 years old, she underwent gastrostomy.

At the first visit in our outpatient clinic at the age of 2 years and 6 months, an improvement in her food intake and articulatory functions that would usually be gained from the treatment of cleft lip and palate could not be expected because of her psychomotor retardation. However, the patient's esthetic issues were creating psychological distress in her parents, and they wanted a certain level of improvement in her appearance. We prepared for the lip repair surgery.

Physical findings at the first visit and preoperative planning

The patient had complete bilateral cleft lip and palate, hypoplasia of the nose, aplasia of the columella, a short prolabium, and a tiny floating premaxilla (Fig. 2a, b, c, and d). Based on her general condition, the attending pediatricians required us to finish the operation within 1 h. Therefore, we coordinated the operation as follows: (1) after the introduction of general anesthesia, we changed the operative process depending on her general condition, (2) in order to shorten the duration of operation, any measurements using a precision gauge for intraoperative design were omitted, (3) in order to shorten the duration of operation, the operation was completed using intradermal

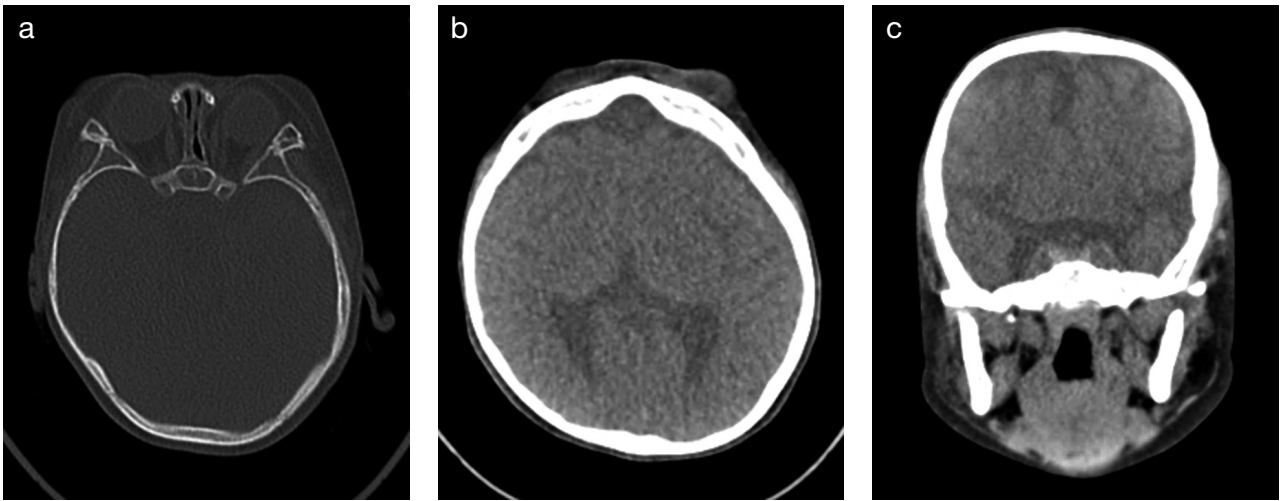


Fig. 1 a. Orbital hypotelorism is obvious in the axial view of the CT examination.
b. Axial view of the brain CT examination. Obscurity of the third ventricle and frontal horn of the lateral ventricle are indicated.
c. Coronal view of the CT examination. Atrophy of the frontal lobe and an incomplete cerebral interhemispheric fissure in lower part of frontal lobe are shown. These findings were compatible with holoprosencephaly.

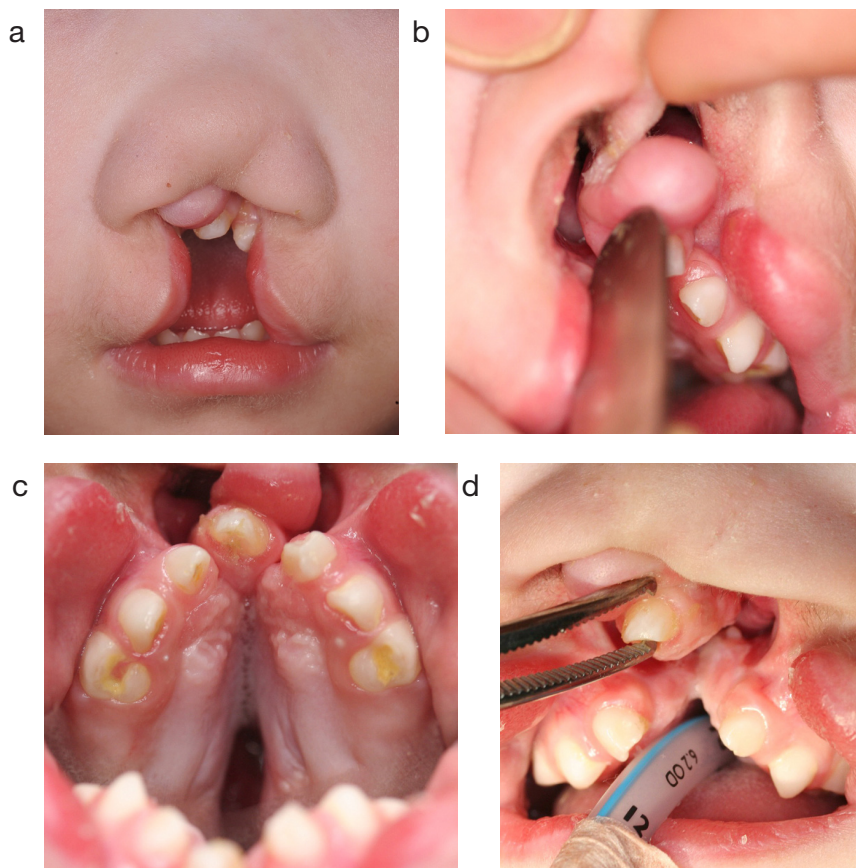


Fig. 2 a. Photograph taken at the first visit in plastic surgery outpatient clinic. Bilateral complete cleft lip and palate, hypoplasia of nose, aplasia of columella, a short prolabium, and a tiny floating premaxilla are noted.
b. An aplasia of the columella is shown.
c. Bilateral complete cleft palate with a hypoplastic premaxilla.
d. A short prolabium and tiny floating premaxilla are shown.

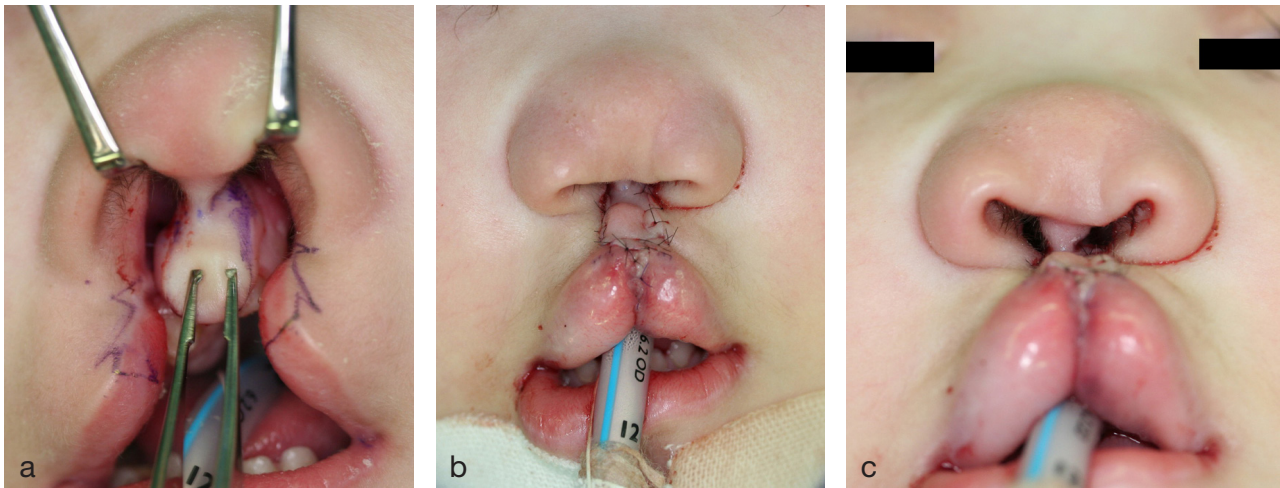


Fig. 3 a. Intraoperative design of the lip repair surgery. A modified De Hann design to supply an appropriate volume of soft tissue to the upper lip from the prolabial skin was planned. In order to shorten the duration of the operation, measurement using a precision gauge for intraoperative design was omitted.
 b. The operation was completed using a modified De Hann design with minimal skin suture. The key sutures and the skin flap from the prolabium is visible.
 c. Hypoplastic columella was left untouched. The operation was completed in 1 h and 21 min without large fluctuations in the patient's vital signs.

suture and minimal skin sutures, and (4) when the blood circulation of the prolabium was insufficient, we changed the operational design and completed the operation without using a skin flap from the prolabium.

Intraoperative findings

At the age of 2 years and 7 months, the lip repair surgery was performed under general anesthesia. After careful inspection of the blood circulation of the prolabium, we decided to use a modified De Hann as the intraoperative design [6], which would supply appropriate soft tissue volume to the upper lip from the prolabial skin (**Fig. 3a, b, and c**). We left the hypoplastic columella untouched. The operation was finished in 1 h and 21 min without large fluctuations in her vital signs.

Postoperative progress

She was extubated in the operating room and had controlled spontaneous respirations in the ICU until the next day. On postoperative day 6, she was discharged from the hospital. Three years have passed since the lip repair surgery, and the patient is still alive. Currently, she is 5 years and 7 months old and still can be taken care of at her home. Unexpectedly, her prolabium has grown long enough to give her upper lip a natural shape. The surgical scar is not conspicuous. The shape of the vermillion border and the Cupid's bow are acceptable (**Fig. 4a and b**). The reconstruction of the aplastic columella and revision of the flat and short nasal shape will be required; however, her parents are satisfied with her appearance.

DISCUSSION

Trisomy 13 (Patau syndrome) is a congenital disorder first described by Patau in 1960, and it is caused by having an extra 13th chromosome [1]. The mean maternal age of these mothers has been reported as 31.3 years old by Baty *et al.* [2] and as 33.03 years old by Drugan [7]. These ages are markedly higher

than the mean maternal age of 26.0 years old in the normal population as researched by Baty *et al.* [2]. The frequency of this syndrome has been reported between 1:3000 to 1:29000 live births [8–11], and the incidence and survival rate in females is higher than those in males at all ages [2, 4]. Trisomy 13 is associated with a variety of characteristic malformations; some typical anomalies are congenital heart disease (59%–78%), central nervous system malformations (82%), polydactyly (50%–64%), scalp defects (29%–44%), an abnormal auricular helix or low-set ears (80%–89%), and cleft lip and palate (45%–71%) [2, 3, 12]. Trisomy 13 has been described as a clinically severe condition. Magenis *et al.* [13] (1968) reported that 28% of these newborns died during the first week, 44% died within the first month, and 86% died within their first year. The most common causes of death were cardiopulmonary arrest (69%), congenital heart disease (13%), and pneumonia (4%) [2]. However, more recent papers have concluded that the life expectancy of patients with trisomy 13 has increased compared with that reported previously. Baty *et al.* [2] suggested a more optimistic prognosis for trisomy 13, with a survival rate at 1 year of 38% and five-year survival of 13%; the mean survival was 12.8 months. Furthermore, Nelson *et al.* [5] found that 46% of birth admissions were discharged alive after a mean stay of 7.8 days, 41% of the hospital records were of children over 1 year of age, and in >10% of discharges, the child was over the age of 8 years at the time of admission. Tunca *et al.* [8] proposed that the presence of non-lethal congenital anomalies and aggressive medical care may positively contribute to the longer survival of patients with trisomy 13.

Holoprosencephaly is a malformation sequence with a basic feature of impaired midline cleavage of the embryonic forebrain [14]. DeMyer *et al.* [15] identified the correlation between the median facial anomalies and degree of holoprosencephaly, and graded the holoprosencephaly as I to V. Our case was classified as a grade V; with this grade, her prognosis was less

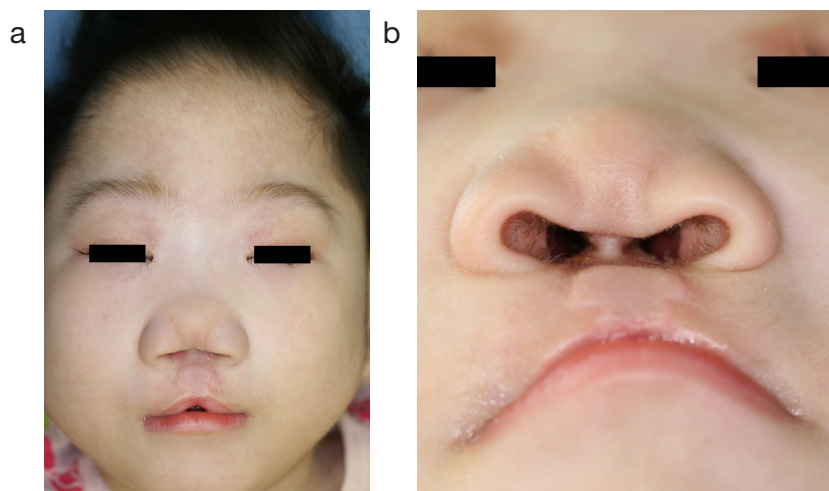


Fig. 4 a. Photograph at 3 years after operation. Surgical scar is not conspicuous. The shape of the vermillion border and the Cupid's bow is acceptable.
 b. Reconstruction of the aplastic columella and revision of the flat and short nasal shape will be required; however, her parents are satisfied with her appearance.

severe. Of the chromosomal syndromes that frequently involve this sequence, trisomy 13 syndrome is the most commonly associated with holoprosencephaly [14]. Warkany *et al.* [16] (1966) observed holoprosencephaly in 78% of patients with trisomy 13, Taylor [17] (1968) reported this condition in 64% of patients, Gullotta *et al.* [18] (1981) reported it in 39% of patients, Rios *et al.* [2] (2004) described the incidence in 40%-70% of patients, and Lin *et al.* [3] (2007) mentioned it in 14% of patients (in 17% of brain lesions).

There have been few reports about cleft lip and palate surgery for trisomy 13 cases [19], despite there being many reports about cardiac procedures [2, 5]. Baty *et al.* showed that the percentage of patients that had surgery for trisomy 13 during the neonatal period was 23% [2]. Nelson *et al.* described that cleft palate surgery represented 3.2% ($N = 1075$) of the total major therapeutic procedures in children with trisomy 13 that were performed from 1997 to 2009 [5]. The prognosis for our case was expected to be better than in other patients with trisomy 13 that had congenital heart defects. On the other hand, her deformity in the cleft lip and palate was severe because of holoprosencephaly. The features of her cleft lip such as hypoplasia of the prolabium or aplasia of the columella made the lip repair surgery more difficult than it would have been in a patient with cleft lip and without holoprosencephaly. However, we finished the operation in the appropriate amount of time by careful preoperative planning. The patient has made steady progress since the operation. Consequently, considering the recent improvement in the life expectancy of patients with trisomy 13, we recommend that surgical treatments for non-life threatening malformations such as cleft lip and palate surgery should be performed for such patients.

CONCLUSION

Despite the conventional understanding of trisomy 13 as a lethal congenital malformation, a substantial number of children are living for longer periods of

time. Surgical procedures for esthetic problems are required as observed in this case. It is possible to perform low-invasive and short surgical procedures with careful consideration during the preoperative planning period.

CONFLICT OF INTEREST

All authors declare no conflict of interest.

ETHICAL STANDARDS

All procedures performed in this article were in accordance with the ethical standards of the institutional ethical board of Tokai University School of Medicine and with the 1964 Declaration of Helsinki and its later amendments.

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