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
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-sagittal 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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**Capsule**

**Fertile ground for cancer proteins**

Leukemia inhibitory factor (LIF) is a secreted glycoprotein first identified, as its name implies, as a regulator of leukemic cell differentiation. More recently, attention has focused on the role of this cytokine in the female reproductive tract. In mice, LIF is one of the few molecules known to be required for implantation of the blastocyst, or earl-stage embryo, into the uterus. Thus, it has been hypothesized that drugs targeting LIF activity could (depending on their mode of action) be used either to enhance fertility by promoting implantation or as contraceptives that prevent implantation. Progress on the latter front is reported by White and colleagues (Proc Natl Acad Sci USA 2007;104:19357) who developed a potent LIF antagonist that is completely effective in blocking blastocyst implantation when administered systemically to mice. Whether this antagonist – a chemically stabilized mutant version of LIF that binds to its receptor but does not trigger downstream signals – has similar activity in primates remains to be explored. In independent work, Hu et al. (Nature 2007;450:721) found that LIF expression in the mouse uterus is positively regulated at the transcriptional level by p53, an intensely studied tumor suppressor protein. Discovery of this link between LIF and p53 raises the possibility that cancer drugs designed to activate p53 might be useful tools for investigating the mechanisms underlying blastocyst implantation or as an alternative means of enhancing fertility.

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