GENETICS AND GENODERMATOSES

NEUROFIBROMATOSIS TYPE 1 AND ELEPHANTIASIS NEUROMATOSA: A RARE ENTITY

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Background: A 24-year-old female with Neurofibromatosis type 1 (NF1) presented to the lymphoedema clinic presenting with left leg lymphoedema in the setting of having had a large plexiform neurofibroma which was previously debulked. She had other features of NF1 including multiple cutaneous neurofibromata, cafe-au-lait macules and axillary freckling. She has had two epiphyseal fusions to stop the growth of the left leg. Her left leg remained 1cm longer than the right leg.
There was an extensive plexiform neurofibroma of the left leg with excessive skin and swelling due to lymphoedema.
Venous duplex scans showed moderate saphofemoral junction reflux in to the lower saphenous vein in the thigh. The deep veins were normal. Plain x-rays showed lamellated periosteal thickening and evidence of bony remodelling.
Histology from the tissue taken from the debulking procedure suggested an extensive neurofibroma with features of both diffuse and plexiform neurofibroma. The diffuse component filled the dermis and extended in fibrous bands into the deep dermis extending around vessels. The plexiform component was present at all levels of the dermis and demonstrated hypertrophic nerve trunks with variable myxoid change.

Observation: First reported in 1947, Elephantiasis Neuromatosa (EN) is a rare complication of NF1, most commonly affecting the lower limbs. EN can arise from plexiform neurofibroma of the superficial and deep nerves due to a hyperproliferation of the perineural connective tissue can affect the limb soft tissues (causing adipocyte metaplasia), bone (bone overgrowth and focal gigantism) and lymphatic system (presenting with congenital lymphangiomatosis and lymphostasis with subsequent lymphoedema). Bleeding is a common complication and malignant transformation can occur in 5% of cases.

Key Message: Elephantiasis Neuromatosa is a rare, but significant, association with Neurofibromatosis type 1. Regular surveillance of these lesions, especially during younger

years, is recommended to monitor for complications and to refer for surgical debulking procedures.