

10 Warning Signs of WHIM Syndrome

Primary immunodeficiencies (PIs), like WHIM (Warts, Hypogammaglobulinemia, Infections, and Myelokathexis*) syndrome, are diseases where the immune system doesn't function properly. WHIM syndrome is predominantly caused by pathogenic variants in the *CXCR4* gene. Patients may experience a wide variety of signs and symptoms, and most do not present with all four features listed in the acronym, making WHIM syndrome challenging to recognize and diagnose. Below are the most common signs and symptoms. **If you know a patient who has two or more of these warning signs, consider a genetic test to inform a diagnosis.**

1 NEUTROPENIA

Low number of neutrophils, a type of white blood cell; may return to normal during infections

2 LYMPHOPENIA

Low levels of lymphocytes, a type of white blood cell, but not due to severe combined immunodeficiency

3 LOW LEVELS OF ANTIBODIES

Low levels of antibodies in blood; also known as hypogammaglobulinemia

4 RECURRING RESPIRATORY TRACT INFECTIONS

Two or more infections such as pneumonia within 1 year

5 FREQUENT EAR INFECTIONS

Multiple ear infections within 1 year

6 SKIN INFECTIONS

(eg, cellulitis, impetigo, folliculitis, abscess)

7 RECURRENT WARTS

Skin or anogenital warts that may be unresponsive to treatment

8 BRONCHIECTASIS

Damage to the airways of the lungs, causing them to widen

9 CANCERS ASSOCIATED WITH HPV

(eg, cervical, head, and neck)

10 FAMILY HISTORY

(eg, neutropenia, recurrent infections, warts)

*Myelokathexis, retention of neutrophils in the bone marrow; **CXCR4**, C-X-C chemokine receptor 4; **HPV**, human papillomavirus.

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For more information about WHIM syndrome, visit [WhatIfItsWHIM.com](https://www.whatiftswhim.com).

For more information about primary immunodeficiencies, visit [info4pi.org](https://www.info4pi.org).

For Healthcare Providers

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