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DOI: [10.1159/000348318](https://doi.org/10.1159/000348318)**Isolated Lacunar Infarct: An Early Clinical Presentation of CADASIL?**C. Carra-Dallière^a, X. Aygnac^a, D. Renard^c,
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A 33-year-old woman, 31 weeks pregnant, with a personal history of migraine without aura, presented with acute right-sided hemiparesis and allodynia. Her mother, who had high blood pressure, showed recurrent lacunar strokes at the age of 57, without any other explanation.

Clinical examination revealed right-sided proportional hemiparesis, brisk deep tendon reflexes on the right side and normal blood pressure. Brain MRI showed a left capsular and thalamic infarction of 12 mm diameter on diffusion-weighted images, with an absence of white-matter abnormalities on FLAIR and T2 sequences (fig. 1) or microbleeds on gradient echo imaging. Transesophageal echocardiography, intra- and extracranial MRI angiography, and ECG were normal. Screening for antiphospholipid antibody syndrome and autoimmune diseases was negative. Because of the absence of cardiovascular risk factors, a genetic condition was suspected. Screening of NOTCH 3 gene was performed and revealed a pathogenic mutation in exon 22 (c.3769C>T).

Comment

To our knowledge, this is the first report of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) revealed by an isolated lacunar infarct, with an absence of coexisting white-matter changes. CADASIL is an inherited small vessel disease, with clinical manifestations confined to the central nervous system, usually characterized by five

Fig. 1. Brain MRI of the patient. **a, d** Left subacute lacunar capsular and thalamic infarct shown on diffusion-weighted brain MRI. **b, c, e, f** Absence of white-matter changes on fluid-attenuated inversion recovery images.

main symptoms, i.e. migraine with aura, subcortical ischemic events, mood disturbance, apathy and cognitive impairment [1]. Diagnosis is supported by MRI findings including diffuse white-matter hyperintensities on FLAIR or T2-weighted images, lacunar infarcts and microbleeds [2]. White matter hyperintensities presumably appear during the third decade and are constant after 40 years. They progressively become confluent and diffuse.

In our report, CADASIL is likely the cause of the lacunar infarct. CADASIL is an increasingly diagnosed monogenic cause of lacunar infarct. It has been reported to account for 11% of cases of lacunar infarcts with coexisting leukoencephalopathy in patients younger than 50 years [3]. However, in the absence of leukoencephalopathy, screening of CADASIL in patients with lacunar infarct is considered not worthwhile [3]. In a recent study, among 30 patients under 65 years of age presenting with cerebral ischemia of the lacunar subtype without hypertension or diabetes mellitus, 2 had pathogenic mutations of the gene encoding for CADASIL [4]. Both of these patients, however, showed white-matter involvement in the anterior temporal pole.

In recent years, the phenotypic spectrum of CADASIL with paucisymptomatic presentation has been widening. Our report suggests that screening for CADASIL may be worth considering in young patients with unexplained lacunar infarct in the absence of cardiovascular risk factors, even with a lack of coexisting white-matter changes.

References

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