Arthrogryposis Multiplex Congenita

Description

Arthrogryposis multiplex congenita is a collective term applied to a very large number of different syndromes characterised by non-progressive, multiple joint contractures present at birth.\(^1\)\(^2\) The joints usually develop normally in early embryonic life but, as gestation progresses, movements are required to facilitate normal development. Where there are abnormalities that prevent this from occurring, such as neurological or connective tissue disorders or physical restriction, the condition forms. The muscles involved are partially or completely replaced by fat and fibrous tissue. The most common form, accounting for 40% of cases, is amyoplasia.

Other secondary problems are associated with generalised fetal akinesia.

Epidemiology

- It may occur to some extent in 1 in 3,000 to 1 in 5,000 live births.\(^3\) The condition is usually detected at birth or before by ultrasound examination.
- It is often secondary to other conditions.
- If they are X-linked this will produce a male preponderance but otherwise there is an equal sex incidence.
- It has been found to be more common in some isolated communities in Finland and Israel.

Aetiology\(^4\)

Over 150 different syndromes may cause this symptom complex; therefore, careful history and examination are important to try to elucidate the underlying cause. The basic cause is fetal akinesia (reduced fetal movements).

The underlying cause can be environmental (lack of ability to move) or genetic (single gene conditions). The underlying cause may be part of a more complex disorder affecting muscles, nerves or connective tissue. A variety of maternal illnesses may result in arthrogryposis. This varied aetiology has contributed to a more complex classification of the symptom complexes.

Amyoplasia is the most common type of arthrogryposis and this accounts for about one third of cases. The conditions may be classified as below and broadly under the following headings:

- Disorders characterised mainly by limb involvement.
- Disorders that involve the limbs and other body parts.
- Disorders with limb involvement and central nervous system dysfunction.
- Other associated syndromes and conditions.

Classification

Disorders with mainly limb involvement

- Amyoplasia.
  - Sporadic condition.
  - Most common type of arthrogryposis (accounting for one third of cases).
  - Distinct appearance of limbs and joints (including internally rotated, adducted shoulders, fixed extended elbows, pronated forearms, flexed wrists and fingers and bilateral talipes equinovarus).
  - Normal IQ.
  - Oddly, 80% have midline facial capillary haemangioma.

- Other distal arthrogryposes. There are seven subtypes, classified as type I, type II and types IIA to IIE.

Disorders with involvement of limbs and other body parts

- Multiple pterygium syndrome (pterygium meaning 'wing' and referring to triangular membranes affecting the neck, knees, elbows, ankles, etc):
  - Autosomal recessive: multiple joint contractures with marked pterygia (and dysmorphic facies and cervical vertebral anomalies).
  - Autosomal dominant: multiple pterygia (with or without general learning disability).
• Other syndromes:
  • Freeman-Sheldon syndrome.
  • Osteochondrodysplasias.
  • Chromosomal disorders.
  • Cerebro-oculo-facial skeletal syndrome.

Presentation

History
History can be examined in terms of family history, pregnancy and delivery.

Ask about family history of:
• Other affected children or members of the family.
• Consanguinity - increases the risk of rare recessive disorders.
• Increasing parental age - may increase risk, both mother and father.
• A parent who may have a mild form or have had infantile contractures.
• Miscarriages, possibly with fetal abnormalities.
• Maternal disease. This may include myotonica dystrophica that can produce a very severe condition but also myasthenia gravis and multiple sclerosis.

Ask about the pregnancy and delivery:
• Infection with some viruses, including rubella and Coxsackie, can cause neuropathy.
• Prolonged maternal pyrexia can produce contractures due to abnormal nerve growth and migration. This can also be produced by very hot baths, hot tubs and saunas in pregnancy.
• Drugs, including phenytoin and alcohol, can impair fetal movements.
• Oligohydramnios reduces fetal movements. A septate uterus or large fibroids can do the same.
• An abnormal lie is common and this will complicate delivery.
• There may have been amniotic bands or placental abnormality. A short cord or one wrapped around a limb reduced mobility.
• Multiple pregnancy restricts room to move.

Examination
Classical presentation of amyoplasia shows involvement of both upper and lower extremities with the lower extremities typically more involved. Abnormalities can include:
• Shoulders (adduction, internal rotation).
• Elbows (extension or fixed flexion).
• Wrists (deviation).
• Deformity of thumb and palm and rigid interphalangeal joints.
• Hips (with dislocation of one or both sides).
• Knees (fixed extension or flexion).
• Rigid bilateral club feet/vertical tali.
• Other characteristic features include:
  • Thin subcutaneous tissue and absent skin creases.
  • Symmetrical deformities (becoming more severe distally).
  • Rigid joints.
  • Congenital dislocation of the hips (and sometimes the knees).
  • Atrophy or absence of groups of muscles.
  • Normal sensation.
  • Contractures (especially of distal joints).
  • Pterygia (these are winglike triangular membranes occurring typically in the neck, knees, elbows, ankles or fingers).
  • Other deformities of the limbs (including shortening, webs, compression often due to cord wrapping, absent patellae, dislocated radial heads, and dimples).
  • Deformities of face and jaw (including asymmetry, flat nasal bridge, haemangioma, micrognathia and trismus).
  • Scoliosis, genital deformity and umbilical or inguinal herniae are common.
  • There may be many other malformations of the skeleton, respiratory tract, urinary system and nervous system.

Differential diagnosis
There is a wide and varied range of rare conditions to be considered in the diagnosis. Conditions causing arthrogryposis include:

Fetal abnormality
- Neurogenic disorders - eg:
  - Myelomeningocele.
  - Sacral agenesis.
  - Spinal muscular atrophy (anterior horn cell disease of prenatal origin (SMA 0), not Werdnig-Hoffman (SMA 1).
  - Congenital contracture syndrome (lethal).
  - Cerebro-oculo-facial syndrome.
  - Marden-Walker syndrome.
  - Pena-Shokeir syndrome.

- Myopathic disorders - eg:
  - Congenital myopathies.
  - Congenital muscular dystrophy.
  - Myasthenic syndromes.
  - Intrauterine viral myositis.
  - Mitochondrial disorders.

- Connective tissue disorders - eg:
  - Diastrophic dysplasia.
  - Osteochondroplasia.
  - Metatropic dwarfism.

- Mechanical limitations to movement - eg:
  - Oligohydramnios as in Potter's syndrome and multiple births.

Maternal disorders

- Maternal infection (including rubella, Coxsackievirus and enterovirus infections).
- Drugs (including, for example, alcohol, phenytoin and methocarbamol).
- Trauma.
- Intrauterine vascular abnormalities/compromise.
- Other maternal illnesses (including myotonic dystrophy, myasthenia gravis and multiple sclerosis).
Investigations

- X-rays of all joints may show bony abnormalities, including missing bones, skeletal dysplasias, scoliosis, ankylosis and fractures arising from difficult delivery. X-ray of the spine and pelvis should always be included.
- Ultrasound, CT and MRI scans may all be useful to assess muscle mass and abnormalities in other tissues like the central nervous system.
- If muscles are very flaccid, a blood test for creatine kinase may be revealing. Antiviral antibody may also show a cause.
- Cytogenetic studies may be required

Associated diseases

Other problems can develop as a result of fetal akinesia:

- Polyhydramnios
- Pulmonary hypoplasia
- Micognathia
- Ocular hypertelorism
- Short umbilical cord

Management

This is likely to involve several different specialists and therapists. Ideally, management should be co-ordinated by a key specialist (often a paediatrician) who is part of a team looking after affected patients. Broadly speaking, management can be divided into medical, surgical, social and psychological care.

Medical care

Physical therapy to improve the range of motion and stretch surrounding tissues is very useful, especially in amyoplasia and distal arthrogryposis, although in diastrophic dysplasia it may lead to ankylosis instead. It should be started early. Splinting between times can correct deformity, especially in the hands and wrists. Serial casting after physical therapy has achieved maximum usefulness with weekly changes of cast and gentle manipulation.

Surgical care

Surgery is often needed to correct soft tissue contractures and joint deformities. It can also reduce and stabilise dislocated hips, correct foot deformities and stabilise spinal deformities. Anaesthesia can be a problem, as some patients with some forms of arthrogryposis are at risk of malignant hyperpyrexia.

Social and psychological care

This encompasses some fundamental care needs which will be associated with the condition. The extent of the need for care will vary depending on the individual, their circumstances and the impact of the condition on the particular individual. Information is important and support groups provide useful information and advice. The new system of social care and support is called Self-directed Support and this should allow for the varied and complex care needs of this particular condition.

Prognosis

- Prognosis depends on the underlying cause but most have a normal lifespan.
- If, however, there is a central nervous system problem in addition, about half of patients die in the first year.
- Scoliosis is common and can appear at any age but needs correction before it becomes severe.
- The long-term prognosis in terms of dependency is poor in many cases.
- Patients with amyoplasia tend to have a much better prognosis.
## Prevention

Genetic advice may be essential to prevent arthrogryposis. Extrinsically derived contractures have a low recurrence risk but the recurrence risk for intrinsically derived contractures depends on the aetiology.

### Further reading & references


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<th>Author: Dr Colin Tidy</th>
<th>Peer Reviewer: Dr John Cox</th>
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<td><strong>Document ID:</strong> 1822 (v23)</td>
<td><strong>Last Checked:</strong> 23/06/2015</td>
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