



GENETICS AND GENODERMATOSES

AN ELEPHANTIASIS NEUROMATOSA IN A CONTEXT OF NEUROFIBROMATOSIS TYPE I

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Background: Neurofibromatosis type 1 (NF1) or von Recklinghausen's disease is clinically characterized by presence of diffuse plexiform neurofibroma (PNF). A variant of the PNF sits at the lower third of the leg associated with lymphangiomatosis can give elephantiasis neuromatosa (EN). Which is a rare tumor during NF I, we report 1 case.

Observation: 35-year-old woman, no similar case in family, who present a giant tumor taking the right lower extremity of firm consistency that had been evolving for years, for whom the clinical examination revealed multiple and diffuse neurofibroma, lentiginos and coffee stain with milk, suggestive of NF I. The rest was normal. An MRI of the right lower limb looking for signs of degeneration and operability of the patient, revealing radiological signs in favor of an EN with no sign of degeneration with presence of infiltration of the nervous and vascular structures. The patient was therefore inoperable, and we opted for surveillance.

Key message: NF is an autosomal dominant genodermatosis, characterized by the presence of PLN. These are unencapsulated, poorly circumscribed formations that infiltrate nerves, fat, and adjacent muscles. A variant of the PNF: the gelatiniform mixoglioma, is generally soft and is located in the lower third of the leg. When associated with lymphangiomatosis, it may be the cause of EN. The latter is characterized by abnormal soft tissue hypertrophy and bone dysplasia associated with early and excessive bone growth in the affected leg compared to the contralateral leg. Pachidermocele or dermoholysis may be associated with NF1. The etiology of EN has not yet been fully elucidated, but the association of primary lymphatic dysplasia with a proliferative lymphatic process has been proposed. Lymphoscintigraphy and magnetic resonance imaging is a tool for diagnosing and monitoring the patient. The early diagnosis of these pathologies allows monitoring and prevention of complications.

