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Case Report

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Unusual stroke cause: bilaterally fornix infarction in a patient with biotinidase deficiency

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Learning points for clinicians

Fornix is a white matter tract bundle that is a substantial component of the Papez Circuit and has memory and cognitive functions. Isolated bilateral fornix infarction is uncommon, and the clinical presentation is memory impairments in general. Most cases are accompanied by vascular comorbidities such as pulmonary embolism and peripheral venous thrombosis. Here, we presented an unusual case of bilaterally fornix infarction with biotinidase deficiency and presenting with paraparesis and acute-onset anterograde amnesia.

Introduction

Isolated bilateral fornix infarction is very rare in patients presenting with stroke. Very few cases have been defined in this regard, in the literature. Fornix is one of the most significant memory-related structures in the brain. Therefore, in almost all of these cases, the clinical presentation is amnesia.[1–3] In our report, a 20-year-old female patient admitted to the emergency department with paraparesis and tingling in her feet was contrary clinically from the other cases in the literature with bilaterally fornix infarction. Acute onset of anterograde amnesia also accompanied the clinic. In most cases, vascular etiology has been accused due to presence of comorbidities such as pulmonary embolism and peripheral venous thrombosis.[1] In here we reported an infrequent case of isolated bilaterally fornix infarction with biotinidase deficiency and presenting with paraparesis and acute-onset anterograde amnesia. To our knowledge, such presentation of biotinidase deficiency is a first in English literature.

Case report

A 20-year-old female patient was admitted to the emergency department with paraparesis and tingling in her feet. Acute onset of anterograde amnesia was also present in the patient. The patient had a history of she had been diagnosed with biotinidase deficiency 2 months ago. She declared that oral biotin treatment had been initiated, but she did not pay attention to the treatment. There was no history of fever, vomiting or nausea. She did not also have meningeal irritation findings. On physical examination, there were loss of feeling and weakness on her both lower limbs. Her body temperature and other vital signs were normal. She had low levels of biotin. However, other laboratory examinations were normal, including the complete blood cell count, serum electrolytes and liver function tests. Magnetic resonance (MR) imaging and diffusion-weighted imaging (DWI) scans were performed to exclude cerebrovascular accident. MR images showed a thickened cerebral fornix and increased signal in bilateral fornix. DWI revealed an isolated bilateral fornix infarction (Figure 1). In subsequent clinical studies, biotinidase deficiency was detected in the patient. Biotinidase enzyme activity was 2.51% (normal range 8–10%) and biotinidase level was 1.15 mU/ml (normal range 6.9–9.45 mU/ml) to the laboratory estimates. Based on these clinic, laboratory and radiological findings, bilateral isolated fornix infarction associated with late-onset biotinidase deficiency was considered in the patient.

Discussion

Biotinidase deficiency is an autosomal recessive metabolic disorder that may present with severe neurological and cutaneous symptoms in early childhood.[4] Following the oral free biotin administration, symptoms are greatly improved and delay in initiation of the treatment may cause irreversible complications such as optic atrophy and sensorineural hearing loss.[4,5] Delayed-onset form of the disease manifested by the symptoms such as spastic tetraparesis, paraparesis and scotomas may occur in adulthood, although very rarely, and it may be difficult to recognize the disease clinically. In this form, spinal cord or cerebral lesions may also be encountered in MR and this evidence is uncommon also.[6]

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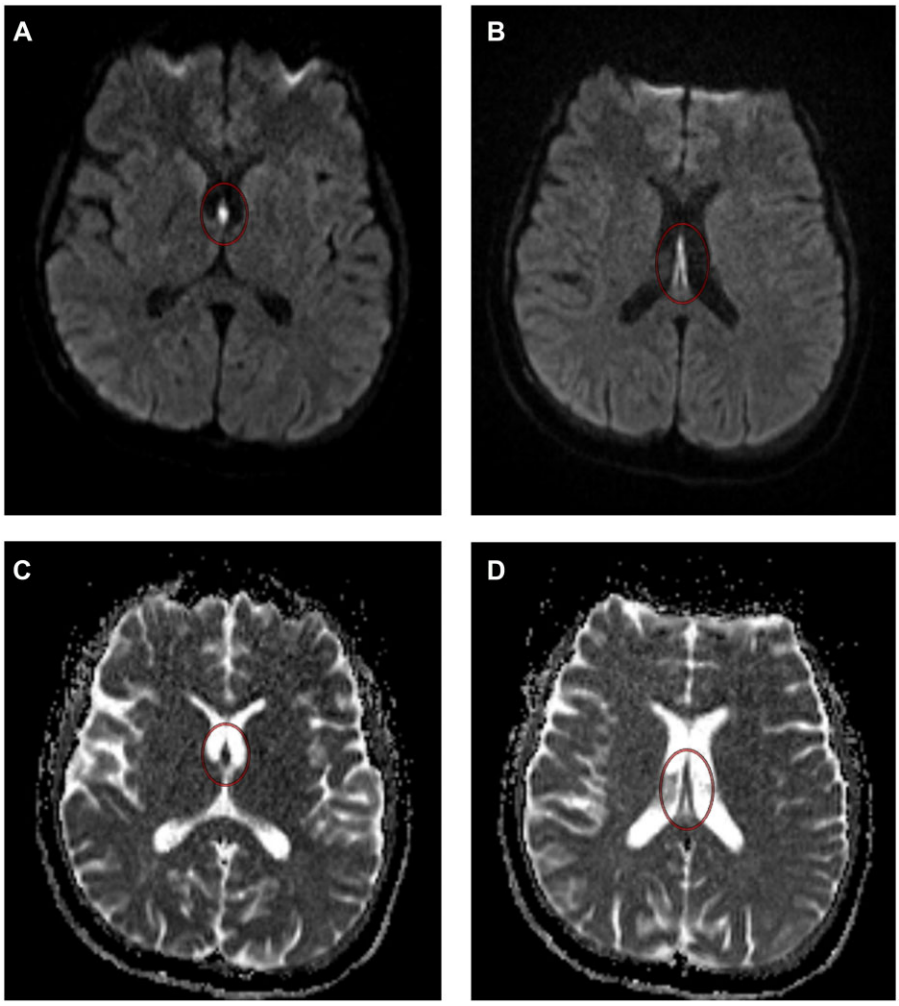

Figure 1. Consecutive axial diffusion-weighted images (**A** and **B**) show increased signal intensities (circle) in the bilateral cerebral fornix. Apparent diffusion coefficient (ADC) maps (**C** and **D**) demonstrate low signal values (circle) compatible with restricted diffusion in the bilateral cerebral fornix.

Bilateral isolated fornix infarction is an extremely rare form of the cerebral infarction. Occlusion in the perforating branches of anterior communicating artery (ACOA) is suggested to be the main cause in the etiology. Because fornix is one of the important structures of the memory formation loop, clinical manifestations are cognitive changes and memory disturbances such as acute amnestic syndrome, transient global amnesia, Korsakoff syndrome and long-term amnesic syndrome.[6] Paraparesis may be a rare clinical manifestation as in our case. However, often fornix infarction is associated with infarction of other structures such as cingulated cortex, septum pellucidum, corpus callosum and isolated cases are infrequent.[2,6,7] These anatomical structures are mostly fed by the subcallosal artery, which is the branch of ACOA, and rarely anterior cerebral artery.[1,8,9] In the majority of cases, vascular etiology has been accused due to the presence of comorbidities such as pulmonary embolism, peripheral venous thrombosis and myocardial ischemia. Nevertheless, biotinidase deficiency may accompany the clinic rarely, as in our case.

In conclusion, late-onset biotinidase deficiency in adulthood is extremely rare. Bilateral isolated fornix infarction may accompany the disease unfrequently, and patients may present with unusual clinical manifestations such as acute-onset anterograde amnesia and paraparesis.

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Conflict of interest

The authors declare that they have no conflict of interest to the publication of this article.

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