

Fact Sheet

Connective tissue dysplasia



The name "connective tissue dysplasia" covers a wide range of disorders. These disorders are caused by defects in the connective tissues such as bone, ligaments, tendons and skin.

Children with these disorders may have:

- too much joint movement (hypermobility)
- not enough joint movement (joint contractures - hypomobility)
- excessive laxity (looseness) of skin or
- fragile bones, skin, ligaments etc.

The disorders can be variable because many affected people have a mixture of these different symptoms.

Most connective tissue dysplasias follow common patterns of inheritance. These patterns can help genetic counsellors to provide families with information about inheritance in their family.

There are similarities between the disorders, so that research findings in one disorder are often of great help in understanding another disorder.

Kinds of disorders

The disorders can be grouped into:

- Arthrogyryposis and related disorders
- Ehlers-Danlos Syndrome and familial hypermobility
- Marfan Syndrome
- Mucopolysaccharidoses
- Osteochondrodysplasias
- Osteogenesis Imperfecta.

Arthrogyryposis and related disorders

The two most common signs of this disorder are:

- joint hypomobility (not enough joint movement)
- contractures (excessive tightness of the skin or ligaments).

Decreased joint movement is present at birth and in the most severe cases, virtually all joints are affected. However, in most people with Arthrogyryposis, only some joints are affected. The aim of rehabilitation is to restore normal function and develop each individual's strengths.

There are two major subgroups:

- Amyoplasia, where there is a lack of muscle development in most areas and
- Genetic arthrogyryposis which often involves mainly the hands and feet (distal).

Ehlers-Danlos Syndrome

People with this syndrome have hypermobile joints (very loose joints) from birth and throughout life. Some people may have fragile skin, and a tendency to sprains, while others have fragile blood vessels with a tendency to bruising or blood vessel rupture.

There may be deformities of the spine such as scoliosis, or joint contractures due to dislocation of joints with damage to nearby muscles.

Rehabilitation goals include prevention of trauma to skin, protection of loose joints, and treatment of specific complications.

Marfan Syndrome and related disorders

The Marfan Syndrome is associated with tall stature and long arms and legs. There is an increased risk of visual disability and blood vessel rupture in adult life.

People with Marfan Syndrome usually have very loose joints, but some people have very tight joints and ligaments. A special group have the related disorder, Beals Contractural Arachnodactyly.

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Mucopolysaccharidoses (MPS)

This group of inherited disorders are caused by a lysosomal enzyme deficiency. The body does not produce any, or very little enzyme, so complex sugars are stored in the body tissue instead of being broken down and used by the body. These complex sugars are called polysaccharides, which is how this disorder gets its name.

MPS is a progressive disorder, although that progress may be slow. Some children have progressive storage of polysaccharides in the brain, which can be quite damaging.

Most children with various types of MPS have very tight joints (joint contractures), but some have very loose joints (joint hypermobility).

Osteochondrodysplasias

There are about 300 disorders in this group. They are generally diagnosed by an x-ray of the skeleton. They affect bone (bone dysplasias) and/or cartilage (chondrodysplasias).

Most result in very short stature, such as in Achondroplasia, but some have average or tall stature. Joints may be either contracted or hypermobile. Some people have congenital heart defects, cleft palate, extra fingers or visual disabilities.

Osteogenesis Imperfecta (OI)

People with OI have very fragile bones. There are different types of OI and a wide range in severity from person to person. In some people, the whites of the eyes, called sclerae, appear very blue. People with OI may be very short and some develop severe deformity of the limbs and/or spine. Teeth may also be fragile. Some affected individuals develop deafness in later life.

Other connective tissue dysplasias

There are other disorders which are also called Connective Tissue Dysplasias. These include rare disorders such as the Weill-Marchesani syndrome and common disorders such as familial hip dysplasia.

Advice about diagnosis and management of any of these conditions can be sought through your local genetic service.

Remember

- A genetic counsellor will provide you with information about inheritance in your family.

This fact sheet is for education purposes only.
Please consult with your doctor or other health professional to make sure this information is right for your child.

This document was reviewed on Thursday, 29th March 2007.

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