

# CLINICAL ARTICLE

## Arthrogryposis multiplex congenita of the upper limb

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### Introduction

The name *arthrogryposis* is derived from the Greek and means stiff joints (*arthron* = joint and *grypos* = stiff). Arthrogryposis represents a large group of disorders that present with joint contractures at birth. These congenital contracture syndromes total over 65 conditions with different clinical courses and pathological processes. Contracture syndrome groups can be divided into the following:

- Group involving all four extremities – includes arthrogryposis multiplex congenita (AMC) and Larsen syndrome, usually with total body involvement.
- Distal arthrogryposis – group predominantly or exclusively involving the hands and feet. Freeman-Sheldon whistling face is an example in this group.
- Pterygia syndromes – identifiable skin webs cross the flexion aspects of knees, elbows and other joints. Multiple pterygias and popliteal pterygia belong to this group.

**Key words:** Arthrogryposis multiplex congenita, upper limb

### Arthrogryposis multiplex congenita (AMC)

AMC was initially described by Otto in 1841, who declared that his patient was a 'human wonder with curved limbs'. There is no race or gender predilection. The incidence is 1 in 5 to 10 000 live births and the disease does not directly affect the life expectancy of the patient. AMC has the following characteristics:

- The full clinical expression is present at birth (congenital).
- There is usually symmetrical involvement of multiple joints and muscles.
- There is usually no involvement of other systems, e.g. heart, brain, skeleton, GI tract or urogenital tract.
- The intellect is normal.
- It is not inherited; Mennen and Williams (1996) presented a case report of AMC in a monozygotic twin.<sup>1</sup>
- It is not due to an embryologic malformation (not abnormal induction).
- Anterior horn cell numbers are decreased in the spinal cord without an increase in microglial cells.

- Muscle mass is reduced, with infiltration of fibrous and fibrofatty tissue between muscle fibres.<sup>2</sup> Periarticular fibrosis causes a fibrous ankylosis of joints.
- Sensation is normal.
- There is no progression of the condition after birth but secondary changes occur with growth.
- Joint deformities are due to secondary changes from a lack of joint movement.
- The patient learns adaptive movements to compensate for loss of normal function.

### Aetiology

The exact aetiology of the disease is uncertain. The most likely cause is damage to the anterior horn cells of the spinal cord in the developing foetus (Swaiman and Wright, 1994).<sup>3</sup> The suggested cause(s) may include direct damage by a viral infection, e.g. herpes simplex, or indirectly by an increase in temperature due to the infection, placental insufficiency or a stress reaction in a foetus carrying malignant hyperthermia-associated myopathy. Cross-circulation with disturbed foetal thermodynamics may also be implicated.

### Classification

This classification system is based on the amount of anterior horn cell damage and the resultant degree of stiffness due to muscle under-development. Mennen (1993)<sup>3</sup> suggested that pre- and post-operative clinical evaluation could divide patients into the following groups:

- Type I: 'Loose' type has little involvement of anterior horn cells and good functional prognosis. The limbs appear normal and these patients will have little difficulty in walking. Their deformities are correctable pre-operatively and spinal muscles are not involved. Secondary surgical procedures are rarely indicated.
- Type II: 'Stiff' type has very little pre-operative joint movement. The patients' spinal muscles are involved which affects their ability to sit and stand. They present with severe club foot deformities, and hip and knee subluxation or dislocation. There are very few if any muscle fibres found intra-operatively. Joint capsules are thick and contracted, often with intra-articular adhesions and secondary joint deformity.

A new classification system has been proposed by Mennen (2004)<sup>4</sup> that takes the function and age of the patient into account. Passive movement (baby), active movement (young child) and function (older child and adult) are assessed; function is calculated from the ranges of movement (active and passive) and the ability to execute activities of daily living with a specific joint. These values are expressed as a percentage of normal and plotted on a disc-o-gram, thereby creating an image of total body function. Any change in function from therapy or surgery can be plotted on the same disc-o-gram and will thereby change the shape and size of the 'image of function'.

The patients are classified into five types by adding up the values of joint movement or functional ability. These groups are further divided up into three subsections depending on the pattern of limb involvement:

- |     |  |
|-----|--|
| 0-2 | I Rigid  |
|     | A) Both upper limb (UL) and lower limb (LL) involvement (i.e. UL and LL involvement) |
|     | B) Minimal or no LL involvement (i.e. UL involvement)                                |
|     | C) Minimal or no UL involvement (i.e. LL involvement)                                |
| 2-4 | II Minimal mobility  |
|     | A) Both UL and LL involvement (i.e. UL and LL involvement)                           |
|     | B) Minimal or no LL involvement (i.e. UL involvement)                                |
|     | C) Minimal or no UL involvement (i.e. LL involvement)                                |

- |      |  |
|------|--|
| 4-6  | III Moderate mobility                                      |
|      | A) Both UL and LL involvement (i.e. UL and LL involvement) |
|      | B) Minimal or no LL involvement (i.e. UL involvement)      |
|      | C) Minimal or no UL involvement (i.e. LL involvement)      |
| 6-8  | IV Near normal mobility                                    |
|      | A) Both UL and LL involvement (i.e. UL and LL involvement) |
|      | B) Minimal or no LL involvement (i.e. UL involvement)      |
|      | C) Minimal or no UL involvement (i.e. LL involvement)      |
| 8-10 | V Mobile/normal  |
|      | A) Both UL and LL involvement (i.e. UL and LL involvement) |
|      | B) Minimal or no LL involvement (i.e. UL involvement)      |
|      | C) Minimal or no UL involvement (i.e. LL involvement)      |

### Clinical features

The limbs are stiff in varying degrees and appear tubular with smooth skin over joints and absence of normal skin folds. Deep dimples may be seen over the large joints. The muscles are reduced in size and feel firmer than normal. The shoulders are adducted and internally rotated with weak or absent shoulder girdle muscles. The arms may be in such severe internal rotation that the hands may only be used in pronation. The elbows are more often in extension than flexion, with weak or absent biceps and brachialis muscles, while the triceps is less affected. Wrists are usually pronated, in severe flexion and ulnar deviation, lacking wrist extension. The thumbs are adducted across the palms (thumb-in-palm deformity) and the fingers are flexed and rigid. The finger deformities usually involve rigid flexion at the IP joints and neutral to extension position of the MP joints. The fingers are often overlapped and with slight flexion in a 'paw' position.

Patients with AMC are usually pain-free. Complaints that may be present are inguinal hernias due to weakened musculature, or feeding problems due to a stiff jaw and immobile tongue that can lead to respiratory infections and a failure to thrive. The face is not particularly dysmorphic, but may demonstrate a small jaw, facial narrowing and, if the ocular muscles are involved, a limited upward gaze.

Two-thirds of patients have equal involvement of all four limbs, and in one-third lower limb involvement (club feet, flexion deformity of the knee and subluxed or dislocated hips) will predominate. Upper limb involvement rarely predominates. When spinal muscles are involved the child has difficulty with sitting and standing up.

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Up to one-third of patients will develop scoliosis. The joints appear normal on X-ray, and the changes are adaptive and acquired over a period of time due to the joints' fixed position.

The diagnosis of AMC is clinical, but it may be suspected if the prenatal ultrasound demonstrates a decrease in foetal movements, especially in combination with polyhydramnios. Some contractures seem to become stiffer over time but no new joints become involved.

AMC patients may develop compensatory movements to assist activities of daily living such as pushing the forearm against a table to bring the hand close to the mouth to eat, or if the patients are standing they may reach their face by swinging their arms and using lumbar spine lordosis and gravity to assist the movement. These patients may also assist themselves using a cross-arm technique.

### Management

When considering management of the upper limb the whole arm must be taken into consideration and individual joints must not be isolated. The ultimate goal of surgery to the upper limb is to improve the patient's self-care ability, especially eating and hygiene (writing is a bonus).

#### Conservative management

All upper limb deformities must be gently manipulated (muscle and joint stretching) from birth by a qualified hand therapist with the best results achieved if started before 6 months of age.<sup>3</sup> Physiotherapy includes passive manipulation several times a day followed by night-time splinting of the position gained. The therapist may further assist these children by teaching them trick movements to achieve better function. Splinting a patient in a certain position may allow the patient to decide if the new position will be desirable or not before surgery is done. Deformity correction may be attempted by the following measures:

- Intensive exercise programme – usually only results in a slight improvement in ROM with the chances of success declining with age, and little gain expected after 3 years of age.<sup>9</sup>
- Serial casting – this is time-consuming with a high rate of recurrence. If done too aggressively it may cause cartilage necrosis and further stiffness. Smith and Drennan recommended the use of serial casting for wrist flexion deformities,<sup>10</sup> but did show that the classical form of arthrogryposis with rigid wrist deformities was resistant to serial casting. Some feel that serial casting may lessen the extent of surgery, even though the deformity is not completely corrected.<sup>6</sup> Repeat casting is unlikely to be successful if recurrence of the deformity occurs.

If no further correction can be achieved by conservative or surgical means then the patient will benefit from modification of mechanical aids. The following are some examples:

- Chair and tables – these will often need adjustment for feeding and playing.
- Eating and drinking – it may be necessary to fix the plate to the table and adjust the handles of eating utensils.
- Dressing – Velcro can replace buttons, and zips can be fitted with large ring handles. Dressing may further be assisted by using shoes without laces and sticks to assist with the activity of getting dressed.
- Toilet needs – self-cleaning toilets are available, but are expensive and will only be available in the home environment. The height of toilet seats may also need to be adjusted at home to accommodate the patient. Showers may need to be fitted with seats and liquid soap dispensers.

#### Surgical management

Surgery is offered after 6 months if there is a failure to progress with conservative measures. Some of the principles of surgery (Mennen) are the following:

- Early surgery. The ideal time for surgery is between 3–6 months of age.<sup>5</sup> Early surgery is easier, e.g. carpal bones can be removed with a scalpel. A younger child recovers faster, with less scarring and has the ability to remodel joint surfaces. They are also more adaptable, reducing the need for intensive physiotherapy after the procedure. In very young children the remnants of the carpal bones left behind develop ossification centres, which will result in functional carpal bones.<sup>5</sup> The surgery becomes more difficult later, i.e. after 1 year, as contractures become more fixed and joint congruity changes, limiting joint movement. Joint adhesions increase and the skin becomes less pliable adding to the abnormal joint movement.
- One-stage procedures. One-stage procedures give better results than staged procedures<sup>5</sup> and may include surgery to bones, joints and soft tissue rebalancing. However, as the child grows, smaller procedures may be needed to maintain optimum function.
- Osteotomies. Correction of deformity by osteotomy is of limited value in young children as remodelling will cause recurrence of the deformity within 1–2 yrs.

### Management of the hand and wrist in AMC

The wrist is almost always affected with a flexion deformity of up to 90° and ulnar deviation. It is widely agreed that correction of the hand and wrist deformity will improve the overall function of the upper limb.

*The ultimate goal of surgery to the upper limb is to improve the patient's self-care ability, especially eating and hygiene (writing is a bonus)*

### Carpectomy

This procedure was met with mixed results according to earlier literature,<sup>12</sup> but when performed early (3–6 months) and in combination with soft tissue balancing as part of a one-stage procedure it has shown promising outcomes.<sup>3,5</sup> When the carpectomy is performed before ossification of the carpal bones it has the following advantages:

- The unossified carpal bones allow the surgeon to sculpt a wedge-shaped removal of cartilaginous bones with a scalpel.
- The exact anatomy of the carpal bones can be ignored and the surgeon only needs to focus on removing a clearly defined trapezoid wedge from the carpal bones.

In milder forms of the disease the trapezoid should be removed from the mid-section of the carpus, leaving the radio-carpal joint intact. The carpus is not only shortened by the trapezoid wedge, but the following is of importance:

- The volar portion of the trapezoid wedge relaxes the volar capsule and the other soft tissues, e.g. neurovascular structures. When the flexor tendons are relaxed, it allows the fingers to assume a more functional position.
- The dorsal portion of the trapezoid wedge helps correct the wrist flexion deformity. The size of the wedge is determined by the need to achieve 40° of dorsiflexion; the wrist is then fixed with K-wires. In severe cases almost all the carpal bones may need to be included in the wedge and rarely the base of the second to fifth metacarpals.

### Soft tissue balancing

The wrist's dorsal capsule is incised transversely before the carpectomy. These flaps are then sutured tightly overlapping each other. The wrist flexor's flexor carpi ulnaris (FCU), flexor carpi radialis (FCR) and palmaris longus are transferred to the dorsal side to augment the dorsal pull on the metacarpals. The FCU and/or FCR are sutured to the extensor carpi radialis brevis (ECRB) or to the distal capsular flap. Z-lengthening of these flexors may be necessary in order to achieve this.

Wrist extensors may be poorly developed, but extensor carpi ulnaris (ECU) can be centralised to compensate for weak wrist extension.

Mennen recommends the sequence of carpectomy followed by internal pinning, then performing the capsular suturing and finally doing the flexor tendon transfer to protect the volar neurovascular structures, which may be tensioned unnecessarily if another sequence is followed.<sup>3</sup>

Older patients or patients with recurrence of their deformity may benefit from wrist arthrodesis as a salvage procedure to achieve a more functional position of their wrist.

### Thumb adduction

The thumb-in-palm deformity is the result of a combined thumb adduction and first MCP joint flexion contractures and prevents the hand from grasping normally. If the thumb fails to correct with passive and dynamic first web space manipulation, then a thumb adduction release is indicated which may need to be combined with an opponensplasty.<sup>3</sup> The flexor pollicis longus musculotendinous complex is also released to achieve adequate correction. Williams recommended a combined first web space release with transfer of a superficial flexor tendon (usually the ring finger's superficial tendon) dorsally to replace the typically absent thumb extensors and abductor. Drummond *et al.* suggested a Z-plasty for the first web space and release of adductor pollicis with or without MPJ fusion.<sup>11</sup>

### Finger stiffness

Improvement in ROM is seen with gentle manipulation. It is also noted that if the wrist is placed in 40° of dorsiflexion before 12 months of age the finger and metacarpophalangeal joints are more mobile and normal skin folds over the joints can develop.<sup>3</sup> Occasionally contractures may need to be released and skin grafted. Williams described an intrinsic release for patients with MP joint flexion contracture and extension of the IP joints. If the IP joints have an extension contracture then a dorsal release can be done with a flexor tendon shortening.<sup>11</sup>

After correction of wrist and hand deformities the patient is usually splinted in a functional position until skeletal maturity.

### Management of the elbow in AMC

Elbow flexion is particularly important in these patients in order to achieve independent function in feeding and care of the face and hair. Extension of the elbow is required for toilet and transfers if the lower limbs are severely affected. Ideally one arm (dominant arm) should be able to function in flexion to perform feeding activities, and one arm should be able to function in extension for hygiene purposes.

Goals of treatment are to achieve at least 90° flexion from a fixed extended position. If both elbows are equally involved, surgery to increase flexion should only be done on the one side.

### Elbow flexorplasty

Arthrolysis and capsular release are indicated if passive manipulation has not achieved more than 90° of elbow flexion by 6 months' of age. The triceps can be lengthened by a Z- or V-Y lengthening procedure if necessary. If active elbow flexion is lacking the surgeon will need to do a flexorplasty at the same time as the joint release procedure, bearing in mind that passive elbow flexion to 90° is a prerequisite. Various options are available for an elbow flexorplasty:



- Steindler flexor origin transfer – The flexor origin is released from the medial epicondyle and transposed proximally and anteriorly on the humerus. It is seldom recommended in AMC as the flexor muscles are shortened, fibrotic and have poor excursion and may further tighten wrist and finger flexors in a patient with existing wrist and finger flexor contractures. The flexor group of muscles is also too weak to achieve active elbow flexion.
- Clark pectoralis major muscle transfer: Here 2½ inches of the sternal head of pectoralis major is detached, tubed and attached to the biceps tendon at the elbow. Schottstaedt, Larsen and Bost modified the technique by detaching the entire sternal head of pectoralis major. The muscle is completely mobilised on its neurovascular pedicle, the muscle insertion reattached to the acromion process and the sternal origin to the biceps tendon or the radius with rectus fascia. The muscle is seldom functional but occasionally it may be powerful enough for elbow flexion.<sup>3</sup>
- Latissimus dorsi muscle transfer (Hovnanian): The origin of latissimus dorsi is detached and the muscle belly mobilised on the long thoracodorsal nerve, passing it subcutaneously down the anterior aspect of the arm and suturing it to the biceps tendon. Like pectoralis major this muscle is often non-functional in AMC but if it is available it is the best option for elbow flexion.<sup>3</sup>
- Triceps tendon transfer: This is a viable option for tendon transfer to achieve elbow flexion if the triceps muscle strength is at least a grade 4/5. The technique of Carroll and Hill involves detaching the triceps aponeurosis and periosteum from the olecranon and proximal ulna, which is passed subcutaneously around the lateral aspect of the elbow and attached to the proximal radius or biceps tendon. The disadvantage of the procedure is that if an undesirable flexion contracture of the elbow is created, it will be nearly impossible to correct. If a flexed elbow of more than 90° occurs in one arm and the other arm is in extension, the patient loses the ability to transfer objects from one hand to the other, losing the bimanual function. A flexed elbow has the functional advantage of being able to reach the mouth and the perineum and performing most other activities of daily living. A gutter crutch may also be used if the patient has difficulty with walking and stability.
- Van Heest *et al.* demonstrated that elbow capsulotomy and triceps lengthening alone without tendon transfer improved passive elbow flexion and the arc of elbow motion to enable hand-to-mouth activities.<sup>7</sup> Twenty-nine elbows were operated in 23 children and an average of 33° of passive motion was achieved, changing the arc of motion to a more flexed position. The authors felt that the risk of tendon transfer after capsulotomy may outweigh the benefits if the patient could achieve functional independence by other means such as compensatory movements.

In the older patient an elbow arthrodesis (Kelikian) or an anterior closing wedge osteotomy of the distal humerus may be used to place the patient's limited arc of motion in a more functional position.

If the radial head is dislocated it should not be excised until growth is completed to prevent a progressive cubitus valgus or tardy ulnar nerve palsy developing.

## Management of the shoulder in AMC

Management of the shoulder is seldom needed in AMC as it usually functions satisfactorily without treatment. Flexion and abduction (active and passive) are usually sufficient to allow the patient to reach the mouth or perineum. If the shoulders are in severe internal rotation the hand function may be limited and forced to function in a back-to-back fashion or crossover style to hold objects. Toilet usage may be a problem as the dorsum of the hand presents to the perineum, and walking may be restricted by inability to grasp crutches or a walking frame.

If the hands can be made functional enough an external rotation osteotomy of either the proximal or distal humerus can be performed.

## Prognosis

The skin folds develop over joints as soon as movement around that joint starts. Mennen<sup>5</sup> reported the expected functional improvement around joints, after early one-stage corrective surgery (before 1 year) in 47 limbs operated:

Elbow:	30–100° flexion (average of 49 degrees)
Wrist:	10° flexion 30° extension (average 27° degrees active motion)
Fingers:	MCPJ: 20–85° flexion (average 65° degrees active flexion)
PIPJ:	20–80° flexion (average 45° active flexion)
DIPJ:	15–35° flexion (average 20° active flexion)

## Conclusion

To achieve the best results for this complex condition of the upper limb, manipulation of deformities is recommended as soon as possible after birth. If surgery is required to gain function then it should be done as an early one-stage procedure between the ages of 3 months to 1 year.

*No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.*

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