



Arteriovenous Malformations in the Setting of Giant Elephantiasis Neuromatosa

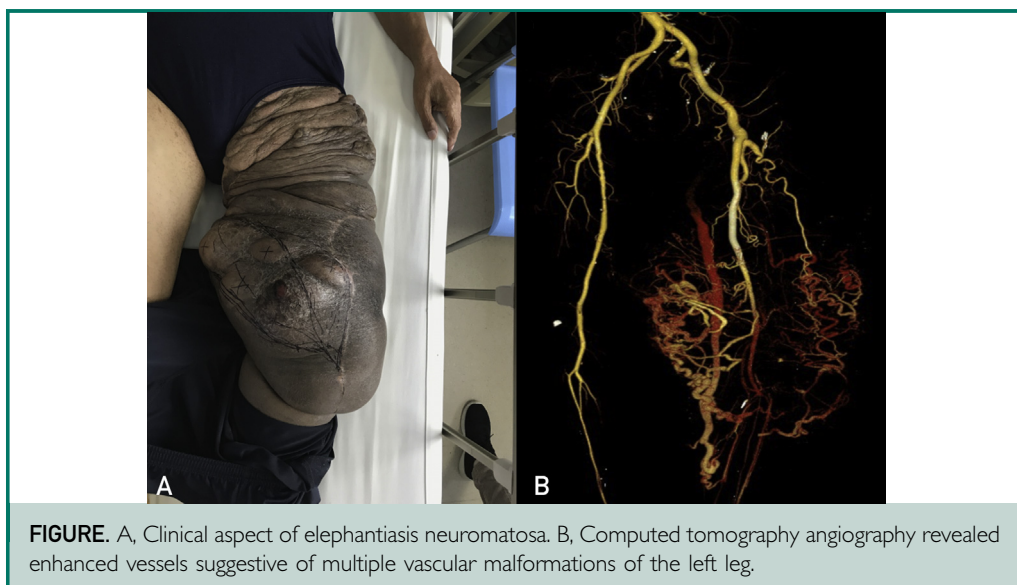
Chunni He, MD, and Jun Li, PhD

From the Department of Radiology, Yantai Affiliated Hospital of Binzhou Medical University, Yantai, China.

A 65-year-old man presented with giant elephantiasis of the left leg, accompanied by skin pigmentation and local ulceration (Figure A), which had started to grow since he was 4. Numerous café-au-lait spots were found across the trunk. The patient had previously undergone surgery 4 times during recent 10 years and had been diagnosed with neurofibromatosis type 1 (NF-1) based on clinical symptoms and histopathological results. Magnetic resonance imaging revealed extensive inhomogeneous signal intensity masses consisting of multiple flow void vascular structures. Computed tomography angiography (Figure B) revealed multiple enlarged, mesh-like vascular malformations, involving popliteal and femoral arteries draining into the popliteal and femoral veins. The patient underwent surgical resection under combined spinal-epidural anesthesia. A fusiform incision centered in the masses was made.

Surgical findings revealed affluent nourishing vessels of subcutaneous and deep soft tissue and extensive grayish black tissues, with local necrosis and cystic degeneration. Peripheral tissues were separated, and the masses were completely resected. Pathological examination confirmed neurofibromatosis, with some areas transformed into a malignant peripheral nerve sheath tumor.

Neurofibromatosis type 1 is a rare autosomal dominant disease, which is characterized by multiple neurofibromas and pigmented lesions. Eight percent to 15% of patients with NF-1 have a malignant peripheral nerve sheath tumor during their lifetime. Abnormal soft tissue hypertrophy and deformed masses growing along the length of nerves can occur and affect surrounding tissues such as skin and muscle. This condition is termed *elephantiasis neuromatosa*, which can cause changes in the vascular and lymphatic system.¹ Although no more than 40 cases of



elephantiasis neuromatosa are described in the literature, the real incidence is estimated to be higher. Vascular abnormalities are a well-recognized manifestation of NF-1. The renal artery is the most frequent site of involvement. These abnormalities may be caused by Schwann cell proliferation, dysplastic smooth muscle, or proliferation of neurofibromatosis in the vascular wall.² Its genetic basis is a loss-of-function germline mutation in the NF1 gene at 17q11.2.

Potential Competing Interests: The authors report no competing interests.

Correspondence: Address to Jun Li, PhD, Department of Radiology, Yantai Affiliated Hospital of Binzhou Medical University, No. 717, Jinbu St, Yantai, Shandong 264100, China (junli@bzmc.edu.cn).

ORCID

Jun Li:  <https://orcid.org/0000-0003-1389-558X>

REFERENCES

1. Martínez-García S, Vera-Casaño A, Eloy-García Carrasco C, del Boz-González J, Martínez-Pilar L, Crespo-Erchiga V. Elephantiasis neuromatosa in a patient with neurofibromatosis type 1. *J Eur Acad Dermatol Venereol*. 2008;22(1):103-105.
2. Oderich GS, Sullivan TM, Bower TC, et al. Vascular abnormalities in patients with neurofibromatosis syndrome type 1: clinical spectrum, management, and results. *J Vasc Surg*. 2007;46(3):475-484.