THE MARFAN FOUNDATION

Know the Signs. Fight for Victory.

LOEYS-DIETZ SYNDROME FOUNDATION

A Division of the Marfan Foundation

Dural Ectasia

in People with Marfan Syndrome and Loeys-Dietz Syndrome

Background

Dural ectasia (DE) refers to the widening or ballooning of the dura, the normally-tough membrane that surrounds the spinal cord. Dural Ectasis is frequently associated with connective tissue disorders like Marfan syndrome and Loeys-Dietz syndrome. In these conditions, the connective tissue of the dura is weak, leading to dilatation, often in the lumbar and sacral spine region. This can cause a range of symptoms that may vary in severity and often overlap with other issues common in connective tissue conditions.

Symptoms of Dural Ectasia

1. **Lower Back Pain and Discomfort:** Chronic low back pain is common and may worsen with prolonged sitting, standing, or activity. Pain can radiate to the hips or legs or perineum. Yet some patients with DE have no pain.
2. **Leg Pain or Weakness:** Due to nerve compression or stretching from the dural enlargement, patients may experience radicular pain, tingling, or numbness in the legs. Severe cases can lead to weakness and impaired mobility.
3. **Headaches (Often Positional):** Dural ectasia may cause positional headaches, similar to those experienced with CSF leaks. These headaches may worsen upon standing and alleviate when lying down.
4. **Bowel or Bladder Dysfunction:** In cases where the spinal nerves are compressed, patients may experience difficulty controlling bladder or bowel function. This may range from urinary urgency to incontinence.
5. **Scoliosis or Kyphosis:** Although not caused by DE, like scoliosis or kyphosis, are more common in people with Marfan and Loeys-Dietz and can worsen the symptoms of dural ectasia by altering spine dynamics.
6. **Pelvic Masses** in the sacral dura may expand into the pelvis, and may be mistaken for an ovarian mass.

Diagnosis

Identifying dural ectasia in people with Marfan or Loeys-Dietz syndrome involves a combination of clinical evaluation and imaging studies:

- **MRI of the Spine:** MRI is the preferred method to evaluate the extent of dural enlargement and detect any associated nerve compression.
- **CT +/-Myelography:** This test may be used to further assess the spine and dura, especially if MRI results are inconclusive. Myelography should be performed with reluctance, as it may lead to CSF leak

Management and Treatment

1. **Conservative Management:**
    - **Pain Management:** Pain is typically managed with medications such as acetaminophen gabapentin. Non-steroidal anti-inflammatory drugs (NSAIDs) should be used cautiously in patients with connective tissue disorders.
    - **Physical Therapy:** Targeted exercises to strengthen core muscles can reduce strain on the lower spine and may alleviate pain symptoms. Additionally, lumbar braces may help.
    - **Lifestyle Modifications:** People with Marfan syndrome and Loeys-Dietz syndrome are advised to avoid heavy lifting, prolonged standing, or activities that may strain the spine.
2. **Surgical Intervention:**
    - Surgery is considered in severe cases where there is progressive nerve damage, loss of bladder or bowel function, or when pain is unmanageable. Techniques like laminectomy or dural reduction can reduce the dura's size or decompress the nerves.
    - **Spinal Fusion:** In people with severe spine deformities, fusion may help stabilize the spine and improve symptoms.
3. **Monitoring and Regular Imaging:** Regular follow-ups with imaging may be required to monitor the progression of dural ectasia, especially in people with worsening symptoms or new neurological findings.
4. **Multidisciplinary Management:** Given the complexity of connective tissue conditions, care typically involves a team approach, including a geneticist, neurosurgeon physiatrist, physical therapist, pain management specialist, and orthopedic surgeon.

Other Considerations for People with Marfan and Loeys-Dietz Syndrome

Management of dural requires a nuanced approach due to the fragile connective tissue and the potential for associated cardiovascular issues, especially in Loeys-Dietz syndrome. Close monitoring, symptom management, and minimizing surgical interventions when possible are critical for maintaining quality of life and managing this condition's long-term effects.

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