

## USEFUL REFERENCES

- 1 **De Paepe A**, et al *Revised diagnostic criteria for the Marfan syndrome. Am J Med Genet.* 1996 Apr 24;62(4):417-26.
- 2 **Villeirs GM**, et al *Widening of the spinal canal and dural ectasia in Marfan's syndrome: assessment by CT. Neuroradiology.* 1999 Nov;41(11):850-4.
- 3 **Knirsch W**, et al *Dural ectasia in children with Marfan syndrome: a prospective, multicentre, patient-control study. Am J Med Genet* 140(7):775-781.
- 4 **Foran JR**, et al *Characterization of the symptoms associated with dural ectasia in the Marfan patient. Am J Med Genet.* 2005 Apr 1;134A(1):58-65.
- 5 **Laccasie HJ**, et al (2005). *Dural ectasia: a likely cause of inadequate spinal anaesthesia in two parturients with Marfan's syndrome. Br J Anaesth*;94:500-4.
- 6 **Ahn NU**, et al *Dural ectasia in the Marfan syndrome: MR and CT findings and criteria. Genet Med.* 2000 May-Jun;2 (3):173-9.
- 7 **Smith HP**, et al *Anterior sacral meningocele: two case reports and discussion of surgical approach. Neurosurgery.* 1980 Jul;7(1):61-7.
- 8 **Chung SJ**, et al (2000). *Syndrome of cerebral spinal fluid hypovolemia: clinical and imaging features and outcome. Neurology.* Nov 14;55(9):1321-7.
- 9 **Cheuret E**, et al. *Intracranial hypotension in a girl with Marfan syndrome: case report and review of the literature. Childs. Nerv Syst.* 2008 Apr;24(4):509-13. Review.
- 10 **Comeglio P**, et al *The importance of mutation detection in Marfan syndrome and Marfan-related disorders: Report of 193 FBN1 mutations. Hum Mutat.* 2007 Sep;28(9):928.

## Contacts

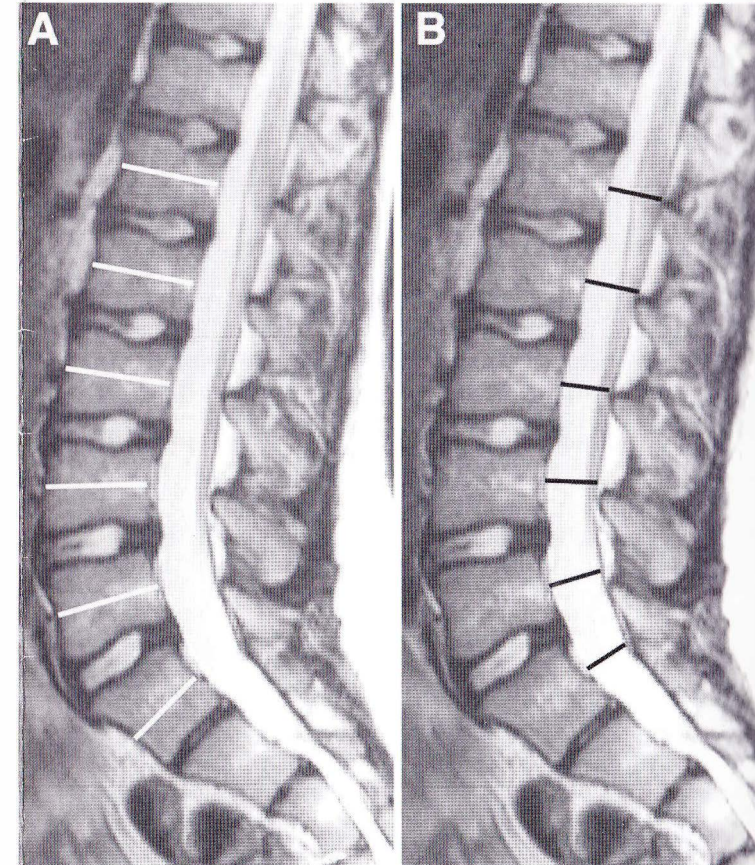
**Dr. Anne Child MD FRCP**  
(Medical Director, Marfan Trust)  
Guy Scadding Building  
Dovehouse Street  
London, SW3 6LY  
Tel: 020 7594 1605  
email: info@marfantrust.org

**Marfan Trust**  
Guy Scadding Building  
Dovehouse Street  
London, SW3 6LY  
Tel: 020 7594 1605  
email: info@marfantrust.org

[www.marfantrust.org](http://www.marfantrust.org)

# Dural Ectasia in Marfan syndrome

Written by **Dr Anne H Child MD, FRCP**  
**Dr Nick Bunce MBBS, MD, MRCP**



Magnetic resonance images of the lumbosacral spine show measurement lines of A, vertebral body diameters, and B, dural sac diameters.

**Marfan Trust**  
Supporting Research into Marfan Syndrome

With support from the Marfan Trust  
and the Bluff Field Charitable Trust

## WHAT IS MARFAN SYNDROME?

An inherited disorder of connective tissue that affects many organ systems including the skeleton, eyes, heart and blood vessels.

- Caused by a mutation in the gene for fibrillin-1 on chromosome 15.
- Can affect both males and females of any race or ethnic group.
- Around 18,000 people in the United Kingdom have Marfan syndrome (1 in 3,300 of the population).

## SALIENT FEATURES

### Skeletal

Tall, thin physique, with long limbs and fingers, scoliosis, narrow chest with breastbone (pectus) deformity, joint hypermobility and dislocations. Dilation of the lumbar dural sac occurs in about 65% of patients.

### Cardiovascular

Dilatation of ascending (and sometimes descending) aorta, incompetence of aortic and mitral valves, aneurysm and rupture of aorta.

### Respiratory

Pneumothorax, bronchiectasis, emphysema and asthma.

### Ocular

Dislocation of lens, myopia and unstable refraction, detachment of retina, squint (strabismus), glaucoma.

### Dental

High arched palate, crowding of teeth.

### Genetic

Males and females are affected equally frequently. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

In 25% of cases neither parent is affected; however, apparently unaffected parents should be screened carefully as the severity and pattern of disease are variable, even within one family.

## DIAGNOSIS

Diagnosis is made after careful physical examination and echocardiography, demonstrating classical features in two out of three major systems (eyes, heart, skeleton), preferably with a family history. Mutations can be found in the fibrillin-1 gene in 92% of patients, assisting with screening of family members.

## DURAL ECTASIA

Dural ectasia is defined as enlargement of neural canal anywhere along the spinal column. The dura (envelope) surrounding the spinal cord enlarges, especially in the lower lumbosacral regions where cerebrospinal fluid pressure is greatest. Diagnostic criteria for Marfan syndrome are set out in the Ghent Nosology<sup>(1)</sup> requiring major involvement in two out of three systems (eyes, heart, skeleton) with minor involvement in the third system. Dural ectasia has been added as a major diagnostic feature<sup>(2)</sup>.

### Incidence

Dural ectasia is a condition which appears over time. Even in childhood, dural ectasia can be detected<sup>(3)</sup> with a frequency of 40%. Adult Marfan syndrome patients with back pain show a higher incidence of dural ectasia than patients without symptoms<sup>(4)</sup>. On average dural ectasia is found in 65% of adult patients. It is also found in other connective tissue disorders such as Ehlers–Danlos syndrome.

## Symptoms

The most common associated symptoms are low back pain, headache, weakness, loss of sensation above and below the affected area, occasional rectal pain, genital pain and abdominal pain. The symptoms are aggravated by lying face downward and are relieved by lying on the back. Dural ectasia can cause inadequate spinal anaesthesia during delivery<sup>(5)</sup>.

## Measurement

CT scan can be used especially if the patient has a pacemaker, but images are not as accurate as in magnetic resonance imaging (MRI)<sup>(6)</sup>. Assessment is based on measurements of vertebral body diameter compared to dural sac diameter at the levels of the third lumbar and first sacral vertebrae<sup>(6)</sup>. In children, measurements of greater than one standard deviation above the mean of the healthy control group are considered abnormal.

## Treatment

If symptoms are related to a meningocele or cyst, surgical decompression or excision of the cyst is necessary<sup>(7)</sup>. Postural headache may be due to cerebrospinal fluid leak<sup>(8)</sup>. Headache increases when standing and decreases when lying down. Epidural blood patching may be necessary<sup>(9)</sup> or surgical repair considered<sup>(7)</sup>.

## Recommendation

In a patient without dural ectasia symptoms, if the diagnosis of Marfan syndrome requires confirmation, search for a mutation in the fibrillin-1 gene (92 % success rate) should be made using dHPLC in the molecular genetic laboratory. This provides a specific diagnosis, and also a rapid screening tool for close family members<sup>(10)</sup>. The cost of this test is equivalent to that of an MRI scan.

If the patient has the above symptoms, an MRI scan for dural ectasia will aid diagnosis, especially in adulthood, as the test has high sensitivity and specificity in Marfan syndrome.