REVIEW

Journal of International Surgery and Clinical Medicine (*JISCM)* 2022, Volume 2, Number 2: 31-35
E-ISSN: 2807-7008

Published By :
Surgical Residency Program
Universitas Syiah Kuala

Profile of Arthrogryposis Multiplex Congenita (AMC) patients on surgery treatment at single centered study in orthopaedic hospital and a brief review

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Received: 2022-10-30
Accepted: 2022-12-06
Published: 2022-12-25

ABSTRACT

Introduction: Arthrogryposis multiplex congenita (AMC) is a complex abnormal condition characterized by multiple congenital contractures in the body's joints. Although this condition is not progressive, if not treated properly, it will increase the degree of disability in children. There are two types of AMC. The most common type of AMC is amyoplasia. The second type is distal arthrogryposis (DA), a genetically inherited type characterized by congenital contractures in multiple joints and no neurologic or muscular abnormalities. The incidence of AMC 1:3000 births with clubfeet and congenital hip dislocation is the most common. In this retrospective study, the authors described the profile of AMC patients who underwent surgery at our center with the approach and management.

Method: The study design was a descriptive retrospective. The population of this study is all AMC patients who underwent surgery at the orthopedic hospital of Prof. Soeharso Surakarta, Indonesia. We used the total sampling technique who underwent surgery in the 2018-2021 period.

Result: In this study, we got eleven AMC patients who underwent surgery. Distribution based on sex found six boys and five girls. Half of the patients underwent surgery under the age of three years, while the other half underwent surgery after the age of five. The presence of patients who are consulted at an older age indicates a lack of public literacy about the abnormal condition of AMC or a lack of concern for the condition. Almost all AMC patients who undergo surgery are in the lower extremities.

Conclusion: Rehabilitation plays a very important role in improving the quality of life for AMC patients. A multidisciplinary approach is needed in the treatment of AMC patients. Surgery is performed if stretching and external support splinting/casting cannot achieve functional ROM and repair the deformity.

Keywords: Arthrogryposis, Child, Contracture, Physical and Rehabilitation Medicine.
Cite This Article: Kumara, H.C., Satriadi, A.B., Saddalqous. 2022. Profile of Arthrogryposis Multiplex Congenita (AMC) patients on surgery treatment at single centered study in orthopaedic hospital and a brief review. *Journal of International Surgery and Clinical Medicine* 2(2): 31-35. DOI : 10.51559/jiscm.v2i2.27

INTRODUCTION

Arthrogryposis Multiplex Congenital (AMC) is a complicated clinical condition. Arthrogryposis is taken from Greek, which can be interpreted as curved or hooked joints.[1] It refers to the condition of multiple contractures in many joints present at birth.[1,2] Abnormalities occur not only in the joints or musculoskeletal system, but can also be accompanied by other organ systems, such as; the nervous system and cardiovascular.[2] Although this condition is not progressive, it will increase disability in children.[3]

AMC is a congenital abnormality closely related to genetics. There are two types of AMC. The first type, amyoplasia, is the most common type of AMC. This type is characterized by sporadic muscle hypoplasia.[4] The second type is distal arthrogryposis (DA), a genetically inherited type characterized by congenital contractures in multiple joints and no neurologic or muscular abnormalities.[5] Experts have conducted genetic mapping studies of AMC-type DA. Available online Mendelian Inheritance in Man (OMIM) (see Table 1).

The etiology of this abnormality occurs during intrauterine fetal development.[3] It can be simplified as anything that causes limited fetal movement since eight weeks gestation or even more severe akinesia resulting in fetal akinesia deformation sequence (FADS).[6] Apart from abnormalities of the neuro-cardiovascular system, limitations of movement also occur in oligohydramnios, multiple pregnancies, and other conditions.

The previous study has stated that the incidence of AMC 1:3000 births with syndromic clubfeet and congenital hip dislocation is the most common form.[3] Another study found an incidence AMC of 5:10000 births.[1] Amyoplasia is the most

common type that occurs with a rate of 1:10000 events, whereas in the same literature, the general incidence rate of AMC is 1:3000 births.[4]

AMC can be detected since the fetus is intrauterine with the help of ultrasound.[1] Doctors can immediately start a physical medication and rehabilitation (PMR) program when the baby is born, which is ideal for treatment.[2] serial casting of the affected limb is common. Physiotherapy on stiff joint range of motion (ROM) must be followed regularly. However, it is not uncommon for patients to require surgery. The action that is often taken is the release of contractures in the joints to obtain a sufficient range of motion. There is also an osteotomy if the contracture has caused a severe deformity.[7,8] In this retrospective study, the authors described the profile of AMC patients who underwent surgery at our center. This study also described the approach and management of AMC patients.

METHODS

This study design was a descriptive retrospective. The population of this study is all AMC patients who underwent surgery at the orthopedic hospital Prof. Soeharso Surakarta Indonesia. We used the total sampling technique who underwent surgery in the 2018-2021 period. All of these patients were admitted through outpatient care.

This research data is obtained by searching for the medical record file and electronic database from the department of orthopedic and traumatology, Universitas Sebelas Maret/Orthopedic Hospital Prof. Soeharso. AMC epidemiology data were obtained from patient data entered in the hospital, which has been assessed as an AMC based on physical examination and imaging support. The data is parsed for further analysis based on age, gender, and the type of surgery the patient underwent in the upper or lower extremities.

Data processing has first performed the selection, editing, coding, and tabulation. In this descriptive study, we found that all aspects of the criteria in the form of categories will be presented in a table.

RESULTS

In this study, we got eleven AMC patients who underwent surgery. Distribution based on sex found six boys and five girls. Like previous studies that have been carried out, the distribution of AMC was the same between boys and girls. There is no tendency in either sex.[9]

Based on age, there was no significant pattern in which half of the total patients underwent surgery under three years, while the other half of the patients underwent surgery after the age of five. As is well known, AMC is a congenital condition, so the abnormality's etiology occurs during pregnancy. The presence of patients who are consulted at an older age

Table 1. Syndrome types, labels, and OMIM numbers for the distal arthrogryposis (DA).[2]

Syndrome	Label	OMIM Number
Distal Arthrogryposis type 1	DA1	108120
Distal Arthrogryposis type 2A (Freeman-Sheldon syndrome)	DA2A	193700
Distal Arthrogryposis type 2B (Sheldon-Hall syndrome)	DA2B	601680
Distal Arthrogryposis type 3 (Gordon syndrome)	DA3	114300
Distal Arthrogryposis type 4 (scoliosis)	DA4	609128
Distal Arthrogryposis type 5 (ophthalmoplegia, ptosis)	DA5	108145
Distal Arthrogryposis type 6 (sensorineural hearing loss)	DA6	108200
Distal Arthrogryposis type 7 (trismus-pseudocamptodactyly)	DA7	158300
Distal Arthrogryposis type 8 (autosomal dominant multiple pterygium syndrome)	DA8	178110
Distal Arthrogryposis type 9 (congenital contractural arachnodactyly)	DA9	121050
Distal Arthrogryposis type 10 (congenital plantar contractures)	DA10	187370

Table 2. Data distribution of AMC patients.

No	Sex	Age (Month)	Surgery	Additional event
1	M	24	Reconstruction of right congenital hip dislocation	Congenital scoliosis
2	M	18	Reconstruction of right dislocation knee	Syndromic TEV bilateral
3	M	7 (y.o)	Reconstruction contracture of right wrist	Bilateral syndromic TEV
4	F	12	Release contracture of left ankle	Flexion contracture wrist bilateral
5	F	8	Release of left syndromic TEV	Flexion contracture of left wrist
6	F	36	Reconstruction of left dislocation knee	Left syndromic TEV
7	M	19 (y.o)	Osteotomy of the right foot	Syndromic TEV bilateral
8	F	5 (y.o)	Osteotomy of the right foot	Syndromic TEV bilateral
9	M	5	Release of right syndromic TEV	Left syndromic TEV
10	F	12	Release of bilateral syndromic TEV	Flexion contracture wrist bilateral
11	M	13 (y.o)	Osteotomy of the left foot	Flexion contracture wrist bilateral

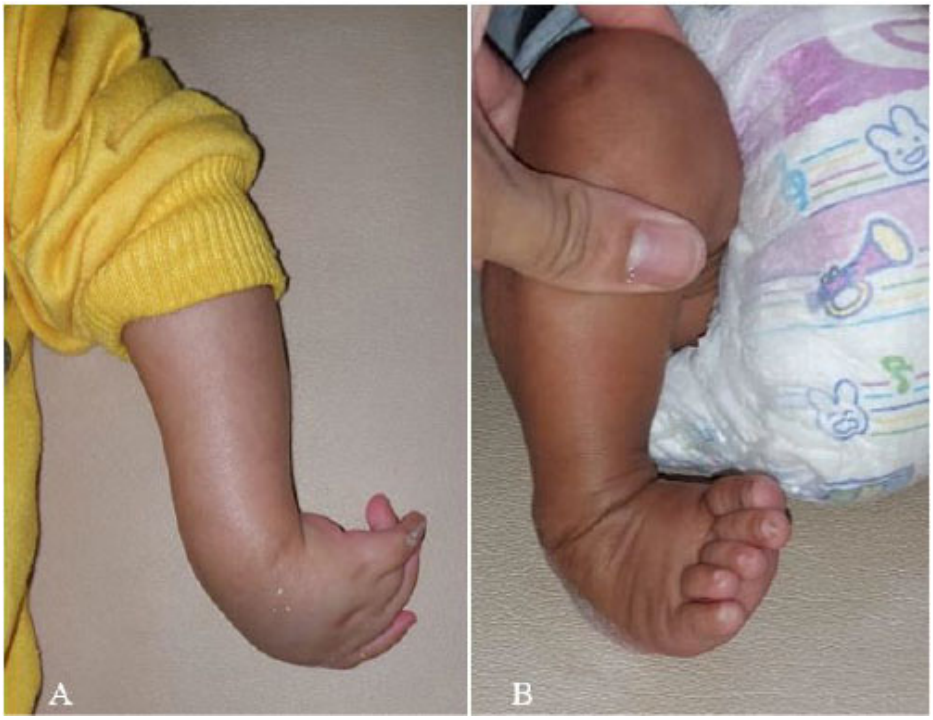

Figure 1. Shows the characteristics of deformity in the upper extremity (A) and lower extremity (B).

Figure 2. A child with AMC underwent Ponseti treatment on both his lower limbs, while both patient's upper extremities were stretched and splinted.

indicates a lack of public literacy about the abnormal condition of AMC or a lack of concern for the condition. Whereas early rehabilitation programs will increase the positive prognosis in patients with AMC. The data of this study can be seen in Table 2.

Almost all AMC patients who undergo surgery are in the lower extremities. This procedure is aimed at pursuing the child's standing and walking functions. Patients should be optimized and mobilized according to their age. However, there was one patient who underwent surgery on the upper extremity. This patient also had an additional event in the form of bilateral syndromic talipes equinovarus (syndromic TEV). We have done a serial ponsetti cast for that condition, and the condition is improving, so it was determined that no surgery is needed. Therefore, we performed surgery on the upper extremity. Here, all patients suffering from AMC will undergo the PMR program immediately after admission as outpatients at our hospital.

Table 2 showed that five patients with AMC who underwent surgery had contractures in the lower extremities; the remainder were combinations of the upper and lower extremities. No previous literature stated that AMC is more likely to occur in the upper or lower extremities. Generally, previous literature states that in AMC, contractures occur symmetrically, with both lower extremity, upper extremity or upper-lower extremity combinations on the same side. Additional contractures in more than two joints can occur anywhere.[3,9]

DISCUSSION

The diagnosis of AMC in infants is done clinically. Obtaining contractures in more than two joints with both lower extremities, upper extremities, or a combination of upper and lower extremities on the same side can be said to be an AMC condition.[3,9] In the upper extremity, the deformity that occurs is shaped; by internal rotation of the shoulder, elbow extension-pronation, wrist palmar flexion-ulnar deviation, fingers flexion, and thumb in the palm. Meanwhile, in the lower extremities, hip flexion- external rotation- abduction, knee flexion, and ankle talipes equinovarus occur (see Figure 1).[1]

The role of ultrasound is quite large in observation during pregnancy. Limited fetal movement space with loss of fetal movement (akinesia) and abnormal form of fetal extremities indicates that AMC is established.[10,11] If this condition is found, it becomes a vigilance for doctors and parents of the fetus. This section has multi-field collaborations; obstetrician, pediatrician, orthopedic, and medical rehabilitation.

Amyoplasia is the most common type of AMC. This situation can be interpreted as not developing muscle tissue.[4,5] There are reports of studies performing MRI examinations on infants. The study stated that the MRI evaluation findings could confirm the amyoplasia condition in AMC. There was a picture of drastically reduced muscle tissue in both of these patients' femurs compared to normal infant femurs. In addition, at the hip level, it is more emphasized with almost lost muscle tissue.[12] Muscle tissue that is reduced or even lost is replaced with fatty-fibrous tissue.[13]

Previous literature mentions three groups in AMC.[3] The first group disorders with mainly limb involvement. AMC with the absence of muscle tissue formation or called amyoplasia. The second group

is AMC, characterized by the formation of pterygium formations in several parts of the patient's body. The third group is disorders with limb involvement and central nervous system dysfunction. These three groups in the latest literature are simplified into AMC types: amyoplasia and distal arthrogryposis (DA).[1,2,5] The author in this study uses the division of AMC based on these two types. Pterygium is common in AMC with the amyloplast type. A loss of joint lines on the skin accompanies this web-shaped tissue. In comparison, AMC patients with central nervous system disorders will be discussed in certain syndromes.

It is known that arthrogryposis multiplex congenital (AMC) is a non-progressive syndrome characterized by multiple congenital joint contractures. Babies with AMC will experience disabilities in the course of their lives. Rehabilitation plays a very important role in improving the quality of life for AMC patients. There is no need to wait for a certain age to start the PMR program. Rehabilitation measures, such as stretching, and external support, such as splinting and casting, are carried out immediately. The first three weeks of age is the golden period for doctors and teams to start PMR. If this opportunity is used properly, it will reduce the possibility of surgery at a young age (see Figure 2).[14]

There are approaches to facilitating rehabilitation programs for AMC patients. We can divide therapeutic attainment into three stages.[15] The first stage is improvement in body function and structure. At this stage, focus on pain as well as muscles and joints. Pain will be felt by the patient when passive stretching is done. Physical medication is needed to reduce these complaints.[14] The first stage is also to repair muscles and joints. Usually, external support such as splinting, and casting is done.

The second stage is activity. At this stage, the physician will rehabilitate to increase the patient's self-care ability and mobility. The third stage is advanced in the form of the patient's ability to participate in society. These two stages, which play a large role, are specialist pediatricians and the rehabilitation team.[15]

The operation is in the first stage, namely improvement in body function and structure, especially in muscles and joints. Surgery is a last resort if the deformity or range of movement (ROM) does not improve with stretching and external support. For that, we need to know the normal range of motion and the functional position of these joints. In the elbow joint, as stated in the literature, the ROM of 30 to 130 degrees of elbow flexion and 50 to 50 degrees of pronation/supination is sufficient to achieve most positional and functional tasks in children and adolescents.[16] Furthermore, the ROM required for a functional wrist joint is 40 degrees dorso/palmar flexion and 40 degrees radial/ulnar deviation.[17] In the hip joint, in order for a person to be able to do a squatting position, ROM flexion is 110 degrees, abduction is 9 degrees, and external rotation is 18 degrees.[18] In the knee joint, to carry out the function of walking, a minimum ROM of extension/flexion of 0-60 degrees is required.[19] This condition is, of course, with the ankle and foot planty grade position.

These positions need to be pursued in the rehabilitation process for AMC sufferers. If there are difficulties, surgery can be performed. Prioritizing stretching, splinting, and casting will simplify surgery, hoping that single-event multilevel surgery (SEMLS) can be performed. Ultimately, it will accelerate the achievement of treatment goals for AMC patients.

There are limitations to this study, such as the small number of cases found. This could be due to the medical record system which does not display AMC as a diagnosis but is described according to each contracture or deformity present in the patient. So there is a bias in the actual number of AMC cases.

CONCLUSION

Arthrogryposis multiplex congenital (AMC) is a complex abnormal condition characterized by multiple congenital contractures in the body's joints. A multidisciplinary approach is needed in the treatment of AMC patients. Obstetrics, pediatrics, orthopedics, and rehabilitation have a stake in managing PMR and surgery. Three stages are the goals of treatment, the first is improvement in body function and structure, the second is activity, and the third is participation.

DISCLOSURE

Author Contribution

Author 1: Acts as a doctor and surgeon who performs actions on patients. Determine the theme of the case series, study concept, and design as well as make corrections to the writing.

Author 2: Acts as doctors and surgeons who perform actions on patients. Make corrections to the writing of the study.

Author 3: Acts as the data collector, data analysis and interpretation as well as writing the paper.

Ethical Consideration

The ethical commission of Medical Faculty, Universitas Sebelas Maret has approved this study. The patients also have been permitted to join in the research after giving informed consent to the parents.

Conflicts of interest

All authors state that there are no financial and personal relationships with other people or organizations that could inappropriately influence (bias) for this study.

Sources of funding

In this study, we use our resources without any financial assistance from other parties.

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