# Dural Ectasia as Presenting Symptom of Marfan Syndrome

Arie Altman MD PhD[1,2], Livnat Uliel MD MSc[3] and Liron Caspi[1,2]

[1]Center for Autoimmune Diseases and Departments of [2]Medicine B and [3]Radiology, Sheba Medical Center, Tel Hashomer, and Sackler Faculty of Medicine, Tel Aviv University, Ramat Aviv, Israel

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Marfan syndrome is a genetic, dominant, systemic connective tissue disease with variable clinical features, some of which are life threatening. The prevalence is 1:10,000 live births [1]. The syndrome results from mutations in the FBN1 gene, on chromosome 15, which encodes for the fibrillin 1 protein. Fibrillin is an important compound in the extracellular matrix and the main structural protein in microfibers of elastin [2]. There is a second gene of fibrillin, the FNB2 gene, which is responsible for the congenital contractural arachnodactyly, known as Beals syndrome [3].

In 2005 Loeys and Dietz described an autosomal dominant disease similar to Marfan syndrome (aortic aneurysm, dural ectasia, drum stick fingers), which is the result of mutations in the transforming growth factor-beta receptor (TGFBR)-1 and TGFBR2. However, there are several features specific to this disorder, such as hypertelorism, craniosynostosis, cleft palate, double or huge uvula, and large convoluted arteries [4].

The structural defect in the fibrillin 1 protein is the cause of Marfan syndrome features like the cardiovascular system pathology, and skeletal, ocular and central nervous system manifestations [5]. Since 1996 the diagnosis of Marfan syndrome has been based mainly on symptoms and according to the Ghent nosology [6]. Dural ectasia is considered one of the major criteria in the

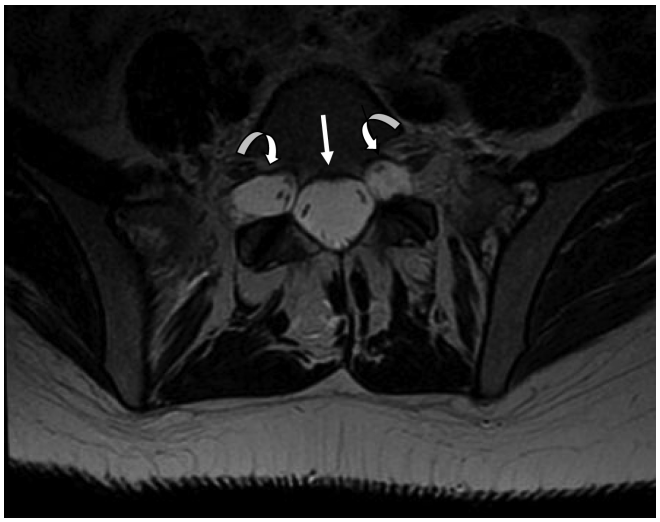

**Figure 1.** Axial T2-weighted image at the level of the upper sacrum shows dilatation of the spinal canal (straight arrow) and neural sleeves (curved arrows).

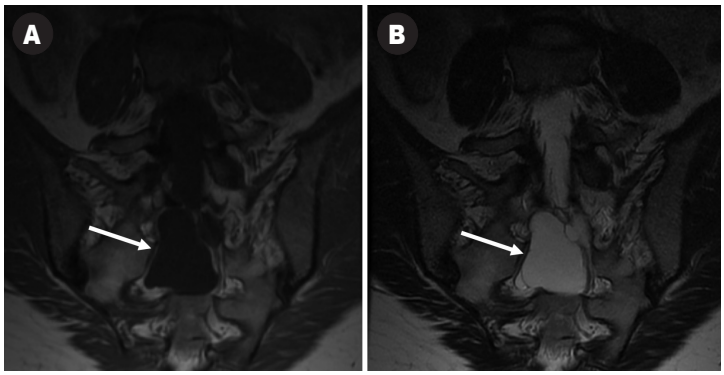


**Figure 2.** Coronal T1-weighted **[A]** and coronal T2-weighted **[B]** MR images at the same lumbosacral level show ballooning of dural sac (arrows) in the sacral region.

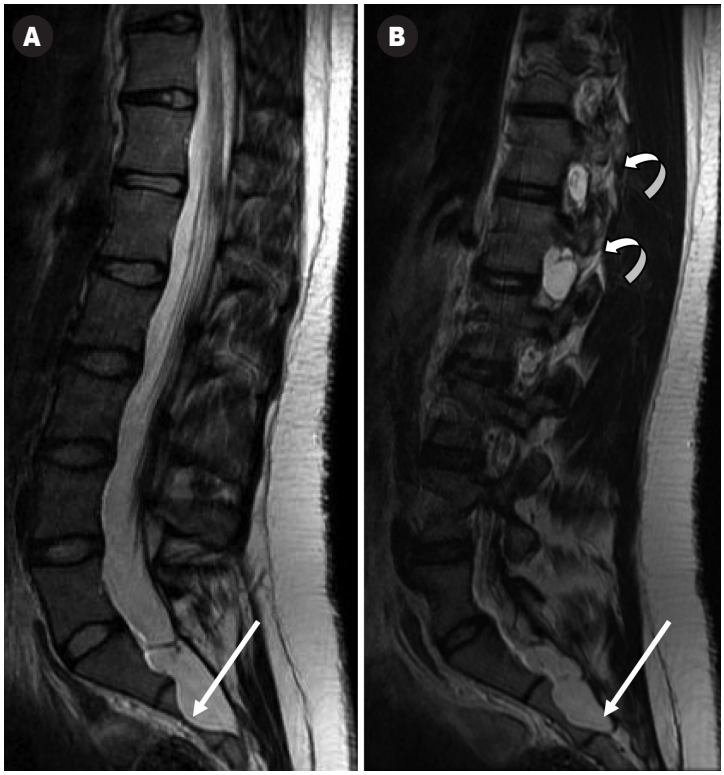


**Figure 3.** Mid-sagital T2-weighted **[A]** and parasagital T2-weighted **[B]** MR images of the lumbosacral spine show scalloping of S2 (straight arrows) and dilatation of the neural sleeves (curved arrows, B).

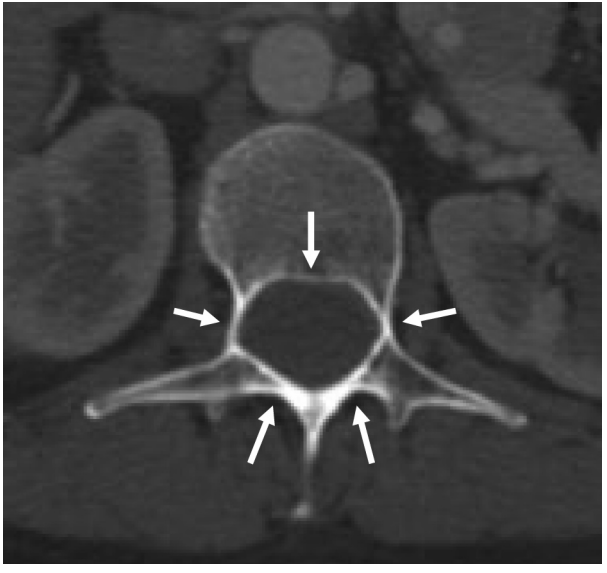

**Figure 4.** Axial computed tomography scan of the bone window shows dural ectasia of lumbar vertebra. The dilated canal is marked by arrows.

diagnosis of Marfan syndrome and is prevalent in more than two-thirds of affected adults (63–92%); the prevalence in children is unknown.

Dural ectasia is an enlargement of the dural sac and the spinal canal and sometimes with enlarged nerve sleeves [Figures 1-4]. Dural ectasia can affect the spinal canal in any plane, but the most common sites are the lumbosacral region [Figures 2,4]. Therefore, the most common clinical symptoms are low back pain, headache, weakness, and loss of sensation above and below the affected limb, occasional rectal pain and pain in the genital area. The symptoms are aggravated mainly in the supine position and are relieved by lying on the back [7].

The radiological definition of dural ectasia has not been standardized. The most accurate method is magnetic resonance imaging [8]. The antero-posterior plane of the vertebra and the diameter of the dural sac in mid-sagital position at the level of S1 and L3 are measured [Figure 3]. Those parameters are used to calculate the ratio of the dural sac; the common dural sac ratio in Marfan syndrome is 0.75 at the S1 level and higher than 0.47 at the L3 level [9]. In Marfan patients with severe low back pain and radicular manifestations, dural ectasia should be suspected as the etiology.

## References


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**Correspondence:** Dr. A. Altman, Dept. of Medicine B, Sheba Medical Center, Tel Hashomer 52621, Israel.
Phone: (972-3) 530-2637
Fax: (972-3) 535-2637
email: arie.altman@sheba.health.gov.il