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CASE REPORT / ПРИКАЗ СЛУЧАЈА

Rehabilitation of children with arthrogryposis

Рехабилитација деце са артрогрипозом

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ABSTRACT

Arthrogryposis is a congenital, multiple and non-progressive deformation of the joints, which is characterized by a multiple contractures of the joints at the child's birth. Stiff joints are cylindrical in shape, retaining usual contours, with partially erased muscle and skin outlines. The classic form of arthrogryposis is usually referred to as amyoplasia in the current literature. The case presented here is of a female child aged 17, with the diagnosis of Arthrogryposis multiplex, categorized as mildly mentally disabled, semi-mobile and using wheelchair. The female subject is with deformities of both upper and lower extremities and is completely dependent on the help of another person in matters of usual daily activities, such as maintaining her personal hygiene and satisfying her basic living needs, also in practicing her basic skills. The rehabilitation plan is grounded upon the diagnosis of disease, as well as on the results based on a functional assessment. The rehabilitation plan included daily exercises, passive exercises for lengthening of the soft tissue structures, active sustained exercises, active exercises and those with load on all joints according to the estimation made by the manual muscle test. The plan was being implemented during the research over the course of one whole month. The patient cooperated greatly and was motivated to work.

Treating children with arthrogryposis requires cooperation between physiatrists, surgeons, physiotherapists, occupational therapists, social workers, orthotics, with early and complete engagement of parents. Therefore, the treatment of arthrogryposis necessitates to be approached by all aforementioned experts as a team.

Key words: congenital abnormalities; musculoskeletal abnormalities; arthrogryposis; physical and rehabilitation medicine; adolescent.

АПСТРАКТ

Артрогрипоза је урођена, вишеструка и непрогресивна деформација зглобова, које се карактеришу мултиплим контрактурама зглобова на рођењу детета. Укочени зглобови имају цилиндричан облик, задржане контуре и делимично избрисан рељеф мишића и коже. Класичан облик артрогрипозе се у новијој литератури обично зове амиоплазија. Приказан је случај женског детета узраста 17. година са дијагнозом Arthrocryposis multiplex, категорисана као лако ментално ометена, полупокретна и користи инвалидска колица. Код испитанице присутни су деформитети горњих и доњих екстремитета и у потпуности је зависна од помоћи друге особе када су у питању свакодневне уобичајне активности одржавања личне хигијене и задовољења основних животних потреба и вештина. План рехабилитације се заснива на дијагнози болести, као и на резултатима добијеним на основу функционалне процене. План рехабилитације обухватао је примену свакодневних вежби, пасивних вежби за истезање мекоткивних структура, активно потпомогнутих вежби, активних вежби и активних вежби са оптерећењем у свим зглобовима према оценама на мануелном мишићном тесту. План је спровођен за све време истраживања у трајању од месец дана. Испитаница је одлично сарађивала и била мотивисана за рад.

Лечење деце са артрогрипозом захтева сарадњу физијатра, хирурга, физиотерапеута, радног терапеута, социјалног радника, ортотичара, уз рано и потпуно укључивање родитеља. Дакле, лечење артрогрипозе захтева тимски приступ свих стручњака.

Кључне речи: конгениталне абнормалности; мишићноскелетне абнормалности; артрогрипоза; физикална и рехабилитациона медицина; адолесцент.

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INTRODUCTION

Morphological changes of locomotor apparatus which propagate bigger or smaller functional distresses are called body deformities. Deformities are not so rare phenomenon in children. According to a research carried out in conformity with the recommendations of World Health Organization, 1000 examinees had been scrutinized, with results pointing out that 36% of children is without deformities, 55% with one deformity, and 9% with more than one deformity detected. By type of causer, body deformities are divided into: congenital (inborn) and acquired deformations. Congenital disorders are structural defects of tissues and organs which originated during morphogenesis, the ones that are present and visible at birth. Different kinds of anomalies are conditioning or following different disorders in physical and mental functioning, reflecting on one's social status and function. Congenital deformities of newborn child which occur in praxis (minor or major malformations) are not directly life endangering and they do not require urgent therapeutic measures. Nevertheless, certain congenital deformities ought to be recognized immediately after birth, already in delivery room, so the proper therapeutic measures could be taken in order to prevent serious endangering of a child's life.[1-3]

Arthrogryposis is a congenital, multiple and nonprogressive deformation of the joints, which is characterized by a multiple contractures of the joints at the child's birth. Stiff joints are cylindrical in shape, retaining usual contours, with partially erased muscle and skin outlines. The classic form of arthrogryposis is in current literature usually referred to as amyoplasia.[3-5] The main cause of arthtrogryposis is not quiet known, but there are several hypothesis, which include viral infections during pregnancy, fetal hyperthermia (overheating of an embryo), disorders in blood provision of an embryo, decreased amount of amniotic fluid, usage of some medicaments, alcohol and drugs during pregnancy. According to some theories, influenza virus, measles and other viral infections in the first months of pregnancy are responsible for the occurrence of arthrogryposis. There are two clinical forms, neuropathic and myopathic. Tendon reflexes in both clinical forms are lowered or silent, while sensibility is preserved. Small, vaguely bounded area of mildly red pigmented skin can be seen between eyebrows, a so-called stork bite. Skin above the joints caught by arthrogryposis can be writhed, while it is as a whole thin, due to a reduced amount of subcutaneous tissue. Extremities are fusiform or cylindrical in shape, and muscles are atrophic, while some muscle groups can be completely missing. Joints are subluxated, or completely luxated, especially hips and knees.[6-8]

Arthrogryposis is a deformity which, in addition to surgical interventions, immediately after delivery, also requires a long process of rehabilitation as one of the most important components of a treatment. In the process of rehabilitation of body deformities, early and continuous rehabilitation is of utmost significance. Rehabilitation treatment is preceded by scrutinizing of psychological, physical, physical, social and professional consequences of deformities. [9] There are no universal methods of rehabilitation, as they are being adapted to a given case with an importance of continuously carrying out the process of rehabilitation. Early rehabilitation can lead to significant functional capacitating altogether with greater abilities of a child for daily life activities (DLA). Continuous treatment can contribute to an education process and later employment of such children. Family is also to be included in rehabilitation process and by the methods of counselling, sociotherapy and education of parents are being applied. Education refers to a training in properly doing the exercises, including their appropriate duration, to possible combinations of exercises and requisites to be used, as well as to various kinds of improvisation in exercising in domestic conditions. Parents should be advised to accept their child as it is and guided to acknowledge it's real abilities. Rehabilitation is an interdisciplinary activity in which all members of a team are equal. Though the process of rehabilitation lasts longer than other forms of medical treatment, at one moment it must come to an end. Usually it's the moment when therapeutic goals are achieved, and the patient had been made feasible up to the maximal limit.[10-12] Nevertheless, if the achieved status is not being kept up, body deformity could easily be reverted to a previous condition, sometimes resulting in an even more severe clinical picture than before, especially in children. It is necessary to educate the patient and to give him knowledge from the area that his deformity belongs to, to inform him how the rehabilitation is going to be carried out, which are possible complications and consequences of absence of therapy. Thereby the patient is aware of situation and actively takes sides by the physiotherapist.[13-16]

Nowadays body deformities are great disturbing factor in child's growth and development. Besides esthetical problems, they cause many other health problems to a man, which are ought to be in due time therapeutically taken care of. Treating of body deformities requires close cooperation of physical medicine therapist and physicians in order to diagnose and administer adequate therapy (corrective exercise, using of certain medical apparatus, surgical intervention).[17-20]

CASE REPORT

Psychophysical status

Female patient, aged 17, categorized as mildly mentally disabled, semi-mobile and in wheelchair. Due to a degree of her invalidity and the fact that she's mentally challenged, her hygiene habits and habits of self-help are not developed. She does not control her physiological needs. Her verbal abilities are well developed and, pursuant to her age and social setting, it can be said that has got a rich vocabulary. She understands instructions and requests and responds in a verbally adequate manner. She's a sweet, smiling girl, always in a mood for social communication. She is interested in all happenings inside the group, in the beginning a little bit insecure at the first contact with previously unknown people, but capable of quick accommodation with a bit of a support. She shows initiative and motivation for taking part of the group activities, putting an effort into it. Regardless of hand deformities, she's trying to carry out her activities succsesfuly; she can differentiate and name colors, she's well oriented in space and time, she can enumerate days of the week and she knows of how many seasons a year consists. She's not a conflictive person. She's fond of listening to music, likes to look nice and neat, loves to spend time with adults who take care of her. Most of the day she spends in the wheel chair, communicating with other children and watching TV. One comes away with an impression that the patient possesses solid socially cognitive and psychic abilities which, unfortunately, hadn't been adequately developed due to a non-stimulating and untimely working with her.

Health status

The patient was diagnosed with arthrocryposis multiplex at birth. She has deformities of upper and lower extremities, she's semi-mobile and uses wheelchair. Her occasional ear inflammation is of chronic character. She doesn't control her sphincters. She is overweight. Her menstrual cycle is regular. She doesn't have any other health issues.

Legal status

She was abandoned by both of her parents right after her birth. Since she was born, until 2005, she resided in an asylum in Kulina. Since 2005 she had been transferred to a Home for uninhabited children in Niš, where she is still a resident. Her parents are completely devoid of any parental right.

Self-dependence and self-care

Completely dependent on the help of other person in matters of usual daily activities, self-hygiene maintenance and satisfying basic life needs and abilities. Deformities of upper extremities restrict her in self-dependant nourishment, so she is usually being fed by a nurse. She recognizes all the cutlery during the meal, but she's not capable of using it. She can't self-dependently maintain her personal hygiene, she's not able to wash her face, brush her teeth, wash her hands, nor to take a bath. She likes to be neat and if her clothes get dirty, she asks to change; she recognizes the clothes. She knows how to combine her clothing with weather conditions, but, because of aforementioned deformities, can't dress herself. Having adopted the circadian rhythm, she knows the succeeding of the activities, taking an active participation in workshops. With the support and help from an adult, she get to see a final object of workshop's task, her reactions are in elated and euphoric manner, so that she has always wanted to work some more. When the action is three times repeated, this patient knows by the fourth time to independently iterate it by the sequence of execution, which speaks to a distinct degree that the epatient possesses clearly developed scheme of actions necessary for performing certain activity.

Social status

The patient easily establishes social contacts, considering the fact that her verbal abilities are well developed; she actively interacts with adults and her age-mates from group. She positively accepts socially eligible behaviour, she's adapted to an environment, very companionable, always in cheerful mood, and when something bothers her or she doesn't find it suitable for her, she doesn't verbalize that, but withdraws to herself and bursts into tears. The patient takes an educational group as her own, developing distinctive feeling of belonging and she grows a protective attitude towards each member of the group. She spends most of the time with the patient, who occasionally gives her support and helps her with feeding or by passing her things she makes a bid for. The patient knows names of every member of the group, names of all adults in Home, her attention is properly directed and she easily makes a direct contact. Because of her inability to shake hands she utilizes usual stereotypes: winking and blowing kisses.

Table 1. Patient's problems

Problems			
Social skills	Communication skills	Self-dependence and self-care	General health
	Occasional stereotypical communication	Completely dependent on other people's care and attendance	Chronic ear inflammations
		Limited coordination of upper and lower extremities	Increased body mass
Priority problem: due to a constant confinement to the wheelchair, together with good appetite, possible risks are burdening of spinal column because of a body mass increase, by which is her health condition endangered			
Goal: preserving health condition and maintaining body mass at lower level, as well as preserving and improving communication and social skills			

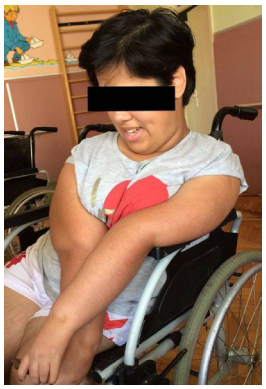

Figure 1. Patient in wheelchair

Figure 2. Stationary position

PLAN OF REHABILITATION

Plan of rehabilitation is based upon primary diagnose as well as upon results pursuant to functional estimation. It had been carried out over the course of the research lasting one month. The patient cooperated greatly and had been motivated to work. On the first day, after getting acquainted with the patient, a functional procedure had been done, including identifying contractures, estimating muscle strength, measuring an amplitude of joints movements, measuring size and length of extremities, estimating motor self-dependence. After determining existing abilities of the patient concordant to existing deformities, the plan of rehabilitation had been made, meaning a continuous treatment. The rehabilitation plan included daily exercises, passive exercises for lengthening of the soft tissue structures, active sustained exercises, active exercises and those with load on all joints according to the estimation made by the manual muscle test. The plan for muscles of head and neck meant daily performing of active exercises with growing number of iterations for increasing the muscle strength. The plan for muscles of upper extremities meant performance of passive exercises for lengthening of the soft tissue structures, reaching the pain limit, as well as performing active sustained exercises for increasing the amplitude of movements. The plan for muscles of lower extremities meant performing of active exercises for tight muscles, passive lengthening by positioning relying on a gravity in the knee joint, as well as sustained active exercise for foot. Dail agenda of breathing exercises, as well as turning on the side and positioning, had been planned. Daily increase of resistance, where it is possible, as well as number of repeating of movements, had been also planned. The plan for improvement of transfer from bed to wheelchair had been also made

CARRYING OUT OF REHABILITATION

Rehabilitation treatment had been carried out over the course of 30 days. Treatment included performing of kinesiotherapy by the patient in her bed, in different positions (mostly in a supine position and side position), with verbal communicating and explaining how is each of exercises to be done and why should it be done, on demand of the patient (figures 1-5). The progress could be noticed on daily basis as well as the importance of continuous work concerning rehabilitation treatment.

Passive exercises, sustained active exercises and active exercises of upper and lower extremities, in all areas where it was possible to perform any kind of a movement because of existing deformities together with great degree of contractures in certain joints, had been done with the patient.

Figure 3. Performing a movement of extension in both arms shoulders, a passive movement

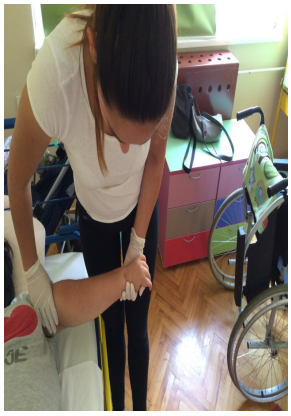

Figure 4. Performing a movement of abduction in left arm shoulder, a passive movement

Figure 5. Performing a movement of extension in hip with existing contracture in knee

Finer handling was also exercised with the patient, which she managed to improve up to limits of her possibilities, meaning thus much as her existing deformities could allow. (figures 6 and 7)

Figure 6. Exercise, part 1

Figure 7. Exercise, part 2

MENTAL ESTIMATION

The patient had shown significant communication abilities, she had been very curious, wanting to know everything related to rehabilitation treatment. Through conversation, Jane Doe had displayed various interests for the world around her as well knowledge of her own disease. It's been noticed that the patient is of a lower average of intelligence but with a significant educational negligence. Insufficient work together with relatively recent starting with school concerning her age point out why the patient still hasn't got knowledge from certain social fields. She's currently attending School for mentally disabled children in Niš, she's in the fifth grade of primary school and an excellent pupil with proper behavior. Over the course of the research the patient made an extraordinary advance in both mental and social aspects, indicating the significance of continuous treatment.

Figure 8. The patient is drawing

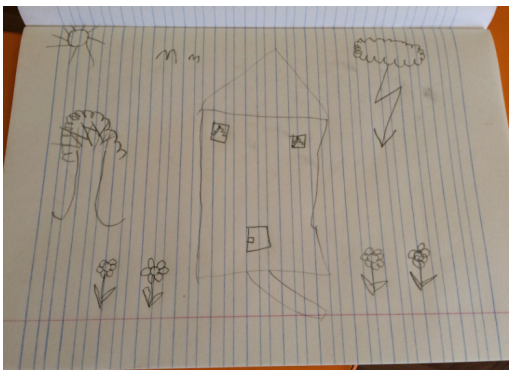

Figure 9. The drawing of the patient

COMPARATIVE STATUS

Table 2. Comparative status of the research according to the functional estimation 1

Parameters of the research		Before the research	After the research
Inspection of the deformities	hip	luxation	the state remained unaltered
	knee	flexion contracture	the state remained unaltered
	foot	equinus – right equinovarus - left	the state remained unaltered
	shoulder	adduction contracture	the state remained unaltered
	elbow	extension contracture	the state remained unaltered
	hand	flexion contracture	the state remained unaltered
	spinal column	lumbar scoliosis and thoracic kyphosis	the state remained unaltered
Inspection of the position	supine	conditioned by deformities	alleviated
	prone	somewhat difficult	alleviated
	on side	conditioned by deformities	alleviated
	sitting	in the wheelchair	in the wheelchair, alleviated
		impossible	the state remained unaltered
Transfer	from bed to wheelchar	with a help from another person	alleviated
	from wheelchair to bed	with a help from another person	alleviated

Table 3. Comparative status of the research according to the functional estimation 2

Parameters of the research		Before the research	After the research
Manuel muscle test	muscles of head and neck	flexion 3+ extension 3+ lateral flexion 3+	flexion 4 extension 4 lateral flexion 4
	muscles of upper arm	flexion 2- extension 1 abduction 2- adduction 1 external rotation 1 internal rotation 1 horizontal abduction 2- horizontal adduction 2-	flexion 2 extension 1 abduction 2 adduction 1 external rotation 1 internal rotation 1 horizontal abduction 2 horizontal adduction 2
	muscles of fore arm	flexion 1 extension 1 supination 1 pronation 1	flexion 1 extension 1 supination 1 pronation 1
	muscles of hand	flexion 1 extension 1 abduction 1 adduction 1	flexion 2- extension 1 abduction 1 adduction 1
	muscles of trunk	flexion 2 extension 2 lateral flexion 2 rotation 2-	flexion 2+ extension 2+ lateral flexion 2 rotation 2
	muscles of upper leg	flexion 3- extension 3- abduction 3 adduction 3 external rotation 2- internal rotation 2-	flexion 3 extension 3 abduction 3+ adduction 3+ external rotation 2- internal rotation 2
	muscles of lower leg	flexion 1 extension 1	flexion 1 extension 1
	muscles of foot	flexion 1 extension 1 abduction 1 adduction 1	flexion 1 extension 1 abduction 1 adduction 1

Table 4. Comparative status of the research according to the functional estimation 3

Parameters of the research		Before the research	After the research
After the research	head and neck	flexion 25° extension 20° lateral flexion 23° rotation 50°	flexion 32° extension 26° lateral flexion 30° rotation 60°
	hand	shoulder flexion 70° shoulder extension 10° shoulder abduction 30° shoulder adduction 10° shoulder internal rotation 15° shoulder external rotation 15° forearm flexion 20° forearm extension 20° forearm pronation 10° forearm supination 10° hand flexion 15° hand extension 5° hand abduction 2° hand adduction 3°	shoulder flexion 76° shoulder extension 12° shoulder abduction 32° shoulder adduction 10° shoulder internal rotation 15° shoulder external rotation 15° forearm flexion 20° forearm extension 20° forearm pronation 10° forearm supination 10° hand flexion 15° hand extension 6° hand abduction 2° hand adduction 3°
	leg	hip flexion 50° hip extension 15° hip abduction 30° hip adduction 15° hip external rotation 15° hip internal rotation 10° knee flexion 8° knee extension 3° foot flexion 2°f foot extension 2° foot eversion 2° foot inversion 2°	hip flexion 60° hip extension 17° hip abduction 32° hip adduction 16° hip external rotation 16° hip internal rotation 11° knee flexion 8° knee extension 3° foot flexion 2° foot extension 2° foot eversion 2 ° foot inversion 2°
	trunk	flexion 35° extension 10° lateral flexion 5° rotation 5°	flexion 45° extension 15° lateral flexion 5° rotation 5°

Table 5. Comparative status of the research according to the functional estimation 4

Parameters of the research		Before the research	After the research
Size of extremities	upper arm	upper-level – 32 cm lower-level -30 cm	upper-level - 30 cm lower-level -29,5 cm
	elbow	29 cm	29 cm
	fore arm	30 cm	29 cm
	wrist	15,5 cm	15,5 cm
	hand	13,5 cm	13,5 cm
	upper leg	upper-level - 65 cm middle-level - 52 cm lower-level - 46 cm	upper-level - 63 cm middle-level - 51cm lower-level - 44 cm
	knee	47 cm	46 cm
	lower leg	42 cm	41 cm
	hock	29 cm	29 cm
Length of extremities		arm	the state remained unaltered
		leg	the state remained unaltered

DISCUSSION

Multiple congenital contractures or arthrogryposes are visible state of limited motility of joints at child's birth. There are more than 150 diseases associated with similar clinical picture. Amyoplasia congenita is today mostly used as a name for a classical arthrogryposis. It covers 43% of all multiple congenital contractures. The incidence of arthrogryposis is 1:3000 newborns by us, as well as it is throughout the world. The manner of inheritance remains unknown. Two types are distinguished: neuropathic type, which is present in 90% of cases, and myogenic type, which is present in 10% of cases of children with arthrogryposis. In 60% of patients there is an equal hold on all limbs which are in a characteristic symmetrical position. Both sexes are equally frequent.

Deformities of a body, including arthrogryposis, are today a widespread problem, especially in youth. When deformities of a skeleton are taken into consideration altogether, it is even one third of children of school age that have some kind of a deformity, and there are even more deformities occurring at the same time in a certain number of children. For instance, deformities of spinal column are stated in 15,4% of school children, so that they represent one of primary health problems in that category of citizens. Deformities of a foot are registered in 17,6% of pupils, meaning that more than every tenth child has that kind of a deformity. 95% of children suffering from arthrogryposis have some kind of a spinal column deformity beside the joints contracture; mostly it is scoliosis, as well as foot deformities, prevalently in a form of an equinovarus component. Research carried out by the

request of World Health Organization has confirmed that in a treatment of deformities, after surgical interventions, the utmost efficiency is provided by medical rehabilitation, and if it is carried out on a regular and continuous basis, 50% of children is 100% efficient.

Rehabilitation treatment of arthrogryposis is the same, both throughout the world, and with us, and it is in the first place based upon early and continuous rehabilitation, and also upon a teamwork and an approach to a patient. The purpose of treatment is to achieve the best motility of joints or to accomplish the position of joints suitable for their function. Medical rehabilitation is being started with as early as possible. It is necessary to carry out the joints' motility exercises, immobilization should be avoided, and light splints for the maintenance of the accomplished level of joints' motility. For the purpose of making walking possible, surgical treatment is mostly performed on feet. Treatment is lasting and complicated, and recidives of deformities are frequent.

Motoric capacitating level of children with arthrogryposis depends on a great number of factors like duration of a rehabilitation treatment, motivation of parents for improving the state of their child, degree of deformity's severity, formation of a rehabilitation team. On the other hand, a degree of a mental development of children with arthrogryposis has much better prognosis, because, according to statistical data, 90% of children with arthrogryposis has their intelligence preserved and if they are don't get to be uncared-for they can achieve significant results.

Rehabilitation process is to be started from getting acquainted with a structure of a person with a handicap. Sometimes, during the rehabilitation, it is necessary to make an impact on those elements of a personality which are alterable, and therefore to act upon a structure of a handicapped personality, in order to realize an adaptation on a handicap more effectively and to end an integration process more successfully. Educational aspect, meaning education, refers to acquiring knowledge, changing attitudes towards oneself and regarding social values, as well as to realizing education and professional ccapacitating programs. Speaking of handicaped persons, a continuous treatment of rehabilitation, starting from their birth, should be comprehensive and appropriate in order to make a contribution to an improvement of everyday life of handicaped persons up to maximum limits of their recovery. If a handicaped person is enabled to produce a capital gains, or to produce enough to keep it's needs catered, it is considered that the economical aspect is fully satisfied, because being that way, one becomes useful member of a community. Persons with disturbances in their development, bearing in mind that this group is primarily consisted of children and young adults, should be included into school, work and wider social setting inside the frame of society. Throughout the world, as well by us, thanks to medical interventions and advanced scientific achievements, mortality is decreased, but the number of children born with anomalies and disturbances in development is still growing. Arthrogryposis is a very complex disease which brings out a whole scope of problems not only to a child who suffers from it but also to people in its surrounding. Nevertheless, from a humane point of view, we should never give up on persons such as these, but put an effort to help them, as much as possible, to integrate themselves into a society, as well as work on developing the rest of their functions, especially intellectual.

CONCLUSSION

As it can be seen from the research on a display, continuous working with those children, even over the course of one month, could make a great contribution and have significant results regarding questions of muscle strength, range of movements, transfer, daily life activities, as well as communication and intellectual skills, especially when it concerns a child who hadn't got an opportunity to make an advancement in education. Working with children who suffer from arthrogryposis requires a lot of patience and love, but it must be pointed out that even small steps mean great success for them.

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