

A Unique Case of Marked Myeloproliferative Eosinophilia in an Infant of Unknown Etiology

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Abstract

Hyper eosinophilic syndrome is a rare disorder, often reported in adults but rarely in children, defined by elevated blood eosinophils (greater than 1500 eosinophils/ μ L) for at least 6 months, signs of organ involvement, and absence of other eosinophilia causing conditions. We present an infant with marked eosinophilia of unknown cause that had variable response to systemic steroid.

Methods:

4 month old Caucasian female, ex-35 week preemie with history of uniparental disomy of chromosome 14 and G-tube dependence admitted for vomiting, later developed diarrhea and increased work of breathing which required oxygen supplementation. Initial lab showed leukocytosis (WBC 124,300 TH/cmm) and absolute eosinophil count (AEC) of 96,500 TH/cmm which peaked at 110,000 TH/cmm.

Results:

Work up looking in to different end organ damage was negative. Gastrointestinal biopsy was not obtained. Quantitative immunoglobulins, tryptase, B12, ESR, ANA and ANCA, and extensive parasitic work up were unremarkable. Bone marrow biopsy showed markedly increased eosinophilic precursors. Flow cytometry and T and B cell clonality studies were inconsistent with malignancy. FISH for PDGFRA, PDGFRB, FGF21, and PCM1-JAK2 were negative. CHIC2 (4q12) deletion, *DOCK8*, *SPINK5*, *STAT3*, *TYK2* were normal. She showed variable response to systemic steroids (measured by AEC), requiring approximately 2 months of therapy. Her gastrointestinal and respiratory symptoms improved.

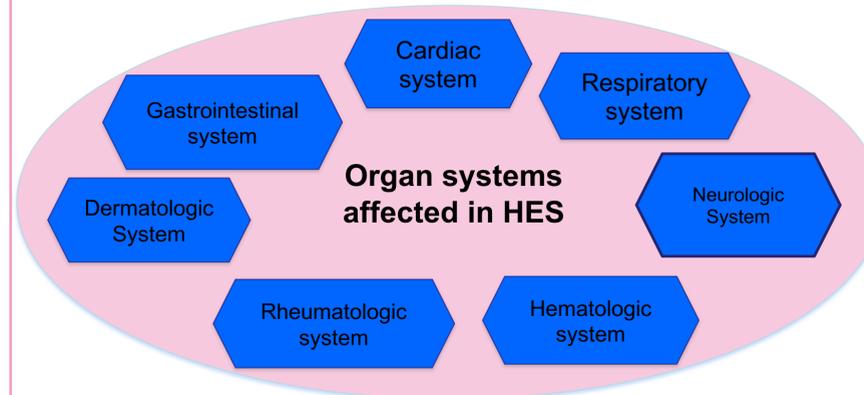
Conclusions:

This case demonstrates a rare presentation of marked myeloproliferative eosinophilia in infancy of unknown cause. We have not found a case presentation in the literature with these findings. Although no underlying etiology was found; this case may help those caring for patients with similar presentation.

Background

Hyper eosinophilic Syndrome (HES)

- **Persistently elevated eosinophil count greater than 1500/ μ L without any recognizable cause**
- **End organ damage present**



Case

HPI: 4 month old Caucasian female born at 35w 2d with history of uniparental disomy of chromosome 14 and G-tube dependence admitted for vomiting. Patient has history of failure to thrive at birth that required hospitalization and genetic work up. Due to persistent feeding issues, she presented to outside hospital and labs obtained revealed leukocytosis and patient was transferred to UMMC. Labs showed WBC 124,300 TH/cmm with 78% eosinophils, absolute eosinophil count 96,500 TH/cmm.

PE: HR 148 RR 42. CV and Lung exam benign. Abdomen-gtube in place, poor abdominal musculature. Skin, Lymph and Neuro exam unremarkable.

Differential Diagnosis/Work Up

1. Neoplastic disorders

Bone marrow biopsy showed markedly increased eosinophilic precursors. Flow cytometry and T and B cell clonality studies were inconsistent with malignancy. FISH for PDGFRA, PDGFRB, FGF21, and PCM1-JAK2 were negative.

2. Infectious Diseases

Extensive parasitic work up and HIV negative.

3. Immunodeficiency

CHIC2 (4q12) deletion, *DOCK8*, *SPINK5*, *STAT3*, *TYK2* were normal. Quantitative immunoglobulins normal.

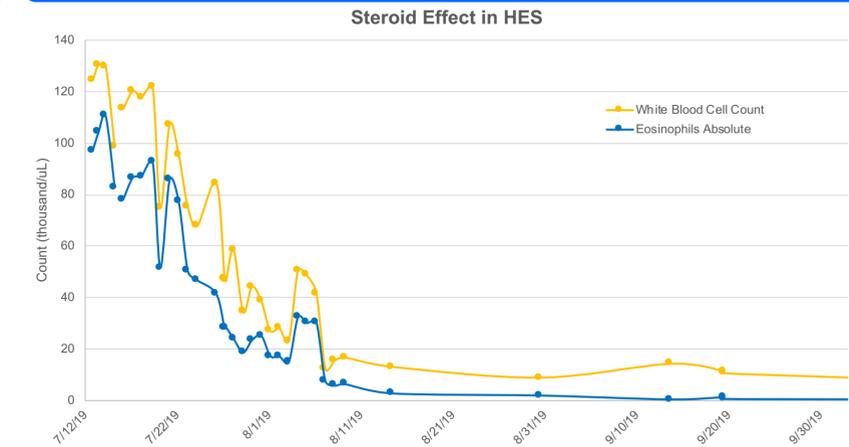
4. Immunologic

Tryptase, B12, ESR, ANA and ANCA within normal limits

Clinical Course

- During hospitalization, AEC peaked at 110,000 TH/cmm
- Extensive work up initiated with no identifiable cause of hypereosinophilia identified.
- Patient was healthy prior to presentation, history of uniparental disomy was felt to be non contributory to her presentation.
- Besides tachypnea, brief supplemental O2 requirement and intermittent diarrhea no other evidence of end organ damage found.
- Started PO oral steroids with no significant response. PO steroid was switched to IV steroid with some improvement of AEC.
- Patient discharged home on PO oral steroid with a prolonged taper over 2 months and discontinued when AEC was 450 TH/cmm
- AEC 200 TH/cmm, one month after systemic steroid discontinued and patient doing well clinically.

Lab Evaluation



Discussion

- This case describes an infant with marked eosinophilia.
- This patient had extensive work up with no etiology for eosinophilia found.
- This patient showed suboptimal response to systemic steroid and required prolonged course and taper to improve eosinophilia.
- Once prolonged steroid course discontinued patient continued to do well, without rebound of eosinophilia.

References

- Gleich, G. J., & Leiferman, K. M. (2009). The hypereosinophilic syndromes: current concepts and treatments. *British Journal of Haematology*, 145 (3), 271-285.
- Lawrence B. Schwartz, Javed Sheikh & Anish Singh (2010) Current strategies in the management of hypereosinophilic syndrome, including mepolizumab. *Current Medical Research and opinion*, 26:8, 1933-1946.