Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

MANAGING MARFAN & ITS FUTURE

See the Signs, Save a Life

A rare, genetic condition, Marfan syndrome is a lifelong challenge. It is a disorder of the body's connective tissue and belongs to a wider family of such conditions that includes Loeys-Dietz syndrome. Connective tissue maintains the structure of the body and supports the internal organs. In Marfan, the body is deficient in the protein fibrillin-1, weakening the cells that support its structure. This has implications for the skeleton, the eyes, and most life-threateningly, the heart. It is estimated that 18,000 in the United Kingdom have Marfan syndrome. Of these, 8,000 are undiagnosed. Early diagnosis of Marfan syndrome saves lives.

Spanish actor Javier Botet (above) has Marfan syndrome

"Once a doctor meets a person with Marfan syndrome, the doctor never forgets. Otherwise a doctor may not know about this unusual condition, and awareness is so important, as early diagnosis of Marfan syndrome can ensure correct management and save lives".

Dr Anne Child, Medical Director, Marfan Trust

Marfan syndrome is 'democratic' and can affect any gender or ethnicity. The severity varies from person to person. 75% of patients inherit the condition whilst 25% develop it as a result of a spontaneous (new) gene change. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

The condition unites a motley mix of famous people including English poet Edith Sitwell, French President Charles de Gaulle, and basketball player Isaiah Austin.

- Approximately 200 new cases of Marfan syndrome are identified every year
- Half of sufferers remain undiagnosed
- 25% of calls to the Marfan Trust Helpline are from those who suspect they or a family member has the syndrome but do not know how to approach a diagnosis

Signs, Symptoms, and Salient Features of Marfan Syndrome

The syndrome can manifest in many disparate ways which sometimes makes it difficult to diagnose. But there are classic characteristics which are easy to recognise once they are known. By virtue of the weakened connective tissue, everything in the Marfan body is stretchier and patients often grow to excessive height while remaining very slender. Limbs and fingers are disproportionally long, the spine is sometimes curved and the chest can protrude inwards, or stick outwards.

A GP or a doctor in A&E is in a position to assess and to make sense of these different signs and symptoms, and join the dots.

Skeleton

Musculo-skeletal problems are common and troublesome in MFS, with Marfan patients often growing to excessive height with long fingers and hammer toes while developing curvature (scoliosis/kyphosis/lordosis) of the spine. Skeletal problems also involve abnormally shaped chest (pectus deformity) and loose joints, often causing pain and dislocation

- Recognition from a RHEUMATOLOGIST could be the first vital step towards diagnosis of the underlying condition.

the classic Marfan hand and wrist signs

Eyes

People with Marfan syndrome are generally myopic, with some experiencing dislocation of the ocular lens and retinal detachment. Strabismus (squint) and glaucoma are also ocular signs.

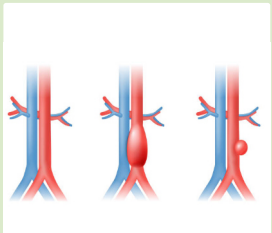
- Recognition from an OPTICIAN is sometimes the first vital step towards diagnosis of the underlying condition.

Heart (cardiovascular system)

Ballooning and potentially fatal tearing of the aorta and backward billowing of the heart's valves. These symptoms can cause death at an early age unless diagnosed in good time and treated

medically and surgically. Doctors must listen to the heart and send patients for an echocardiogram.

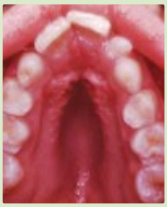
- Recognition from a CARDIOLOGIST can set you on the path to diagnosis.



Aortic aneurysm

Dental

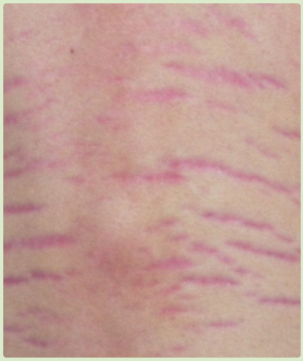
A high-arched palate and crowding of teeth with a history of tooth extraction.



- Recognition from a DENTIST is another a route to diagnosis of Marfan syndrome.

The lack of elasticity skin in Marfan syndrome means that patients often develop stretch marks in odd places such as on the back.

Skin



The lack of elasticity skin in Marfan syndrome means that patients often develop stretch marks in odd places such as on the back.

Every aspect of Marfan syndrome is treatable. As Dr Reed E. Pyeritz said many years ago: “30 years of research [into Marfan syndrome] equals 30 years of additional life expectancy.” Once diagnosed, the patient is monitored closely by the relevant teams, given the correct medication and surgery (if necessary), and can expect to live a normal lifespan.

“Jonny was tall and thin, his rib cage stuck out (pigeon chest), his toes curled, he had stretch marks on his back and he was able to touch his wrist with his thumb. His wing span was longer than hisheight. Had we heard of Marfan syndrome we may have been able to join the dots. Jonny’s heart would have been tested and he probably would have undergone surgery. He would be alive today”.

[Jonny’s mother]

Jonathan ‘Jonny’ Edwards died at just 23 of an aortic dissection. He bore the physical hallmarks of Marfan syndrome and had even spent time in hospital, but his signs & symptoms remained unrecognised. Had Jonny been monitored in cardiology, his life could perhaps have been spared.

The Marfan Trust

The Marfan Trust is the only charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome. We provide personalised support, conduct cutting-edge research, and continue to educate the world about the condition. You can help to secure our future by becoming a member today for just £3 per month, or by donating to our charity.