Attending a genetic counselling appointment

In the past people tended to be told that they had Arthrogryposis Multiplex Congenita (AMC), rather than be given a specific diagnosis. Meanwhile, AMC is a description of joint contractures present at birth in more than one body area – it is not a diagnosis. There are over 200 specific conditions which come under the AMC umbrella.

In the last few years there have been significant developments in the field of genetics and an increasing number of genes have been identified as being associated with different conditions that come under the AMC umbrella – that is to say conditions that have multiple joint contractures as one of the symptoms. Consequently, there is generally greater understanding about the different types of Arthrogryposis today and people are more likely to be given: a specific diagnosis; a clearer idea of what inheritance pattern to expect.

Why ask to be referred to a genetic counsellor?

AMC is a rare condition and even medical professionals may not have come across it. As a result you may not have been advised to seek genetic counselling or you may not have felt it had any relevance, especially if you were told that it would not change the approach to treatment, or perhaps if you were not thinking of having anymore children. Some people are told it may not be worth seeking genetic counselling until the child affected with AMC approaches adulthood.

Nevertheless, it has become clear that the prognosis for different conditions under the AMC umbrella can vary. According to Judith Hall, Professor Emeritus and one of the main authors of the book Arthrogryposis – A text atlas, prognosis depends on the specific diagnosis and the natural history of that disorder. Also, a child's prognosis is usually clear by the age of 3 or 4.

Reasons for seeking genetic counselling include:

- Giving birth to a baby with Arthrogryposis
- Wanting to have more children
- The affected child is approaching adulthood
- Only the name of Arthrogryposis has been used – no specific diagnosis ever given
- The diagnosis of Distal Arthrogryposis has been given but the actual type not specified

What questions might I/we be asked?

The consultant will take into consideration the physical symptoms i.e. which joints are affected and how; whether there is scoliosis, respiratory or other problems. The consultant is also likely to ask many questions regarding the pregnancy and medical history of the affected child and mother as well as questions on the family history through the generations.

The pregnancy and medical history

- Was there maternal fever or viruses during pregnancy?
- Was there oligohydramnios (too little amniotic fluid) or an unusually shaped uterus?
- Was the baby active throughout the pregnancy?
- Are there any problems with hands, wrists, elbows, shoulders, knees, jaw, or spine?
- Is there any muscle weakness or hypotonia (poor muscle tone)?
- Did the baby have any muscle biopsies or other blood tests to rule out possible disorders?

Family history

- Is there anyone else with club foot, joint dislocations or scoliosis?
- Are there any individuals who are short in stature?
- Is there anyone with muscular dystrophy, muscle disease, muscle weakness?
- Is there anyone with cleft lip and/or palate, hearing loss, learning difficulties?

How can I/we prepare for the appointment?

If you are able to answer some of the questions above at your first appointment you are going to give the geneticist information that may help her or him to establish a better picture sooner. So it might be a good idea to do a bit of ‘family’ research beforehand and also to think a little about the pregnancy.

Remember this is also your opportunity to ask questions too and so it is a good idea to prepare a list before you go. It is very easy to forget things once you are in the appointment, especially if you are having to digest a lot of technical information or you are having to deal with information that affects you emotionally.

Not just for genetic appointments but as a matter of course Professor Judith Hall advises families to always: Ask questions; take photographs; make a notebook; keep records; ask questions!

Useful Organisations

The Genetic Interest Group (GIG)
www.gig.org.uk Tel: 020 7704 3141

Unique (Rare Chromosome Disorder Support Group)
www.rarechromo.org
Tel: 01883 330766

The Arthrogryposis Group