FACTSHEET

Connective tissue dysplasia

The name "connective tissue dysplasia" covers a wide range of disorders. These disorders are caused by a weakness 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)
- fragile bones, skin, blood vessels or ligaments
- degenerative joint disease
- short stature
- spinal complications

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 provide families with information about inheritance in their family.

Kinds of disorders

The disorders can be grouped into:

- Ehlers-Danlos Syndromes (EDS)
- Marfan Syndrome and related disorders
- Skeletal Dysplasias
- Brittle Bone Disorders
- MPS Disorders (Mucopolysaccharidoses)

Ehlers-Danlos Syndrome

The more severe forms of Ehlers-Danlos syndromes (EDS), all have some degree of tissue fragility from birth and throughout life. Some people may have fragile skin (classic type), while others have fragile blood vessels with a tendency to bruising or blood vessel rupture (vascular type). Rarely, the corneas of the eyes are brittle and this may result in visual loss.

Management goals include prevention of trauma to skin, blood vessels and eyes, protection of loose joints, and treatment of specific complications.

Joint Hypermobility Syndrome, also known as Ehlers Danlos syndrome of the Hypermobile Type, is the combination of widespread joint hypermobility (double jointedness) and chronic musculoskeletal issues. Please see the separate factsheet titled 'Joint Hypermobility'.

Marfan Syndrome and related disorders

Marfan Syndrome can be associated with tall stature and long arms and legs. There is an increased risk of visual disability in childhood from dislocation of the lens of the eye. Aneurysm of the main blood vessel (aortic aneurysm) may also occur, but serious health complications can be avoided through regular health and heart check-ups.

Skeletal Dysplasias

There are about 400 disorders in this group. They affect bone (bone dysplasias) and/or cartilage (chondrodysplasias).

Most result in short stature and intelligence is usually normal. Where short stature is present there are different patterns with some people having normal upper body height and short limbs, and others with a short trunk. Some skeletal dysplasias are associated with problems with internal organs and visual and hearing loss. Each disorder or group of disorders has specific requirements for management.

Build-up of complex sugars in the body can cause problems with the growth and function of the skeleton and connective tissue as seen in the Mucopolysacccharidoses (MPS) disorders.

There can be a range of manifestations, depending on the type of MPS.

Brittle Bone Disorders

People with Brittle Bone Disorders have fragile bones that fracture easily. There are different types of Brittle Bone Disorders and the range and severity vary from person to person.

Osteogenesis Imperfecta (OI)

Osteogenesis Imperfecta (OI) is the most common type of Brittle Bone Disorder. In some people with OI, the whites of the eyes, known as sclerae, appear blue.   People with severe forms of OI may have short stature and some develop deformity of the limbs and/or spine. Teeth may also be fragile. Children with Brittle Bone Disorders need specialised multidisciplinary assessment and treatment.

> **Remember:**
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> If you have concerns about your child please discuss them with your GP or Paediatrician, most public hospitals in NSW have a Genetics Clinic where advice on diagnosis can be obtained.