

Plexiform neurofibromatosis with peripheral malignant nerve sheath tumor and scoliosis - more surveillance imaging needed?

ABSTRACT

Malignant peripheral nerve sheath tumors (MPNSTs) are rare but aggressive neoplasms associated with neurofibromatosis type 1. Specifically, children with deep plexiform neurofibromas are 18 times more likely to develop MPNSTs compared to the general population. However, there is currently no standard surveillance imaging protocol for children diagnosed with deep plexiform neurofibromatosis. We present a case of a boy with neurofibromatosis type 1 and scoliosis, who later developed MPNST. This case highlights the need for more frequent surveillance imaging and the challenges of diagnosing MPNST in a patient with scoliosis. In order to facilitate early detection of malignant transformation, we suggest annual surveillance MR imaging for patients known to have deep plexiform neurofibromatosis.