Hemophagocytic Lymphohistiocytosis Triggered by Systemic Mastocytosis

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INTRODUCTION

Hemophagocytic Lymphohistiocytosis (HLH) is a rare, inappropriate immune response causing excessive inflammation and multiorgan failure if left untreated.

CASE PRESENTATION

A 79-year-old female presents with fatigue and asymptomatic hyperbilirubinemia for 6 months. She also developed anemia and thrombocytopenia.

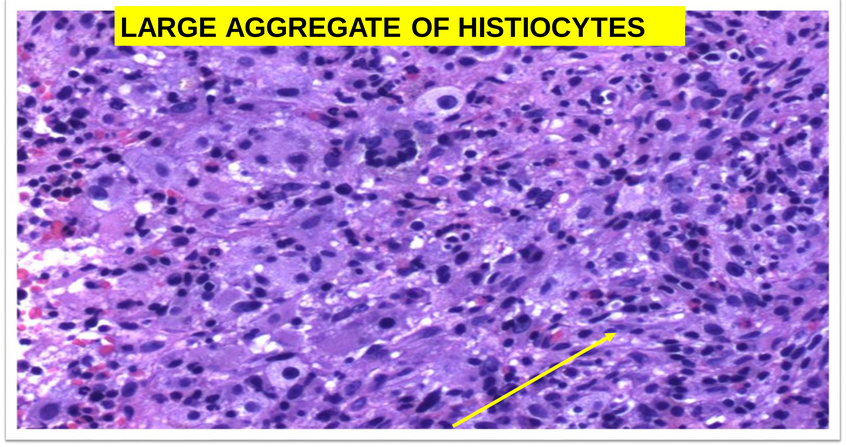
DIAGNOSTICS

- Bone marrow biopsy: SM with hematological neoplasm and large aggregates of histiocytes.
- Molecular testing: KIT D816V mutation ++
- Elevated: Ferritin, CRP, CXCL9, IL-18, soluble IL-2 levels
- Low: Tryptase levels
- CT Abdomen: Splenomegaly

TREATMENT

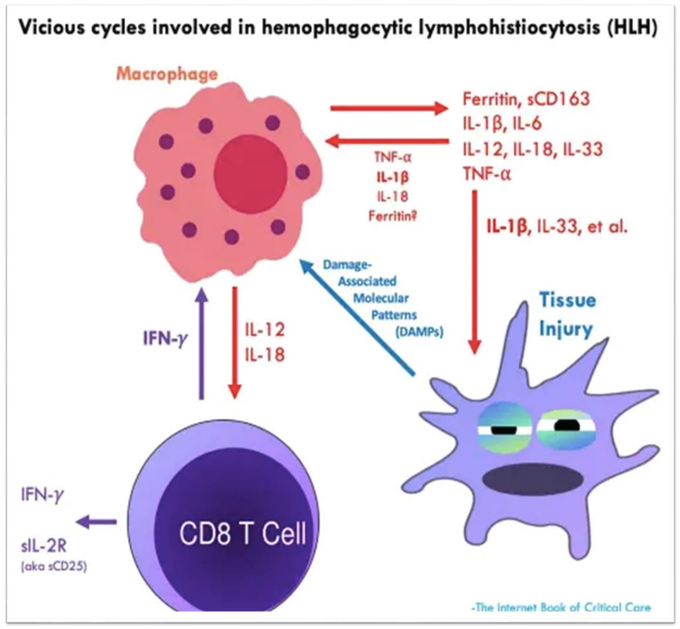
- Steroids
- Midostaurin

BONE MARROW BIOPSY



DISCUSSION

PATHOPHYSIOLOGY



HLH DIAGNOSTIC CRITERIA (5 of 9)

- FEVER
- SPLENOMEGALY
- CYTOPENIAS- ATLEAST 2
- HEPATITIS
- SOLUBLE IL-2
- FERRITIN >500
- BONE MARROW WITH HISTIOCYTES
- NK CELL ACTIVITY
- HYPERTRIGLYCERIDEMIA
- HYPOFIBRINOGEN
- IL-18 <450
- CXCL9 <647

Primary HLH

- Presents in early childhood
- Associated genetic mutations or clinical syndromes

Secondary HLH

- Presents in adults
- Triggered by acute illness or infection, malignancy and autoimmune disorders

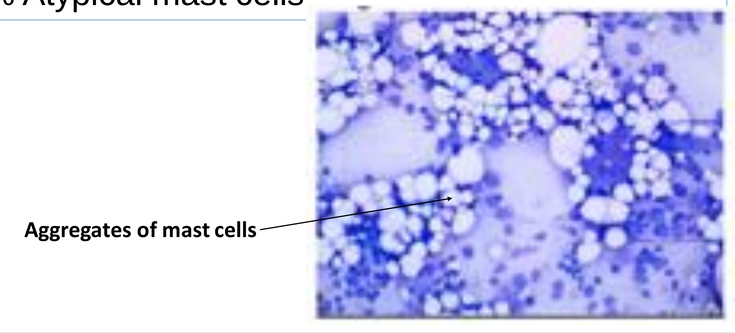
SM DIAGNOSTIC CRITERIA

Major

>15% of mast cells in clusters on bone marrow biopsy/extracutaneous organs

Minor

Tryptase>20
Abnormal CD25 expression
KIT D816V Mutation
>25% Atypical mast cells



PATIENT'S HLH WAS TRIGGERED BY SM

Treatment: trigger avoidance, symptomatic management, antihistamines and Midostaurin.

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