Arthrogryposis multiplex congenita

Arthrogryposis (synonyms: *arthrogryposis multiplex congenita (AMC), multiple joint contracture syndrome, amyoplasia congenita, arthrogryposis universalis congenita*[1]) is a comprehensive syndrome characterized by **non-progressive multiple joint contractures** (congenital joint stiffness) caused by muscle fibrosis and shortening and thickening of the joint capsule and ligaments.[2]

Etiopathogenesis

By affecting the soft tissues of the joint, it leads to a concentric limitation of the movement of the joints of the limbs. The basis is probably a **binder failure**, the actual **cause is unknown**.

(note: Fetal akinesia due to various fetal or maternal abnormalities, neurotrophic disorders caused by ganglion cell differentiation disorders, or primary muscle aplasia is considered.) [3]

In the course of the disease, *fibrous joint changes of the joint capsules* appear with the development of contractures. There are also progressive forms that are lethal.[2]

Classification

According to probable cause[2]

- **neuropathic form** (95%) – accompanied by degeneration of the cells of the anterior horns of the spinal cord (lack of neurotrophic influence → lack of muscle development, amyoplasia), non-genetically linked
- **myopathic form** (5%) - primary breakdown of muscle fibers, genetically linked

According to anatomical involvement (Brown et al.)[2]

- **type I** and **II** – impairment of upper limbs (segments C5–C8), elbows in extension (type I, does not eat) / in flexion (type II)
- **type III'** to **VIII** - involvement of the lower limbs (lumbosacral segments)

Complex division according to Hall [2]

- **group 1** - predominantly limb involvement (main focus of treatment, incidence 1:10,000)
- **group 2** - involvement of limbs and visceral organs, craniofacial malformation
- **group 3** - involvement of limbs and CNS (fatal in early childhood)

Another used division[2]

- **distal arthrogryposis** - affects the peripheral parts of the limbs, hereditary
- **classic arthrogryposis** - affects large joints

Clinical picture

It manifests itself in varying degrees of "stiffness of the joints of the limbs", mainly of the hip, knee, wrist and hand joints[1]. Stiffness is not caused by primary joint changes, but by **muscle changes** (see above)[1]. Children's appearance can be compared to **wooden dolls**[2]. The contours of the limbs are elongated, cylindrical, with accentuated skin eyelashes[3]. There is a weakening of muscle and joint relief (*amyoplasia*), reduction of subcutaneous fat[2].Joints affected symmetrically[3].Upper and lower limbs mostly affected (*tetramel form*)[2]. Hearing and intelligence are usually normal[2].

They are often associated with **pedes equinovari**, **luxation of the hip**, **outstretched upper limbs**, **``waiter-tip *wrist position*[2].**

We distinguish between the "flexion" type (joints stiff in flexion) and "extension" joints stiff in extension, most common on the upper limb[1]. Limitation of movement during joint contractures (more pronounced distally), fine motor skills preserved[2].

Stationary changes begin to manifest after childbirth, they do not show a tendency to spontaneous improvement or deterioration[3].

X-ray image

Initially, the finding is negative. Later we observe a '*narrowing* of the joint spaces up to joint ankylosis[3].

Therapy

- **rehabilitation** - exercises, positioning, loosening of joints, improvement of muscle strength, casting, application of orthoses and corrective splints[2]
- **arthrolysis, osteotomy** - Ch. elbow ankylosis in extension[3]
- '*primary task* is to ensure walking within 18 months[2]
- then we try to ensure '*self-sufficiency* in food, hygiene and dressing (upper limb surgery around 4 years of age)[2]

Treatment of extension contractures of the upper limbs: ***Conservative*** (splitting) or ***surgical therapy*** (dorsal release of the elbow joint) with the aim of obtaining passive flexion of the elbow greater than 90°. The goal is to restore active elbow flexion while maintaining sufficient extension for hygienic tasks. The so-called "tendon transfers" are performed, i.e. the bilateral transposition of three fifths of the "M. pectoralis major *instead of* M. biceps brachii *and* M. brachialis *(fixation of the transferred parts into the M. flexor carpi ulnaris tendon) by modifying Clark's operation according to Chomiak and Dungl 2002, 2003*[2]*.*

Differential diagnosis

- multiple pterygium syndrome
- Freeman-Sheldon Syndrome
- Baels Syndrome
- diastrophic dwarfism[2]

Links

Related Articles

- Congenital limb defects
- Developmental hip dysplasia
- Dog equinovarus congenitus
- Diastrophic Dwarfism
- Congenital developmental defects

External links

- Arthrogryposis multiplex congenita (Czech Wikipedia)

- Wikipedia EN (https://en.wikipedia.org/wiki/Arthrogryposis)
- AMC Support (https://amcsupport.org/) (USA)
- The Arthrogryposis Group (http://www.tagonline.org.uk) (Velká Británie)

Reference

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2. DUNGL, P.. *Orthopedics.* 1. edition. Grada Publishing, 2005. ISBN 80-247-0550-8.
3. SOSNA, A. – VAVŘÍK, P. – KRBEC, M.. *Basics of orthopedics.* 1. edition. Triton, 2001. ISBN 80-7254-202-8.

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