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Author's Contribution
A – Study Design
B – Data Collection
C – Statistical Analysis
D – Data Interpretation
E – Manuscript Preparation
F – Literature Search
G – Funds Collection

Hana Samekova[1], Viktor Bialik[2], Gad M. Bialik[2]

[1] *University Department of Pediatric Orthopedics, Comenius University, Bratislava, Slovak Republic*

[2] *Pediatric Orthopedics Unit, Rambam Medical Center and Faculty of Medicine, Technion-Israel Institute of Technology, Haifa, Israel*

Arthrogryposis multiplex congenita – local experience

Key words: *congenital defects of the musculoskeletal system, distal arthrogryposis, Larsen's syndrome*

SUMMARY

Background. *Arthrogryposis is a heterogeneous group of problems of the musculoskeletal system, clinically characterized by multiple contractures of the joints of at least two anatomical regions, presenting at birth. The aim of our study was to evaluate the results of a therapeutic approach to patients at the University Pediatric Orthopedic Clinic in Bratislava.*

Material and methods. *Nineteen patients (13 girls, 6 boys) were treated and followed up between 1993 and 2004.*

Results. *Six patients had a clear clinical appearance of classical arthrogryposis multiplex congenita, six had distal arthrogryposis, two girls had Larsen's syndrome, and three had other forms of joint involvement. At a mean age of 18.4 months (range, 9-52 months), 77 surgeries had been performed. Most of the surgical procedures were for clubfoot-like deformities and vertical talus (45 operations) and for hip dislocations (20 operations). All patients who underwent surgery for hip dislocations achieved a full range of hip movement after subsequent physiotherapy. Surgery on the other hip also had good functional outcome. At the last examination, all patients were independently ambulatory, but some (mainly the younger ones) needed help in feeding, hygiene or dressing.*

Conclusions. *In our experience with this group of patients, physiotherapy and occupational therapy supported by orthotic equipment is an inseparable part of their lives. Complementary surgery gives an opportunity to improve the correction of deformities in cases where conservative treatment does not meet the needs of the patient, because their normal intellect gives them the potential for an almost normal adult life.*

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Adres do korespondencji / Address for correspondence
Dr. Hana Samekova
Detska ortopedicka klinika LFUK a DFNsP, tel./fax: +42 028-384-05-14
Limbova 1, 833 40 Bratislava, Slovak Reppublic, e-mail: kokavec@dfnsp.sk

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BACKGROUND

Arthrogryposis multiplex congenita (AMC) is a term describing the presence of multiple joint contractures at birth. The severity and number of joints involved can vary from mild (only a few joints involved with near to normal motion) to severe (almost all joints involved, including the spine, with severely limited motion). Recently, syndromes previously unknown and not described have been added to this disabling disorder. This problem was first described by Adolf Wilhelm Otto in 1841 as a „human wonder with curved limbs" [1]. Another description was by Stern in 1905. Gordon [2] and Hall [3] believed that the reason for AMC was restricted fetal movement (fetal akinesis). We believe that in a small clinic such as ours we would expect to see patients with the most common forms of this syndrome, and this report can

Tab. 1. Patients with Arthrogryposis

No.	Patient	Age [2004]	Lower Limb Findings	Upper Limb Findings	Other Findings	No. of Surgeries	Mental Function	Skin Healing	Elevated Laboratory Parameters	Genetic Type
1.	EM	16	Lt DDH CF	Ulnar deviation of wrist; Campt.		7	+	Keloid		AMC
2.	PR	4	Extended knee CF	Elb. Flex; Ulnar deviation of wrist	Hypospadias	5	+ +	Keloid		AMC
3.	PS	3	DDH	Ulnar deviation of wrist	Anteposition	6	+		Uric acid	DA
4.	KH1	4	DDH Inst. pat. Talus vertical	Dermato glyphs	Polystigmatic face Microgenia Brachycephaly Hemangiomas Short neck	2	+/-	Keloid		Larsen's syndrome. Mother with DDH; Sister with Larsen's syndrome
5.	KH2	2	DDH Vertical talus Extension knee contracture	Ulnar deviation of wrist	Polystigmatic face VCC Diaphragmatic hernia Inguinal hernia	4	+/-			Mother with DDH; Sister with Larsen's syndrome
6.	VV	4	CF	Extension elbow contracture; Ulnar deviation of wrist	Ankylosis of jaw Micrognathia	4			Uric acid	DA
7.	DD	4	CF	Ulnar deviation of wrist	Polystigmatic face; Nevus flammeus	2	+/-	Good		
8.	EB	12	Rt DDH Extended knee contracture; CF		Late motor development	3	+/-			
9.	LF	9	CF			1	+			DA I.
10.	EŠ	9	CF	Brachial plexus lesion; Flexion contracture of elbow; Ulnar deviation of wrist (bilat.)	Hemangiomas of arms	5	+	Keloid		
11.	L'D	21	CF	Ulnar deviation of wrist (bilat.)		4	+			DA I.
12.	NL	12	CF	Extension contracture of elbow (bilat.)	Polystigmatic face	2	+	Keloid		

shed some light on questions regarding the epidemiology of this problem.

MATERIAL AND METHODS

Between 1992 and 2004, we treated 19 patients diagnosed as having arthrogryposis in the broad sense of the term. We were able to divide them into four groups according to clinical findings:

– 'classical' arthrogryposis multiplex congenita (AMC) – 7 cases;
– distal arthrogryposis – 6 cases;
– Larsen's syndrome – 2 cases;
– 4 other patients who could not be classified more precisely, as the parents refused more detailed genetic studies.

In all, we treated 13 girls and 6 boys. The lowest number of surgical procedures was one, while one boy had nine operations. Table 1 shows patient details.

All patients now walk independently, despite the fact that the lower limbs were usually more severely involved. They also usually display normal intellect. None of this group of patients had spinal problems needing orthosis or surgery, but two required hand surgery. We found a problem with wound healing and a tendency to develop keloid scars (see Table 1).

Tab. 1. Patients with Arthrogryposis (c.d)

No.	Patient	Age [2004]	Lower Limb Findings	Upper Limb Findings	Other Findings	No. of Surgeries	Mental Function	Skin Healing	Elevated Laboratory Parameters	Genetic Type
13.	AK	14	Circumduct. Knee recurv. CF	Ulnar deviation of wrist; Flexion contracture of elbow (bilat.)	Hyperlaxity	5		Keloid		
14.	MZ	11	Coxa valga; Knee flexion contracture; CF	Arm hypotrophy Ulnar deviation of wrist	Lumbar lordosis Pigeon chest	3	+/-	Keloid		
15.	PH	11	DDH; CF; Knee flexion contracture	Short arm Camptodactyly	Hypoplasia of larynx; Pigeon chest; VCC	9	+	Keloid	Uric acid	
16.	RB	10	Flexion contracture of the knee; Hip flexion contracture	Ulnar deviation of wrist; Camptodactyly		4				Myogen type according to EMG
17.	ZL	11	DDH Rt Vertical talus	Camptodactyly Aplasia of thumb; Ulnar deviation of wrist	Polystigmatic face; Meningomyelocele; Umbilical hernia	4				
18.	DM	12	DDH Lt. Hip flexion contracture; CF	Brachial plexus paresis Dysplastic index finger	Polystigmatic face; Strabismus; Palatoschisis; Narrow larynx; Incontinence	7			Urea creatinine Uric acid	
19.	KČ	10	Rt CF Lt vertical talus		Polystigmatic face	6	+			
Total No. of Surgical Procedures						83				

Glossary:
DDH - Developmental dysplasia of the hip
Inst. pat. - Instability of patella
AMC - Arthrogryposis multiplex congenita
Elb. - Flexion contracture of elbow
CF - Club foot
VCC - Vitium cordis congenita
DA - Distal arthrogryposis

In our experience, treatment should be conservative. Manipulative treatment followed by plaster casts or orthotic immobilization must be introduced during the first days of life. It is not the aim of this paper to assess conservative treatment, only surgical treatment.

RESULTS

The youngest child in this group (KH2) is a 2-year old girl, sister to another girl with Larsen's syndrome. This girl was investigated for this syndrome intrauterinely (because of the older sister's condition), but no restricted intrauterine movement or deformity was found. The parents refused amniotic fluid examination. Because of multiple dislocations, the delivery was complicated, and a birth fracture of the right femur was diagnosed (Fig. 1). After the age of one year, we gradually performed surgery on both knees and the hip dislocations. Because of hyperextension contracture of both knees with subluxations, Z-plasty of quadriceps, release of the sartorius muscle, capsulotomy and medial collateral ligament were performed, with subsequent subluxation repositioning. The fixation of the reduced knee was accomplished with two Kirschner wires, and plaster casts were installed for six weeks. Open reduction was performed, including capsulorrhaphy with shortening of femur and various derotation osteotomies for both hips.

The oldest patient was a 21-year-old woman (LD) with classical distal arthrogryposis with less severe involvement. She had a full range of movement and was of normal intellect. She underwent surgery in early childhood, and, at the age of 16, had talo-tibial arthrodesis of the right ankle.

At the last examination, all patients were independently ambulatory, but some (mainly the younger ones) needed help in feeding, hygiene or dressing.

Only two patients required surgery on the upper limbs. Both had lengthening of the triceps brachii tendon with subsequent capsulotomy to improve the extension contracture of the elbow to permanent flexion. Improvement of 30o enabled independent feeding by bringing the hand close enough to the mouth.

Patients with arthrogryposis usually have normal intellect, and one of these patients (PS) has an IQ of 145. At the age of 3 years, he was able to deal with simple mathematical problems (counting) and, at the age of four, was able to read fluently. He has classical AMC with severe club feet which did not respond to conservative treatment. At the age of 3 years, he had surgical release of extension contracture of both knees with quadriceps lengthening.

As mentioned above, 80 surgeries were performed on the lower limbs while only two were performed on the upper. None of the patients was treated for spinal or hand problems.

Most of the surgical procedures were required on the lower limbs: 53 for club feet, 7 for vertical talus (Achilles tenotomy), dorsal capsulotomy and postero-medial release (according to Brockmann, Carrol or McKay) for club feet and Harcke procedure for

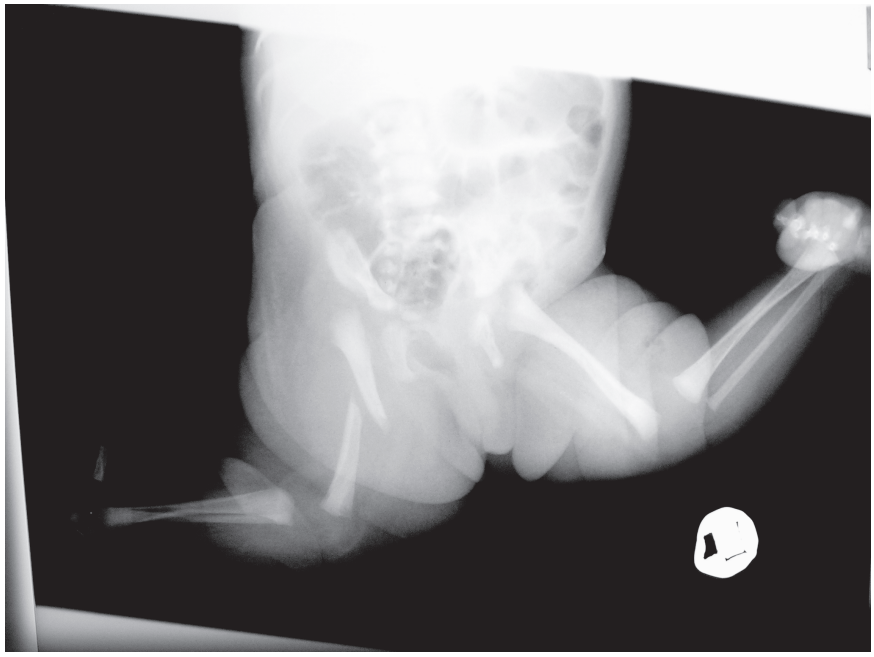

Fig. 1. KH2 – 2 year old girl with Larsen's syndrome, with multiple joint dislocations and birth fracture of right femur

vertical talus, while two patients are well after talectomy. Ten knees were treated in six patients, five for extension (two with rotational dislocation of the knee joint). They underwent quadriceps lengthening and capsular release. Dislocations after reduction were fixed with Kirschner wires. One patient with flexion contracture of the knee underwent surgery at the age of 14 months; the surgery was repeated two years later, with adhesions and keloid scars causing relapse. Despite release of the soft tissue, the contracture persisted at the age of 14 years.

Open reduction was performed on eight hip joints of six patients, with an additional shelf procedure according to Lance for two patients, while four patients had additional proximal femoral osteotomies. For one of the girls with Larsen's syndrome, closed reduction was performed before open reduction. Other osteotomies (except for upper femur) or arthrodeses were performed on the lower limbs in ten instances.

DISCUSSION

Despite the fact that this paper does not bring a detailed assessment of conservative treatment, in our experience with this group of patients, physiotherapy and occupational therapy supported by orthotic equipment is an inseparable part of their lives. Despite their disabilities, most of their life spans can be compared to that of the general healthy population. In our practice, conservative treatment begins as early as birth and continues, if necessary, until adulthood.

Single surgery correcting all deformities in one session at the age of 3-5 months demonstrated the best results. This is also in agreement with other authors [4,5]. We believe that new diagnostic methods, especially in genetics, will shed more light on this diagnosis and the subsequent treatment of this disabling disease. The cooperation of geneticists, orthopaedic surgeons, paediatricians, neurologists, physical therapists and gynaecologists in a multi-team approach is necessary for the best prevention-diagnostic-treatment programme [6].

In conclusion, we believe that the main effort in the treatment of patients with arthrogryposis should be conservative. Complementary surgery gives an opportunity to improve the correction of deformities in cases where conservative treatment does not meet the needs of the patient, because their normal intellect gives them the potential for an almost normal adult life.

CONCLUSIONS

1. We believe that the main effort in the treatment of patients with arthrogryposis should be conservative.
2. Complementary surgery gives an opportunity to improve the correction of deformities in cases where conservative treatment does not meet the needs of the patient, because their normal intellect gives them the potential for an almost normal adult life.

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