Marfan Syndrome Awareness Month

FIND THE MISSING 15,000

[The American actor Vincent Schiavelli, above, had Marfan syndrome]

Travelling frequently unnoticed through everyday life is a potentially fatal condition. Marfan syndrome is a rare genetic disorder of the body's connective tissue, affecting approximately 1 in 3,000 people. It is estimated there are 18,000 people living in the United Kingdom with the condition, 3,000 of whom are known to our medical charity. We hope to find "The Missing 15,000" as it is probable that half of these remain dangerously undiagnosed.

"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

What is Marfan Syndrome?

Put simply, in Marfan syndrome the body is missing the glue that binds it together, weakening its very structure. The condition can affect three main systems – the skeleton, the eyes, and, most life-threateningly, the heart – and any gender or ethnicity. The severity varies from person to person. 75% of patients inherit the condition whilst 25% occur 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 an unlikely mix of famous people including the original Chewbacca, and French President Charles de Gaulle, pictured.

- Approximately 200 new cases of Marfan syndrome are identified every year in the UK
- 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 obtain 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.

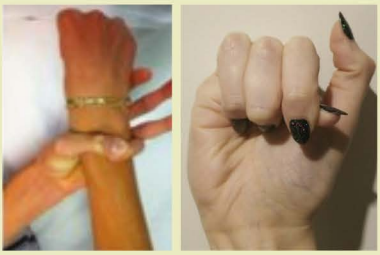
"Jonny was tall and thin, his rib cage stuck out (pigeon chest), his toes curled, he had stretch marks on his back and he was able to touch his wrist with his thumb. His wing span was longer than his height. Had we heard of Marfan syndrome we may have been able to join the dots. Jonny's heart would have 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, a ticking time bomb that had been hiding in plain sight. He bore the standard physical hallmarks of Marfan syndrome and had even spent time in hospital. Had his signs and symptoms been recognised earlier, and had Jonny been monitored in cardiology, his life could perhaps have been spared.

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.



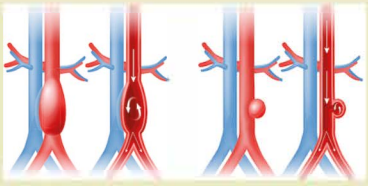
[pictured: the classic Marfan hand and wrist signs]

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.



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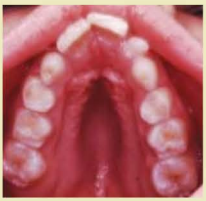
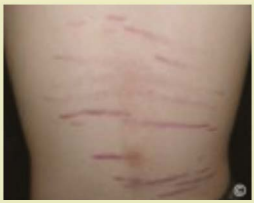


DENTAL: A high-arched palate and crowding of teeth with a history of tooth extraction.

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



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

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, receiving the correct medication and timely surgery (if necessary), and can expect to live a normal lifespan.

The Marfan Trust is the sole 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.

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Marfan Trust

Supporting Research into Inherited Aortic and Related Disorders