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Hemophagocytic Lymphohistiocytosis: A Case Report

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Abstract: Hemophagocytic lymphohistiocytosis (HLH) is a rare but potentially fatal hyperinflammatory syndrome characterized by excessive immune activation. The condition may be genetic (primary HLH) or secondary to infections, malignancies, or autoimmune diseases. Early diagnosis and prompt treatment are essential to prevent multi-organ failure and mortality. We present a case of HLH diagnosed based on clinical and laboratory criteria, highlighting the diagnostic challenges, treatment approach, and patient outcome.

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I. INTRODUCTION

Hemophagocytic lymphohistiocytosis (HLH) is a life-threatening syndrome of excessive immune activation leading to severe inflammation and multi-organ dysfunction. It may be **primary** (familial HLH due to genetic mutations) or **secondary** (triggered by infections, malignancies, or autoimmune conditions). HLH remains a diagnostic challenge due to its nonspecific symptoms, which can mimic severe infections or malignancies. The HLH-2004 diagnostic criteria help identify cases, but a high index of suspicion is required for timely diagnosis and treatment.

II. CASE PRESENTATION

A 45-year-old female presented with **persistent fever, fatigue, and weight loss** for 7 days. On examination, the patient had **hepatosplenomegaly and generalized lymphadenopathy**. Laboratory tests showed:

- **Cytopenia**: Hemoglobin: 8.7 g/dL, WBC: 2.7 $\times 10^9$/L, Platelets: 14 $\times 10^9$/L
- **Hyperferritinemia**: 34088 ng/mL
- **Hypertriglyceridemia**: 371 mg/dL
- **ANA profile:** Positive for nRNP, SM, SS-A, RIB
- **Bone marrow biopsy**: Evidence of hemophagocytosis

The patient met the HLH-2004 criteria, confirming the diagnosis. Given the severity, treatment was initiated with IV pulse steroid therapy in the form of Injection Methylprednisolone 500mg OD for 5 days followed by oral Tab. Prednisolone 1mg/kg along with antibiotic coverage. Supportive care included antimicrobial prophylaxis and organ function monitoring. Despite initial complications, the patient showed **clinical and biochemical improvement** with therapy.

III. DISCUSSION

HLH is a **hyperinflammatory syndrome** triggered by **defective immune regulation**, leading to excessive cytokine release ("cytokine storm"). The hallmark features include fever, cytopenia, hepatosplenomegaly, and laboratory markers of immune dysregulation. **The HLH-2004 criteria remain the gold standard** for diagnosis, requiring at least five of the following:

- Fever
- Splenomegaly
- Cytopenia in ≥2 cell lines
- Hypertriglyceridemia or hypofibrinogenemia
- Hemophagocytosis in bone marrow, spleen, or lymph nodes
- Hyperferritinemia (>500 ng/mL)
- Elevated soluble IL-2 receptor
- Absent/low NK cell activity

- **Primary HLH** is caused by mutations in genes like PRF1, UNC13D, or STXBP2, often manifesting in childhood.
- **Secondary HLH** is triggered by infections (e.g., EBV, CMV), malignancies (e.g., lymphoma), or autoimmune diseases (e.g., systemic lupus erythematosus).

Treatment is based on the HLH-94 and HLH-2004 protocols, which include corticosteroids, etoposide, and cyclosporine. In refractory cases, newer biologics like **emapalumab (anti-IFN-γ monoclonal antibody)** or hematopoietic stem cell transplantation (HSCT) may be needed.

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IV. CONCLUSION

HLH is a life-threatening syndrome requiring **early diagnosis and aggressive treatment**. This case underscores the importance of recognizing HLH in patients with prolonged fever, cytopenia, and organomegaly. Timely intervention can improve survival outcomes.

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