Neurofibromatosis Type-1 with Unidentified Bright Objects in Advancing Age

**ABSTRACT** Neurofibromatosis type 1 (NF1) is the most common autosomal dominant neurocutaneous syndrome. Unidentified bright objects (UBOs) are the most common neuroimaging feature of NF1 and usually expected to disappear in adulthood. In our case, we wanted to draw attention to the fact that some of the UBOs can continue in adulthood without any clinical signs.

Keywords: Neurofibromatosis type 1; unidentified bright objects; advancing age; cognitive dysfunction

Neurofibromatosis type 1 (NF1) is the most common autosomal dominant neurocutaneous syndrome and affecting approximately 1 in 2700 newborns.\(^1\) NF1 has high variability of expression, for that reason different clinical manifestations are seen on patients with the same NF1 gene mutation. Unidentified bright objects (UBOs) which is the most common neuroimaging feature of NF1, especially in childhood.\(^2\) Generally, UBOs are transient and disappear into adulthood but some of them persist.\(^3\)

We discuss the neuroimaging findings of 52-year-old man with NF1 who demonstrated UBOs on the right side of the hemispheric white and gray matter.

**CASE REPORT**

52-year-old right handed man referred for the complaint of dizziness to Haydarpaşa Numune Training and Research Hospital Neurology Service. He did not have NF1 diagnosis before. However, he represented clinical features consistent with NF1 such as axillary and inguinal freckling, café-au-lait spots, Lisch nodules, neurofibromas. In family history, his mother, who had died, had neurofibromas and other clinical findings not known. As a result of evaluations, NF1 was detected in his two sisters and daughter who were not diagnosed before. In neurological examination, he had a complaint of dizziness which increased with movement, but the complaint was existed at rest. Cognitive functions, cerebellar tests and the rest of examination was normal, he had no ataxia or gait abnormalities.
Magnetic resonance imaging (MRI) demonstrated hyperintense foci, termed UBOs or focal areas of signal intensity (FASI) on T2 weighted Figure 1, and fluid attenuated inversion recovery (FLAIR) MRI scans (Figure 2). They were isointense on T1 Figure 3, and without contrast enhancement (Figure 4). UBOs were found in cortico-subcortical region of fronto-parietal white matter. A consent form was obtained from the participant.

**DISCUSSION**

NF1 is related with somewhat neuroimaging findings such as white and gray matter volumetric changes. UBOs are the most common neuroimaging feature in NF1 patients which frequently presented in thalamus, internal capsule, basal ganglia, brain stem, cerebellum and subcortical hemispheric white matter. These lesions indicate high signal on T2 and FLAIR sequences, isointense to hyperintense on T1, no contrast enhancement and no mass effect. UBOs are defined by vacuolar/spongiotic changes of myelin without inflammation and no marked demyelinization in the around tissue. UBOs noted benign and generally transient which happened in patients with 4-12 aged. Expecting to regress in adulthood and not associated
with focal neurological deficits, some persist and their presence, number and location correlated with cognitive dysfunction in recent studies. Barbi et al. reported that in 50% to 100% of the NF1 patients, MRI shows hyperintensities on T2-weighted sequences which are associated with learning disabilities. In the study of Cabellero et al., 31 patients with the NF1 were evaluated, 10% of patients had mild intellectual disability and UBOs were found in most of this subgroup. In our case UBOs were found in cortico-subcortical region of fronto-parietal white matter. Despite being seen in advanced age, they did not cause cognitive dysfunction. Mini Mental State Examination (MMSE) score was 30.

Optic nerve gliomas, sphenoid wing dysplasia, parenchymal gliomas, dural ectasia of the optic nerve sheath and spinal canal are the common manifestations central nervous system manifestations which were not seen in our patient.

Our case highlights the importance of presence, number and location of UBOs in advancing age without cognitive dysfunction. Long term prospective studies can express the reasons of regression of UBOs as age progresses and their relation with cognitive functions.

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Conflict of Interest
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