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Case Report

Arthrogryposis Multiplex Congenita (AMC): Functional Improvement of a Seven-Year-Old Girl after 3 Years of Physical Medical Rehabilitation Intervention

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ABSTRACT

Arthrogryposis multiplex congenita (AMC) is a genetic disease typified by non-progressive and multiple congenital limb contractures. The incidence of AMC is 1 in 3,000 live births, occurring in two or more areas of the body. The etiology of the syndrome is largely unknown and is multifactorial. The aim of this article is to report a patient diagnosed with arthrogryposis multiplex congenita who received physical medical rehabilitation intervention. We report a seven-year-old girl who frequently had symptoms of joint deformities, movement limitation, and walking difficulties. The patient's fingers and toes also had muscle hypoplasia and pterygium. She was diagnosed with arthrogryposis when she was born. Several surgical interventions had been performed. Since four years old the patient started the physical medical rehabilitation programs. Owing to the integrated orthopedic and medical rehabilitation therapy, it showed some improvements. The patient could walk by holding onto a bench and eat or drink by herself. The patient had obtained the daily living exercises using the correct compensation technique using modified tools. Physical medical rehabilitation programs can improve the functional condition of AMC patients. The goals of a physical medical rehabilitation treatment are to gain the walking ability and improve the ability to independently perform activities of daily living. Stretching exercises, ROM exercises, splints, modified tools, and orthopedic surgery are essential for arthrogryposis patients.

Keywords: *Arthrogryposis multiplex congenita, Physical medical rehabilitation, Surgery.*

Introduction

Arthrogryposis multiplex congenita (AMC) is a hereditary disorder characterized by non-progressive multiple congenital limb contractures. This disease often improves gradually with appropriate management.[1] AMC is usually a feature of neurodevelopmental abnormalities or primary intrauterine muscle disease. Arthrogryposis is a term for multiple contractures related to many different diseases. Specific prenatal diagnosis is hard to be performed because more than 400 conditions are associated with these findings.[2]

The etiology of arthrogryposis syndrome is largely unknown and is multifactorial. Hall et al classified the causes of AMC into neuropathic disorder, structural (muscle function) disorders, connective tissue disorders, space limitation disorders, maternal diseases, and disorders of intrauterine or fetal vascularity.[3,4] The neuropathic disorder is associated with the loss of motor neurons due to changes in the spinal cord that lead to muscle atrophy. In addition, 95% of arthrogryposis is caused by neuropathic disorders.[5,6] When the joint is not moved for a long time, connective tissue can grow around the joint. This mechanism incurs the movement limitation. The difficulty of joint motion can occur when the tendons that connect to the joint do not stretch to their proper length. The lack of joint movement of the fetus in the initial developmental stage can lead to collagen proliferation, muscle fibrosis, and thickening of the joint capsule.[2,7]

Several types of arthrogryposis are caused by certain single-gene diseases (X-linked inheritance, autosomal recessive or autosomal dominant inheritance), metabolic diseases, chromosomal aberrations, microduplications, and microdeletions.[4,8] Amyoplasia as the classic form of AMC is usually sporadic. Arthrogryposis is common in only one of the monozygotic twins.[4] Distal arthrogryposis is probably inherited by autosomal dominant. It is a specific subgroup of disorders. AMC occurs with phenotypic variation between families and even between members of the same family. The distal arthrogryposis syndrome is caused by several different gene mutations, which take a role in the contractile apparatus.[5,9]

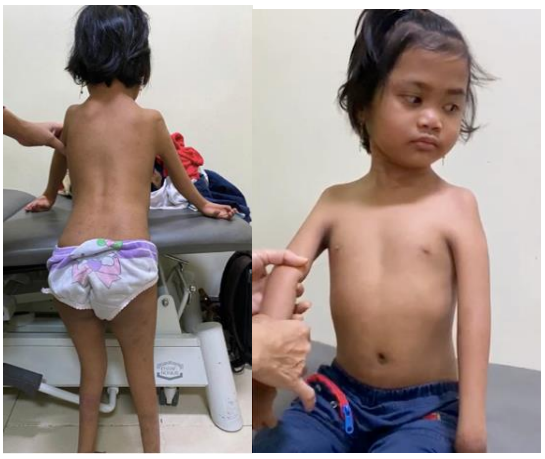
Material & Method (Case Report)

Case Report

A seven-year-old girl was admitted to the outpatient clinic with complaints of joint deformities, movement limitation and walking difficulties. The patient was born with fractures of both femur necks, and three days later hip reconstruction surgery was performed at the Kariadi Hospital. She was the first daughter of her family and had no siblings. The multiple contractures of the joint were developed since she was born. As a result, she was diagnosed with arthrogryposis multiplex congenita (AMC) with symmetrical shoulder, internal rotation and adduction of the shoulder, lumbar hyperlordosis, elbow extension, wrist flexion, and ulnar deviation.

Objectives

The examination revealed generalized muscle atrophy, weakness in the body, elbows and knee semi-flexion, toes pterygium, clubfoot deformities of both feet, and mild scoliosis (Image. 1).



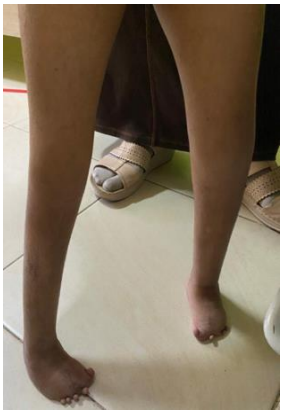


Image 1. There was generalized muscle atrophy, weakness in the body, elbows and knee semi-flexion, toes pterygium, clubfoot deformities of both feet, mild scoliosis, and lumbar hyperlordosis

In this case, all limbs are involved in symmetrical patterns. Note the elbow's extension and contracture, the wrist's flexion, and the ulna's deformity. The fingers were thinner and longer. Both hands had flexion contracture at the distal and proximal interphalangeal joints. The fingers also had ulnar deviation. The patient had contractures of the shoulders, elbows, wrists, fingers, knees, and ankles. The hands were hook-shaped, with skewed fingers and ulna (Image. 2). No pathological findings were found in the internal organs of the patient.

The contractures of the metacarpophalangeal and interphalangeal joint cause a hook-like appearance of the hands.

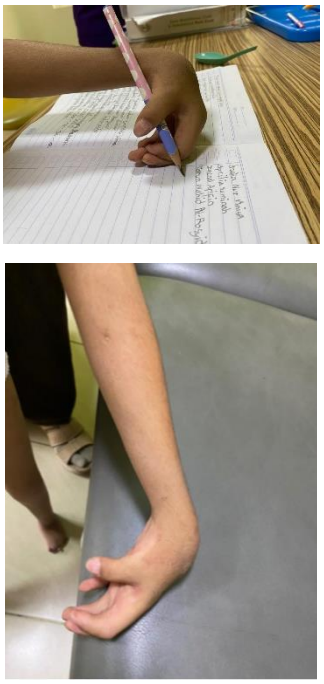


Image 2. The fingers and the ulna were skewed, and both hands were hook-shaped

Intervention

The Physical Medical Rehabilitation program was conducted in Kariadi hospital four years ago. Her past surgical histories were the soft-tissue release surgical of both ankles. When she was three years old, the surgery was extended to the wrist and finger flexors. The patient had 11 surgical interventions, and the orthopedist decided that they would be continued until the patient is around 18 years old. Since the age of 3.5 years, the patient was referred to the Medical Rehabilitation doctor at Kariadi Hospital. However, the patient did not come regularly due to limitation of her mother's job. From the age of four years, she began to regularly visit the doctor and joined the routine therapy at Kariadi Hospital.

The patient received stretching exercises, ROM exercises, splints, modified tools, and orthopedic surgery, which are essential for arthrogryposis patients. The physical medical rehabilitation programs were performed twice a week. The initial programs were a range of motion and stretching exercises. Those continued until now. Since the age of five years, the patient had received ADL and fine motor skill exercises. The ADL programs consist of feeding, dressing, and grooming exercises. Fine motor skill exercises were performed twice a week. These include writing, drawing, and buttoning exercises.

Result

The admission to the hospital made her meet the other patients with the same diagnosis, which surprisingly increased her self-confidence. She had regular physiotherapy and occupational therapy programs once a week. Hand wrist splint and ankle-foot orthosis (AFO) were given to prevent further deformity. The patient could walk by holding onto a bench. She also could eat alone with her arms supported by her legs. The patient was given the exercise activities of daily living using the correct compensation technique. She obtained walking exercises and a modified walker for indoors and outdoors by wheelchair. Furthermore, the patient attended Special School C and had many good records such as being the winner of various poetry reading and storytelling competitions for children with disabilities.

Discussion

Children with AMC require interdisciplinary programs such as pediatricians, rehabilitation professionals, and orthopaedists.[10] No major malformations were found in the internal organs of the AMC patient and there were no abnormalities in their intelligence or

pathological findings in the internal organs. AMC is accompanied by several symptoms. Our patient had generalized muscle atrophy, weakness in the body, elbows and knee semi-flexion, toes pterygium, clubfoot deformities of both feet, mild scoliosis, and lumbar hyperlordosis. In several studies, the AMC symptoms include thinning of the skin, muscle atrophy, and limb abnormalities. Sometimes, they have also abnormalities of the face and jaw, scoliosis, and various abnormalities of the respiratory, urinary, and nervous systems.[3,11]

In the case discussed here, we could observe several joint deformities. The contractures made it hard to determine the location of each anatomical site during clinical evaluation. Clinical examination showed knee subluxations and hip joint dislocations. There was a need for effective treatment without the risk of joint instability and minor diagnostic errors. The orthopedic intervention of children with arthrogryposis can give satisfactory results and decrease the limitations in daily activities. AMC patients should receive multidisciplinary integrated examinations.[8,10]

Various motion exercises, passive stretching, and serial casting are recommended for treating the contracture in AMC patients. The therapy program may become a long-term rehabilitation program due to the patient's condition. Using modified eating and drinking utensils is very helpful for AMC patients.[2,3,11] The patient was currently able to perform eating and drinking activities independently without leg movements. The patient could also write very well even though the movement of her fingers was very limited. Exercise like writing is also essential for them to gain a better future. Several patients may need help from other people even if their independence returns.

The prevention of deformity by early surgical release of contracted tissue should be considered. The patient had continued the rehabilitation program at Kariadi hospital since early childhood. She got her first surgery for the contractures when she was three years old. The passive range of motion exercises and stretching were performed to treat her contractures and scoliosis. The goal of the walking exercise for the patient was to improve scoliosis, walking ability, and balance. Thus, the patient's performance and ambulatory were getting better.[9,12]

Most patients with AMC disease develop progressive scoliosis. Radiological follow-up of scoliosis should be carried out regularly. If the curve size exceeds 40 degrees, surgical intervention is currently recommended.[8,13] Some AMC patients need intensive rehabilitation and surgery for articular deformations. More than 70% of patients with arthrogryposis foot joints need surgical correction. In addition, 39% of patients with knee joints need intervention.[3,10] Before the children learn to walk, it is recommended to perform surgical release of the soft tissues with the total release of the tendons. The use of plasters in arthrogryposis does not provide permanent results.[2]

Comprehensive rehabilitation methods and orthopedic treatments showed significant improvements in range of motion and daily activities. Functional evaluation and joint deformation evaluation were performed from the patient's birth. Currently, she continues the rehabilitation programs, close follow-up, and orthopedic consultation. Several studies state that the best results will be obtained in the first months of life if the corrective action starts as early as possible. Developmental stimulation is also an essential program that should not be neglected. The articular limitation of the patient with arthrogryposis can influence their cognitive development and motor function.[2,8,14]

Conclusion

The physical medical rehabilitation programs can improve the functional condition of AMC patients. The goals of AMC treatment are to gain the walking ability and improve the ability to independently perform activities of daily living. Stretching exercises, ROM exercises, splints, modified tools, and orthopedic surgery are essential for arthrogryposis patients.

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The authors declare that they have no conflict of interest with the publication of this study.

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