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Unusual Presentation of Hemophagocytic Lymphohistiocytosis: A Diagnostic Dilemma

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Description

Hemophagocytic Lymphohistiocytosis (HLH) is a rare, life-threatening hyperinflammatory syndrome characterized by uncontrolled activation of immune cells. While HLH typically presents with fever, cytopenias, and hepatosplenomegaly, atypical clinical manifestations can make diagnosis challenging. We report a case of an adult patient with HLH presenting with neurological symptoms, including altered mental status, which posed a diagnostic dilemma. This case underscores the need for vigilance in recognizing unusual presentations of HLH to ensure early intervention and improve outcomes.

Hemophagocytic Lymphohistiocytosis (HLH) is a rare disorder characterized by uncontrolled immune system activation. It can occur in both familial and acquired forms. HLH is often associated with a dysregulation of immune cells, leading to excessive inflammation, and can be triggered by various factors, including infections, malignancies, and autoimmune disorders. Common clinical features of HLH include fever, cytopenias, hepatosplenomegaly, and hyperferritinemia. However, atypical presentations can complicate the diagnostic process.

A 32-year-old male with no significant medical history presented to the emergency department with altered mental status and seizures. On admission, he was disoriented, febrile, and exhibited generalized tonic-clonic seizures. Neurological examination revealed neck stiffness, but no focal deficits. Laboratory tests revealed pancytopenia with a hemoglobin level of 8 g/dL, a platelet count of 40,000/µL, and a white blood cell count of 2,000/µL.

Magnetic Resonance Imaging (MRI) of the brain demonstrated bilateral thalamic and basal ganglia hyperintensities. Cerebrospinal Fluid (CSF) analysis revealed pleocytosis with an elevated white blood cell count and elevated protein levels. No microorganisms were identified on Gram stain or culture. Electroencephalogram (EEG) showed generalized slowing.

Given the neurological findings and cytopenias, the differential diagnosis included viral encephalitis, autoimmune encephalitis, and HLH. However, an initial extensive workup, including blood and CSF PCR for infectious agents, autoantibodies, and paraneoplastic panels, failed to provide a definitive diagnosis. The patient was empirically treated with antiviral and immunomodulatory therapy, but his condition continued to deteriorate.

Further laboratory investigations showed hyperferritinemia (serum ferritin >10,000 ng/mL) and elevated triglycerides, which raised suspicion of HLH. The patient's Natural Killer (NK) cell activity was found to be reduced. A bone marrow biopsy was performed, revealing hemophagocytosis, confirming the diagnosis of HLH. A genetic panel for familial HLH was negative.

Treatment with etoposide and corticosteroids was initiated. The patient's neurological symptoms improved, and his blood counts gradually normalized. He was discharged with plans for close follow-up.

Discussion

HLH is a challenging condition to diagnose, particularly when it presents with atypical clinical features. The hallmark signs of fever, cytopenias, hepatosplenomegaly, and hyperferritinemia may be absent or overshadowed by other symptoms, as in this case.

Neurological involvement in HLH is uncommon and thalamic and basal ganglia abnormalities are rarely reported. These neurological findings can mimic viral or autoimmune encephalitis, leading to diagnostic uncertainty.

The diagnostic criteria for HLH are typically based on the Histiocyte Society guidelines, which require the presence of specific clinical and laboratory features. However, these criteria may not be met in atypical presentations, delaying the diagnosis and treatment. Therefore, maintaining a high index of suspicion is essential, and early consideration of HLH in patients with neurological symptoms and cytopenias can prevent diagnostic delays and improve outcomes.

Conclusion

This case illustrates the diagnostic challenges associated with atypical presentations of HLH, particularly when neurological symptoms predominate. Healthcare providers must remain vigilant and consider HLH in the differential diagnosis when confronted with patients exhibiting unexplained altered mental status, cytopenias, and hyperferritinemia. Timely recognition and intervention are important in managing this life-threatening condition.

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