# Vertebral scalloping in neurofibromatosis-1

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## ABSTRACT

A 39-year-old male patient known to have neurofibromatosis-1, presented with sciatica and low back pain. At the age of 27, an MRI of the spine revealed widening of the lumbar canal due to posterior scalloping of the last 4 lumbar vertebrae. Repeat MRI and simple lateral lumbosacral plain film at the age of 39, revealed significant progression of the lumbosacral lesion. In view of the progression of the disease during the last 15 years, we opted for insertion of a lumbo-peritoneal (LP) shunt to decrease the CSF pressure, which is the most probable cause of scalloping. We shied away from lumbar fixation as we thought that this would not stop the vertebral erosion. The LP shunt affected good symptomatic and radiologic outcome over a 2-year follow up.

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Neurofibromatosis-1 (NF1) is an inherited disorder characterized by formation of neurofibromas in the skin, subcutaneous tissue, cranial nerves, and spinal root nerves.[1-4] We present a case of progressive symptomatic vertebral scalloping successfully treated with lumbo-peritoneal (LP) shunting and followed for 2 years.

**Case Report.** A 39-year-old Lebanese male known to have NF1 presented with sciatica and low back pain. The pain started many years earlier but exacerbated in the last few months. The pain was localized at the lumbar region with irradiation to the anterolateral sides of both thighs. He also complained of paresthesias in the upper and lower limbs and mild motor weakness of the hands. He had numerous congenital café-au-lait spots, many of them more than 2 cm in size and many freckles in the axillary region. At the age of 24 years, the patient noticed a scoliosis; a lumbosacral x-ray revealed scoliosis of the lumbar spine and widening of the spinal canal with posterior scalloping of the lumbar vertebrae, compatible with NF. At the age of 27, an MRI of the spine was carried out and revealed widening of the lumbar canal due to posterior scalloping of the last 4 lumbar vertebrae and the first sacral without evidence of any spinal or paraspinal tumoral syndrome (**Figure 1**). Repeat MRI and simple lateral lumbosacral plain film carried out at the age of 39 (**Figure 2**), revealed significative progression of the lumbosacral lesion, however, the MRI of the cervical and dorsal spine showed no abnormalities. Plain x-ray also revealed the persistence of oily contrast medium within the spinal canal at the level of T11-12 and the sacral region from a previous myelography (**Figure 3**).

**Discussion.** Neurofibromatosis-1 is an autosomal dominant inherited genetic disorder.[3,5-7] It is associated with deletions, insertions, or mutations in the NF1 gene. This gene is a tumor suppressor gene located in the pericentromeric region of chromosome 17.[3,8] The disease affects 1 in every 2500 to 3000 live births. Half of the patients with this disease have affected family members, the remaining patients represent new mutations.[9] Neurofibromas are the hallmark of the disease and usually appear during childhood or adolescence after café au lait spots. Skeletal abnormalities such as scalloping of the vertebrae, congenital bowing of long bones with pseudoarthrosis, unilateral orbital malformations and cystic osteolytic lesions occur in almost 40% of patients with this disease.[3,6] Criteria for the diagnosis of NF1 are met in an individual if 2 or more of the following signs are found:[1,2] Six or more café-au-lait spots or hyperpigmented macules greater than or equal to 5 mm in diameter in children younger than 10 years and 15 mm in adults; multiple freckles (Crowe sign) in the axillary or

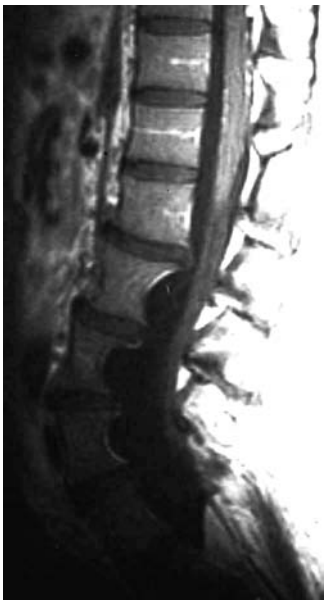

**Figure 1 -** An MRI of the spine reveal posterior scalloping of the last 4 lumbar vertebral.

**Figure 2 -** An MRI revealed significant progression of the lumbo-sacral lesion.

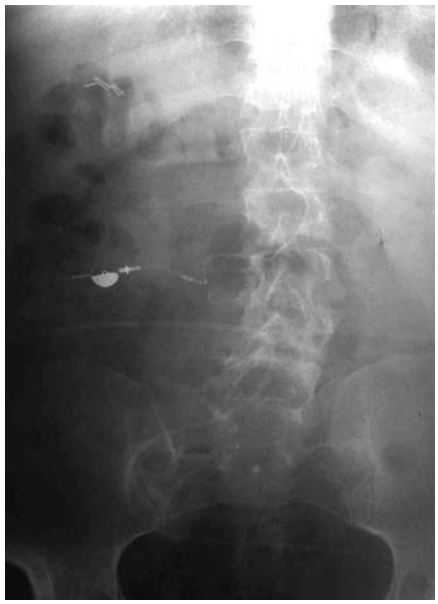

**Figure 3 -** Plain x-ray showing the lumbo-peritoneal shunt.

inguinal region; 2 or more typical neurofibromas or one plexiform neurofibroma; a distinctive osseous lesion, such as sphenoid dysplasia or thinning of long bone cortex, with or without pseudoarthrosis; optic nerve glioma; 2 or more iris hamartomas (Lisch nodules) seen on slitlamp or biomicroscopy examination; and a first-degree relative (parent, sibling, offspring) with NF1, as diagnosed by using the criteria above. Dural ectasia with enlargement of the spinal canal is also a relatively common finding in neurofibromatosis. On plain films, dural ectasia can be suspected on the basis of vertebral scalloping or concavity of vertebral bodies. The cause of dural ectasia is possibly a congenital weakness of the dura, in which case the constant pulsation of cerebrospinal fluid causes progressive enlargement of the dural sac with resultant scalloping of the posterior portions of the vertebral bodies and erosion of the pedicles. Another explanation for scalloping has been thought to be primary bone dysplasia, which results in a softened, weakened centrum, which is slowly eroded by normal pulsations of cerebrospinal fluid. A combined effect of the 2 mechanisms has also been suggested. Schonauer et al,[10] presented in their report on 2 NF 1 patients with lumbosacral dural ectasia, that the dura in the area of the ectasia is extremely thin and fragile and predisposes surgical patients to high morbidity.[10]

Our own patient has at least 3 criteria: more than 6 café-au-lait spots greater than 20 mm, multiples freckles in the axillary region and vertebral dysplasia with scoliosis. Furthermore, his father and his son have the same café-au-lait spots. He has neither neurofibroma nor optic nerve glioma. No other problems or systemic disease were reported. In view of the progression of the disease during the last 15 years (increasing of the scalloping and enlargement of the lumbar canal), we opted for insertion of a LP shunt to decrease the CSF pressure, which is the most probable causative agent of scalloping.[10] We shied away from lumbar fixation as we thought that this would not stop the vertebral erosion. The LP pump used (**Figure 3**) fits with the patient's position better than that used in the ventriculo-peritoneal shunt. After the operation, he presented with a remarkable improvement of the symptoms, and now he can ambulate and has returned to work again.

Although most individuals with NF1 lead relatively long and healthy lives, the overall life expectancy may be reduced by as much as 15 years. The major causes for this increased morbidity and subsequent mortality are hypertension, sequelae of spinal cord lesions, and malignancy. Prompt attention to complications of NF1 and early detection of medical problems may significantly reduce the overall morbidity, and mortality rates.[2,4]

In conclusion, we report a case of vertebral scalloping and scoliosis with NF1, causing sciatica and low back pain, operated by lumboperitoneal pump to hold back the progression of the disease. On follow-up, the symptoms were greatly improved.

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