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Post-operative cerebral fat embolism in a patient with previously undiagnosed CADASIL

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Introduction

Ischemic stroke in young adults requires a thorough etiological investigation, particularly when traditional vascular risk factors are absent. In such cases, MRI characteristics of the acute lesion, along with the evaluation of chronic MRI changes, can provide insights into the underlying stroke mechanisms and covert comorbidities. We report a case in which a rare embolic post-surgical stroke prompted further imaging and diagnostic workup, ultimately revealing an undiagnosed genetic small vessel disease.

Clinical Presentation and Diagnostic Workup

A 39-year-old woman presented to the emergency department with generalized weakness, speech disturbance and gait instability, which appeared two days after autologous fat grafting for breast augmentation. Medical history included only hypothyroidism. On admission, she was alert but exhibited moderate ideomotor slowing, dysarthria, and right-sided hemiparesis.

Brain CT revealed multiple, bilateral supratentorial hypodensities predominantly located in the subcortical white matter.

The MRI confirmed the subacute ischemic nature of these lesions characterized by restricted diffusion and involvement of multiple vascular territories in a characteristic multifocal (so-called “starfield”) pattern, highly suggestive of a widespread embolization. Such pattern has been typically described in cerebral fat embolism.

In addition, in **Figure 1**, it can be appreciated the presence of T2 hyperintensities throughout the bilateral subcortical white matter (A), multiple and diffuse DWI-bright spots (B) and DWI-negative WMLs involving the temporal poles (arrows), the juxtacortical white matter (curved arrows), and the left external capsule (dotted arrow) (C).

These latter features were highly suggestive of CADASIL and led to genetic testing that showed the presence of a heterozygous c.1282T>C missense mutation in exon 8 of NOTCH3 gene, confirming the diagnosis.

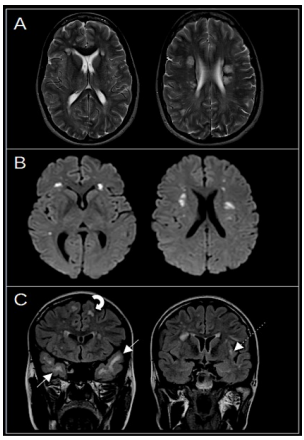

Figure 1.

Discussion and Conclusion

Cerebral fat embolism, though an infrequent sequela of trauma or surgery, manifested with radiological features classically described in literature[1].

The concomitant presence of imaging stigmata of CADASIL, subsequently corroborated by genetic analysis, delineates a singular pathological convergence.

This case compels consideration of whether chronic microangiopathy may render the brain more susceptible to embolic insult, underscoring the necessity of a nuanced approach to cerebrovascular diagnostics[2].

References:

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[2] Manini A, Pantoni L. Genetic Causes of Cerebral Small Vessel Diseases: A Practical Guide for Neurologists. Neurology. 2023 Apr 18;100(16):766-783. doi: 10.1212/WNL.0000000000201720. Epub 2022 Dec 19. PMID: 36535782; PMCID: PMC10115494.