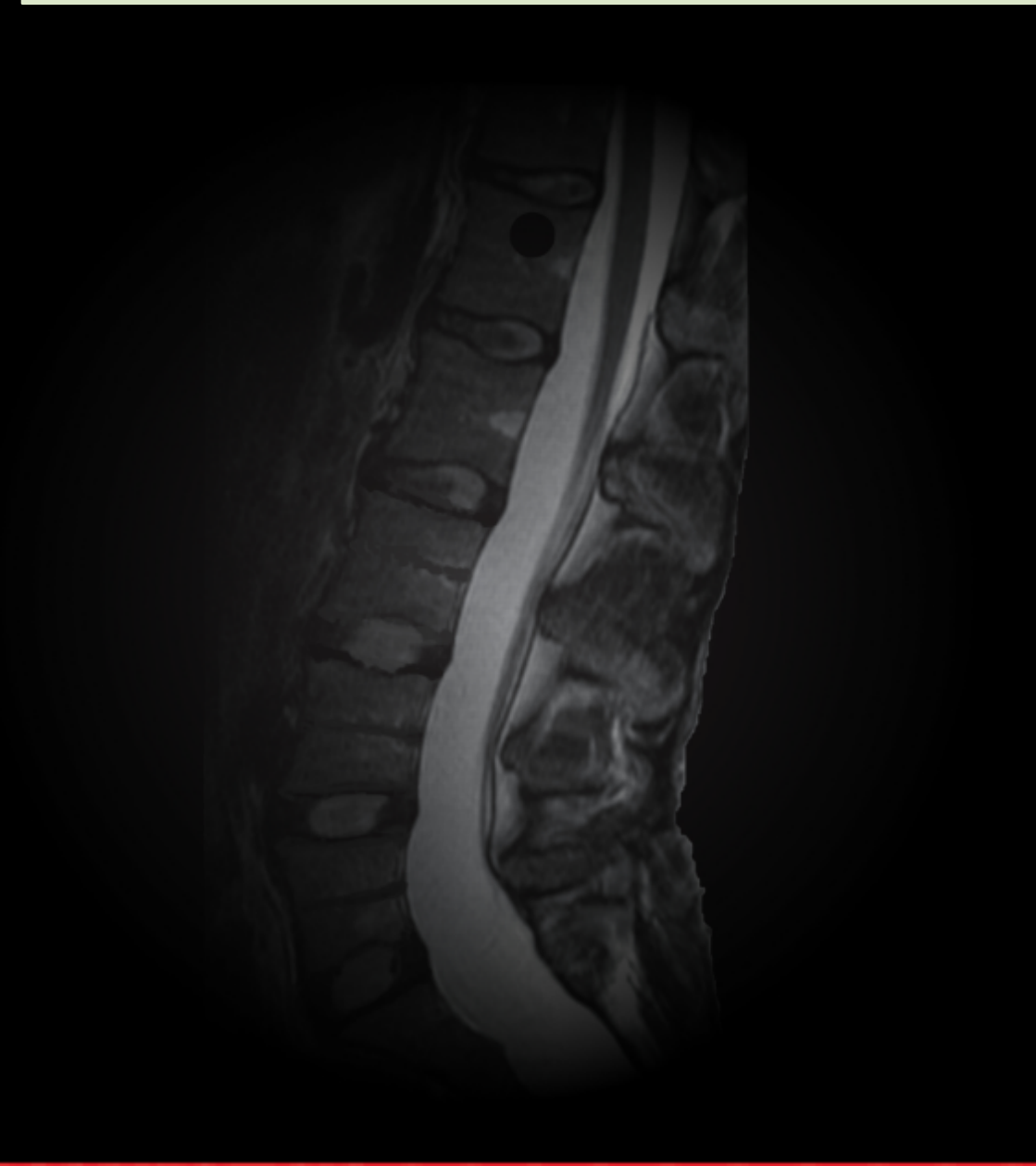
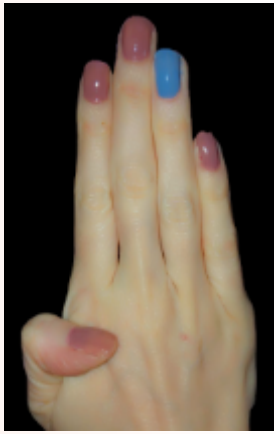
Guide to Dural Ectasia



Dr Anne Child (MD, FRCP) Joanne Jessup (RGN)
Reviewed by Dr Fariborz Neirami, Consultant in Pain Medicine and Spinal Intervention

What is Marfan Syndrome?



Marfan syndrome (MFS) is a genetic disorder of the body's connective tissue that affects any gender, race or ethnic group. Connective tissue helps provide structure to the body, binding skin to muscle and muscle to bone. It provides the stretchy strength of tendons and ligaments around joints and in blood vessel walls. It also supports the internal organs. The tissue Is made of fine fibres and 'glue'. One fibre is called fibrillin.

In MFS, a change in the fibrillin-producing gene, FBN1, means that this protein is deficient in connective tissue throughout the body, creating an unusual stretchiness and weakness of tissues. This has far-reaching implications and can affect the eyes, lungs, gut, nervous system, skeleton and, most dangerously, the cardiovascular system. Symptoms can vary widely from person to person with people experiencing mild to severe manifestations.

MFS affects roughly 1 in 3,000 people which means that approximately 18,000 people in the UK have MFS. We estimate that half remain undiagnosed. 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.

Lucy has Marfan syndrome

Dural Ectasia

Dural ectasia (DE) is defined as enlargement of the neural canal anywhere along the spinal column. The dura (envelope) surrounding the spinal cord enlarges, especially in the lower lumbosacral region where cerebrospinal fluid pressure is greatest.

Although this manifestation of Marfan syndrome is not as life threatening as the vascular problems that people may experience, it can still cause significant problems with quality of life AND as the Marfan population ages. Due to greatly improved survival into old age, these issues will become more prevalent (Pollock et al, 2021). The systemic features of MFS (such as DE) can be a clue to diagnosis and should trigger further investigations for a connective tissue disorder. DE is included in the Ghent nosology (2010) and adds two points to the Systemic Features score if it is present.

DE is seen in other connective tissue disorders e.g. Ehlers Danlos syndrome (EDS) or Loeys Dietz syndrome (EDS). A recent literature review suggests a prevalence of between 63-97% in adults with Marfan syndrome (Pollock et al, 2021).

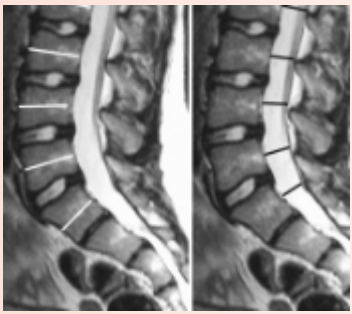
Symptoms

In many individuals DE may be present but without symptoms(asymptomatic). If symptoms are present, the most common of these are:

- Low back pain
- Headache
- Radicular pain in the legs or buttocks (nerve pain radiating from the back)
- Loss of sensation above and below the affected area (occasional)
- Rectal pain/genital pain

Symptoms can be aggravated by lying face downward and are relieved by lying on the back. Pain can also be postural (worse when standing up) as the cerebrospinal fluid will pool in the dilated area.

Diagnosis



Magnetic resonance imaging (MRI) is usually used to diagnose DE as it is better at imaging the dural sac (Pollock et al, 2021) than other imaging techniques like computed tomography (CT) although these can be used if MRI is contraindicated.

Current research suggests that DE can develop later in life. The study by Boker et al (2019) which followed patients for 10 years found that the severity of DE can increase with age; the same was found in the cohort with LDS. This finding was supported by Sheikhazedah et al (2014). This has implications for the growing population of older people with Marfan syndrome in whom a diagnosis of DE should continue to be considered as they age, even if it wasn't previously detected.

MRI findings include anterior meningoceles, nerve root sleeve dilatation, a wider dural sac at the level of S1 compared to L4, and increased dural sac ratio (DSR), which assesses the relationship between the dural sac diameter (DSD) and the vertebral body diameter (VBD) at the same vertebral level (Lundy et al, 2009).

Assessment is based on measurements of vertebral body diameter compared to dural sac diameter at the levels of the third lumbar and first sacral vertebrate. In children, measurements of greater than one standard deviation above the mean of the healthy control group are considered abnormal.

Management

Most of the treatment for DE is conservative and involves pain medication and physiotherapy. Those affected may require a referral to a neurologist or a pain specialist. Physiotherapy needs to be part of the care planned and the therapist needs to be aware of the DE and the connective tissue disorder.

Severe postural headaches may be due to a cerebrospinal fluid leak; a tear can occur in the dura due to the weakened tissue. In these cases, an epidural blood patch may be considered (injection of a small volume of the patient's own blood into the epidural space with the aim of creating a blood clot to seal the tear).

If symptoms are related to a meningocele or cyst, surgical decompression or excision of the cyst is necessary.

Spinal Anaesthesia

This needs to be carefully considered for individuals with Marfan syndrome. There are case reports in the literature that point to problems with the effectiveness of spinal anaesthesia in Marfan syndrome, which are thought to be due to DE and the resulting increased volume of lumbar cerebrospinal fluid.

Anaesthetic review prior to surgery is important for cases in which spinal anaesthesia may be used.

Pregnancy

Pregnancy needs to be carefully planned and monitored in Marfan syndrome to keep an eye on the aorta and ensure that there are no significant changes. Adequate delivery planning is vital and regional anaesthesia is an important strategy to try and reduce the extreme fluctuations in blood pressure and heart rate linked to pain.

As part of preconception counselling for pregnancy in Marfan syndrome, imaging of the spine is recommended to check for DE. DE can be present without symptoms. Predelivery consultation with the anaesthetist is important to plan for the delivery if DE is present (Whelan et al, 2023)

Dural Ectasia in Children

A study by Knirsch et al (2006) found that around 40% of the children with Marfan syndrome in their study population had signs of DE. It is usually asymptomatic in children.

Veldhoen et al (2014) found that in their study population the prevalence of dural ectasia in children with MFS amounted to 90%.

DE has been downgraded in the Ghent nosology from a major diagnostic criterion to part of the Systemic Score with a score of 2 for the presence of DE. The presence of DE can be a useful diagnostic tool in the absence of some of the other major criteria (i.e. aortic root dilatation, ectopia lentis, family history).

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How you can help

The Marfan Trust relies solely on the goodwill of its unstoppable supporters who tirelessly raise funds and awareness, allowing the charity to continue its good work and lift the shadow from this condition.

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month:

www.marfantrust.org/pages/10-membership

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- BANK: Charities Aid Foundation (CAF) ACCOUNT NAME: Marfan Trust
- SORT CODE: 40-52-40
- ACCOUNT NUMBER: 00017677
- REFERENCE: Your Name (plus campaign name if relevant)

By donating to the Marfan Trust, you are contributing to an ever-growing body of knowledge on the condition, allowing more doctors and medical specialists to deliver the best possible treatment to patients affected by Marfan syndrome.

You can also help to fund a piece of equipment in Dr José Aragon-Martin's Sonalee Laboratory. Email info@marfantrust.org to find out more.

Raising Awareness

Critical to a healthy life is early diagnosis. Once MFS is diagnosed, a treatment plan is put in place and every aspect of the condition will be managed. But, as a little-known, rare condition, Marfan syndrome is often missed. You can change this. Opportunities exist everywhere in everyday life to recognise the syndrome, from opticians and GP surgeries to rheumatologists and dentists.

Spread the word by distributing our leaflets. Follow us on social media and share our posts.

You can also hold a fundraising event and here's a link to our web page, a trove of helpful ideas. www.marfantrust.org/pages/3-fundraising-ideas

All this will help to lift the shadow of Marfan syndrome from the estimated 18,000 affected people in the UK.

Thank you!

Supported by The Hospital Saturday Fund