Neurofibromatosis Type 1: Focal areas of signal intensity in the brain

A 15-year-old female patient is being followed up for neurofibromatosis type 1. Cranial MR examination was taken for control purposes.

Imaging Findings:

In cranial MRI, a pathological signal with a diameter of 5-6 mm is observed at the globus pallidus level. The signal area is observed as a slight central hypointense signal showing slight peripheral hyperintensity in T1 images, and as a hyperintense signal in T2 and FLAIR images.

Discussion:

Background

Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen’s disease, is a multisystem neurocutaneous disorder. It is the most common neurocutaneous disorder. NF1 is an autosomal dominant disorder. Half of all cases are spontaneous mutations. The diagnosis is made based on clinical manifestations. Its estimated incidence is 1/3500 live births [1]. Diagnosis requires the presence of 2 or more major criteria: 6 or more café au lait spots, axillary or inguinal freckling, 2 or more cutaneous neurofibromas, 1 plexiform neurofibroma, characteristic bony lesions (thinning of long bone cortex with or without pseudarthrosis, sphenoid wing hypoplasia), an optic glioma, 2 or more iris Lisch nodules, or a first-degree relative with NF1 [1]. Focal abnormal signal intensities (FASI) on brain MRI occur commonly in patients with NF1. FASI is present in varies from 43 to 93% of pediatric patients with NF1. [2]

Clinical Perspective

Most of the time, FASI is an incidental lesion in cranial MR. They do not constitute a clinical finding, but some studies have found a relationship between cognitive dysfunction and the number and location of T2-hyperintensities, particularly in the cerebellum and the thalamus. [3]

Imaging Perspective

The most common central nervous system abnormalities in neurofibromatosis are magnetic resonance imaging (MRI) T2-hyperintensities. These features most commonly occur in the basal ganglia, brainstem, thalamus,
cerebellum, and hippocampus; they display no mass effect, do not enhance with contrast, are not associated with focal neurological deficits, and resolve by adulthood. [4,5]

**Outcome**

As a central nervous system finding in NF type 1; In addition to FASI, optic nerve glioma or optic pathway glioma, progressive sphenoid wing dysplasia, lambdoid suture defects, dural calcification at the vertex, rarely moya-moya phenomenon, buphthalmos can be seen [1].

MRI is a valuable method for their identification. When FASI is seen in a patient with NF type 1 on MRI, it should usually not be a cause for concern. It should be kept in mind that thalamic FASI’s may be associated with cognitive disorders. [2-5]

Follow-up should be recommended when FASI is detected in cranial MRI in patients with NF type 1. Although rare, FASI has the potential to show a mass effect and / or increase in contrast. Rarely, such features are a sign of a malignant change. [6]

Written informed patient consent for publication has been obtained.

**Differential Diagnosis List:** Cranial focal area of signal intensity in NF type 1

**Final Diagnosis:** Cranial focal area of signal intensity in NF type 1

**References:**


**Figure 1**

**Description:** Axial cranial T1 MR; The pathological signal at the left globus pallidus level can be difficulty selected in T1 image. **Origin:** Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020
Description: Axial cranial T2 MR; A hyperintense pathological signal is observed at the left globus pallidus level

Origin: Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020
Description: Axial cranial FLAIR MR; A hyperintense pathological signal is observed at the left globus pallidus level

Origin: Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020
Description: Sagittal cranial T2 MR; A hyperintense pathological signal is observed at the left globus pallidus level

Origin: Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020
**Description:** DWI MR; There is no evidence of diffusion restriction at the left globus pallidus level

**Origin:** Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020
Description: ADC MR; There is no evidence of diffusion restriction at the left globus pallidus level
Origin: Department of Radiology, Bursa Yuksek Ihtisas Training and Research Hospital, Turkey, 2020