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Segmental Neurofibromatosis (NF-5): A Rare Form of Neurofibromatosis

Neurofibromatosis is a common disorder that affects approximately 1 in 3000 persons [1]. Segmental neurofibromatosis is a rare form of the disease in which the cutaneous and neural changes are confined to one region of the body. Because the natural history and genetics of the segmental type may be different from those of the more common forms, it is important to become familiar with this unusual disorder.

Case Report

A 28-year-old woman had had vague pain in the right lower back and pruritus above the right buttock for several years. The pain was nonradiating and was exacerbated by supine positioning. The results of physical examinations and radiographs of the lumbar spine performed over the preceding 2 years were normal. A routine gynecologic examination revealed fullness in the right ischioanal fossa, and pelvic MR images were obtained. The study, performed on a Philips superconducting magnet operating at 0.5 T showed multiple rounded hypointense masses in the right lumbosacral plexus on T1-weighted images (750/20/4) (TR/TE/excitations). The masses (Fig. 1A) extended along the surface of the piriformis muscle, involving the right ischioanal fossa and exiting the true pelvis via the greater sciatic foramen. The lesions were homogeneous and well-defined. They became hyperintense on T2-weighted images (2347/100/2) (Fig. 1B). A small lesion in the left lumbosacral plexus was seen also. CT showed identical morphologic findings; the masses were of low attenuation (Fig. 1C), and no bone erosion was detected. A diagnosis of plexiform neurofibroma was made, and the most superficial lesion was partially resected. Pathologic examination showed a hypocellular benign neurofibroma. No mitotic figures were seen. Additional clinical examination of the patient revealed one small (less than 1 cm) café-au-lait spot on each lower extremity. Slit-lamp examination showed no Lisch nodules, and the patient had no family history of neurofibromatosis. The results of a neurologic examination were completely normal, and no additional neurofibromas could be palpated.

Discussion

Neurofibromatosis is a complex disorder involving cells derived from the neural crest. Tremendous clinical variability is seen; eight types of the disorder were defined at a National Institutes of Health

(NIH) consensus conference [1, 2]. Classical peripheral neurofibromatosis (von Recklinghausen disease, NF-1) is characterized primarily by café-au-lait spots of the skin, Lisch nodules (pigmented hamartomas in the iris), and neurofibromas that may involve virtually any portion of the body. Less common manifestations include CNS tumors (especially optic nerve gliomas and astrocytomas), kyphoscoliosis, pseudoarthrosis, pheochromocytoma, and medullary carcinoma of the thyroid. The presence of bilateral acoustic neuromas establishes a diagnosis of a separate entity of central neurofibromatosis (NF-2), which differs from NF-1 in the distribution of cutaneous neurofibromas, prevalence of CNS tumors, and pigmentation manifestations.

Segmental neurofibromatosis (NF-5 in the NIH classification) [2] is a rare form of neurofibromatosis in which the cutaneous and neural changes are limited to one sector of the body and the patient usually has no family history of neurofibromatosis. In 1956, Crowe et al. [3] first introduced the concept of segmental neurofibromatosis. They established the diagnostic criteria as (1) unilateral neurofibromas limited to one or adjacent nerve roots, (2) café-au-lait spots either absent or limited to the involved region, and (3) a cause presumed to be due to a somatic mutation with a resultant absence or marked reduction in genetic transmission. Lisch nodules are not present. In their original description of four patients, Crowe et al. described one person who had disease limited to the back in a bilateral distribution. Since then, scattered reports of cases of segmental neurofibromatosis have appeared in the literature [4–8], some with exceptions to these criteria. In fact, segmental neurofibromatosis itself may be a heterogeneous disorder [7]. For example, in 1984, Takiguchi and Ratz [6] described a patient with cutaneous neurofibromas restricted to the lower portion of the back but in a bilateral distribution; at least one other report of bilateral segmental involvement has been published [7].

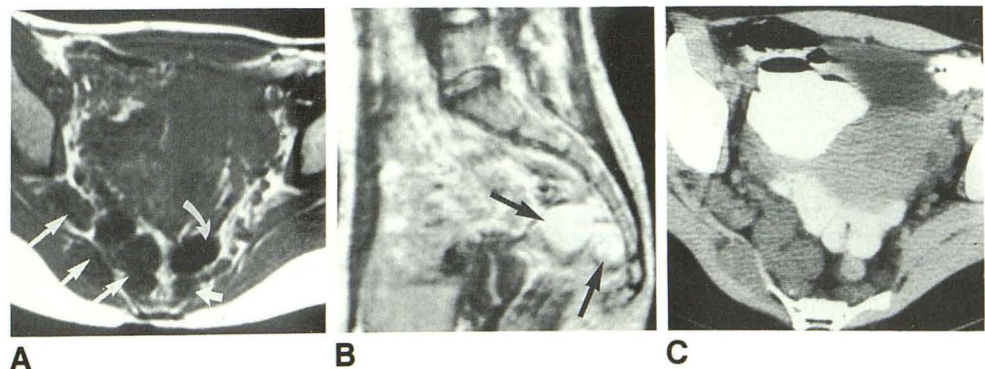
The patient described here had no family history of neurofibromatosis and no Lisch nodules. The plexiform neurofibromas involved the right lower segment almost exclusively (lumbosacral plexus and its more peripheral branches); the presence of a small lesion in the same location on the contralateral side was not completely unexpected in view of the experiences of others [3, 6, 7]. The two small café-au-lait spots may have been incidental findings, as persons without neurofibromatosis may have spots of this size and number [1]. The imaging features on MR (Figs. 1A and 1B) and CT (Fig. 1C) were typical of plexiform neurofibromas.

Fig. 1.—Segmental neurofibromatosis.

A, T1-weighted axial MR image shows multiple hypointense pelvic lesions involving branches of right lumbosacral plexus (*thin straight arrows*), a small neurofibroma on left side (*thick straight arrow*), and a loop of rectosigmoid colon (*curved arrow*).

B, On T2-weighted sagittal MR image, visualized portion of plexiform neurofibroma (*arrows*) becomes hyperintense.

C, On CT scan, masses are of low attenuation (approximately 20 H), typical of neurofibromas.



The natural history of segmental neurofibromatosis is quite variable and depends on the severity of involvement; slow growth of the tumors is the norm. Pain is the most frequent symptom; neurologic deficit is much less common. In 3–13% of patients with the NF-1 type, malignant peripheral nerve-sheath neoplasms develop, usually after latent periods of 10–20 years. However, none of the patients reviewed for this report had malignant tumors. Pruritus, which was present above the right buttock in the patient described here, has only recently been recognized as a feature of neurofibromatosis. The presence of large numbers of mast cells in neurofibromas and the ameliorative effect of antihistamines suggest that these agents may account for pruritus in neurofibromatosis [1]. As previously mentioned, segmental neurofibromatosis generally is thought to be a somatic mutation and therefore not transmitted to offspring [3]. This is of great significance in genetic counseling, because NF-1 and NF-2 are autosomal dominant traits, and offspring of affected persons are at a 50% risk of acquiring the disease. Nonetheless, a few reports [8] have indicated that patients with segmental neurofibromatosis can have offspring who have neurofibromatosis [8]. Segmental neurofibromatosis may, in some cases, occur as a heritable disease characterized by an increased susceptibility to somatic mutation at the locus for neurofibromatosis [7].

In conclusion, the rare entity of segmental neurofibromatosis can be diagnosed when the appropriate clinical and radiologic information is available. This is of great importance as the natural history, clinical

manifestations, and inheritance of this form are probably different from those of the more familiar forms of neurofibromatosis.

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