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A CASE OF ATYPICAL PRESENTATION OF HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS

General Medicine

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KEYWORDS

INTRODUCTION

Hemophagocytic lymphohistiocytosis (HLH) is a rare and life-threatening syndrome of immune system dysregulation characterized by the phagocytosis of various cells by histiocytes in the bone marrow. HLH is divided into primary or genetic HLH and secondary HLH. Secondary or acquired forms of HLH occur in association with infections, autoimmune diseases, malignant diseases, immune deficiencies. Both sporadic and familial cases are often precipitated by acute infections.

HLH usually presents as an acute febrile illness associated with multiple organ involvement in the form of hepatitis, renal failure, ARDS, encephalitis, seizures ,skin changes such as rashes, purpura, petechiae.

Hlh-2004 Criteria

At least five of the following eight should be present:

1. Fever ≥38.5°C
2. Splenomegaly
3. Peripheral blood cytopenia, with at least two of the following: hemoglobin <9g/dL; platelets <100,000/microL; absolute neutrophil count <1000/microL
4. Hypertriglyceridemia (fasting triglycerides >265 mg/dL) and/or hypofibrinogenemia (fibrinogen <150 mg/dL).
5. Hemophagocytosis in bone marrow, spleen, lymph node, or liver
6. Low or absent NK cell activity
7. Ferritin >500 ng/mL
8. Elevated soluble CD25 > 2400U/ml

CASE

A 50 year old female presented with complaint of high grade fever with chills and rigors since 3 months associated with weight loss and anorexia with no significant past history of any major disease or hospitalization.

Her mother and father had history of TB and some malignancy.
Patient's following work up was done:

- CBC - Hb - 6.6gm/dl, WBC – 2600 cells/dl, Platelets – 94000 cells/dl Retic count – 0.5, ESR - 12
- RFT and S.Bilirubin – within normal limit
- AST - 186 U/L, ALT – 139U/L
- Fasting triglyceride – 572mg/dl
- S. Ferritin 1063ng/ml
- S. Fibrinogen – 299mg/dl
- Serum ANA, RA , ASO negative
- Coombs test – Negative
- Cultures for blood, urine and sputum were Negative
- Investigations for brucellosis, pulmonary TB, dengue, malaria ,EBV, infectious hepatitis, HIV were negative.
- Radiological screening of abdomen KUB pelvis and thorax showed mild hepatosplenomegaly with mild ascites.
- 2D ECHO within normal limit.
- Bone marrow aspiration and biopsy was S/O normocellular marrow with evidence of HEMOPHAGOCYTOSIS.
- Fungal KOH and culture and geneXpert for M. Tuberculosis were negative.
- Pt was given supportive treatment with antipyretics, IV antibiotics, IV fluids and was discharged with oral steroids(tablet prednisolone 1mg/kg) with stable vitals.

CONCLUSION

Most patients with HLH are acutely ill with multiorgan involvement, cytopenias, liver function abnormalities and neurologic symptoms. This case presented with slow and smoldering course as exception to normal acute and sudden deteriorating course of the disease. It fulfilled 6 out of 8 criteria of HLH and score was 239. HLH should be suspected in an infant, child, or adult with unexplained cytopenias, hepatitis, or inflammatory central nervous system findings who has unexplained fever, hepatosplenomegaly, prior HLH-like episodes, a family history of HLH, or a known genetic disorder associated with HLH. Prompt treatment is critical, but the greatest barrier to a successful outcome is often a delay in diagnosis due to the rarity of this syndrome, variable clinical presentation, and lack of specificity of the clinical and laboratory findings.

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