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INTERESTING MEDICAL IMAGE

Don't Miss the Target

A case of neurofibromatosis type 1

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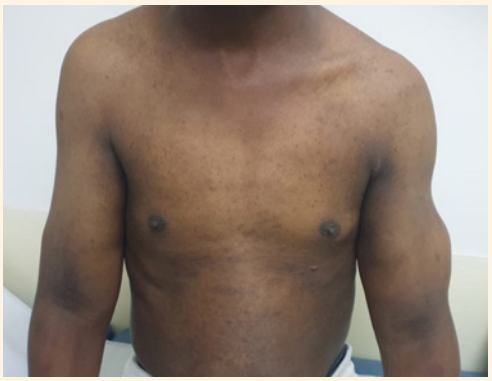

Figure 1: Photograph of the chest of a 27-year-old male patient showing café-au-lait macules and multiple swellings in the arms.

A 27-year-old male patient, with no diagnosed comorbidities, presented to a vascular surgery clinic in Muscat, Oman, in 2021 referred as a case of lymphangioma, after an ultrasound was done elsewhere. He presented with a history of a progressively enlarging left lateral arm swelling, that he noticed two years earlier when receiving an intramuscular injection for analgesia, following an injury to the leg while playing football. Examination revealed hyper-pigmented spots, cafe-au-lait macules, distributed over his body [Figure 1]. The swelling was overlying the lateral aspect of the left arm, measuring approximately 12 × 7 cm. It was non-tender and firm with limited mobility. There were similar smaller swellings over his chest, back and other limbs. Magnetic resonance imaging (MRI) was performed after a clinical diagnosis of neurofibromatosis type 1 (NF-1). This revealed a classical appearance of neurofibroma known as the target sign [Figure 2]. Patient's consent was obtained prior to taking and using his photos and images for publication purposes.

Comment

Neurofibromatosis is an autosomal dominant neurocutaneous disorder; there are two types: NF-1 and NF-2. NF-1 comprises approximately 96% of all cases and affects 1 in 3,000 individuals worldwide and presents with cutaneous and non-cutaneous manifestations.[1] Commonly, it presents with café au lait spots, neurofibromas, axillary or groin freckling and optic manifestations. Other complications of the disease include seizures, problems with digestion, psychological burdens and developmental delay including delay in learning to walk and talk. NF-2 usually presents with schwannomas and meningiomas.[2,3] There are three varieties of neurofibroma's: localised, diffuse and plexiform.

Classically, radiologists have recognised two types of neurofibroma: the discrete mass/nodular form and the diffuse or plexiform variety.[2] On MRI, plexiform neurofibromas present with the pathognomonic 'target sign' depicted as lesions with central hypointensity and surrounding hyperintense rim on T2-weighted images, representing fibro collagenous and myxoid components, respectively. A 'reverse target sign', on T1-weighted images after intravenous (IV) gadolinium-based contrast administration, is seen as central enhancement with peripheral hypointensity.[4] Venous malformations mimic the classical 'target sign' seen in plexiform neurofibromas, whereby the central hypointense focus represents a phlebolith in a dilated venous channel.

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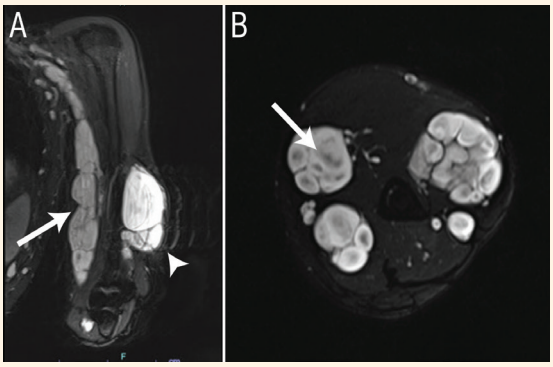

Figure 2: Magnetic resonance imaging of the left arm of a 27-year-old male patient showing (**A**) the localised lesion laterally (arrowhead) with plexiform lesion medially (arrow) and (**B**) an axial view showing the target sign (arrow). The target sign refers to a central area with low signal intensity, surrounded by a high signal intensity and is sometimes referred to as a bull's eye sign.

Phleboliths are also noted as a focal hyperintensity on gradient-recalled echo sequences. Other MRI-differentiating features of venous malformations include a hyperintense, lobulated and septated lesion on T2-weighted images and IV contrast enhancement of its cavernous section without arteriovenous shunting on dynamic MRI.[4] NF-1, with its various manifestations, is a clinical diagnosis and its management requires the involvement of a multidisciplinary team. Patients are referred to surgeons to excise the neurofibroma when the patient is symptomatic with pain, there has been a recent increase in size, the tumour results in a neurological deficit or has a high risk of rupture and bleeding. Plexiform neurofibromas have significant morbidity due to the way it infiltrates surrounding structures, it's unpredictable growth and its potential to bleed.[5]

AUTHORS' CONTRIBUTION

ES conceptualised the manuscript. OH, BY, IA and RS performed the literature review. OH, BH, IA, HM, RS and KW drafted the manuscript. ES revised the manuscript. All authors approved the final version of the manuscript.

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