A 7-year old boy was admitted to our Department as an outpatient due to the presence of a palpable lesion of bimalleolar localization in the right ankle, with a growing tendency over the previous two years. The boy complained of vague localized pain, aggravated by physical exercise. ECHO, MRI (Panel A) and MRA tests were conducted and the lesion was evaluated as a low flow vascular abnormality. As a result scheduled surgery was performed, and five atractoid shaped lesions were removed (Panel B), starting from the subcutaneous to the articular sac of the right ankle. The postoperative course was uneventful and the boy was discharged home on the 2nd postoperative day in excellent general condition. Histopathological and immunohistochemical examination revealed a plexiform neurofibroma (PN). The lesion consisted of spindle cells arranged in bundles, fascicles and whorls, with minimal cytologic

PLEXIFORM NEUROFIBROMA AS A FIRST MANIFESTATION OF NEUROFIBROMATOSIS TYPE 1 IN A 7-YEAR OLD BOY

Dimitrios PATOULIAS¹, Maria KALOGIROU², Ioannis PATOULIAS³

¹Department of Internal Medicine, General Hospital of Veria, Veria, Greece, ²1st Department of Pediatric Surgery, Aristotle University of Thessaloniki, GHG. Gennimatas, Thessaloniki, Greece
Atypia. Mitotic figures were rare. No necrosis or invasion in the surrounding tissues was observed (HE × 40) (Panel C). The patient’s uncle had been diagnosed with Neurofibromatosis type 1 (NF1) during his adolescence, while his twin sister and the mother of our patient, after a detailed diagnostic procedure, was found in early childhood to have freckles on the groin bilaterally. PN is usually diagnosed before the age of 5 years. It is symptomatic in 55% of all patients (1), while in 17-64% of cases it is located in an anatomic area where it may cause functional disorders (2). A rapid increase in size, intense local pain and neurological manifestations should always be evaluated carefully by the clinician, as they may indicate the development of a malignant peripheral nerve sheath tumor (MPNST) (3). The latter usually develops during adulthood. Meticulous follow-up of these patients is vital for exclusion of MPNST development. 18FDG-PET is crucial for diagnosis of malignancy. Annual follow up consists of: physical examination, blood test, ophthalmological examination and evaluation of the patient’s psychomotor development. After a 4-year follow up period post-operatively, our patient remains asymptomatic, while there is no evidence of recurrence. The patient is due to undergo a genetic test for identification of a NF1 gene mutation, after detailed investigation of his family history. Prenatal diagnosis by amniocentesis or a chorionic villus sample, or preimplantation genetic diagnosis in the case of in vitro fertilization, aimed at avoiding passing on this disorder to his children in the future, is the basis of genetic counseling.

Key words: Plexiform neurofibroma • Child • Malignant peripheral nerve sheath tumor.

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Correspondence: dipatoulia@gmail.com
Tel.: +231 0225 083; Fax.: +231 0225 083

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