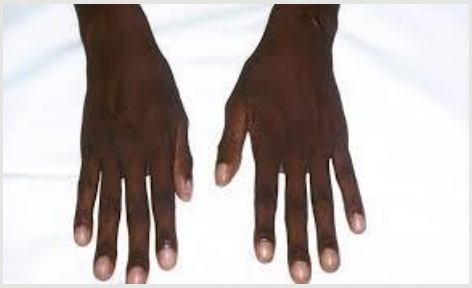

Hampshire Services
SPECIALIST TEACHER ADVISORY SERVICE

MARFAN SYNDROME

What is Marfan Syndrome?

Marfan syndrome is an inherited condition that affects the connective tissue. The primary purpose of connective tissue is to hold the body together and provide a framework for growth and development. In Marfan syndrome, the connective tissue is defective and does not act as it should. Because connective tissue is found throughout the body, Marfan syndrome can affect many body systems, including the skeleton, eyes, heart and blood vessels, nervous system, skin and lungs.

How does Marfan Syndrome affect the child?

Marfan syndrome affects people in different ways. Some people have only mild symptoms, while others are more severely affected. In most cases, the disorder progresses as the person ages. The body systems most often affected by Marfan syndrome are:

- **Skeleton** - People with Marfan syndrome are typically, but not always, very tall or taller than unaffected people in their family, slender and loose jointed. Marfan syndrome affects the long bones of the skeleton. Therefore the arms, legs, fingers, and toes may be disproportionately long in relation to the rest of the body.
- **Eyes** - More than half of all people with Marfan syndrome experience dislocation of one or both lenses of the eye. The lens may be slightly higher or lower than normal and may be shifted off to one side. The dislocation may be minimal, or it may be pronounced and obvious. Retinal detachment is a possible serious complication of this disorder. Many people with Marfan syndrome are also shortsighted (myopic), and some can develop early glaucoma (high pressure within the eye) or cataracts (the eye's lens loses its clearness).
- **Heart and blood vessels (cardiovascular system)** - Most people with Marfan syndrome have problems associated with the heart and blood vessels. The valve between the left chambers of the heart is defective and may be large and floppy, resulting in an abnormal valve motion when the heart beats. In some cases, the valve may leak,

creating a "heart murmur," which a doctor can hear with a stethoscope. Small leaks may not cause any symptoms, but larger ones may result in shortness of breath, fatigue and palpitations (a very fast or irregular heart rate). Because of faulty connective tissue, the wall of the aorta (the large artery that carries blood from the heart to the rest of the body) may be weakened and stretched, a process called aortic dilation. Aortic dilation increases the risk that the aorta will tear (dissect) or rupture, causing serious heart problems or sometimes sudden death.

- **Nervous system** - The brain and spinal cord are surrounded by fluid contained by a membrane called the dura, which is comprised of connective tissue. As people with Marfan syndrome get older, the dura often weakens and stretches, then begins to weigh on the vertebrae in the lower spine and wear away the bone surrounding the spinal cord. These changes may cause only mild discomfort or may lead to radiated pain in the abdomen or to pain, numbness or weakness of the legs.
- **Skin** - Many people with Marfan syndrome develop stretch marks on their skin, even without any significant weight change or pregnancy. These stretch marks can occur at any age and pose no health risk.
- **Lungs** - Restrictive lung disease occurs in 70 percent of persons with MFS. Spontaneous pneumothorax (collapse of the lung in the absence of trauma) and early emphysema, without a history of smoking, may be consequences. Sleep-related breathing disorders, such as snoring and sleep apnea, are also associated with Marfan syndrome, even when the person is not overweight.

What can be done to help?

For both children and parents, appropriate medical care, accurate information and social support are key to living with the condition. Genetic counselling may also be helpful in understanding the disorder and its potential impact on future generations.

Marfan Syndrome can affect visual acuity and functional vision in different ways. Your Specialist Teacher Adviser (Visual Impairment) will be able to advise on implications for your particular pupil/child.

Useful websites:

RNIB	**www.RNIB.org.uk**
VI Scotland	**www.viscotland.org.uk**
The Marfan Trust	**www.marfantrust.org**