Clinical pictures

An unusual haversack on the shoulder

A 59-year-old Caucasian male presented to our department complaining of progressive developing of severe disfigurement and deformity of trunk by a massive swelling (Figure 1) for ~20 years.

A large pendulous mass with folds of loose and thickened skin, appearing as a big ‘haversack’ on the left shoulder, arose from the anterior chest wall, hung down to the waist and completely obscured the breast; similarly, deltoidal and scapular regions were covered by the same lesion. It felt like a bag of worms when palpated and was freely mobile over the underlying tissue.

This is a case of type 1 neurofibromatosis, a diagnosis established on patient’s clinical presentation and personal history.

Originally described in 1882 by Friedrich von Recklinghausen,1 neurofibromatosis 1 (NF-1) is a common autosomal dominant disorder, with an incidence of about 1 : 2500–1 : 3300 and accounting for up to 90% of the cases of NF.2

The hallmark of NF-1 is an increased propensity for developing both benign and malignant tumours. A plexiform neurofibroma consists of a poorly circumscribed, locally invasive, non-metastatic proliferation of cells in the nerve sheath, more frequently in trigeminal or upper cervical plexuses, extending across the length of a nerve and involving multiple nerve fascicles; it may be visible from the body surface, or may be internal with no evident superficial extension.

Figure 1. Enormous, smooth, elastic, red-violaceous mass on the scapular area, involving supravacicular and mammary regions with big digitations.
Plexiform neurofibromas are present in ~17% of patients with NF-1, often appear in the first decade and their growth can occur at any time in life, with a preference for the early childhood and puberty. They are usually solitary and need to be monitored frequently because about 2–5% develop into malignant peripheral nerve sheath tumours.3

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References