

소아재활

게시일시 및 장소 : 10 월 19 일(토) 08:30-12:30 Room G(3F)

질의응답 일시 및 장소 : 10 월 19 일(토) 11:00-11:30 Room G(3F)

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Torticollis Management Using Soft Neck Collar in CATCH 22 Combined with Klippel-Feil Syndrome

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Introduction

CATCH 22 syndrome and Klippel-Feil syndrome (KFS) are rare congenital diseases, and genetic association between both syndromes has not yet been reported clearly, but cervical vertebral anomaly is frequently observed in both. We present a case of torticollis managed using neck collar in a girl with both CATCH 22 syndrome and KFS.

Case report

An 11-year-old girl visited outpatient clinic for torticollis. At birth, her pediatrician performed chromosome analysis because of her low ear set, skull deformity. The test result showed deletion of 22q11 and she was diagnosed as CATCH 22 syndrome. She presented tilted and rotated head to the left about 25 degrees (Figure 1A), and reported cosmetic problem and neck pain due to abnormal head posture. X-ray and 3-dimensional computed tomography scan revealed left thoracic scoliosis with 50 degrees of Cobb's angle, and right lateral C3 hemivertebra accompanied by multisegmental fusion of cervical vertebral bodies which implies possibility of KFS (Figure 2). Under the co-work with the pediatricians, we decided to perform whole exome sequencing (WES) to confirm the diagnosis, which is finally revealed as negative study. Since she was unable to fully cooperate with exercise instruction due to intellectual disability, we applied the customized soft neck collar (Figure 3A). The ipsilateral aspect of the neck collar is designed higher to provide support between clavicle and mandibular angle and is made to be adjustable in height. A rough surface using Velcro was attached to the site of contact with mandibular angle at the tilted neck side, which makes the patient uncomfortable, inducing her to tilt neck to opposite side as negative feedback (Figure 3B). We applied the neck collar for 1 hours a day and instructed active stretching exercise of ipsilateral neck muscle and strengthening of contralateral neck muscle. The angle of inclination of the head was measured every visit at outpatient clinic, and the neck collar was serially modified to fit between the ipsilesional clavicle and mandible according to improved neck motion. After a year of treatment, angle of inclination of the head was improved from 25 to 10 degrees (Figure 1B). She had a cosmetic problem due to abnormal tilted head posture before the customized soft neck collar

treatment. She and her family were satisfied with corrected head posture through a negative feedback exercise using neck collar.

Discussion

Negative feedback Exercise using customized soft neck collar can be one of the treatment options of postural management in patient with torticollis in CATCH 22 syndrome combined with KFS.



Figure 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 a year of treatment. In both pictures, long vertical lines are midline of her body and the short ones are midline of her head.

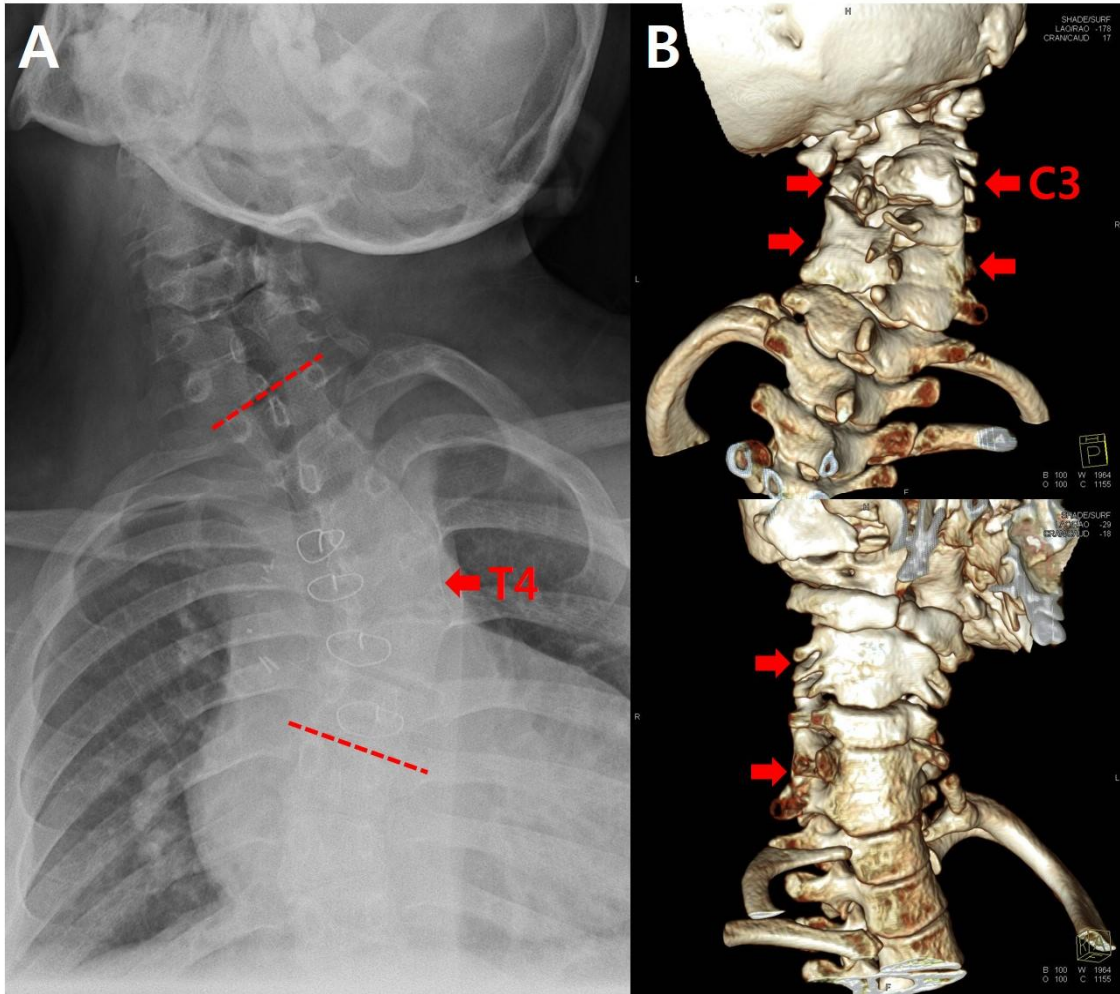


Figure 2. (A) The plain radiography shows left thoracic scoliosis at T4 with 50 degrees of Cobb's angle. (B) Three-dimensional computed tomography representing C3 hemivertebrae and multisegmental fusion, C4 to C6, of cervical spine (indicated by arrows).

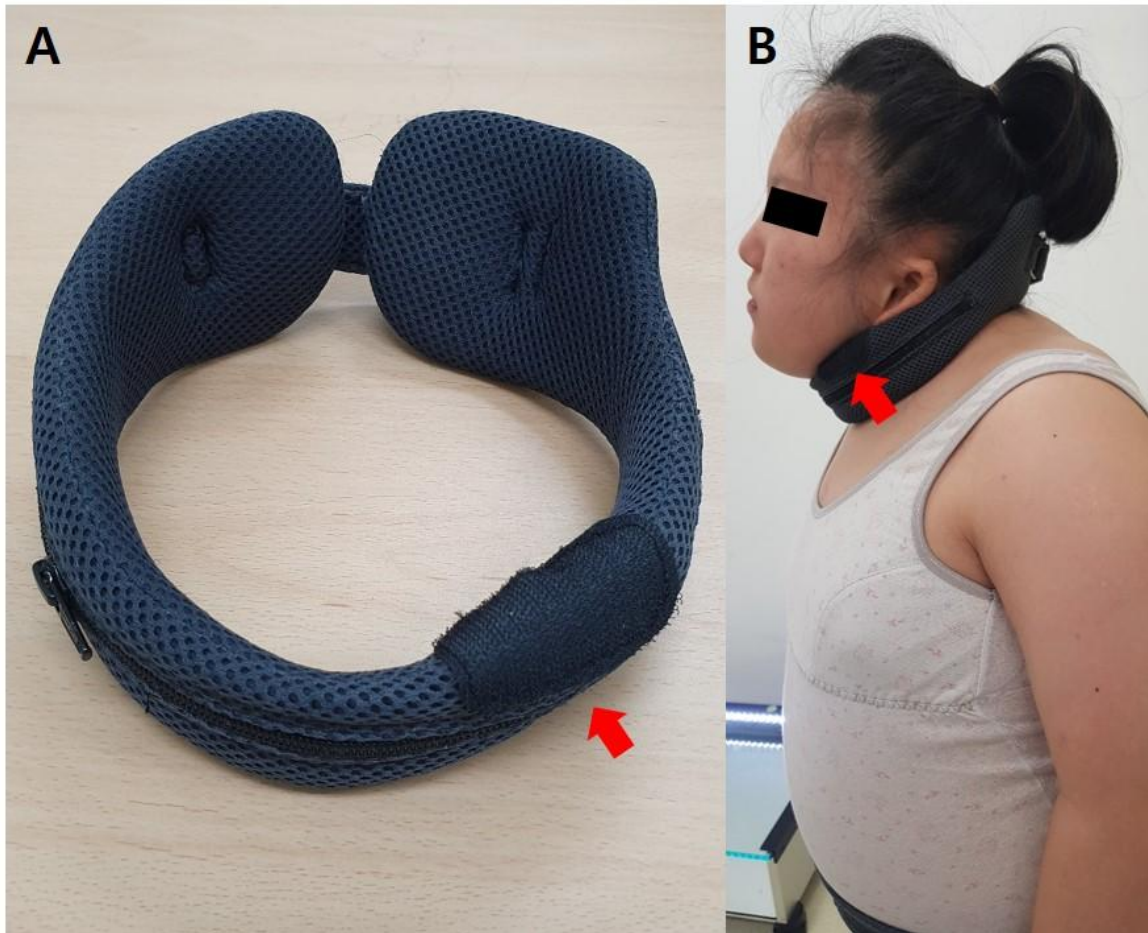


Figure 3. (A) A customized soft neck collar. Velcro is attached to the site of contact with mandibular angle at the tilted head side (arrow). The neck collar is made to be adjustable in height. (B) Because of the discomfort caused by the Velcro, the patient is induced to tilt neck to contralesional side.