EUROPEAN JOURNAL OF BIOMEDICAL AND PHARMACEUTICAL SCIENCES
http://www.ejbps.com

ISSN 2349-8870
Volume: 10
Issue: 4
218-220
Year: 2023

RADIOLOGICAL FINDINGS OF CEREBRAL AUTOSOMAL DOMINANT ARTERIOPATHY WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY (CADASIL): A CASE REPORT

Dr. Ishan Gupta*, Dr. Nitashree Konwar and Dr. Mohidul Islam

Post-Graduate Trainee, Department of Radio-Diagnosis, Tezpur Medical College and Hospital, Assam.

***Corresponding Author: Dr. Ishan Gupta**

Post-Graduate Trainee, Department of Radio-Diagnosis, Tezpur Medical College and Hospital, Assam.

Article Received on 06/02/2023 **Article Revised on 27/02/2023** **Article Accepted on 19/03/2023**

ABSTRACT

Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) is a rare genetic disease that affects small blood vessels in the brain, leading to cognitive decline, stroke, and dementia.2 Diagnosis is challenging, and radiology plays a crucial role in identifying characteristic imaging findings, such as white matter hyperintensities, lacunar infarcts, and microbleeds.5 This paper presents a case of a 53-year-old male with a history of hypertension and recurrent strokes who was diagnosed with CADASIL based on clinical presentation and neuroimaging findings.

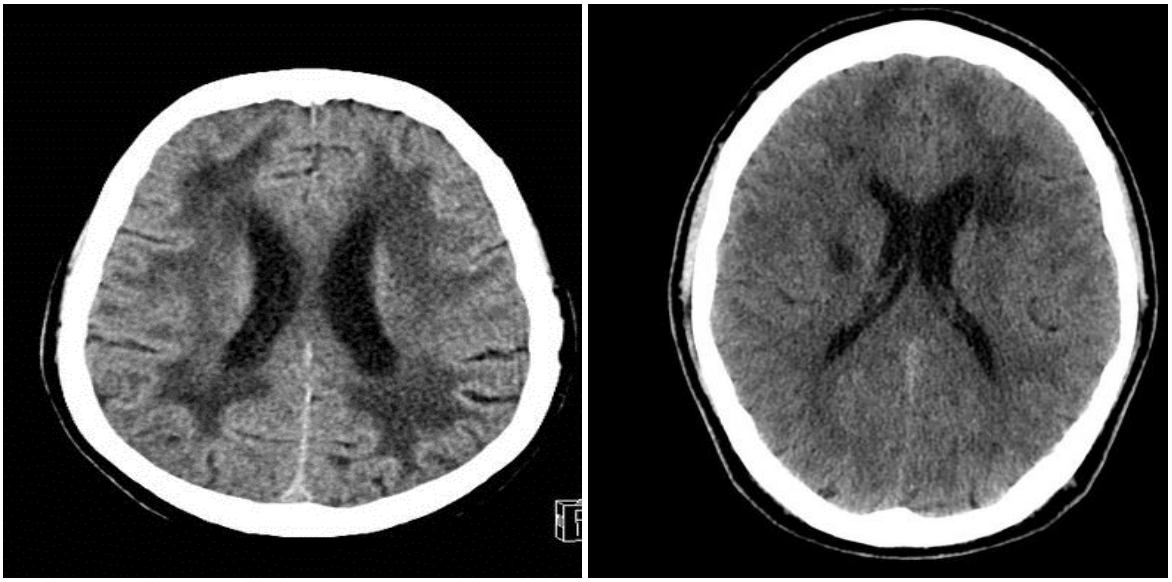
INTRODUCTION

CADASIL is a rare genetic disease that affects small blood vessels in the brain, leading to cognitive decline, stroke, and dementia. The disease is inherited in an autosomal dominant pattern, with a 50% risk of transmission to offspring.[1]

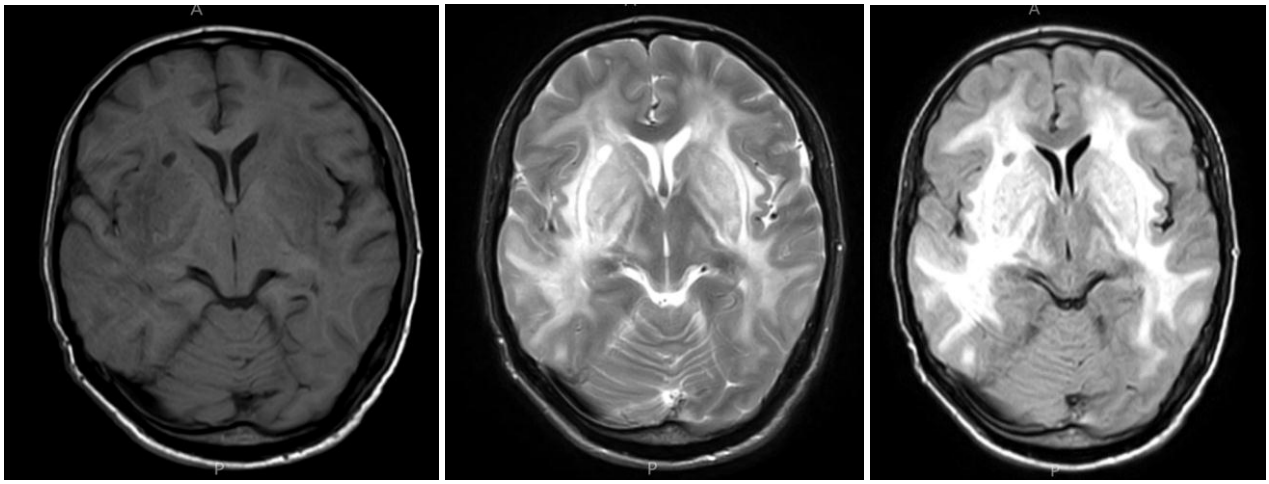
It is caused by mutations in the NOTCH3 gene, which encodes a transmembrane receptor protein that plays a critical role in vascular development and maintenance.3 The pathologic hallmark of CADASIL is accumulation of granular osmiophilic material in the basement membranes of small arteries and arterioles that causes severe fibrotic thickening and stenosis. Long penetrating cerebral arteries and their branches are especially affected. The clinical presentation of CADASIL is heterogeneous, and diagnosis is often challenging, particularly in the early stages of the disease. Radiology plays a crucial role in identifying characteristic imaging findings that aid in diagnosis and disease monitoring.

CASE PRESENTATION

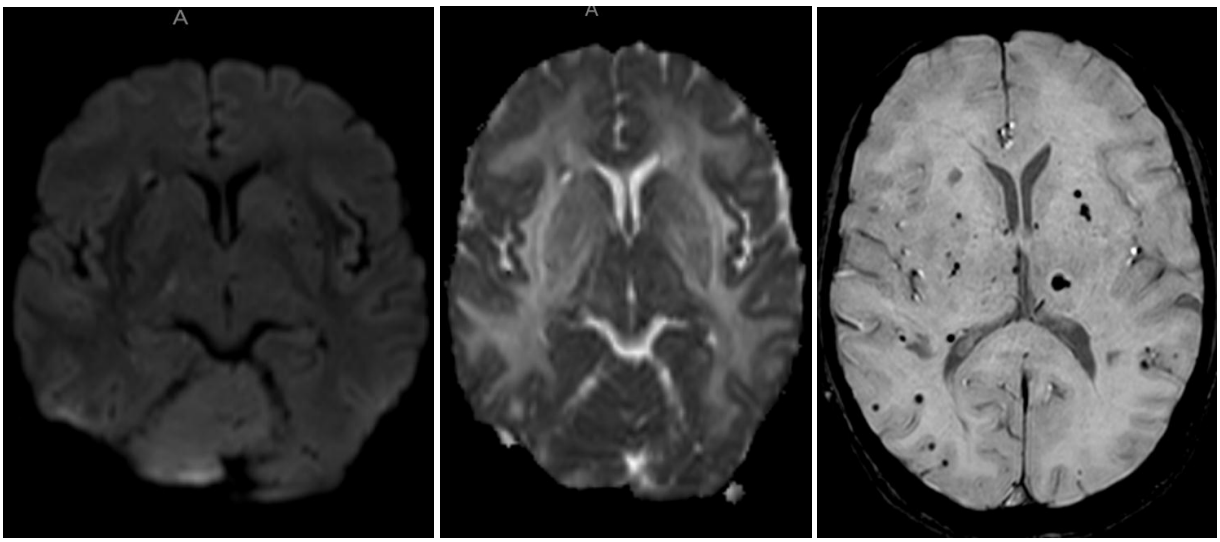
A 53-year-old male with a history of hypertension and recurrent strokes presented to our hospital with progressive cognitive decline, gait disturbance, and urinary incontinence. Neurological examination revealed mild dysarthria, gait ataxia, and bilateral upper extremity dysmetria. Magnetic resonance imaging (MRI) showed T2/FLAIR hyperintensities in bilateral periventricular, deep and subcortical white matter with a T2 hyper and FLAIR hypointense lesion in right external capsule. Multiple foci of microhemorrhages showing blooming on SWI is seen diffusely distributed in bilateral cerebral lobes. Genetic testing confirmed a NOTCH3 gene mutation, which is the hallmark of CADASIL.[6]



The non-contrast enhanced CT of the patient done at the time of presentation showed diffuse hypodensities in bilateral periventricular, deep and subcortical white matter. A hypodense lesion is seen in right internal capsule.



MRI revealed T2/FLAIR hyperintensities in bilateral periventricular, deep and subcortical white matter with a T2 hyper and FLAIR hypointense lesion in right external capsule. The lesion in right external capsule was chronic lacunar infarct.



No diffusion restriction was seen on Diffusion weighted image. Multiple foci of microhemorrhages showing blooming on SWI were seen diffusely distributed in bilateral cerebral lobes.

DISCUSSION

CADASIL is a rare and underdiagnosed disease that is often mistaken for other neurological disorders, such as multiple sclerosis or Alzheimer's disease. Radiology plays a crucial role in identifying characteristic imaging findings, such as white matter hyperintensities, lacunar infarcts, and microbleeds.[4] The sensitivity and specificity of MRI for CADASIL diagnosis are high, and MRI can also be used to monitor disease progression and treatment response. Other imaging modalities, such as transcranial Doppler ultrasound and magnetic resonance angiography, may also be useful in detecting early-stage disease and assessing cerebral blood flow abnormalities.

CONCLUSION

CADASIL is a rare genetic disease that presents with cognitive decline, stroke, and dementia. Radiology plays a crucial role in identifying characteristic imaging findings that aid in diagnosis and disease monitoring. MRI is the most sensitive and specific imaging modality for CADASIL diagnosis, and other imaging modalities may also be useful in detecting early-stage disease and assessing cerebral blood flow abnormalities. Early diagnosis and treatment can improve outcomes and prevent disease progression in patients with CADASIL.

REFERENCES

1. Chabriat, H., Joutel, A., Dichgans, M., Tournier-Lasserve, E., & Bousser, M. G. Cadasil. The Lancet Neurology, 2009; 8(7): 643-653.
2. Desmond, D. W., Moroney, J. T., Lynch, T., & Chan, S. The natural history of CADASIL: a pooled analysis of previously published cases. Stroke, 1999; 30(6): 1230-1233.
3. Joutel, A., Corpechot, C., Ducros, A., Vahedi, K., Chabriat, H., Mouton, P., ... & Tournier-Lasserve, E. Notch3 mutations in CADASIL, a hereditary adult-onset condition causing stroke and dementia. Nature, 1996; 383(6602): 707-710.
4. Lesnik Oberstein, S. A., van den Boom, R., van Buchem, M. A., van Houwelingen, H. C., Bakker, E., Vollebregt, E., ... & Ferrari, M. D. Cerebral microbleeds in CADASIL. Neurology, 2001; 57(6): 1066-1070.
5. O'Sullivan, M., Jarosz, J. M., Martin, R. J., Deasy, N., Powell, J. F., & Markus, H. S. MRI hyperintensities of the temporal lobe and external capsule in patients with CADASIL. Neurology, 2001; 56(5): 628-634.
6. Tikka, S., Mykkänen, K., Ruchoux, M. M., Bergholm, R., Junna, M., Poyhonen, M., ... & Kalimo, H. Congruence between NOTCH3 mutations and GOM in 131 CADASIL patients. Brain, 2009; 132(4): 933-939.