Case Report

Ann Rehabil Med 2016;40(5):938-942
pISSN: 2234-0645 • eISSN: 2234-0653
https://doi.org/10.5535/arm.2016.40.5.938

Annals of Rehabilitation Medicine

Rehabilitation Treatment of a Child Diagnosed With Duplication of 1q42-q44: A Case Report

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Trisomy 1 is a rare chromosomal anomaly and has never been reported in Korea. Clinical features of trisomy 1 include macrocephaly, prominent forehead, flat nasal bridge, low set ears, and micrognathia, all of which result in a very distinguishable facial structure. A child with trisomy 1 also suffers from mental retardation and/or developmental delays. In this case report, the child was diagnosed with *de novo* trisomy 1 without receiving any treatment until visiting our hospital. The child suffered from foot and ankle deformities, leading her unable to stand independently. Here we report the surgical treatment and rehabilitation treatment that enabled the child to walk independently.

Keywords Trisomy, Orthopedic procedures, Rehabilitation

INTRODUCTION

There have been several reports about structural variations and anomalies of chromosome 1, although partial duplication of the long arm is a rare anomaly that has been reported only a few times internationally [1-3]. Structural defects of chromosome 1 can lead to distinguishable facial characteristics with mental retardation and/or developmental delays. Hypotonia can also occur, changing the biomechanics of the foot, which in turn affects the whole kinetic chain [4]. Since the foot cannot withstand the whole body weight when a baby begins to stand, a baby may develop postural anomalies temporarily. Developmental delays can hinder the timing of the foot being properly adapted. In this case report, the characteristics and musculoskeletal problems of the first trisomy 1 patient in Korea are described with comparisons to relevant references.

CASE REPORT

A 7-year-old female visited the outpatient clinic of Rehabilitation Medicine of National Health Insurance Service Ilsan Hospital for evaluation of her current status and management. The child did not have any past history or family history of chromosomal anomalies. When the

Received December 24, 2015; Accepted March 4, 2016
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child was 18 months old, she visited a pediatrician with complaint of developmental delay. Brain magnetic resonance imaging revealed no abnormalities. Chromosome analysis conducted at that time showed band duplication at the end of the long arm (46,XX,dup(1)(q42,1q44)). However, the child did not receive any further treatment.

At the time of the visit, the child's head circumference was 52 cm (75–90%tile), height was 113 cm (10–25%tile), and weight was 16.5 kg (<3%tile), with the head circumference being relatively large compared to her height or weight. The child's face was triangularly shaped with wide and protruding forehead, low set ears, flat nasal bridge, long philtrum, and micrognathia (Fig. 1).

No weakness was observed during physical examination, although the child's muscle tone was slightly hypotonic and her overall muscle bulk was decreased. There was no evidence of any neurological defect. Both forefeet were found to be subluxated at the talonavicular joint with heels at valgus position. Severe equinus deformity was also observed at the left ankle (Fig. 2). Functionally, the child was able to stand with the feet positioned abnormally with her hands held.

According to the Bayley Scales of Infant Development-II version test, the child had a score of mental developmental index (MDI) <50 and a score of psychomotor developmental index (PDI) <50. She had the mental level of a 5-month old and motor level of a 6-month old, indicating profound mental retardation. Speech and language

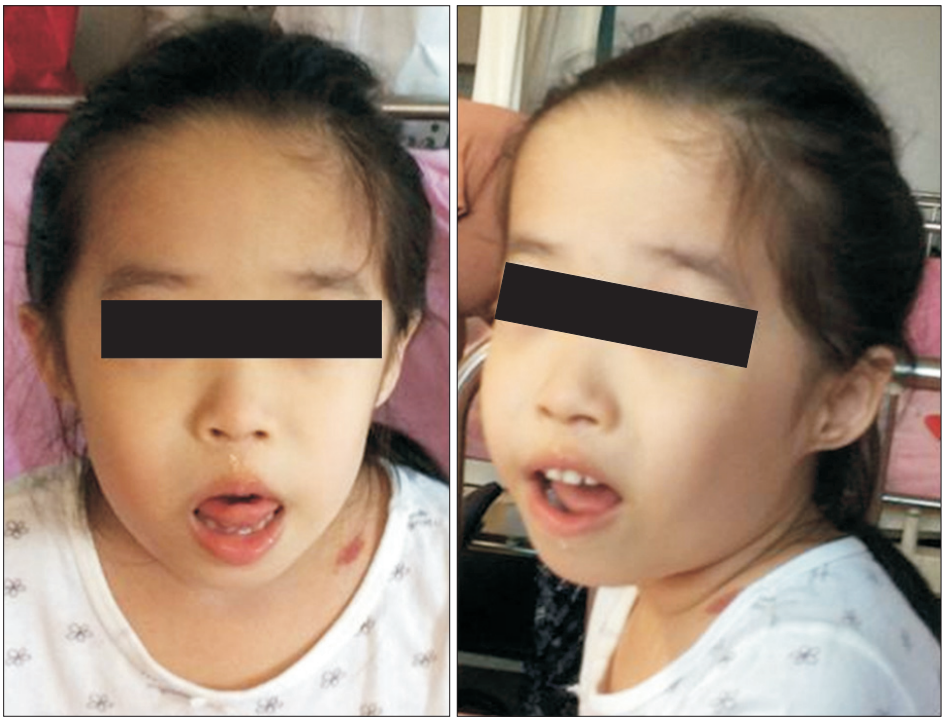

Fig. 1. Facial dysmorphic features. Trigonocephaly, large and prominent forehead, depressed nasal bridge, irregularly positioned teeth, and micrognathia were noted.

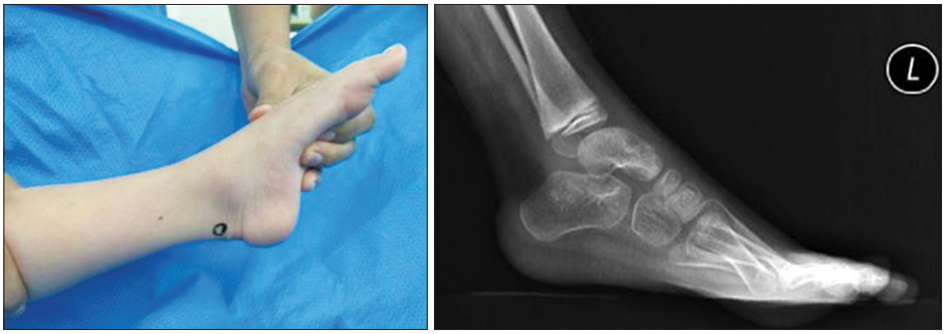

Fig. 2. Pre-operation condition of the left ankle. Equinus deformity and plantar flexion contracture were noted.

evaluation using Sequences Language Scale for Infants (SELSI) showed that her receptive language was similar to that of a 4-month old. Her expressive language was at a level similar to that of a 3-month old. Both were at less than 1%tile compared to others of the same age.

Further testing was conducted to rule out any complications associated with trisomy 1. The child was evaluated with small atrial septal defect (ASD) through ultrasonography at 18 months old. However, there was a follow-up loss. Additional study at our facility showed that the defect was spontaneously closed. Abdominal ultrasonography showed that the child had no urogenital problems. Regarding ophthalmologic issue, only mild hypermetropia was found. We worked together with the Department of Orthopedic Surgery. We concluded that if the mechanical problems of the ankle were solved, it would greatly improve the child's functional levels. Therefore, left tendo-achilles lengthening was performed on pes planovalgus and equinus deformity. To relieve toe flexion contracture, tendon lengthening was performed at the master knot of Henry.

Fig. 3. Post-operation condition of the left ankle. The patient was able to contact both heels completely. She was able to stand and walk independently after the orthopedic surgery.

The child was admitted to the Department of Rehabilitation Medicine after the cast was taken off 6 weeks after the operation. Before the surgery, the child's contact points while standing were the left toes. After the surgery, her contact points while standing were both heels. The child was able to stand independently (Fig. 3). However, since the child lacked experience standing independently, she avoided bearing weight on her left side. Although the child could walk independently, she showed difficulty in controlling her left knee because she had no experience with gait before. To solve this problem, left side weight-bearing and knee grading exercises were implemented. Throughout the therapy, the child showed improvement on bearing weight on both sides. She was able to walk independently at 1 week after starting the rehabilitation treatment. Although the child swayed while walking at first, her stride was shortened and her stability was improved over the course of treatment. Eight weeks into the therapy, even though the child showed a high guard posture with wide base, she could independently walk 50 m and climb up and down the stairs with one hand held.

DISCUSSION

Partial duplication of the long arm of chromosome 1 is a very rare disease. It affects only 200 or so patients worldwide [5], with the affected locus slightly differ from one case to another. There have been familial cases and *de novo* cases, all of which were previously reported outside of Korea. This case is deemed to be *de novo* duplication as both parents are normal in phenotype without showing any evidence of abnormalities in chromosome analysis.

According to various references, trisomy 1 patients have characteristic dysmorphic face and mental retardation, regardless of the length of the trisomic segment [6]. Among clinical features of trisomy 1q syndrome mentioned by Kulikowski et al. [1], our patient also showed macrocephaly, trigonocephaly, large and prominent forehead, prominent glabella, epicanthic folds, hypertelorism, depressed nasal bridge, long philtrum, micrognathia, irregularly positioned teeth, macrostomia, and arachnodactyly. However, metopic ridging, synophrys, or capillary nevus were not observed in our patient. Our patient also showed speech difficulties, hypotonia, de-

velopmental delay, mental retardation, and locomotion difficulties. Other comorbidities associated with trisomy 1 include ocular anomalies, cardiac malformations, and urogenital anomalies [2]. Ocular anomalies in previously reported trisomy 1 patients include hypoplasia of the optic disc, persistent vascularization of the lens, and emergence of hyaloid vessels [6,7]. The patient in our case report did not exhibit any of the above features. However, she did have mild hypermetropia. Related cardiac abnormalities include dilated heart, high ventricular septal defect, dextrocardia, insufficient tricuspidal valve [3], and patent ductus arteriosus [6]. The patient in this study had small ASD. However, it was found to be spontaneously closed. Urogenital anomalies in trisomy 1 patients include urinary tract malformation [3], horse-shoe kidney, and cryptorchidism [6]. Such anomalies could not be found on the abdominal ultrasound or CT scan of the patient in our case. Watanabe et al. [8] have reported that chromosomal translocation can be often found in 1q partial duplication/triplication syndrome cases with 1q41-qter being the most frequently involved region. In addition, anomalies of eyes, ears, nose, and mouth and recurrent respiratory tract infection are common regardless of the translocation among those patients. The patient in our study had duplication of the similar locus, thus showing symptoms similar to those of a previous study [8].

There is a case report showing that the patient has delayed motor development [5]. However, the patient started to independently walk at 3 years old [5]. However, musculoskeletal problems have not been reported. The patient in our study could not stand independently even when she was 7 years old. The hypotonia that the patient developed when she was an infant and the constant asymmetric weight-bearing pattern might have contributed to her secondary heel cord tightness and contracture. This made it even more difficult for the patient to stand independently. After surgical correction of mechanical problems, the patient could independently stand immediately. Further physical rehabilitation treatment allowed the patient to walk independently as well. George and Elchert [4] have reported that patients with developmental delay and hypotonia have improved functional ability after using foot orthosis. Livingstone and Hirst [9] have shown that surgical procedures can help Down syndrome patients with decreased levels of stability of the hip, knee, and foot joints. Guidera et al. [10] have reported that Rett syndrome patients along with scoliosis, contraction of lower limb joints, and misalignment of hip joint can be improved with surgical procedures and orthosis. The musculoskeletal problem in the patient of our case might be a secondary problem as she didn't receive proper treatment on time. It should not be considered as a particular anomaly of trisomy 1.

In conclusion, trisomy 1 can be diagnosed through chromosome analysis if the patient shows particular dysmorphic features or other suspicious signs. There are no specific treatment methods for trisomy 1 yet. The best options available currently are to treat comorbidities and complications, manage any symptoms that may arise such as the musculoskeletal issues in this case, and keep the patient in the best condition possible.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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