Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Registered Charity Number 328070

Marfan Trust, Guy Scadding Building, Dovehouse Street, London, SW3 6LY

W: www.marfantrust.org **E:** info@marfantrust.org **Patrons:** Sir Magdi Yacoub FRCS, Lady Maryanna Tavener, and Professor Marjan Jahangiri FRCS, FRCS (CTh)

MAKE MARFAN MATTER: Make Medicine Aware

Passing frequently unnoticed through GP surgeries and A&Es is a potentially **life-threatening condition**. **Marfan syndrome** is a rare disorder of the body's connective tissue, affecting 1 in 3,000 people. It is estimated there are **18,000 people** living in the United Kingdom with the condition, with **half of these living undiagnosed**, as Marfan hides in plain sight.

The American actor Vincent Schiavelli, below, had Marfan syndrome.

A disorder of the connective tissue means the body is missing the glue that binds it together. Quite simply, the body disconnects and in Marfan syndrome this can affect **three main systems** – the **skeleton**, the **eyes**, and most life-threateningly, the **heart**.

As our **Medical Director, Dr Anne Child** says: **"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".**

What is Marfan Syndrome?

Marfan syndrome (MFS) is an inherited disorder of the body's connective tissues that affects **men and women of any race or ethnic group**. It was identified in 1896 by a French physician Antoine Marfan. The severity varies from individual to individual and 75% of patients inherit the condition whilst 25% occur as a result of a spontaneous (new) mutation. Each child of an affected parent has a 50% chance of inherited Marfan syndrome.

- ***200 new cases are identified every year***
- ***Half of sufferers remain undiagnosed***
- ***25% of calls to the Marfan Trust Helpline are from those left undiagnosed by their GP and seeking support from our Charity***

Signs, Symptoms, and Salient Features of Marfan Syndrome

The syndrome manifests in many disparate ways which makes it difficult to diagnose.

As the cousin of a young man lost to Marfan syndrome said: "***He 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 his height. Had we heard of Marfan syndrome we may have been able to join the dots and he would have been tested and undergone surgery***". **A GP or a doctor in A&E is in a position to assess and to make sense of these different signs and salient features, and join the dots, in doing so potentially saving a life.**

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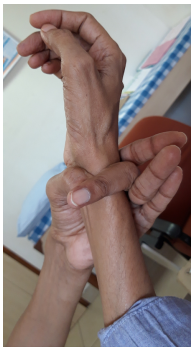
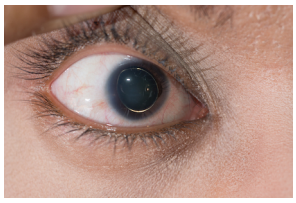


SKELETON: musculo-skeletal problems are common and troublesome in MFS, with Marfan patients often growing to **excessive height** with **long fingers** and **hammer toes** while developing **curvature** (scoliosis/kyphosis/lordosis) of the spine. Skeletal problems also involve **abnormally shaped chest**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

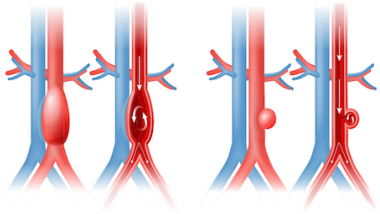
- **Recognition from a RHEUMATOLOGIST** could be the first vital step towards diagnosis of the underlying condition.



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.

AORTIC ANEURYSM



HEART: 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 the patient for an echocardiogram.

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.

SKIN: The lack of elasticity skin in Marfan syndrome means that patients often develop stretch marks.

The Marfan Trust

Marfan Trust is a charity dedicated to improving, and **saving the lives** of those with **Marfan syndrome**. We are not government funded but rely purely upon goodwill and public donation. Our Medical Director Dr Anne Child formed the international consortium that discovered the causative gene in the condition. We receive up to three calls a week from someone who suspects they may have Marfan syndrome.

E: info@marfantrust.org **T: 020 7594 1605** **www.marfantrust.org**