



Thumb in Arthrogryposis

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Abstract

Arthrogryposis is a congenital disorder, a set of conditions of different aetiologies that are characterized by joint stiffness and contractures affecting at least two different areas of the body. Thumb in arthrogryposis is clasped with a flexion adduction deformity. It is characterized by deficiency of thumb extensors, flexion contracture of the metacarpophalangeal joint with possible instability, narrowing of the first web space due to different contractures of the web structures and lack of skin. Thumb function, its position, its length, its stability and its strength are essential for a proper grip. The loss of these features due to contracture in arthrogryposis reduces significantly patients' function and dexterity. Manipulation of the deformities starting soon after birth can improve the range of motion, which, if surgery needs to be done, makes the operation less extensive. Release of structures that are contracted, skin correction, gain of

stability, good position and, if possible, replacement of structures that are weak or absent are the aim of the surgical treatment.

Keywords

Thumb · Arthrogryposis · Thumb contracture
Thumb correction · First web contracture ·
First web release

Arthrogryposis is a descriptive term used to describe a host of clinical conditions resulting in nonprogressive multiple congenital joint contractures [1]. Once arthrogryposis was considered a distinct diagnosis for multiple joint contracture, it is now understood that arthrogryposis is a description of a heterogeneous group of some 300 different conditions with a variety of aetiologies including gene mutations [2].

The term arthrogryposis derives from the Greek words *arthros* (joint) and *grypon* (hooked), and it was coined by Rosencrantz [3]. In 1923, Stern used the term arthrogryposis multiplex congenita to describe the observed multiple joint involvement at birth [4]. Then the term amyoplasia appeared (literally, “a” means no, “myo” means muscle, “plasia” means development); it was created by Sheldon underling the thought that the primary cause for the condition was poor foetal muscle development [5].

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All forms are associated with foetal akinesia; in most cases, joint development is normal during embryogenesis, but at a certain moment, there is a movement decrease. Foetal movements are crucial to the correct development of joints; a reduction leads to contracture. An early onset of akinesia and the severity of contracture are related, as demonstrated in several *in vivo* studies [6, 7]. All factors that reduce foetal movements are involved in arthrogryposis, and its aetiology is multifactorial. Possible causes of akinesia include myopathic processes, neuropathic processes, neuromuscular endplate abnormalities, abnormalities of connective tissue, limitations of *in utero* space, maternal illness, maternal exposures, compromise of blood supply to placenta and foetus, metabolic disturbances and epigenetic disorders like part of complex syndromes [8].

In utero decreased movement leads to an increase of connective tissue around the joints that limits the joint movement, and it increases the contractures [9], muscle atrophy of the muscles associated with the joint due to disuse and abnormal joint surfaces that appear more squared.

Thumb in arthrogryposis is in palm with a narrow first web and soft tissue and muscle contractures (Fig. 21.1). The problems associated with joint contractures are a lack of skin, short tendons, joint stiffness with instability and weak muscles [10]. Particularly thumb is characterized by deficiency of the extensors (Fig. 21.2), flexion contracture of the metacarpophalangeal joint that can be also instable, skin deficiency of the first

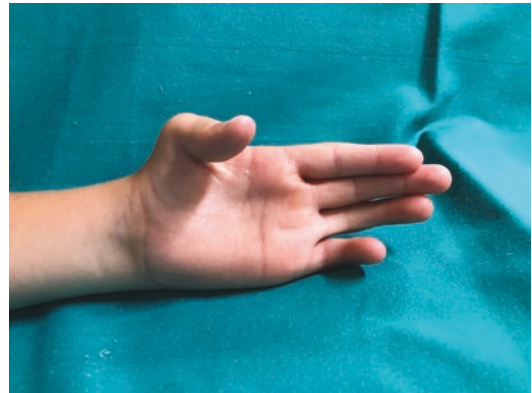


Fig. 21.2 Lack of extension

web and narrowing of the first web space due to variable combinations of contracture of the web structures. Also flexor pollicis longus can present a contracture. Thenar muscles are hypoplastic with fibrosis. Grasp and pinch are limited or absent by the severity of these deformities.

Goal of treatment is to improve thumb mobility and function in order to increase independence with activities of daily living. Occupational and physical therapies are essential components of treatment, and they begin at a very young age. Surgery became important when poor results are achieved with physiotherapy. Every treatment, surgical and nonoperative, must be tailored to each patient. A realistic family and patient expectation is also essential because it is not possible to restore a normal digital motion; function can just be improved [11]. For the assessment, hand X-rays are required.

First approach with thumb in palm is a gentle manipulation and stretching since birth, repeating them a lot of times during the day. The use of customized splints to continue stretching and maintain results during the night is also indicated. Free use of the upper limbs during the day is essential to allow children to explore and find their own way of managing activities. Parents need the support of trained hand therapists to learn the correct way of manipulating and stretching the thumb. Occupational therapists can assist in providing orthoses or adaptive equipment to support some activities, especially as children get older.



Fig. 21.1 Typical aspect of thumb in arthrogryposis



Fig. 21.3 Skin drawing



Fig. 21.4 Muscle release

If adequate progress is not achieved, surgery becomes necessary. Timing of surgery is controversial, but early management is recommended to have a minimal impact on child development. Surgery should be done before contractures become more fixed and joint surface anatomy and joint congruity change, making joint movement more difficult and limited. With time also intra-articular adhesions increase, preventing normal gliding surfaces; the skin becomes less pliable, further preventing normal joint movement [12].

The techniques of reconstruction of clasped thumb in arthrogryposis have not been widely discussed in the literature.

The aims of thumb correction are to release the narrow web structures, to augment the skin of the first web and to obtain extension and stabilization of the metacarpophalangeal joint. Different techniques of skin flaps have been described for skin augmentation of the narrow web, using of four-flap z-plasties [13] or a local flap [14, 15] (Fig. 21.3).

After web skin incision, the tight fascia of the first dorsal interosseous and adductor pollicis have to be released. When adductor release alone does not allow proper thumb positioning, also the thenar muscles must be released (Fig. 21.4). An incision is made next the thenar crease, and the origin of the thenar muscles is released after first identifying and protecting the motor branch of the median nerve. A Kirshner fixation of the



Fig. 21.5 Extensor tendon correction

metacarpophalangeal joint can be done for correct skin and thumb position healing. When there is an instability of the metacarpophalangeal joint, a chondrodesis can be performed [16], and sometimes also an opponensplasty is needed.

If the flexor pollicis longus is tight and prevents thumb abduction, a lengthening at the musculotendinous junction can be performed with attention to not weaken the function.

After muscle and soft tissue release, a careful examination can reveal the status of active thumb extension. In some cases, extensor pollicis longus function is present. However, in some cases, the extensor pollicis longus is poor or absent, and it has to be treated to avoid recurrence of thumb deformity. Sometimes, this tendon can be pliated (Fig. 21.5); otherwise a tendon transfer can



Fig. 21.6 Follow-up



Fig. 21.7 Improvement in thumb extension

be considered to increase extensor power [17]. Unfortunately, in many cases, no adequate tendon is available for transfer.

After skin healing and Kirshner wire removal, physiotherapy and splinting are essential. The risk of recurrence or development of worse contractures is high without a correct postoperative protocol compliance.

Thumb in arthrogryposis treatment is a challenge to the hand surgeon and therapist. An early non-operative approach with manipulation and splints is necessary and can avoid surgery. Surgery becomes indicated when an adequate

progress is not achieved. Its aim is to improve thumb position (Fig. 21.6) and function (Fig. 21.7) to allow patient independence.

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