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Radiological Characteristics and Diagnosis of Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL): Case Report

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Abstract: ***Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a hereditary small vessel disease caused by mutations in the NOTCH3 gene. Radiological imaging plays a crucial role in diagnosing and understanding the progression of CADASIL. This manuscript presents a case of 45 years old male presenting with headache, slurring of speech and forgetfulness for last 1 month underwent MRI which shows imaging findings associated with CADASIL. With this, we will be focusing on MRI as the primary diagnostic tool.***

Keywords: Cerebral autosomal dominant arteriopathy, CADASIL, NOTCH3 gene, MRI diagnosis, hereditary small vessel disease

1. Introduction

CADASIL is the most common form of hereditary stroke disorder and is characterized by recurrent ischemic events, migraine with aura, psychiatric disturbances, and cognitive decline. CADASIL is an inherited autosomal dominant vascular dementia caused by mutation in the Notch3 gene of chromosome 19.

The pathophysiology of CADASIL involves the degeneration of vascular smooth muscle cells and the accumulation of granular osmiophilic material (GOM) within the walls of small arteries. This leads to progressive white matter damage, lacunar infarcts, and leukoencephalopathy, which are hallmark findings on neuroimaging studies. This results to progressive symptoms of transient ischemic attacks, strokes, and vascular dementia.

Clinical diagnosis of CADASIL is challenging due to its variable presentation and overlapping features with other cerebrovascular diseases. Early and accurate diagnosis is essential for managing symptoms and improving patient outcomes. Radiological imaging, particularly magnetic resonance imaging (MRI), is instrumental in diagnosing CADASIL and monitoring its progression.

2. Case Presentation

A 45 - year - old, right - handed male presented with headache, slurring of speech and forgetfulness for last 1 month. There was no medical history such as diabetes or hypertension. There was no family history of dementia or similar complaints.

Clinical examinations revealed normotensive, intact cranial nerves, decreased power over the left upper limb (4/5), deep tendon reflexes +3, normal sensory exam and normal regular and tandem gaits. Laboratory findings on admission did not reveal any significant finding.

Patient underwent MRI non contrast on 1.5 T machine. T2/FLAIR images showed widespread confluent white matter hyperintensities, predominantly in the bilateral frontal and temporal lobes. Symmetric confluent areas of white matter hyperintensity were seen in the peri - ventricular parietal, frontal and temporal regions extending from the subcortical to the periventricular region (Figure 1). All lesion shows diffusion restriction and pseudonormalization of ADC values (Figure 2). These features suggestive subacute infarcts. No stenosis on MR angiography. Venogram was also normal.

With all these characterstic findings diagnosis of CADASIL was made. Patient was managed on protocol of stoke management. He improved, was discharged, and followed up on outpatient basis.

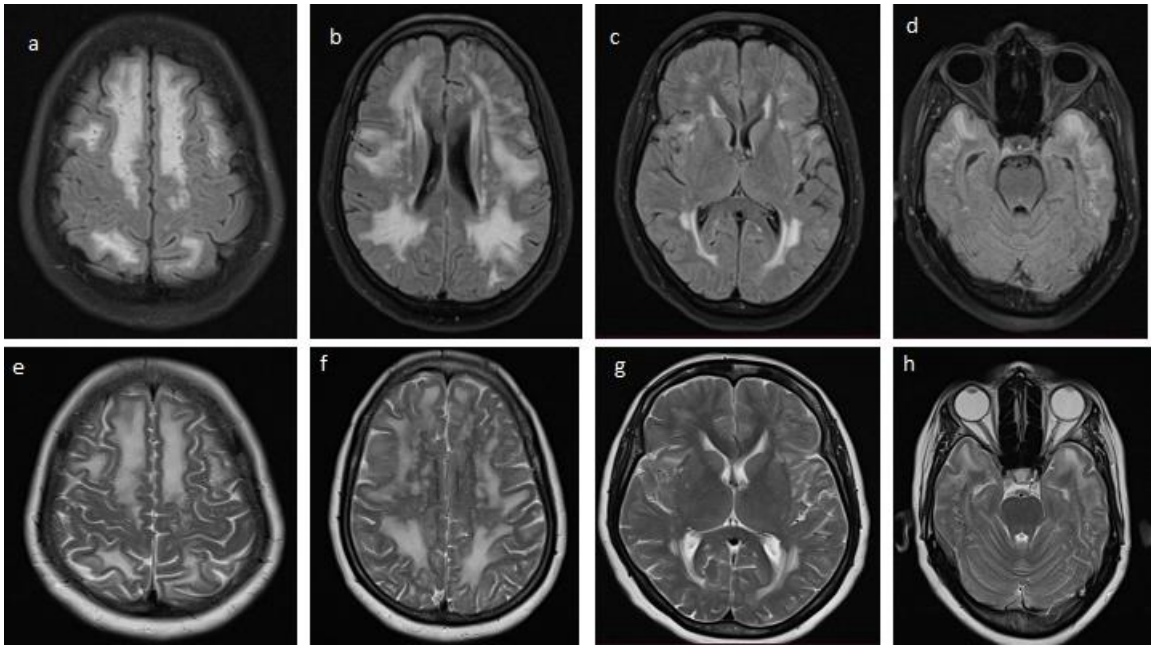

Figure 1: MRI Brain axial (a to d) - FLAIR sequences and (e to h) - T2 weighted sequences

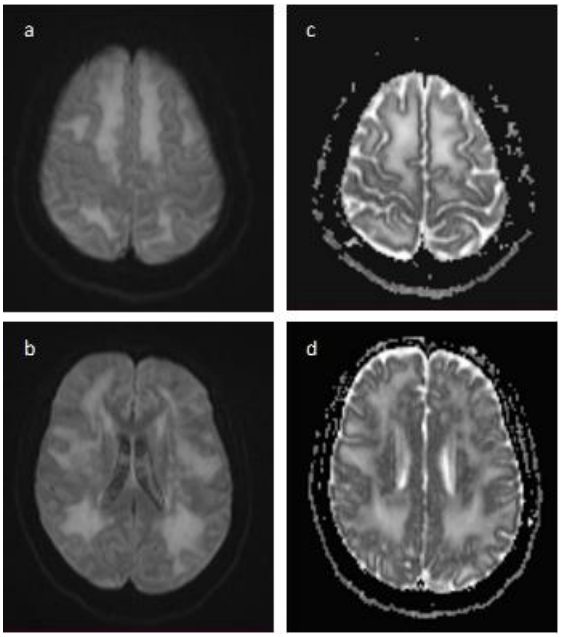

Figure 2: MRI brain axial (a - b) – DWI images and (c - d) – ADC images

3. Discussion

The presented case highlights several key aspects of CADASIL, underscoring the complexity and variability of its clinical presentation.

Radiological Features

Magnetic Resonance Imaging (MRI)
MRI is the gold standard for imaging in CADASIL due to its ability to detect early and subtle changes in brain structure. The typical MRI findings in CADASIL include:

a) White Matter Hyperintensities (WMHs):
T2/FLAIR WMHs are typically found in the periventricular, deep white matter, and subcortical regions. Characteristic regions include the anterior temporal lobes and the external capsule.
Clinical Correlation: The extent and distribution of WMHs correlate with clinical severity, cognitive impairment, and disease progression.

b) Lacunar Infarcts:
- Lacunar infarcts are commonly located in the basal ganglia, thalamus, and brainstem which appear small, cavitated lesions appear as hypointensities on T1 - weighted images and hyperintensities on T2 - weighted images.
- Clinical Correlation: The presence of multiple lacunar infarcts is associated with an increased risk of stroke and cognitive decline.

c) Cerebral Microbleeds:
- Frequently found in the thalamus, basal ganglia, brainstem, and subcortical white matter which appear are best detected using susceptibility - weighted imaging (SWI) or T2 - weighted gradient - recalled echo (GRE) sequences.
- Clinical Correlation: The number of cerebral microbleeds is correlated with the severity of the disease and risk of hemorrhagic stroke.

d) Subcortical Infarcts:
- Typically seen in the deep white matter and subcortical regions. Infarcts appear as small, cavitated, or non - cavitated lesions with signal changes on T1 and T2 - weighted images.
- Clinical Correlation: These infarcts contribute to motor and cognitive deficits seen in CADASIL patients.

e) Brain Atrophy:
Global brain atrophy, particularly involving the subcortical regions.
Detected as a generalized reduction in brain volume on MRI.
Clinical Correlation: Brain atrophy is associated with disease progression and cognitive decline.

Additional Imaging Modalities:
While MRI remains the primary imaging modality for CADASIL, other techniques can provide supplementary information:

1) Computed Tomography (CT):
- Less sensitive than MRI in detecting WMHs and lacunar infarcts.
- May be used in acute settings to rule out hemorrhage.

2) Positron Emission Tomography (PET):
Can provide metabolic information and detect changes in glucose metabolism associated with cognitive impairment.

3) Diffusion Tensor Imaging (DTI):
Offers insights into white matter integrity and connectivity, which are often disrupted in CADASIL.

4. Conclusion

Radiological imaging, particularly MRI, is critical in diagnosing and managing CADASIL. Characteristic findings such as WMHs, lacunar infarcts, cerebral microbleeds, subcortical infarcts, and brain atrophy provide valuable information for the diagnosis and monitoring of disease progression. Advances in imaging techniques continue to enhance our understanding of CADASIL, contributing to improved patient care and outcomes.

Multidisciplinary management, including neurology, cardiology, and rehabilitation services, is essential to address the diverse manifestations of CADASIL and to provide comprehensive care. Regular follow - up and supportive therapies, including cognitive rehabilitation and physical therapy, plays an integral part in managing the patient's symptoms and maintaining functional independence.

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