



# British Society for Paediatric Endocrinology and Diabetes

## MARFAN'S SYNDROME

Marfan's syndrome is a genetic condition in which the connective tissue of the body, which is an important structural component in all organs, is abnormally weak and elastic. Marfan's syndrome affects about 1 in 5000 people, and has variable features. It mainly affects the eyes, skeleton, heart and major blood vessels.

### Cause

Each human cell contains 23 pairs of chromosomes, one of each pair inherited from the mother and one from the father. Each chromosome carries many genes along its length and sometimes faults in genes are expressed as faults in the structure of body chemicals.

Marfan's syndrome is caused by a fault in a single dominant gene. This means that Marfan's syndrome is present when one of a pair of genes, inherited either from the mother or the father, is affected. In about 25% of people with Marfan's the fault in the gene has occurred spontaneously before birth (a mutation) and in 75% it is inherited. Males and females are equally likely to be affected and, whether the gene was inherited or not, have a 50% chance of passing on the abnormal gene, and so the condition, to children of either sex.

The gene for Marfan's syndrome lies on chromosome 15 and carries the code for the structure of fibrillin that is an important part of collagen, the main component of connective tissue.

### Features of Marfan's syndrome

People with Marfan's syndrome are tall and usually of very slim build with especially long limbs, fingers and toes. They may have a high palate (roof of the mouth), either prominence or depression of the breast bone and sometimes curvature of the spine, especially in adolescence. They tend to have an early puberty and stop growing early so the final height of children is seldom as tall as seems likely through the school years. There is usually a degree of hypotonia (floppiness) of the joints, often with "double jointedness".

These features may be apparent from birth or appear later in childhood. In the eyes the most common feature is short sightedness but there may be dislocation of the lens or a blue colour of the white of the eye. The changes in the heart and blood vessels cause the most serious potential complications. The heart valves may be abnormally floppy and this can cause them to become leaky. There may also be widening of the aorta, the major vessel carrying blood from the heart to the body, and this can develop a tear in the wall (aortic dissection). Some children with Marfan's syndrome have mild learning problems and there may also be a tendency to overactive behaviour.

### Diagnosis

The diagnosis is made by looking for the specific clinical features of Marfan's syndrome. Some people may have some of the physical features but do not have Marfan's syndrome. A genetic test may be used to confirm the diagnosis if there is uncertainty or to screen other family members.

### Management



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It is doubly important that people with Marfan's do not smoke because this further damages collagen tissue. Although there is no curative or specific treatment much can be done to assist with the correction of any visual problems, the development of strength and movement skills and the prevention of progressive skeletal abnormality.

The early detection and possible prevention (using beta blockers, a group of drugs which reduce the strain on the aorta) of progressive dilatation of the aorta is important. If progressive aortic dilatation or dissection does occur this section of the aorta may need to be replaced surgically. Affected people and their families need to be fully informed about this problem.

## **Outlook**

In spite of the rather daunting list of possible complications it is important to appreciate that most people with Marfan's syndrome live full lives with few restrictions. Life expectancy is more than 60 years and improving all the time with better treatment.

The Marfan Association, 5 Aldershot Road, Fleet, Hants GU51 3NG (01252-810472) is an excellent source of information and support for families.

<http://www.marfan-association.org.uk/>