An Overview of WHIM Syndrome for patients and caregivers

What is WHIM syndrome?

WHIM syndrome is a specific type of primary immunodeficiency (PI)—a genetic condition that weakens the body's immune system, making it difficult to fight infections.

It is named for 4 signs and symptoms, but despite its name, **only 23% of people with WHIM syndrome have all 4**:

Warts | **H**ypogammaglobulinemia* | **I**nfections | **M**yelokathexis†

*Hypogammaglobulinemia is the medical term for low levels of antibodies in the bloodstream.
†Myelokathexis is the medical term for when infection-fighting white blood cells called neutrophils become trapped in the bone marrow. It is discovered through a bone marrow biopsy.

What are the signs and symptoms of WHIM syndrome?

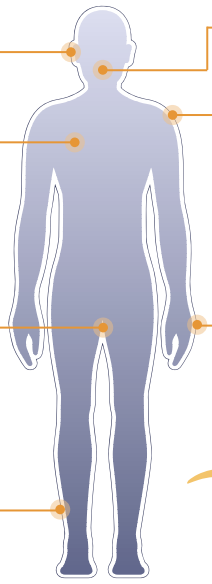
Below are the signs and symptoms that are commonly found in people with WHIM syndrome, including several types of infections, but you do not need to have all of them to have WHIM syndrome.

Ear infections, permanent hearing loss

Sinus infections, recurrent lung infections, pneumonia, permanent lung damage

Genital warts, HPV, cancer

Skin infections, cellulitis, warts



Dental issues, gingivitis

Impaired immune system function:

- Neutropenia
- Lymphopenia
- Hypogammaglobulinemia

Frequent infections that:

- May be bacterial or viral
- May be treated at home or require hospitalization
- Could result in organ damage
- Could be life-threatening

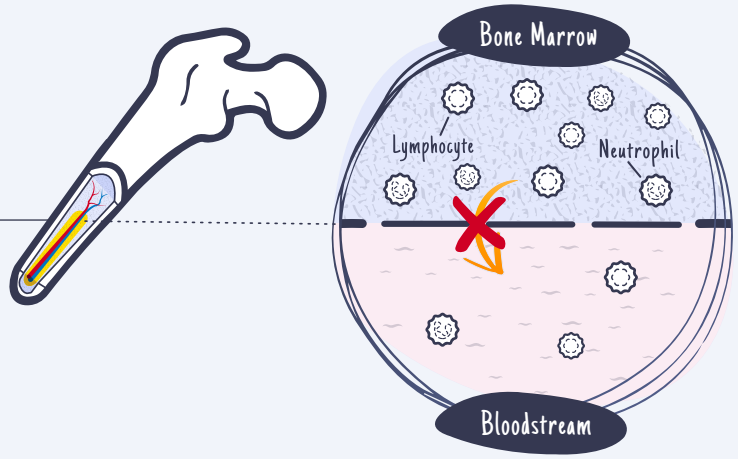
If you've experienced 2 or more of these, consider genetic testing for WHIM syndrome.

What causes WHIM syndrome?

The main cause of WHIM syndrome is a variation in the *CXCR4* gene, which is part of your DNA. This variation affects the CXCR4 pathway, which is responsible for moving infection-fighting white blood cells, including neutrophils and lymphocytes, from the bone marrow into the bloodstream.

In WHIM syndrome, the CXCR4 pathway is not working properly and white blood cells become stuck in the bone marrow.

With low levels of white blood cells in the bloodstream, the body is immunocompromised and vulnerable to infections.



How is WHIM syndrome diagnosed?

Confirming a WHIM syndrome diagnosis can be a challenge because it is rare, its signs and symptoms can be similar to other primary immunodeficiencies like common variable immunodeficiency (CVID) or common neutropenias like chronic neutropenic disorder, and it can vary in presentation and severity, even within the same family. There are different ways to inform a WHIM syndrome diagnosis:

Clinical Evaluation

Reviews signs and symptoms such as recurring infections and warts, your medical history, and family medical history

Bloodwork

Tests for low levels of infection-fighting white blood cells (neutrophils and lymphocytes) and antibodies

Genetic Test

Done using a simple cheek swab to look for variations in the *CXCR4* gene associated with WHIM syndrome

Bone Marrow Biopsy

Looks for a high number of white blood cells called neutrophils trapped in the bone marrow, a condition known as myelokathexis

> "Genetic testing answered so many questions for me. Finally, I know what it is and I have a name for it now...and it makes life so much easier."
>
> — Kirsty

The importance of seeking a WHIM syndrome diagnosis as soon as possible:

- **It is treatable.**
- **It can lead to serious long-term health issues, including cancer.**
- **It can run in families.**

Get support from an X4 Nurse Educator*

X4 Nurse Educators specialize in WHIM syndrome and can provide one-on-one educational support and resources.

Learn about no-cost genetic testing

If you think you may have WHIM syndrome, a genetic test using a simple cheek swab can help you find answers.

To learn more about WHIM syndrome, visit **WHIMsyndrome.com**.

*X4 Nurse Educators are employees of X4 Pharmaceuticals and do not work under the direction of a healthcare professional; they do not offer medical or treatment-related advice. For treatment and medical questions, you should contact your healthcare provider.

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