Referring Doctor Newsletter: Neurofibroma

Clinical presentation

Neurofibromas are flesh-colored lesions that can occur anywhere in the body. They can vary in their prominence in the skin. Lesions can present as masses deep in the subcutaneous plane to projecting as exophytic pedunculated papules. Their sizes also can vary greatly from small lesions 2-3 mm in diameter to larger growths several cm in diameter.

Neurofibromas can present as solitary lesions or as multiple lesions. Multiple neurofibromas can be associated with generalized syndrome of neurofibromatosis (usually neurofibromatosis type 1 [NF-1], also called von Recklinghausen disease of the skin). Patients with NF-1 also present with classic brown colored macules called *café-au-lait* spots. Some lesions often can be invaginated when compressed with a fingertip (known as the *buttonhole sign*). Cutaneous neurofibromas usually remain asymptomatic, but neurofibromas that develop in major peripheral or central nerves can cause motor or sensory dysfunction.

Aetiology

The pathogenesis of both neurofibromatosis and non-neurofibromatosis neurofibromas is not clearly determined.

Histology

Neurofibromas are composed of Schwann cells, fibroblasts, perineurial cells, masts cells, and axons embedded in extracellular matrix. The lesions present as discrete but unencapsulated tumors. A neurofibroma is composed of interlacing fascicles of Schwann cells that have irregular, wavy, elongated nuclei. The nerve's perineurium is disrupted, which suggests a breakdown of appropriate signaling between Schwann cells, fibroblasts, and perineurial cells. The part played by each cell type in neurofibroma formation is unclear. On gross inspection, the neurofibroma tumor usually has small fibers coursing through its substance.

Treatment

Cutaneous neurofibromas can be excised if they cause deformation of surrounding tissues or have become symptomatic. Also, any lesions suspicious for malignant transformation should also be surgically excised.

Individuals with NF-1 harbor a 7–13% lifetime risk of developing a malignant peripheral nerve sheath tumor, which usually arises in preexisting plexiform or focal subcutaneous neurofibromas. Using the normal clinical signs to evaluate for a potential malignant or transforming premalignant lesion is challenging. In patients with NF-1, new lesions develop and grow continually, so normal surveillance recommendations may not detect a potentially malignant lesion.

Neurofibroma can occur in major peripheral nerves, which can cause disruption of nerve function.[7] Neurofibromas commonly occur within the substance of the nerve which creates a severe challenge to remove the tumor without causing further nerve dysfunction. Internal neurolysis and nerve grafting are performed as indicated for neurofibromas resection.

For more information on this or any other skin cancer enquiries,
please contact Dr Ian Katz, mobile 0416 165 459.

Southern Sun Pathology Suite 6, Level 1, 32 Florence St, Hornsby, NSW 2077

Phone 9482 1050 www.southernsun.com.au