Turkish Journal of Cerebrovascular Diseases 2023; 29(2): 102-105 Türk Beyin Damar Hastalıkları Dergisi 2023; 29(2): 102-105
Turk J Cereb Vasc Dis
doi: 10.5505/tbdhd.2023.81904

LETTER TO EDITOR **EDİTÖRE MEKTUP**

HIGH RESOLUTION MAGNETIC RESONANCE IMAGING IN CADASIL: A CASE VIGNETTE

CADASIL'DE YÜKSEK REZOLÜSYONLU MANYETİK REZONANS GÖRÜNTÜLEME: OLGU RESMİ

Tuğba ÖZÜM[1], Mehmet Akif TOPÇUOĞLU[1], Rahşan GÖÇMEN[2], Ethem Murat ARSAVA[1]

[1]Hacettepe University Faculty of Medicine, Department of Neurology, Ankara, TÜRKİYE
[2]Hacettepe University Faculty of Medicine, Department of Radiology, Ankara, TÜRKİYE

Keywords: Small vessel disease, Cerebral, microbleeding, dementia, hypertension.
Anahtar Sözcükler: Küçük damar hastalığı, Serebral, mikrokanama, demans, hipertansiyon.

Dear Editor,

CADASIL is a hereditary small vessel disease that typically manifests in early adulthood and is characterized by migraine headaches, recurrent lacunar strokes, epilepsy, vascular dementia, apathy, pseudobulbar palsy, and organic psychiatric symptoms (1). If there is disproportionate white matter disease on magnetic resonance imaging (MRI) in patients with a positive family history and relatively mild or no vascular risk factors, CADASIL is the diagnosis that comes to mind. In the last 20 years, many cases and case series have been reported in our country, and it is not rare (2,3).

CADASIL has very specific MRI features; in many cases, it is possible to identify the syndrome based on the lesion pattern. In the family of the presented 59-year-old man, a C.3043T>C (p.Cys1015Arg) mutation was found in exon-19 of the NOTCH3 gene. The diagnosis was made after recurrent strokes, that had first occurred 8 years ago. In his family, grandmother, aunt, siblings, and children have a diagnosis of CADASIL. The 3T MRI findings of the patient who presented with acute right-sided hemihypoesthesia while continuing to smoke and use clopidogrel are presented. (Figures 1 and 2). CADASIL's most striking MRI findings are symmetric, coalescing and diffuse hyperintense lesions (ischemic leukoencephalopathy) in T2-weighted and FLAIR sequences (4). Basal ganglia and thalamus are frequently affected. The pons and corpus callosum are also not infrequently affected. A well-documented typical feature is that the anterior temporal lobe and the external capsule are affected first and become completely affected over time. Both areas were severely affected in about 90% of cases (5). The cortex and U fibres are typically spared in CADASIL. Occipital and orbitofrontal subcortical white matter involvement is also relatively rare (6).

Lacunar infarcts correlate closely with clinical symptoms and are often located in the marginal zones of leukoencephalopathy (peripheral lacunae). This is a distinctive feature (7). In CADASIL, lacunae tend to be variable in shape and large. Acute lacunar lesions may be symptomatic or detected on follow-up MRIs of completely asymptomatic cases.

Cerebral microbleeds occur in up to 45% of CADASIL cases and do not have a typical distribution (6).

The number and volume of perivascular spaces were significantly increased in CADASIL. A frequently observed finding is an état criblé (or

Address for Correspondence: Prof. Mehmet Akif Topçuoğlu, M.D. Hacettepe University Faculty of Medicine, Department of Neurology, Ankara, Türkiye.
Phone: +90 312 305 18 06 **E-mail:** mat@hacettepe.edu.tr
Received: 09.07.2023 **Accepted:** 08.08.2023
ORCID IDs: Tuğba Özüm 0009-0000-7635-9034, Mehmet Akif Topçuoğlu 0000-0002-7267-1431, Rahşan Göçmen 0000-0002-0223-9336 , Ethem Murat Arsava 0000-0002-6527-4139.
Please cite this article as following: Özüm T, Topçuoğlu MA, Göçmen R, Arsava EM. High resolution magnetic resonance imagıng in cadasil: A case vignette. Turkish Journal of Cerebrovascular Diseases 2023; 29(2): 102-105. doi: 10.5505/tbdhd.2023.81904.

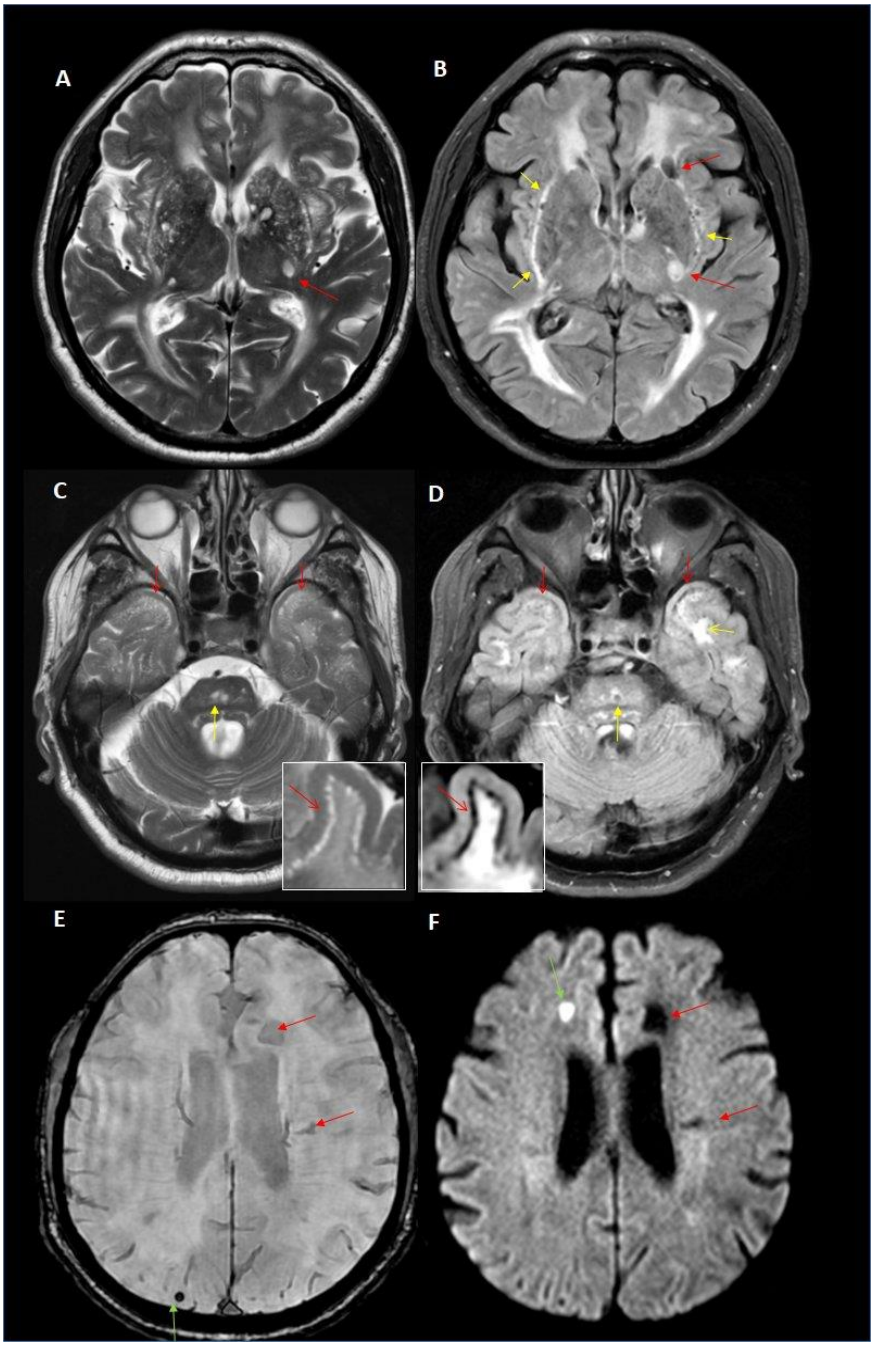
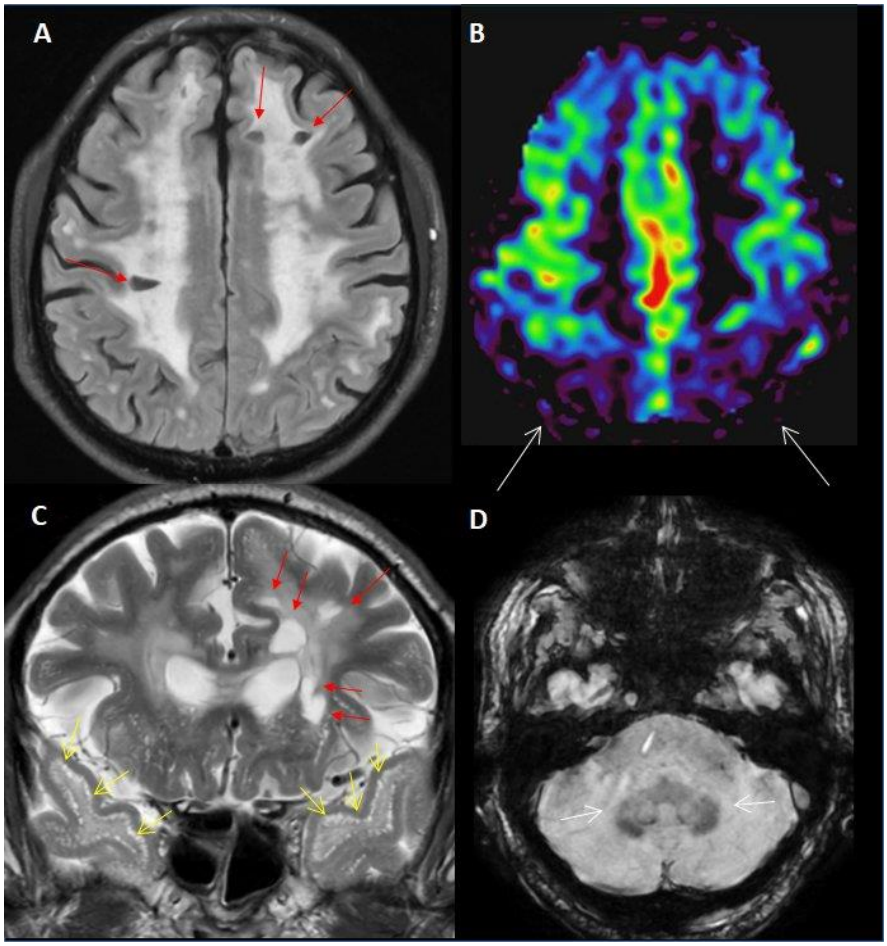


Figure 1. A: T2 axial: diffuse subcortical leukoencephalopathy with preservation of arcuate (u) fibres, lacunar infarcts with peripheral localization [red arrow], multiple central lacunar infarcts, and etate crible; B: FLAIR (fluid-attenuated inversion recovery): Diffuse leukoencephalopathy, symmetric involvement of external capsules [yellow arrows], chronic and subacute central lacunar infarcts [red arrows]; CD. T2 and FLAIR axial: anterior temporal leukoencephalopathy [yellow arrow with open end], marginal perivascular spacing fields (box: an upper section, red arrows with open end), lacunar infarcts in the pons [yellow arrow]; E: SWI (Susceptibility weighted imaging): right occipital microhemorrhage [green arrow], central and peripheral lacunae [red arrow]; F: DWI (diffusion-weighted imaging): Right frontal subcortical acute lacunar infarct [Green arrow], central and peripheral lacunae [Red arrow]. 3T magnetic resonance imaging..



Resim 2. A: FLAIR (Liquid attenuated inversion healing): Diffuse subcortical leukoencephalopathy with preservation of arcuate (u) fibres, lacunar infarcts with peripheral localization [red arrow]; B:ASL (Arterial spin labelling MR perfusion): Bilateral parietal cortical hypoperfusion [white open arrows]; C: Coronal T2 transmission: Diffuse leukoencephalopathy, chronic peripheral and central lacunar infarcts with multiple conglomerates [red arrows], enlarged peri-spaces in a chain-like arrangement [yellow open sell arrows]; D: SWI (Susceptibility weighted imaging): Peri-dentate hypointensity [White arrows]. 3T magnetic resonance imaging..

status cribrosum) localized in the basal ganglia. Corticosubcortical rim location of perivascular spaces, especially of the temporal pole, and the formation of chain-like structures are the the most specific imaging findings of CADASIL (6). Cerebrocortical hypometabolism and associated hypoperfusion due to cortical disconnection caused by subcortical lesions are other guiding imaging findings in CADASIL (8). Disproportionate involvement of the temporal pole and external capsule, micro haemorrhages, and multiple and enlarged perivascular spaces suggest a diagnosis of CADASIL. However, peripheral lacunae, perivascular spaces that chain together, and cortical hypometabolism are almost diagnostic of the disease, especially when they occur together. So, once seen, never forgotten.

REFERENCES

1. Chabriat H, Joutel A, Dichgans M, et al. CADASIL Lancet Neurol 2009; 8(7): 643-653.
2. Bolayir E, Taş A, Kaloğlu C, et al. CADASIL: A Case Report. Turk J Neurol 2002; 8(1): 125-130.
3. Utku U, Celik Y, Uyguner O, et al. CADASIL syndrome in a large Turkish kindred caused by the R90C mutation in the Notch3 receptor. Eur J Neurol 2002; 9(1): 23-28.
4. Bersano A, Bedini G, Markus HS, et al. The role of clinical and neuroimaging features in the diagnosis of CADASIL. J Neurol 2018; 265(12): 2934-2943.
5. O'Sullivan M, Jarosz JM, Martin RJ, et al. MRI hyperintensities of the temporal lobe and external capsule in patients with CADASIL. Neurology 2001; 56(5): 628-634.

6. Jouvent E, Duering M, Chabriat H. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Lessons from neuroimaging. Stroke 2020; 51(1): 21-28.
7. Duering M, Csanadi E, Gesierich B, et al. Incident lacunes preferentially localize to the edge of white matter hyperintensities: Insights into the pathophysiology of cerebral small vessel disease. Brain 2013; 136(Pt 9): 2717-2726.
8. Tatsch K, Koch W, Linke R, et al. Cortical hypometabolism and crossed cerebellar diaschisis suggest subcortically induced disconnection in CADASIL: An 18F-FDG PET study. J Nucl Med 2003; 44(6): 862-869.

Ethics

Ethics Committee Approval: Since this study is a letter to the editor, ethics committee approval is not required.

Informed Consent: The authors declared that they have obtained signed consent from the case for this publication.

Authorship Contributions: Surgical and Medical Practices: TÖ, MAT, RG, EMA. Concept: TÖ, MAT, RG, EMA. Design TÖ, MAT, RG, EMA. Data Collection or Processing: TÖ, MAT, RG, EMA. Analysis or Interpretation: TÖ, MAT, RG, EMA. Literature Search: TÖ, MAT, RG, EMA. Writing: TÖ, MAT, RG, EMA.

Copyright Transfer Form: Copyright Transfer Form was signed by all authors.

Peer-review: Internally peer-reviewed.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.