ARTHROGRYPOSIS - CASE REPORT

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Abstract. *Arthrogryposis* is a congenital disorder characterized by multiple joint contractures found throughout the body at birth. We present the case of a 3.5 year-old girl with multiple congenital defects: arthrogryposis involving bilateral hip, knee and ankle joints, together with sacral agenesis and with lumbar dysmorphism, anorectal agenesis, hydronephrosis of the left kidney as the result from reflux, right kidney hypoplasia, renal fusion and heart anomalies: tetralogy of Fallot. Immediately after birth, in several steps, colostomy, left ureterocystoneostomy and suprapubic cystostomy were performed. Later in infancy corrective surgery for the heart defect was required. At the age of 2 years, surgery for the equinovarus deformities and for left genu flexum was performed. Medical rehabilitation tries to maximize independent function. The main goals were increasing the muscle tonus of upper-limbs, increase the rate of motion of the joints, establishment of stability for ambulation, learning different schemes of movement according to her needs, obtaining of a functional independency. The therapies were successful, after 10 weeks an improvement of the moving capacity and of the transfer in orthostatism with minimal external assistance, with the obvious increasing of the patient's satisfaction, were noticed.

Keywords: arthrogryposis, medical rehabilitation, physical therapy

INTRODUCTION

Arthrogryposis, also known as *arthrogryposis multiplex congenita*, is a rare congenital disorder that is characterized by multiple joint contractures, muscle weakness and fibrosis. It is a non-progressive condition that affects both males and females. It is estimated that the incidence of the disorder is 1 in 3,000 live births of all ethnic backgrounds [1, 3, 5]. In the most common type of *arthrogryposis* hands, wrists, elbows, shoulders, hips, feet and knees are affected. In the most severe types, almost every joint is involved. Some infants with *arthrogryposis* have facial deformities, curvature of the spine, genital deformities, cardiac and respiratory problems and skin defects [9, 12]. It represents a very heterogeneous group of disorders that can be a component of many conditions caused by environmental agents, single gene defects (with autosomal dominant, autosomal recessive, X-linked recessive inheritance), chromosomal abnormalities, known syndromes, or unknown conditions [4, 7, 10, 12]. Males are primarily affected in X-linked recessive disorders [8]. The molecular basis of most genetic causes is not known yet. Joint contractures are caused by limitation of joint motion before birth. Prenatal limitation of joint mobility may be the result of different neurological deficits such as anencephaly, defects of the spine such as spina bifida or nerve deficiencies. It may also occur due to muscle deficits: agenesis of muscle, fetal myopathies, myotonic dystrophy, or myasthenia gravis, and to connective tissue and skeletal defects such as different synostosis, failure of a joint to develop, prenatal fixation of a joint, excess laxity and of dislocation of joints, fixation of soft tissue around the joint [6]. Fetal crowding or constraint is another cause [2, 7]. *Arthrogryposis* is detectable at birth or during prenatal life using ultrasonography. Regarding the prognosis, the life span for an individual with *arthrogryposis* is usually normal, but may be altered by heart defects or central nervous system problems [1]. Medical rehabilitation tries to maximize independent function [5].

CASE REPORT

We present the case of a 3.5 years old girl, born from healthy unrelated parents. Immediately after birth she was diagnosed with a severe plurimalformative syndrome (**Figure 1** and **2**):

- multiple *arthrogryposis* involving bilateral hip, knee and ankle joints, together with sacral agenesis and with lumbar dysmorphism
- hydronephrosis of the left kidney as the result from reflux, right kidney hypoplasia, renal fusion;
- anorectal agenesis;
- ambiguous external genitalia;
- heart anomalies: tetralogy of Fallot

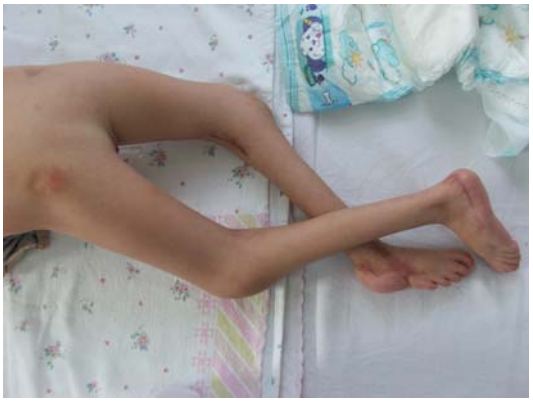

Figure 1.

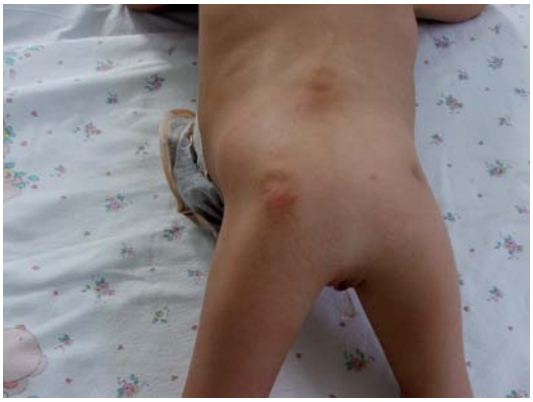

Figure 2.

Clinical examination revealed perineum with blind cloacal orificium, without exteriorizing urine. Immediately after birth suprapubic laparoscopy and Mc Kulicz colostoma on the splenic angle, exploratory cystoscopy and left ureterostomy with cystofix were performed.

Cardiac auscultation revealed harsh systolic murmur, along the sternal border, radiating in wheel's spoke, grade 4.

Evaluation of locomotory system revealed that the muscles in the legs and arms were thin and weak, with the following aspects of the joints: hip-flexed, abducted and externally rotated, knee- flexion deformity and feet with clubfoot deformity.

Paraclinical investigations were performed in order to establish and evaluate the congenital defects.

Urography: revealed hydronephrosis, short urethra with abnormal opening in the urinary bladder. The left kidney was projected in the small pelvis at the level of the left iliac bone, with delayed secretion and excretion; marked dilatation of the pyelocaliceal system.

MRI of the pelvis showed fused kidneys in their internal part, with left hydronephrosis, very thin cortical of 2-4 mm and the right kidney with fine pyelocaliceal system, the renal structures being in the left lumbar and iliac fossae regions and in the paravertebral region. The left urethra is dilatated up to the level of the ureterovesical junction. The right urethra is visualised para-renal, near the urinary bladder, seeming to implant under the vesical floor and has a reduced diameter. Pelvic girdle with a much reduced transversal diameter, with fusion of the pubic bones, horizontalized iliac wings, without the joint with the sacrum, which is present only at S1. The lumbar column has dextroconvex scoliosis, with some vertebral hemibodies at L2-L3 on the right side. The vertebral channel is much enlarged at the lumbar level with the dural sac ended at L5 level and the medullar cord at L3-L4 with phylum terminal attached to the posterior dural sac. Obturatory muscles are present on the right side and absent on the left side.

Mictional cistography revealed homogeneous opacification of the urinary bladder, with irregular contour; the contrast dye is exteriorising through a digestive lumen in the colostoma bag; the right vesico-ureteral reflux, with opacity of the right pyelocaliceal space, which appears dilatated, urethra of normal caliber. Irigography marked out a filiform communication between the urinary bladder and rectum; the rectal ampulla seems to have a blind end at about 3 cm under the skin.

Renal scintigraphy revealed left hydronephrosis, grade II, with normal glomerular function and slow excretion, vesico-ureteral reflux, paramedian right kidney, near the left kidney with a partial fusion between them and also hypofunction.

Cardiac ultrasonography revealed perimembranous ventricular septal defect of 0.7 cm with important left-to-right shunt and patent arterial duct.

Psychological investigation: good collaboration and communication capacity, normal speaking; optimal sensorial – perceptive development, optimal level of adaptation and socializing, medium level intellect; good mnesic function and attention; able of self-service; without any affective or behavioural problems.

Cardiac surgery was performed at Heart Institute from Cluj-Napoca, when the subject was 9 months old and the diagnosis of the heart anomalies was ventricular septal defect, anterior malaligniament type with left-to-right shunt that was hemodynamically significant. She also had large subvalvular pulmonary stenosis, heart failure NYHA II.

Cardiac echography after surgery: muscular ventricular septal defect hemodynamically insignificant, pulmonary insufficiency gr. I/II .

In May 2006 a complex surgical team from the County Clinical Hospital Cluj Napoca, performed surgery to correct the urinary tract anomalies with left ureterocystoneostomy and suprapubic cystostomy with favorable evolution.

In August 2007 at the Children Emergency Clinical Hospital “M.S. Curie” from Bucureşti, surgery for the equinovarus deformities and for left genu flexum was performed.

Karyotype (performed in Cluj-Napoca) showed a female constitution 46, XX with a terminal deletion of the long arm of chromosome 3.

Research has shown that anything that prevents normal joint movement before birth can result in joint contractures. The joint itself may be normal. However, when a joint is not moved for a period of time, extra connective tissue tends to grow around it, fixing it in position. Lack of joint movement also means that tendons connecting to the joint are not stretched to their normal length; short tendons, in turn, make normal joint movement difficult.

While there is no cure, symptoms and deformities may still be alleviated with various methods due to multiple contractures and weakness.

The subject made the rehabilitation treatment in the Paediatric Division of the Clinical Rehabilitation Hospital Felix Spa.

The main goals of the treatment were increasing the muscle tonus of upper-limbs, increase the rate of motion of the joints, establishment of stability for ambulation, learning different schemes of movement according to her needs, obtaining of a functional independency (**Figure 3**, **4** and **5**).

Figure 3

Figure 4.

Figure 5.

Physical therapy intervention including stretching (may include casting and splinting program of affected joints), strengthening, mobility training, are undertaken to improve flexion and range of motion. Occupational therapy interventions can include training in ADL and fine motor skills as well as addressing psychosocial and emotional implications of a chronic condition. Splints can also help stretch joints, especially at night.

The therapies were successful, after 10 weeks an improvement of the moving capacity and of the transfer in orthostatism with minimal external assistance, with the obvious increasing of the patient's satisfaction, were noticed.

The subject returns for medical rehabilitation every 2 months, in the meantime continuing at home the established kinetic programme and is reevaluated with every hospitalization.

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