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A Diagnostic Whisper: When Temporal Poles Remain Unscathed in CADASIL

Tanıyı Fısıldayan: CADASIL'de Temporal Kutup Lezyonu Olmadığında

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A 48-year-old individual with a three-decade history of probable Behçet's disease presented to our neurology clinic in May 2023 with acute-onset vertigo. Neurological examination was largely unremarkable apart from bilateral visual impairment attributable to prior episodes of uveitis. Benign paroxysmal positional vertigo was diagnosed during follow-up. Detailed examination and tests revealed no other neurological, cognitive or psychiatric abnormalities. The patient had a prior history of treatment with Azathioprine and Rivaroxaban. Historical records documented active disease characterized by oral and genital aphthous ulcers, recurrent uveitis, and inferior vena cava thrombosis, with remission achieved approximately a decade ago. Since then, no further systemic manifestations of Behçet's disease have been observed.

Three prior cranial magnetic resonance imaging (MRI) studies dating back to 2009 were interpreted as demonstrating non-specific white matter lesions. However, acute high-resolution MRI performed at our institution revealed radiological findings deemed compatible with CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy) (Figure 1 and 2). Corroborating the diagnosis, family history disclosed a sibling with genetically confirmed CADASIL, in whom next-generation sequencing identified two heterozygous NOTCH3 variants: a pathogenic c.317G>A (p.Cys106Tyr) in exon 3 and a variant of uncertain significance, c.1774C>A (p.Arg592Ser) in exon 11. It was subsequently documented that the patient exhibited the same pathogenic mutations. Retrospective analysis of earlier MRIs revealed imaging characteristics consistent with CADASIL, previously misattributed to vascular inflammation associated with diagnosis of Behçet's disease.

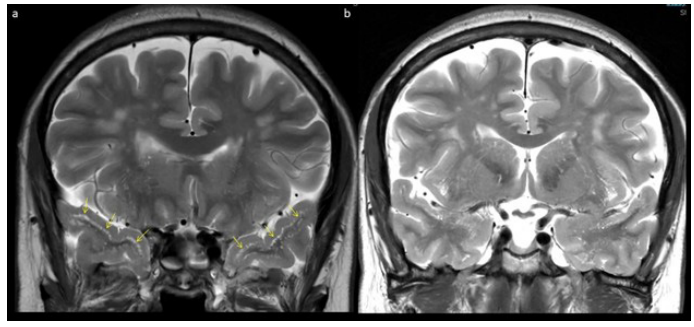

Figure 1a, b. Cranial T2-weighted coronal MR images: Expanded perivascular spaces are arranged like a necklace without leaving any gaps at the cortical-subcortical junction in the temporal lobe (arrows). Numerous bilateral and patchy hyperintense lesions are observed in the white matter of the centrum semiovale. Despite the presence of numerous dilated perivascular spaces in the temporal pole and external capsule, no overt white matter lesions are observed in these regions.

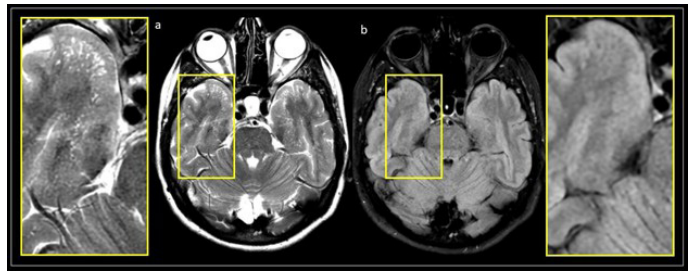

Figure 2a, b. Cranial axial T2 and FLAIR (Fluid-attenuated inversion recovery) images: A distinct clustering of numerous dilated perivascular spaces was noted, with the surrounding regional white matter appearing entirely unremarkable on imaging. The images on either side are magnifications of the encircled area in a and b.

Cranial MRI is an indispensable tool in diagnosing CADASIL. The most emblematic finding is subcortical leukoencephalopathy involving the bilateral temporal poles, external capsule, subinsular regions, and superior frontal gyri.[1] This imaging pattern is highly sensitive and specific, particularly in younger individuals. In contrast, multiple microhemorrhages and enlarged perivascular spaces (also known as état criblé or status cribrosum) in the basal ganglia may be considered nonspecific.[2] Although infrequent, subcortical lacunar infarcts situated at the margins of leukoencephalopathic lesions (named as peripheral lacunes), along with numerous dilated perivascular spaces arranged in a chain- or necklace-like pattern parallel to the ventricular border, particularly in the periphery of the temporal horn of the lateral ventricle, are highly specific for CADASIL.[2-4] Notably, dilated perivascular spaces situated at the junction between the cortical and subcortical white matter have not been described in any condition other than CADASIL.[2,5]

Temporal pole hyperintensity frequently coexists with temporal juxtacortical dilated perivascular spaces in CADASIL.[6] The dominant pathophysiological hypothesis attributes these findings to demyelination driven by impaired interstitial fluid clearance rather than ischemic insult.[7] In our case, although numerous dilated perivascular spaces were evident, the adjacent parenchyma remained structurally intact, suggesting that interstitial drainage may have been sufficiently preserved to prevent myelin injury.

This case reinforces the diagnostic utility of recognizing spatially distributed dilated perivascular space, particularly within the temporal pole and cortical-subcortical junction in CADASIL.[2,3] Even in the absence of hallmark radiologic features such as symmetrical leukoencephalopathy or clustered peripheral subcortical lacunar infarctions, this specific imaging sign warrant consideration of CADASIL and justify genetic testing.[2]

Temporal pole hyperintensity, present in over 90% of CADASIL cases, remains a highly specific marker. Nonetheless, its absence does not exclude the diagnosis, and its presence should be interpreted in the context of the broader radiologic and clinical profile.[5,8]

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