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ELEPHANTIASIS NEUROMATOSA - RIJETKA I EKSTREMNA MANIFESTACIJA PLEKSIFORMNOG NEUROFIBROMA U BOLESNIKA S NEUROFIBROMATOZOM TIPA 1 DIJAGNOSTICIRANA U RANOJ FAZI

ELEPHANTHIASIS NEUROMATOSA - A RARE AND EXTREME MANIFESTATION OF PLEXIFORM NEUROFIBROMA IN PATIENT WITH NEUROFIBROMATOSIS TYPE 1 DIAGNOSED AT AN EARLY STAGE

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SAŽETAK

Plexiform neurofibroma is a rare variant of NF-1 characterized by deformed nerve masses, also affecting all surrounding tissues such as skin, muscle, lymphatics, vessels and bones. Although rare, when abnormal surrounding tissue hypertrophy occurs the entity is termed elephantiasis neuromatosa. It still remains unknow what is the proper case management in order to prevent progression which can cause extensive mutilation of the limb and even lead to amputation. Hereby we present a case of 25-year-old female patient with neurofibromatosis type I that presented to our Lymphoedema Clinic in October 2023 due to the mild swelling of the lower part of the left leg. The swelling appeared at the age of 18 but now it started to progress with development of tingling sensation of the left foot. Clinical examination and tape measurement did not reveal any clinical signs of lymphoedema. Nevertheless, the skin appeared thickened on palpation. Extensive diagnostic

workup was performed that revealed plexiform neurofibromas throughout the periphery. Lymphoscintigraphy revealed indirect sign of surface redistribution of the lymphatic system in both lower parts of the legs. Fortunately, MRI and CT angiography did not reveal involvement of arteriovenous system, nor muscles or bones, just cutis and subcutaneous tissue infiltration. Although elephanthyasis neuromatosa was diagnosed early proper treatment strategy is yet to be determined and negotiated. It has been shown that it is not responsive to any conventional therapies. Surgery, although is the mainstay treatment, is often unsatisfactory and can be associated with life-threatening complications. In young patient MEK inhibitors have shown promising results but are very expensive and not approved nor available for treatment of adult patients in Croatia. A patient with a timely diagnosis of a rare condition such mutilating as elephantiasis neurofibromatosa is deserves a chance for proper treatment.