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Torticollis Management Using the Customized Soft Neck Collar in CATCH 22 Syndrome Combined with Klippel-Feil Anomaly: A Case Report

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CATCH 22 syndrome is rare genetic disease that has various manifestations. Cervical vertebral anomaly, such as Klippel-Feil anomaly, is frequently observed in the patients with CATCH22 syndrome. We present the case of an 11-year-old female patient with CATCH22 syndrome and Klippel-Feil anomaly who had been treated torticollis using the customized soft neck collar. During the patient's first visit to our clinic, she presented with low ear set, skull deformity, intellectual disability, and tilting of the head to the left by approximately 25 degrees. Imaging studies revealed multisegmental fusion and C3 hemivertebrae of the cervical spine and left thoracic scoliosis at T4 with 50 degrees of Cobb's angle. We instructed passive stretching and applied the customized soft neck collar we invented. The ipsilateral aspect of the neck collar is designed to provide vertical support between the clavicle and mandibular angle and is adjustable in height. The Velcro was attached to the neck collar at the point of contact with the ipsilesional mandibular angle, which provides negative sensory feedback, inducing her to tilt neck to the contralesional side. We applied the neck collar for 2 hours a day. After 1 year of treatment, her neck inclination angle improved from 25 to 10 degrees. Providing negative sensory feedback using the customized soft neck collar can be one of the treatment options of postural management in patients with torticollis in cases of CATCH 22 syndrome combined with Klippel-Feil anomaly.

Key words: CATCH 22, Klippel-Feil syndrome, Torticollis, Orthotic device

CASE REPORT

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INTRODUCTION

CATCH 22 syndrome is characterized by cardiac defects, abnormal facial features, thymic hypoplasia, cleft palate, and hypocalcemia, which are associated with the deletion of chromosome 22q11. Since DiGeorge syndrome, velo-cardio-facial syndrome, and conotruncal anomaly face syndrome are all due to the microdeletion in chromosome 22q11, CATCH 22 syndrome encompasses these syndromes [1]. A deletion in chromosome 22q11 is the genetic basis of the most common interstitial deletion syndrome, the 22q11 deletion syndrome in humans, with an incidence of 1 in 4,000–5,000 births [2]. According to the literature, patients with this syndrome are associated with a high incidence of various types of relatively mild cognitive deficits, including learning difficulties, which is sometimes combined with mild learning disability and attention deficit hyperactivity disorder [3].

Klippel-Feil syndrome (KFS) is another rare disease that was initially described by Klippel and Feil [4], and its prevalence is reported to be approximately 1:40,000–42,000 births in the literature [5]. The genetic etiology of KFS is still unclear and had been reported heterogeneously, but defect of the notochord and its signaling with insufficient separation of the cervical vertebrae are considered the

main underlying causes [6]. Partial or complete fusion of two or more cervical vertebrae is the key feature of KFS, frequently associated with further osseous and non-osseous manifestations such as low posterior hairline, torticollis, brevicollis, basilar impression, atlanto-occipital fusion, scoliosis, facial asymmetry, Sprengel's deformity, and other genitourinary, central nervous, and cardiopulmonary system anomalies [7].

There have been a few case reports suggesting genetic association between the CATCH22 syndrome and KFS, but the correlation between two syndromes has not yet been clearly revealed. Nevertheless, cervical vertebral anomaly is frequently observed in both syndromes [8-10]. Since spinal growth originates from the superior and inferior endplates of each body, congenital vertebral malformation with asymmetric shape causes unbalanced vertebral growth. The rate of deterioration and severity of the final deformity vary according to the type and location of the anomaly [11]. Generally, as congenital scoliosis due to vertebral anomaly is typically inflexible, bracing is known to be unresponsive in these cases. Thus, observation is usually indicated in mild congenital scoliosis that does not require surgical treatment [12].

Here, we present a case of torticollis managed using customized neck collar in a female patient with both CATCH22 syndrome and Klippel-Feil anomaly.

CASE REPORT

An 11-year-old female patient was referred to our clinic for torticollis by her pediatricians. She was the firstborn child of a healthy 35-year-old mother and father. She was delivered via cesarean section due to fetal distress after 39 weeks of gestation and weighed 2,180 g. There was no family history of genetic diseases.

At birth, her pediatrician performed chromosome analysis because of her low ear set, skull deformity, and C3 cervical hemivertebrae found on brain magnetic resonance imaging. The test result showed deletion of 22q11, and she was diagnosed with CATCH22 syndrome. Echocardiogram revealed tricuspid atresia combined with several cardiac anomalies. A few days after her birth, cyanosis was observed, and the symptom worsened over time. Her thoracic surgeon planned to perform Fontan surgery, the first step of which was to perform main pulmonary artery banding when the patient was 3 months old. As planned, bidirectional cavopulmonary correction was performed when she was 4 years old, and extracardiac conduit Fontan procedure was finally performed when she was 6 years old.

During her first visit in our outpatient department, she presented with the head rotated and tilted to the left, approximately 25 degrees (Fig. 1A), and prominent Adam's sign was observed on the left posterior upper back. The patient reported

Fig. 1. (A) The angle of inclination of the head on the coronal plane is approximately 25 degrees before treatment. (B) The angle of inclination has improved to 10 degrees after 1 year of treatment. In both pictures, long vertical lines are at the midline of her body, and the short ones are at the midline of her head.

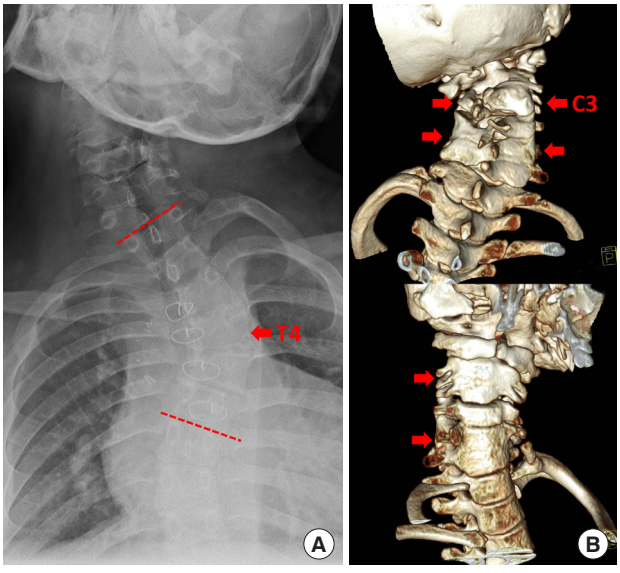

Fig. 2. (A) Plain radiography reveals left thoracic scoliosis at T4 with 50 degrees of Cobb's angle. (B) Three-dimensional computed tomography representing C3 hemivertebrae and multisegmental fusion, C4 to C6, of the cervical spine (indicated by arrows).

Fig. 3. (A) A customized soft neck collar. The Velcro is attached to the site of contact with the mandibular angle at the tilted head side (arrow). The neck collar is adjustable in height. (B) Because of the discomfort caused by the Velcro, the patient tilted her neck to the contralesional side.

cosmetic problems and neck pain due to the abnormal head posture. Plain film radiography and three-dimensional computed tomography (3D CT) scans to obtain an accurate assessment of her spine according to her current age were performed. Her cervicothoracic X-ray revealed left thoracic scoliosis at the T4 vertebral body with 50 degrees of Cobb's angle and right cervical scoliosis due to vertebral malformation (Fig. 2A), and 3D CT scan revealed right lateral C3 hemivertebrae accompanied with multisegmental fusion of the cervical spinal vertebral bodies, C4 to C6, which implied the possibility of KFS (Fig. 2B). Together with her pediatricians, we decided to perform whole exome sequencing to establish the diagnosis, but genetic evidence of KFS was not found.

Since she was unable to fully cooperate with the exercise instruction due to her intellectual disability, we decided to apply the customized soft neck collar specifically tailored to correct torticollis (Fig. 3A). The ipsilateral aspect of the neck collar is significantly designed to provide sufficient vertical support between the clavicle and mandibular angle and is adjustable in height. Additionally, the rough surface was intentionally made by attaching the Velcro to the neck collar at the point of contact with the ipsilesional mandibular angle. This made the patient felt uncomfortable, inducing her to tilt her neck to the opposite side of the lesion, as a negative sensory feedback (Fig. 3B). We applied the neck collar for 2 hours a day and instructed active stretching exercise of the ipsilateral neck muscles and active strengthening exercise of the contralateral neck muscles.

The angle of inclination of her head was measured at each visit in our outpatient clinic, and the neck collar was modified sequentially to fit between the ipsilateral clavicle and mandibular angle according to the improvement of neck motion. After 1 year of treatment, her neck inclination angle improved from 25 to 10 degrees (Fig. 1B). As the abnormal head posture was corrected, cosmetic problems and neck pain were also improved. She and her families were satisfied with the outcome of the treatment.

DISCUSSION

Generally, the initial treatment of juvenile and adolescent idiopathic scoliosis is observation with regular X-ray checkup. It is known that scoliosis with Cobb's angle of 30 degrees or more has a risk of progression into adulthood, and scoliosis with Cobb's angle of 50 degrees or more is certainly going to progress into adulthood [13]. Thus, if there are more than 10 degrees of progression per year or if there is a high risk of progression, nonsurgical treatments, including bracing, stretching, and exercise, are recommended, and surgical treatment is indicated in patients with more than 50 degrees of Cobb's angle [13]. Contrary to idiopathic scoliosis, nonsurgical treatment of congenital scoliosis is rarely recommended because congenital curves, in most cases, are inflexible and unresponsive to bracing, which can cause secondary deformation of the thoracic cage if these are applied forcibly [14]. Surgical procedure is considered depending on the amount and rate of progression and the expected progression of each deformity type [15].

In this case, the child had hemivertebrae of the C3 vertebral body and thoracic scoliosis with approximately 50 degrees of Cobb's angle, which were indications for surgical treatment. However, the patient had an intellectual disability due to CATCH 22 syndrome; hence, difficulty in postoperative care was expected. Therefore, we tried to treat the patient's torticollis by performing nonsurgical treatment as much as possible to improve her quality of life. Bracing was eliminated from our treatment option because it is not effective in treating scoliosis due to congenital vertebral anomaly and difficulty in application of bracing in cervical lesion. Instead, we instructed the patient's family to assist the patient in performing ipsilesional neck muscle stretching and tried to encourage her to perform active strengthening exercise of the contralesional neck muscle. Due to her intellectual disability, it was impossible for her to perform exercise herself. Hence, we designed a customized soft neck collar that fits between her ipsilesional mandibular angle

and clavicle. Moreover, we attached the Velcro to the site of the neck collar that touched the mandibular angle; hence, the patient felt uncomfortable, considered a negative sensory feedback. Thus, the patient tilted her neck to the opposite side.

During her early visits in our clinic, her neck showed a very limited range of motion with an inclination angle of 25 degrees. It was so inflexible that it did not change with active stretching, and we almost considered it a muscular contracture. However, after 1 year of treatment with passive stretching and active strengthening exercise via negative sensory feedback, the neck inclination angle improved from 25 to 10 degrees. Furthermore, her cosmetic problem and persistent neck pain improved. She and her family were satisfied with the result. There has been no report on the management of cervical torticollis, which is a manifestation of congenital anomaly, by providing negative sensory feedback using the customized soft neck collar. The neck collar is a tailor-made novel device that we specifically invented.

The treatment continued afterward, but no further improvement has been observed because the muscular portion had been improved by consistently wearing the customized soft neck collar, but not the underlying bony deformity. Unexpected problems, such as skin irritation and mental stress of the child, have also been observed during the treatment. Despite these fundamental limitations, providing negative sensory feedback using the customized soft neck collar is considered a valuable treatment option because it improves the quality of life of children with cervical scoliosis due to a congenital anomaly that does not require surgery.

CONCLUSION

Providing negative sensory feedback using the customized soft neck collar can be considered one of the treatment options of postural management in patient with torticollis in CATCH22 syndrome combined with Klippel-Feil anomaly.

CONFLICT OF INTEREST

No competing financial interests exist.

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