About Arthrogryposis
Multiplex Congenitae (AMC)

Over 200 different conditions have been identified as being under the Arthrogryposis ‘umbrella’ and all can be divided into one of 3 groups:

1. Amyoplasia – 1/3 of all cases of AMC
2. Heterogeneous group of disorders – 1/3 of all cases of AMC
3. Contractures plus Central Nervous System (CNS) involvement – 1/3 of all cases of AMC

Amyoplasia

The most common form of Arthrogryposis is Amyoplasia, or ‘classic Arthrogryposis’, which accounts for a third of all cases/babies born with Arthrogryposis. Amyoplasia is not a ‘genetic’ condition and so not passed down through other generations of a family. However, parents of a baby with Amyoplasia may have been advised to seek genetic counselling for their child as he/she approaches adulthood.

Distal Arthrogryposis

Distal Arthrogryposis Type I: usually involvement is restricted to the ‘distal’ parts of the limbs, in other words, primarily hands, wrists, ankles and feet. In the newborn the hands have a particular appearance with a clenched fist and overlapping fingers. Feet can be affected in different ways: calcaneas valgus or clubfeet; one of each or sometimes neither foot is involved.

This type has an autosomal dominant* inheritance and, as only one parent must have an abnormal gene in order for the child to inherit the disease, there is a 50/50 chance of passing it on.

Distal Subgroups

Distal Arthrogryposis subgroups tend to have similar distal limb contractures as D A Type I but with additional features. Below are the most common of these different types.

Freeman Sheldon Syndrome

As well as contractures of the hands and feet this condition also has facial involvement. The muscles are contracted in such a way as to give a ‘whistling’ appearance, as the name suggests. In the more severe cases the mouth may be very pursed. When the face is not so affected it might still be difficult for the individual to smile.

Distal Arthrogryposis Type II A

This form of Distal Arthrogryposis Type II A is known as Gordon syndrome and as well as the Distal Type I features there is a cleft palate and short stature. This type is autosomal dominant.

Distal Arthrogryposis Type II B

This Type of D A also has the features of paralysis to all or part of the eye and the eyelids may appear droopy. As well as muscle weakness around the eyes the eyes themselves may...
have limited movement, especially to the sides. Skin and muscles are hard and there is a lack of finger creases. This type is also autosomal dominant and quite common.

** Distal Arthrogryposis Type II C** This Type of D A also has the feature of a cleft lip.

** Distal Arthrogryposis Type II D** With this Type of D A there is also scoliosis (curvature of the spine).

** Distal Arthrogryposis Type II E** This type of D A has a characteristic positioning of the hand in which the wrist is flexed but the metacarpalphalangeal joint (palm to wrist) is extended. There is limited opening of the jaw, trismus*. It is not inherited but sporadic.

* Autosomal dominant inheritance: A single, abnormal gene on one of the autosomal chromosomes (one of the first 22 “non-sex” chromosomes) from either parent can cause certain diseases. One of the parents will usually have the disease (since it is dominant) in this mode of inheritance. Only one parent must have an abnormal gene in order for the child to inherit the disease.

** Pterygium Syndromes**

Many syndromes have the feature of this triangular membrane across body joints such as the neck, knee and elbow. Some of these conditions are autosomal dominant (AD). Others are autosomal recessive** (AR) which means both parents must be carriers in order for the child to have the symptoms of the disease. Blood vessels and nerve run along the edge of this web and so much care is needed when operating.

** Autosomal recessive inheritance: An abnormal gene on one of the autosomal chromosomes (one of the first 22 “non-sex” chromosomes) from each parent is required to cause the disease. People with only one abnormal gene in the gene pair are called carriers, but since the gene is recessive they do not exhibit the disease. In other words, the normal gene of the pair can supply the function of the gene so that the abnormal gene is described as acting in a recessive manner. BOTH parents must be carriers in order for a child to have symptoms of the disease; a child who inherits the gene from one parent will be a carrier.