Musculoskeletal case 12. Diagnosis

Neurofibromatosis type 1

The diagnosis was neurofibromatosis type 1 with associated neurofibromas extending along the axillary and brachial neural plexuses. Because the lesions in the axilla were asymptomatic and appeared stable, the patient was managed conservatively, with clinical and radiologic follow-up to ensure the stability of the lesions. Figs. 1 and 2 (see presentation, page 336), contrast enhanced axial T_1-weighted images, demonstrate discrete enhancing masses medial to the humerus, representing neurofibromas (arrows). Figs. 3 and 4 are coronal T_2-weighted images with fat suppression. Fat suppression nullifies the high signal intensity of fat, making the lesions more obvious. In Fig. 3, the high signal chain of neurofibromas is clearly depicted, extending along the neuronal pathways. Fig. 4 clearly demonstrates one of the larger neurofibromas at the distal end of the lesion complex.

Neurofibromatosis is a multisystemic neurocutaneous disorder, involving both neuroectodermal and mesenchymal derivatives. It is the most common single-gene disorder of the nervous system. The chromosomal defects for at least 2 forms of neurofibromatosis have been delineated and mapped to chromosomes 17 (type 1) and 22 (type 2). Neurofibromatosis is associated with a variety of neoplastic and non-neoplastic manifestations that typically progress in severity during the lifetime of the affected patient.[1] The clinical course for either type is unpredictable. Type 1 (von Recklinghausen's disease or classic peripheral neurofibromatosis) is the more common of the 2 main subgroups, accounting for at least 85% of cases.[2] It is characterized by its cutaneous manifestations, café-au-lait spots, lentigines and neurofibromas. Type 2 (acoustic or central neurofibromatosis) is characterized by bilateral vestibular schwannomas (former acoustic neurinomas) and other tumours of the central nervous system.

The expression of neurofibromatosis is highly variable, even within an affected family, ranging from mild inconvenience with a normal lifespan to serious, progressive manifestations leading to death as early as the perinatal period. The diagnostic criteria for neurofibromatosis in the National Neurofibromatosis Foundation International Database include hyperpigmentation changes (café-au-lait spots and axillary freckling or diffuse duskiness), neurofibromas and melanocytic hamartomas of the iris (Lisch nodules).[3] Other features include seizures and intellectual compromise, optic and acoustic involvement, intracranial and spinal tumours,[4] endocrine disorders, hypertension, vascular anomalies, an increased incidence of malignant lesions, osseous defects and congenital dislocations. The diagnosis is usually made in childhood and may be easy if there is a known family history. Occasionally, the diagnosis can even be made in utero.[5] However, sometimes the diagnosis is difficult and the disease may only be discovered in later life.

Once the diagnosis is made, it is important that a multidisciplinary approach be adopted because of the multisystem involvement of the disease process. Genetic counselling plays an important role in the overall management strategy as the disease is usually inherited as an autosomal dominant trait. Magnetic resonance imaging has rapidly become the investigation of choice in the assessment of both central and peripheral neural manifestations of neurofibromatosis. It has largely replaced computed tomography in this regard because of its superior soft-tissue contrast resolution without the use of ionizing radiation.[6] It may aid not only in the detection and follow-up of neurogenic lesions but is also invaluable in the preoperative evaluation.

References

1. Pollack IF, Mulvihill JJ. Neurofibromatosis 1 and 2 [review]. *Brain Pathol* 1997;7:823-36.
2. Gutmann DH, Aylsworth A, Carey JC, Korf B, Marks J, Pyeritz RE, et al. The diagnostic evaluation and multidisciplinary management of neurofibromatosis 1 and neurofibromatosis 2 [review]. *JAMA* 1997;278(1):51-7.
3. DeBella K, Szudek J, Friedman JM. Use of the National Institutes of Health criteria for diagnosis of neurofibromatosis 1 in children. *Pediatrics* 2000;105(3 Pt 1):608-14.
4. Mukonoweshuro W, Griffiths PD, Blaser S. Neurofibromatosis type 1: the role of neuroradiology [review]. *Neuropediatrics* 1999;30:111-9.
5. Lazaro C, Ravella A, Casals T, Volpini V, Estivill X. Prenatal diagnosis of sporadic neurofibromatosis 1. *Lancet* 1992;339:119-20.
6. Truhan AP, Filipek PA. Magnetic resonance imaging. Its role in the neuroradiologic evaluation of neurofibromatosis, tuberous sclerosis, and Sturge–Weber syndrome [review]. *Arch Dermatol* 1993;129:219-26.