

A case report of fatal familial insomnia with cerebrospinal fluid leukocytosis during the COVID-19 epidemic and review of the literature

Zheng Wang , Yueqi Huang, Shuqi Wang, Jiefang Chen, Gesang Meiduo, Man Jin, and Xiaoying Zhang

Affiliated Mental Health Center & Hangzhou Seventh People's Hospital, Zhejiang University School of Medicine, Hangzhou, China

ABSTRACT

Fatal familial insomnia (FFI) is a rare autosomal dominant genetic neurodegenerative disease. Generally, FFI patients will develop rapidly progressive dementia, sleep disturbance, autonomic dysfunction, and so on. Cerebrospinal fluid examination of FFI patients normally shows no obvious abnormalities. Here, we report a young male patient who was diagnosed with FFI during the COVID-19 epidemic. Clinical symptoms include psychobehavioral abnormality, cognitive decline, sleep disturbance, and autonomic dysfunction. No abnormalities were found in routine examinations after admission. However, the number of white blood cells in the cerebrospinal fluid increased. Though the patient was treated with anti-infection and immunotherapy, the symptoms were not relieved. A lumbar puncture was performed again, and it was found that the total Tau protein in the cerebrospinal fluid was elevated, and PET results showed that brain metabolism decreased. Finally, a genetic test was used to confirm the diagnosis of FFI. This case suggests that patients with FFI may also have elevated white blood cells in cerebrospinal fluid and timely detection of Tau protein in cerebrospinal fluid is helpful for early identification of FFI. And precise diagnosis relies on genetic testing.

ARTICLE HISTORY

Received 4 August 2023
Revised 8 November 2023
Accepted 19 December 2023

KEYWORDS

Clinical features; CSF analysis; fatal family insomnia; PET-CT; PRNP; Tau protein

Introduction

FFI, a rare autosomal dominant disease, is associated with the D178N-129 M missense mutations of the prion protein gene (PRNP) on chromosome 20, and the main pathological feature is scrapie prion protein (PrP^{Sc}) deposition in the brain [1,2]. The average onset age of FFI is about 51 years old, and the course of the disease ranges from 8 to 72 months [3]. Clinical symptoms of FFI patients are variable. Typically, FFI patients are accompanied with rapidly progressive dementia, sleep disturbance, autonomic dysfunction, and so on. Eventually, it will lead to death [4].

In this article, we report one case of a young male patient diagnosed with FFI who had been vaccinated against COVID-19 during the COVID-19 pandemic. This patient had no positive family history but with severe sleep disturbances, progressive cognitive decline, significant changes in autonomic nervous function, and psychobehavioral abnormality. Early detection of cerebrospinal fluid showed increased white blood cells. Multiple magnetic resonance imaging (MRI) and electroencephalography (EEG) examination showed no abnormalities, and the effects of anti-infection and immunotherapy treatment were poor. Finally, the patient was diagnosed with FFI by genetic testing.

Case presentation

The patient, a 28-year-old courier, was admitted to the hospital in July 2021, because of 'Memory impairment and psychotic behaviour disorder for more than one month'. The patient suffered from memory loss one month before admission without obvious incentives. He often made repeated mistakes at work and forgot whether he had eaten. There are often involuntary movements of limbs and frequent changes in his body position during sleep. Later, hallucinations and delusion gradually appeared. When his brother was not at home, he claimed to have seen him. He also said that his child was adopted, and he had another one. The patient had seen a doctor in another hospital before coming to our department. He was diagnosed with 'anxiety disorder' and was treated with 'Olanzapine Tablets 2.5 mg/day'. After treatment, the patient's sleep quality has slightly improved, but he often moves his hands and feet around during sleep and tears the quilt with his hands and teeth.

When admitted to the hospital, the patient's body temperature was normal, his consciousness was clear, his facial expression was stolid, and his feeling of time orientation was not good. His short-term memory was impaired. He could not recall three things (ball, flag,

CONTACT Xiaoying Zhang  xyzhong004@163.com  Department of Neurology, Hangzhou Seventh People's Hospital, 305 Tianmushan Road, Hangzhou 310013, China

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