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Case report

Adult-onset hemophagocytic lymphohistiocytosis type 2 presenting as a demyelinating disease

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ABSTRACT

Familial hemophagocytic lymphohistiocytosis (HLH) is a rare autosomal recessive life-threatening multisystem inflammatory disorder. It is characterized by excessive production of cytokines and uncontrolled activation of lymphocytes and macrophages leading to widespread organ infiltration and tissue destruction. Central nervous system involvement is common occurring in approximately 75% of patients. The neurological symptoms often develop during the course of the disease. However, they can be the initial presenting manifestation. In this article, we describe a patient with adult-onset familial HLH who presented solely with neurological involvement with lack of the initial classical presentation of HLH. He was initially misdiagnosed as acute disseminated encephalomyelitis and later on as multiple sclerosis. This paper indicates that familial HLH may present with pure neurological involvement. A high index of suspicion should be practiced with patients who present with vague recurrent neurological symptoms associated with abnormal non-specific neuroradiological findings. Genetic testing should be included in the investigations of such cases. Steroids and plasma exchange are non-specific therapies that may mask certain conditions including HLH or may be part of the treatment regimen. This may cause a delay in the diagnosis of the underlying causative disease.

1. Introduction

Familial hemophagocytic lymphohistiocytosis (HLH) is a rare life-threatening multisystem inflammatory disorder. It is characterized by excessive production of cytokines and uncontrolled activation of lymphocytes and macrophages leading to widespread organ infiltration and tissue destruction (Elyamany et al., 2016). This disorder is inherited in an autosomal recessive manner. The disease mainly affects young children who present with prolonged fever (>7 days), lymphadenopathy, cytopenias, hepatosplenomegaly, coagulopathy, and infiltration of various organs with benign macrophages that exhibit hemophagocytosis (Esteban et al., 2017). Central nervous system involvement is common occurring in approximately 75% of patients and associated with poor outcome. The neurological symptoms often develop during the course of the disease. However, they can be the initial presenting manifestation. Neurological manifestations may include seizures, ataxia, spastic paraparesis, or even progressive and fatal encephalitis (Cai et al., 2017; Feldmann et al., 2005; Rostasy et al., 2004). In this article, we describe a patient with adult-onset familial HLH who presented solely with neurological involvement with lack of the initial classical presentation of HLH. He was initially misdiagnosed as acute disseminated encephalomyelitis (ADEM) and later on as multiple sclerosis.

2. Case report

A 20-year-old previously healthy male presented to the clinic with painless decrease in vision bilaterally (left more than right) and headache. He had no previous history of visual symptoms, sensory symptoms, walking difficulties, or sphincter control problems. In addition, there were no systemic manifestations including arthritis, fever, night sweat, weight loss, mouth or genital ulcers, and no risk factors for HIV. His parents are non-consanguineous. His family history was remarkable for Wegener's granulomatosis in his brother. His systemic examination including vital signs, eyes, chest, abdomen, and joints was normal. Neurologically, he was conscious and oriented with normal higher mental function. His cranial nerve examination was unremarkable apart from reduced visual acuity in both eyes to finger movement. The rest of his neurological examination including sensory and motor systems, coordination, and gait were all unremarkable. Magnetic resonance imaging (MRI) of the brain showed multiple confluent white matter demyelinating lesions in both cerebral hemispheres mostly involving

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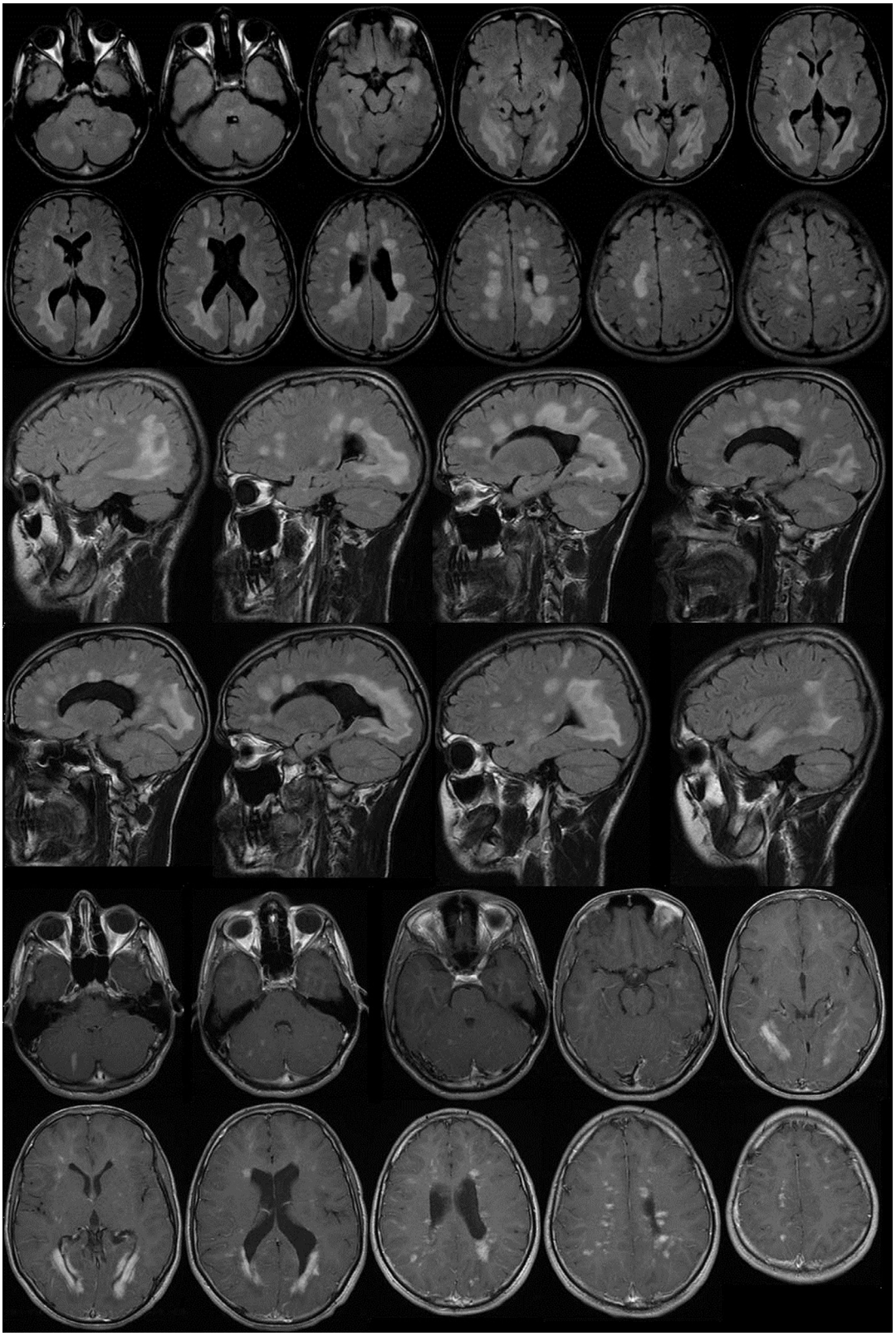

Fig. 1. FLAIR MRI images of the brain showing multiple confluent white matter demyelinating lesions in both cerebral hemispheres (mostly involving the parietal and occipital lobes), corpus callosum, cerebellar hemispheres, and dorsal pons. These lesions demonstrated nodular and linear enhancement in contrast-enhanced T1 images.

the parietal and occipital lobes. In addition, there was demyelinating changes in the corpus callosum, cerebellar hemispheres, and dorsal pons. These lesions demonstrated nodular and linear enhancement (Fig. 1). The spinal cord was absolutely normal. Viral serology was negative including hepatitis, human immunodeficiency virus, cytomegalovirus, Epstein–Barr virus, and herpes viruses. Tumor markers including carcinoembryonic antigen, alpha-fetoprotein, CA-125, CA19-9, and prostate-specific antigen were unremarkable. Cerebrospinal fluid