**Reseberättelse 3rd International Symposium on Arthrogryposis**

**Philadelphia September 24-26, 2018**

This symposium has been organized twice before, in 2007 and 2014, and now was my first opportunity to go. It is very special in the sense that the program includes all aspects of arthrogryposis. There was a part pediatric neurology, genetics, pediatric orthopedics, hand surgery and therapy. The symposium was sponsored by the amc patient-support group and was open to patients to attend. This was a wonderful and very rewarding interaction. Especially for such a rare diagnosis, which is very complex, it was most educational to get an insight in all aspects. Many experts were gathered.

Judith Hall was the very active eminence present. She is a geneticist who has been researching on arthrogryposis since the seventies. She is now 80 years old, but does not seem to be tired ever. She gave a very clear presentation an amyoplasia and was followed by ‘our own’ expert Eva Kimber with a presentation on distal arthrogryposis. Eva Kimber is the pediatric neurologist in our AMC-team in Gothenburg, so I can fully rely on her for the specific diagnosis, which is wonderful. But I feel that I got a much better insight myself after this meeting.

I will write just a short overview for those who are not seeing arthrogryposis-patients regularly. Arthrogryposis or AMC is a ‘symptom’, describing joint deformities. It is a non-progressive disease, meaning it doesn’t get worse than it is at birth and often gets at least somewhat better. There are three groups of diagnoses described within the AMC-spectrum. Amyoplasia is the most common form and stands for about 40% of all AMC-patients, occurs in 1/10.000 births. It is not genetic and usually there has been an occurrence during early pregnancy, leading to a vascular compromise. There probably is damage to the anterior horn cells in the spinal cord, but that has not been proven. The key feature is ‘lack of motion’, which is a common feature to all AMC-diagnoses. Skeletal muscles are weak, usually fairly symmetrical, but can be located in all body areas. This leads to stiff joints, contractures and dislocations. Children can be born with dislocated hips, clubfeet, straight and stiff elbows and knees, wrist and finger contractures. The typical upper limb has weak or no active elbow flexion, but active extension. Wrist contracture either in flexion (more common) or extension, clasped thumb without active extension and narrow first web, stiff fingers in ulnar deviation with some movement in the MCP joint. Any variation is obviously possible. The children have a normal to above average intelligence, maybe due to their need to be ‘problem solvers’.

Distal arthrogryposis (DA) is the second form of AMC. It a group of genetic disorders with variable expression, many of them are autosomal dominant, but a parent can have the gene without symptoms and therefore not knowing. Many genes have been identified, which is obviously very valuable for the families to estimate the recurrence risk and for prenatal testing. The clinical expression in the limb is more limited to the distal ends, but it can be more resistant to treatment. There can also be other associated features like distinct facial features, pain, and tongue atrophy.

The third group of AMC-diagnoses is the neurological type.

There were a few sessions on upper limb surgery and therapy. There was quite some time devoted to therapy and splinting, because this is a very important aspect of the treatment. Also to start as early as possible mobilizing all joints and ensure as much passive range of motion as possible. Many children gain strength in certain muscles, but that requires mobile joints. A common upper limb problem is lack of active elbow flexion. Many times there is passive motion present in the elbow or it can be gained with stretching. It is interesting to see the solutions of patients who never achieved active flexion, but maintained passive flexion. They push for example their forearm to the edge of the table in order to flex the elbow and bring food to their mouth. These children can be candidates for a muscle transfer. Scott Oishi presented on the long head of triceps to biceps transfer, with average age of 11 years old. This technique got negative rumors in the past, because it used to be transferred the entire triceps muscle to biceps, leading to a flexion contracture instead. According to Dr. Oishi, the long head of triceps has a separate nerve, which can be stimulated separately and he showed good results. Other options are LD, pectoralis major and free gracilis. The general recommendation is to do MRI evaluations of the present muscles before deciding which muscle to transfer. Technically, the pectoralis major transfer seems to me the most elegant transfer, but the main drawback is the disfiguring scar on the chest.

The wrist is many times stiff in either flexion or extension. The most common procedure is a wedge excision of the carpus, essentially a midcarpal fusion in neutral position. This seems to have good results. The main question is when to do this. As many children are in a very delicate balance between limitations in both lower and upper limbs, they have developed strategies to move, crawl or walk. Changing the position of the wrist might disturb this balance and should therefore be considered very carefully.

Another common problem is the clasped thumb. This is often a narrow first web and a flexed MCP-joint, because of lack of active extension. A tendon to transfer is often not available and many surgeons tighten the present EPB. Andrea Lester reviewed the patients operated in her clinic and found disappointing results with many recurrences. It seems often to be recommended to fuse the MCP-joint. An exciting technique was presented by Dan Zlotolow, where he does an osteotomy of first metacarpal instead in cases where the CMC joint is flexed and rotated, mainly amyoplasia patients. He also pointed out, that his group found a remarkable difference in results from an elbow release done before or after the age of two. There was a good gain in ROM, if done before 2 years old, and only little gain if done later in life.

There was not so much talk about the fingers. I guess nobody does much to the fingers surgically, since there are no available transfers.

At the end of the meeting, there were discussion rounds, where the participants could choose a session. I went to the session on prenatal diagnosis, with Judith Hall. There were about 20 participants. One of the participants in the group was a young mother of an 8-month-old baby, born with amyoplasia, who happened to be an obstetrician herself. This was extremely valuable for the discussion on what ultrasound can show, how ultrasonografists should be guided, when and how to deliver the babies. There was very little talk about pregnancy termination. Already during pregnancy, experienced mothers can tell that the baby is not moving much. To establish this on ultrasound, the examination should be 45min, which is obviously not standard, but can be suggested when there is a suspicion. Other US features are clenched fists and clubfeet. It is recommended that the babies be delivered with a caesarian section, because of the risk of fractures and umbilical cord problems during delivery. Another suggestion by Judith Hall is to deliver early, within the safe zone, so somewhere between 36 and 38 weeks seems fair. The reason for this is, that the key to recovery is early mobilization.

Interestingly Judith Hall recommended ways to stimulate the baby to move intra-uterine, like drinking coffee, taking regular long walks, playing music.

Overall it has been an extremely valuable meeting, because of the multidisciplinary approach and the high level of experience of the speakers.

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