Case Study

Role of Early Rehabilitation in An Infant with Arthrogryposis Multiplex Congenita: A Case Report with 11 Months of Follow up

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Abstract

Background: Arthrogryposis multiplex congenital (AMC) is etiopathogenetically a heterogeneous disorder which is considered to be a neuromuscular syndrome present at birth. It is characterized by presence of contractures in more than two body areas at the prevalence rate of 1 in every 3000 to 5000 live births.

Purpose: There are no reports in the literature which provides clear guidelines regarding physical therapy interventions for children with AMC. The purpose of this case report is to document the infant's recovery based on the frequency and duration of physical therapy interventions during first 11 months of life.

Key points of case: An infant with arthrogryposis multiplex congenital was followed from day 15 to 11 months of early developmental period. Following continuous and integrated physical therapy, infant achieved normal developmental sequence with visible improvement in joint contractures. Without intervening surgically, there is improvement in club foot of the baby. This article enlightens physiotherapeutic treatment strategies for child with AMC.

Conclusion: Physical therapy of a child with AMC should be multi-centred, holistic and continuous. Early approach to physical therapist minimise the complications following AMC. Early physical therapy interventions can help in prolonging the early need of surgical interventions during developmental age.

Keywords: *Physical therapy, Infant, Early Rehabilitation, Arthrogryposis*

Introduction

Arthrogryposis multiplex congenital (AMC) is etiopathogenetically a heterogeneous disorder which is considered to be a neuromuscular syndrome present at birth. It is characterized by presence of contractures in more than two body areas at the prevalence rate of 1 in every 3000 to 5ooo live births. The primary insult is expected to be present during the first trimester of pregnancy. The severity tapers down if the insult occurs later in the pregnancy.[1-6]

The etiology of AMC is multifactorial & sporadic. The most possible cause could be reduced foetal movements. This foetal akinesia can be the outcome of neuromuscular conditions or uterine abnormalities due to presence of maternal diseases. Twin pregnancy, bicomuate uterus and oligohydroamnios are possible examples of causes reducing the foetal movements.[1, 4, 7, 8]

Arthrogryposis can be in the form of Amyoplasia with characteristic features of shoulders being internally rotated and adducted with elbows extended and wrist flexed with ulnar deviation. Along with it the hips may be

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dislocated with extended knees and feet in equinovarus. This type of AMC children can have normal intelligence. Other forms of AMCs can be related to central nervous system or can be of heterogeneous aetiologies including congenital, chromosomal abnormalities, skeletal dysplasia, and contracture syndromes. [1, 9]

The presentation of symptoms can be different in each patient with AMC which includes difference in severity of contractures in different body regions along with involvement of other systems of the body. Despite of being non progressive condition, the joint contractures can be recurrent. [10-13]

In a growing foetus it is essential to have movements in order to achieve milestones. Restricted movements can become a causative factor for excessive development of the peri articular connective tissues. To achieve this, early and continue physical therapy is a must. This case report enlightens on early intervention for a child with arthrogryposis multiplex congenital. The purpose of this case report is to document the child's recovery based on the frequency and duration of treatment.

Case description:

History and examination: The girl infant was born after second pregnancy to healthy, non consanguineous parents. She was born out of full term pregnancy without any prenatal or natal history. Her birth weight was 2.9 kilograms. She was referred to physical therapy department on day 15 of life with the complaints of joint contractures and the child being less active. No signs of deformations were marked during routine ultrasound examinations. Post-natally during the routine visit the infant was referred to physical therapist.

Systems review:

Musculoskeletal: The infant preferred to keep her head turned to left most of the time with no head control.

Postural observations-

Plagiocephaly (Flat head) is seen on the posterior aspect of the head.

In bilateral upper extremities; shoulders were flexed and internally rotated, forearm pronated with wrist and fingers in flexion. Hands were fisted, ulnarly deviated with thumb in attitude. Bilateral lower extremities were flexed from hip and knee along with severe club feet seen in feet along with overriding of toes.

Initial examination revealed presence of micrognathia & retrognathia with flattened head. Multiple joint contractures were noticed at shoulder, elbow, wrist, hip, knee and ankle along with tightness developing on the left side of neck.

Club feet were marked on dimeglio scale at the grade of III.

Developmental dysplasia of hip was ruled out using x- ray hip.

Neuromuscular: Possible active movements were slight elbow flexion with more of shoulder internal rotation and bilateral kicking in lower extremities with right more than left. Muscle tone, assessed by resistance during passive movement was found to be normal in the available range of motion. Tendon reflexes were found to be normal.

Developmental milestones & reflexes:

Social smile: Present

Sucking: Poor

Rooting: Poor

Reaction to external stimuli: Poor

Palmar grasp: Present

Plantar grasp: Present

Flexor withdrawal: Present

Cognition/ response to pain: Baby had social smile and recognition to mother's touch. Baby used to cry while entering the department and while exposing to new people. Movement in joints stimulated cry in baby.

Integumentary: Issues related to skin irritation, marks or breakdown of skin were absent.

Cardiopulmonary: The baby did not show any positive signs related to cardiopulmonary system. No comments regarding cardiopulmonary status were made by paediatrician of the baby.

Baby was reacting to touch, pain, sound and light.

Description of outcomes:

Passive ROM: For all the joints passive ROM was measured using Gonio-meter. Goniometry is good tool with excellent reliability and validity.[14, 15] ROMs were restricted in bilateral UEs, LEs and cervical spine.

Prone tolerance was measured by keeping the baby in prone position. Initially the time was 10 seconds. Crying was a limiting factor.

Evaluation using ICF model:

Body structure & function impairments: Reduced ROMs in bilateral UEs, LEs and spine, asymmetry at neck

Activity limitations: Decreased movements of all four limbs, Discomfort in prone lying, unable to extend both upper and lower extremities

Participation restriction: Baby had difficulty in exploring the environment in a manner typical of age matched infant.

Description of Intervention:

Therapeutic measures began at the age of 15 days when the infant was referred by orthopaedist to paediatric physical therapy department.

Aims of physical therapy:

To mobilise the foot towards corrective position

To increase the ROMs of joints

To prevent muscle tightness

To enhance neuro sensory motor development

Techniques used:

To increase the mobility of joints

- Gentle massage by using stroking, kneading, picking up and rolling

- Stretching of the muscles of neck , pectorals, elbow flexors, wrist and finger flexors

(30 seconds hold for each stretching position repeated for 3 times)

- Gentle mobilization of foot towards the stretching of ligaments, capsules and tendons

- Passive ROM exercises for each joint (10 repetitions for each joint)

- End range stretch maintained for 30 seconds each time

- Gradual training towards attaining active ROMs with the use of age appropriate toys such as squeaky toys (Minimum 10 repetitions of each movements)

To facilitate neuro sensory motor development:

- Stimulation of sucking and rooting reflexes

- Graded sensory stimulation with the use of different texture and toys in order to overcome thumb-in attitude of hands

(Stimuli- Response- Stimuli sequence)

- Visual tracking of musical and lightening toys

- Different auditory and verbal cues to improve the awareness of external environment

- Prone positioning was advised for most of the time of the day except one hour after each meal. (To promote head holding and avoid flattening of head)

- Positional therapy with the use of bolster, wedges and physioball

- Facilitatory rolling was incorporated on both sides

- Sitting on a bolster roll

- Proprioceptive stimuli for hand opening and foot placement

- Facilitating prone extension while on physioball

- Kinesio taping for correction of scapular position (performed two times in entire therapy duration)

- Counselling to the parents was an important aspect of therapy session. Parents were advised to swaddle the baby with knees and elbows in extended position, carrying the baby on each side simultaneously

in order to avoid fixed position of baby's neck while carrying the baby and repeating the ROM exercises at home.

Frequency of therapy sessions: Initially the infant was seen on everyday basis for a session of 1 hour with small breaks during the sessions. From the age of 3 months the child is been called up thrice a week. Now the child is of 11 months of age and still continuing therapy.

Short term goals of physical therapy sessions were achieved during the first 4 months of therapy and then the baby started achieving different developmental milestones in order to achieve optimal long term goal. The baby is currently able to access the environment by crawling and independent sitting. She is able to stand with support along with the persisting minimal contractures at knees and elbows. No casting is done by the orthopaedist so far but regular consultation is done.

Table 1: Difference in Passive Range of motions at different joints

Joints of the body	**Movement**	**Day 1 rehabilitation**		**11 months post rehabilitation**		**Difference in range of motion**	
		Right	Left	Right	Left	Right	Left
Cervical	Lateral flexion	25º	35º	35 º	40 º	10 º	5 º
	Rotation	60 º	80 º	70 º	80 º	10 º	0 º
Shoulder	Flexion	100 º	100 º	170 º	170 º	70 º	70 º
	Extension	20 º	20 º	80 º	80 º	60 º	60 º
	Abduction	110 º	110 º	160 º	160 º	50 º	50 º
	Internal rotation	90 º	90 º	90 º	90 º	0 º	0 º
	External rotation	60 º	60 º	80 º	80 º	20 º	20 º
Elbow	Flexion	50 º -140 º	50 º -140 º	20 º -140 º	30 º -140º	-	-
Wrist	Flexion	70 º	70 º	70 º	70 º	0 º	0 º
	Extension	60 º	60 º	70 º	70 º	10 º	10 º
Finger	Flexion	Not assessed	Not assessed	FULL	FULL	-	-
	Extension	Not assessed	Not assessed	FULL	FULL	-	-
Hip	Flexion	FULL	FULL	FULL	FULL	-	-
	Extension	50 º	40 º	70 º	70 º	20 º	30 º

Cont... **Table 1: Difference in Passive Range of motions at different joints**

	Abduction	30 º	30 º	40 º	40 º	10 º	10 º
Knee	Flexion	40 º -120 º	40 º -120 º	20 º -120 º	20 º -120 º	20 º	20 º
	Extension	-40 º	-40 º	-20 º	-20 º	20 º	20 º
Ankle	Dimeglio scale grade 3 for club foot (Present Bilaterally)						

Table 2: Summary of Outcome data

	At the age of day 15	At the age of 11 months
Prone positioning time	3s	Able to stay in prone position, crawls nicely
Developmental milestones achieved	Social smile	Head control Rolling bilaterally Prone positioning Crawling Sitting independently Standing with support
Passive ROMs	Restricted	Almost full
ICF model	Participation restricted in environment	Participation improved at environmental, social and functional level

Discussion

Arthrogryposis multiplex congenital is a rare and heterogeneous in nature. Children with AMC should be dealt with interdisciplinary problem solving approach. It is been found that there is very less literature on rehabilitation strategies for treating children with AMC which makes rehabilitation professionals less prepared while treating children with AMC. This case report provides information on rehabilitation sessions and specific treatment strategies for a growing infant with AMC.[16, 17]

For the children with AMC physical therapy play an important role in making these children independent. Recent review of studies on AMC suggests that detailed description of specific therapy session is not clearly mentioned in any literature. Through this report, we tried to convey duration, frequency and therapeutic modalities which were used in treating the girl infant with AMC.[18]

During 11 months of physical therapy there was improvement in joint contractures, limb axis alignment, developmental milestones, postural deviations and environmental participation. The child has become more playful and active. Child has achieved milestones of sitting and crawling independently. Child is able to

stand with support. There were no episodes of joint dislocations or any other unanticipated events during the period. Her casting was delayed as there is noticeable improvement in feet alignment with regular physical therapy. Intensive and continued rehabilitation since the early age helped in prevention of further complications of AMC.

In cases with arthrogryposis surgical correction is required for feet (76% cases), knees (39%) and hips (18%). For the case described no corrections are done surgically instead joints are maintained with physical therapy interventions. This could be possible because of early physical therapy interventions. The possible outcome is supported with the literature stating initial corrective therapy improves the result during first months of life.[19]

As in this infant with AMC neonatal reflexes were found to be poor during initial assessment, specific attention was given in providing sensory stimuli to facilitate sucking and rooting in order to prevent malnutrition. The available literature emphasises that movement restrictions can hamper the ability to explore the world in children with AMC which may affect their cognitive and motor development. Here in this infant the neuro-developmental sequence was equally focused upon.[20, 21]

Along with physical therapy sessions, parents' active participation was commendable during this entire process. Generally parents of children with long term illness experience frustration, depression and disappointments. Their everyday life will change to some extent. Parents of this child were lively, supportive and educated enough to cope up with the ongoing therapy sessions without missing any session except on emergencies.[22]

The goals will change once the child ages. The improvements of ROMs and developmental milestones will help in forming further goals of ambulation and daily life activities.

Conclusion

Physical therapy of an infant with AMC should be multi-centred, holistic and continuous. Early physical therapy interventions can help in prolonging the early need of surgical interventions during developmental age.

Consent: Written informed consent was taken from the parents' for utilisation of informations and images of their child for the publication purpose.

Grant support: This study was not supported by any grants or funding.

Ethical Clearance: It was taken by the Institutional ethical committee.

Conflicts of Interest: Nil

Acknowledgements: I thank parents of the infant for their cooperation and support for providing me their consent for the publication.

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