
Spinal Cord UBOs in NF1 Pediatric Patients: Single Institution Experience

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Introduction: Neurofibromatosis type 1 (NF1) is a neurocutaneous disease that shows specific clinical characteristics and imaging. Being a predisposition syndrome to different types of tumors, neurofibromas and glioma of the optic pathway being the most common lesions. It is essential to know the typical, non-oncological lesions that can be present and diagnosed through neuroimaging, so that they are not misdiagnosed and treated incorrectly. Within these lesions we found UBOs (Unidentified bright objects), located on white matter areas in the parenchyma, but little taken into account at spinal cord level.

Aims: To describe the prevalence of UBOs located in the spinal cord, in patients with NF1 during childhood, based on data obtained at our institution.

Materials and methods: Retrospective, observational and descriptive study of patients between 2 and 21 years old with NF1 diagnosis, evaluated during 2009 to 2023 time period, who had whole brain and spinal cord MRI (n=32). The presence of UBOs was evaluated. We refer to spinal UBO as single or multiple lesions located in the cord, hyperintense on T2/STIR sequences and isointense on T1, without gadolinium enhancement or mass effect.

Results: Male patient 58.3%, with an average age of 13.1 years. Twenty seven patients (79%) had brain UBOs and 9 (26.4%) Spinal cord UBOs. Of the latter, 44.4% presented a single spinal cord lesion, the rest had more than one. The most common location was the subaxial cervical (C3-T1) area, followed by the cervical superior (C1-C2) area, dorsal with 2 lesions and conus medullaris in only one case. In all of them, hypersignal was found in T2 sequence and isointense signal in T1; no case showed enhancement with intravenous contrast or mass effect. There was coexistence of brain UBOs in all our patients. None of the patients presented clinical manifestations associated with these lesions.

Conclusion: There is little literature regarding the existence of spinal UBOs. It is important to recognize the existence and diagnostic imaging characteristics of these in order to avoid diagnostic and therapeutic errors regarding the presence of spinal cord lesion in NF1 patients.

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Risk of Myocardial Infarctions in Individuals with Neurofibromatosis Type 1

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Background: Neurofibromatosis type 1 (NF1) may be associated with comorbidities not yet recognized. Case reports of myocardial infarctions related to coronary artery aneurysms and pheochromocytomas have been reported in NF1, while very few epidemiologic data on the incidence of heart attack in NF1 has been available.

Methods: To investigate the incidence of myocardial infarctions among individuals with NF1, we utilized a comprehensive Finnish cohort comprising 1,811 NF1 patients, drawing on nationwide data. For comparison, we selected a control group at a ratio of 10 persons for each patient, matching them by sex, date of birth, and municipality. We tracked the occurrence of myocardial infarctions using data from the Finnish Care Register for Health Care, which is overseen by the Finnish Institute for Health and Welfare, and the Causes of Death Register maintained by Statistics Finland. The Finnish Care Register for Health Care compiles detailed records of inpatient care and hospital-based outpatient visits. We specifically searched the registers for the ICD-10 codes I21 and I22, along with the ICD-9 code 410, to identify individuals with myocardial infarction.

Results: We observed 42 individuals with myocardial infarction and NF1, leading to a hazard ratio (HR) of 1.4 (95% CI 1.0-1.9) when compared with the matched control group. Six individuals with NF1 died of myocardial infarction with no prior hospital visit related to myocardial infarction. The mean age at the time of the first diagnosis for a myocardial infarction among patients with NF1 was 66.2 years (SD 12.8), while in controls the mean age was 69.7 years (SD 12.5). Among the NF1 patients who experienced a myocardial infarction, 19 were women, with a HR of 1.6 (95% CI 1.0-2.6), and 23 were men, with a HR of 1.2 (95% CI 0.8-1.9). Diagnoses preceding the myocardial infarction in NF1 patients included chronic ischemic heart disease (38%), angina pectoris (19%), disorders of lipoprotein metabolism (19%), heart failure (14%), cerebral infarction (14%), and essential hypertension (29%), yet the proportions did not differ from the controls with myocardial infarction.

Conclusions: Our preliminary findings suggest that at least women with NF1 may be at an elevated risk for myocardial infarction, and the underlying causes of myocardial infarctions in NF1 require further investigation. These results highlight the importance of customized follow-up care for patients with NF1 to better address and mitigate their specific health risks.

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