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Recurrent spontaneous intracerebral hemorrhage in CADASIL patient; A case report

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Background

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a rare hereditary, autosomal dominant, cerebral small vessel disease caused by mutations in the NOTCH3 gene. The disease is clinically characterized by migraine, subcortical ischemic events, psychiatric disorders, and cognitive impairment eventually leading to dementia and disability. While CADASIL is considered as a primarily ischemic form of vascular dementia, spontaneous intracerebral hemorrhage (ICH) has recently been reported in association with CADASIL. In this study, we report CADASIL patient presenting with recurrent ICH.

Case description

A 39-year-old right-handed man was admitted to our hospital with chief complaint of right side motor weakness and motor aphasia. He also showed pseudobulbar affect in which he showed uncontrollable episodes of laughing in inappropriate situation. Initial computed tomography (CT) scan showed a left thalamic ICH (Figure 1A). History taking revealed that this was his second attack that he had developed a spontaneous hematoma in the right basal ganglia five months ago and recovered without prominent neurological deficit after weeks of conservative care. He had had no specific underlying disease or trauma history previously, except that he had family history of stroke. His guardian reported that his mother, maternal aunt and uncle had cerebral stroke and disability in their late 40s (Figure 2). At this hospitalization, his initial blood pressure was 160/110 mmHg and anti-hypertensive medication was taken thereafter. All the other clinical and laboratory examination including HbA1c, coagulation profile, and lipid profile except for triglyceride level were normal. Considering relatively young age, identification of the underlying cause of stroke was essential to prevent further strokes. For thorough evaluation of brain parenchyma, brain magnetic resonance imaging was performed. MRI scans documented chronic ischemic change, leukomalacia in both periventricular white matter and microbleeds at both basal ganglia and thalamus (Figure 1B, 1C). Due to the characteristic MRI imaging, family history, and emotional incontinence, the suspicion of CADASIL was raised, and genetic testing was conducted, revealing heterozygous mutation of the NOTCH3 gene (c.994C>T mutation in exon 6), which leads to an arginine-to-cysteine substitution (p.Arg332Cys). The Result of genetic testing was consistent with the diagnosis of CADASIL.

Conclusion

CADASIL can manifest with atypical clinical findings suggesting that CADASIL should always be considered in the differential diagnosis of young patient with recurrent ICH, mood disorder, and positive family history.

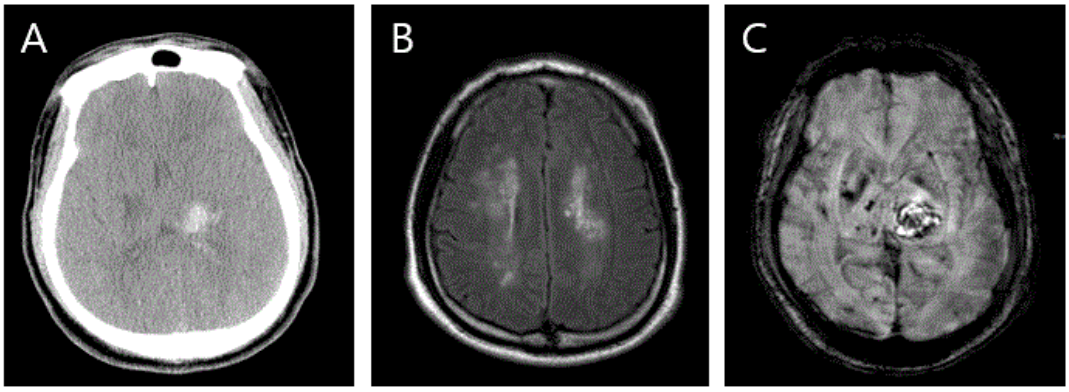

Figure 1. CT and MRI findings in CADASIL patient (A) CT scan shows intracerebral hemorrhage in left thalamus (B) MRI (T2 weighted image) shows chronic ischemic change and leukomalacia in both periventricular white matter (C) MRI (Susceptibility Weighted Image) shows several microbleeds at both basal ganglia and thalamus

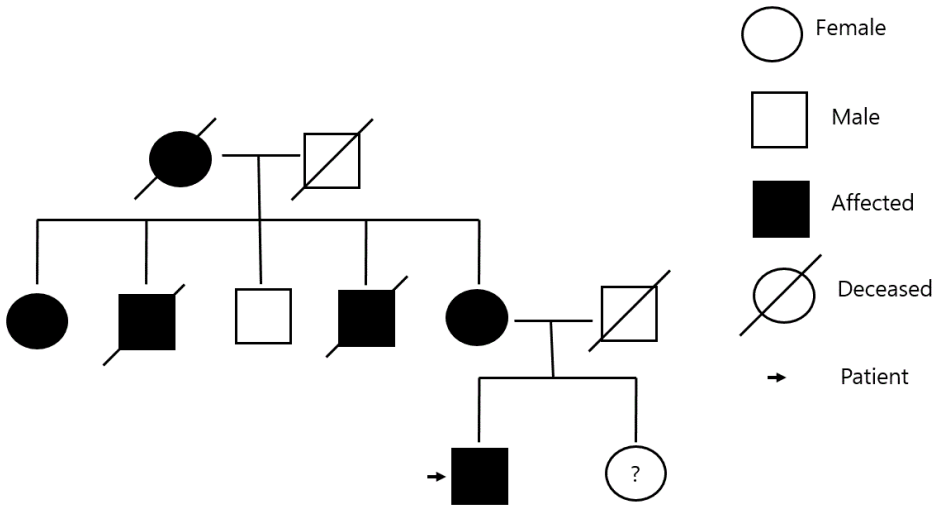

Figure 2. Pedigree tree of CADASIL patient