A Guide to Discuss WHIM Syndrome With Your Healthcare Provider

If you have been diagnosed with an immunodeficiency but have questions about your condition, talking to your healthcare provider can be an important step in finding answers.

This guide can help you start a discussion with your healthcare team about WHIM syndrome, signs and symptoms, getting diagnosed, and treatment goals.

How to Use This Guide

Step 1:
Print out this guide, read through the questions, and fill out the information about your symptoms and how you manage them.

Step 2:
Bring this to your next appointment with your healthcare provider to guide a conversation about WHIM syndrome and discuss next steps for seeking a diagnosis.

Being open and honest helps your healthcare provider understand how your symptoms and current treatment impact your day-to-day life. Having this information can help them find a management plan that works for you.

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
Hypogammaglobulinemia*
Infections
Myelokathexis†

This guide will help you reflect on the signs and symptoms of WHIM syndrome, including those not in the acronym.

*Hypogammaglobulinemia is the medical term for low levels of antibodies in the bloodstream.

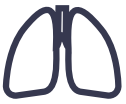
†Myelokathexis is the medical term for when infection-fighting white blood cells called neutrophils become trapped in the bone marrow. This is discovered through a bone marrow biopsy.

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.

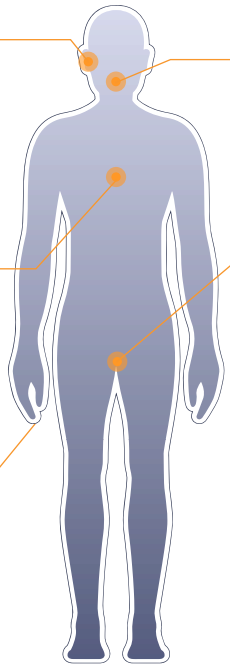
Please check off all signs and symptoms that you are experiencing now or have experienced in the past:

- ☐ Ear infections
- ☐ Permanent hearing loss



- ☐ Sinus infections
- ☐ Lung infections
- ☐ Pneumonia
- ☐ Permanent lung damage

- ☐ Skin infections
- ☐ Cellulitis
- ☐ Warts



- ☐ Dental issues
- ☐ Gingivitis

- ☐ Genital warts
- ☐ HPV/cancer

Abnormal lab findings (add values if available)

- ☐ Neutropenia _____
- ☐ Lymphopenia _____
- ☐ Hypogammaglobulinemia _____

Reflecting on Infections

Because WHIM syndrome can affect your body's ability to fight infections, it's important to discuss your own experience with them.

On a scale of mild to severe, rate the average severity of your infections:

Mild — Moderate — Severe

Over the past 6 months, how many infections have you experienced? _____

About how long did these infections last?

☐ 1 week or less ☐ 1–2 weeks ☐ 2–3 weeks ☐ 3–4 weeks ☐ 1 month or more

List any family members who have a history of recurrent infections:

_____

_____

WHIM syndrome is genetic and can be inherited. Family members with WHIM syndrome may have different signs and symptoms.

Current Symptom Management Plan

Consider how you and your healthcare team have approached managing your symptoms. This can include prescription and/or over-the-counter medications, and those administered by your healthcare team.

List all your current treatments and your experience with them:

Current Treatments	Your Experience

Based on your current experience, what are your goals for treatment in the future?

Describe the impact infections or any of the symptoms you identified on Page 2 have had on your daily life and activities, for example, missing out on school, work, traveling, or other important life events:

If you are still seeking answers about your immune deficiency or continue to experience infections even while on treatment, talk to your healthcare provider about WHIM syndrome.

Finding a Diagnosis

If you suspect you could have WHIM syndrome, your healthcare provider has several ways to confirm a diagnosis:

Clinical Evaluation

Review of:

- Current health and symptoms
- Bloodwork results (levels of antibodies and white blood cells [neutrophils and lymphocytes])
- Your clinical history
- Family medical history

Genetic Testing

- Looks for variations in the *CXCR4* gene (unique to WHIM syndrome)
- Done with a simple cheek swab
- Available for no cost

Learn more about how to order a test.

Bone Marrow Biopsy

- Used to look for myelokathexis—a hallmark feature of WHIM syndrome
- If you have had a bone marrow biopsy done in the past, those results can be reviewed again

Get Support From an X4 Nurse Educator*

X4 Nurse Educators specialize in WHIM syndrome and can help you find answers through one-on-one education and resources.

Scan the QR code here or connect today at 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.

If your healthcare provider would like to learn more about WHIM syndrome, they can visit **WhatIfItsWHIM.com**.