

# WHIM Syndrome: A Chronic Neutropenic Disorder

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### **Resource Information**

#### **About This Resource**

These slides are one component of a continuing education program available online at MedEd On The Go titled WHIM Syndrome (A Chronic Neutropenic Disorder): Uncouple the Complex for HCPs and Patients

#### **Program Learning Objectives:**

- Gain an understanding of WHIM syndrome as a rare PID/ chronic neutropenic disorder with diverse clinical presentations
- Implement strategic measures to improve the early identification of WHIM syndrome patients for prompt assessment and diagnosis to avoid potential complications and long-term sequelae
- Understand the CXCR4 pathway dysregulation and how it relates to the underlying causes of WHIM syndrome
- Garner an understanding of the limitations of current approaches for WHIM syndrome and potential new approaches for patients

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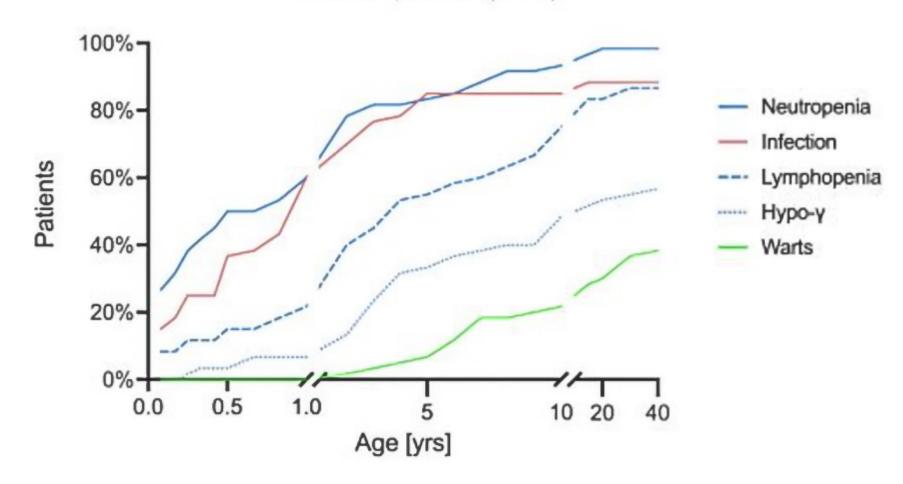
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## WHIM Syndrome: Poorly Functioning Immune System from Birth

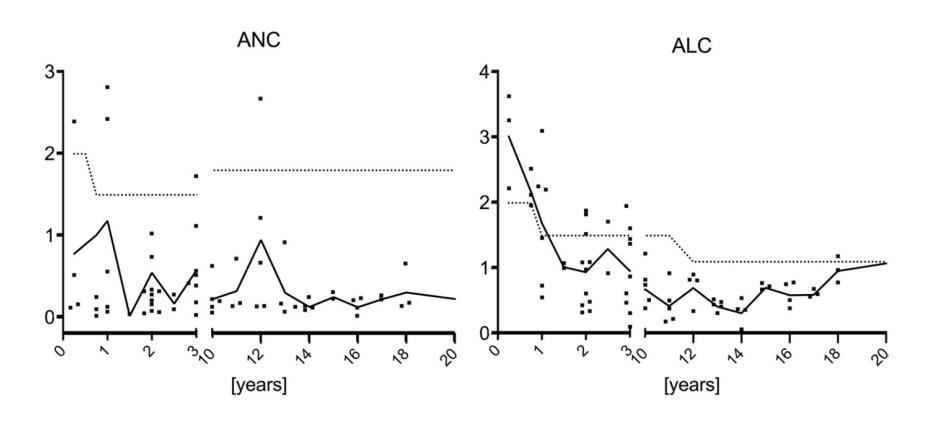
All WHIM patients (n=66)



Neutropenia is the earliest and most consistent finding

WHIM, warts, hypogammaglobulinemia, infections, and myelokathexis. Geier CB, et al. *J Clin Immunol*. 2022; 42(8): 1748–1765.

## WHIM Syndrome: Poorly Functioning Immune System from Birth



Neutropenia is the earliest and most consistent finding

### WHIM Syndrome: A Chronic Neutropenic Disorder

Consider WHIM syndrome in:

- Chronic neutropenia with onset in early infancy
- Chronic neutropenia with recurrent infections

i.e., likely congenital neutropenia

Definitely test for WHIM in:

- Chronic neutropenia with lymphopenia
- Chronic neutropenia with hypogammaglobulinemia
- Family history of WHIM

### WHIM Syndrome: A Chronic Neutropenic Disorder

- Genetic testing is the most direct approach
- Early diagnosis is paramount to prevent irreversible tissue damage and long-term sequelae
- Early diagnosis also informs genetic counseling and family planning

### WHIM Syndrome and Research

- Multiple Research Programs Worldwide
- SCNIR-
  - Clinical Course, Treatment and Disease Outcomes Monitored
  - Collects General and Specific information on patients with chronic neutropenia
  - WHIM syndrome studied in the context of other neutropenic disorders
  - Has resources for patients and families



Severe Chronic Neutropenia International Registry

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