WHIM Syndrome

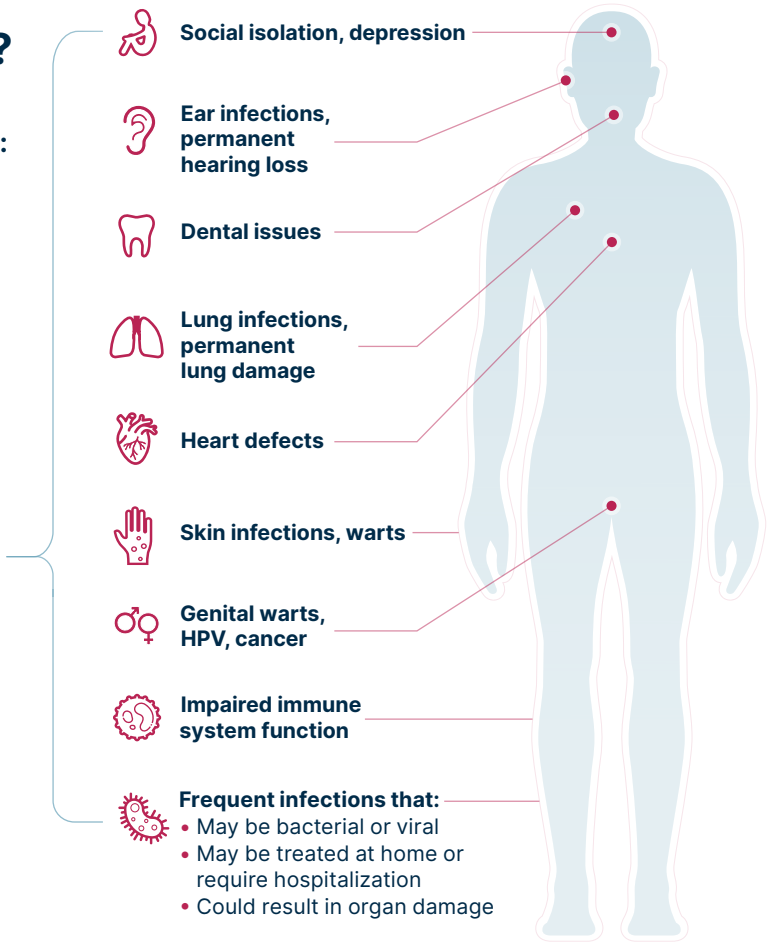
WHIM syndrome is a rare and chronic immune deficiency in which the body's immune system does not function properly and has trouble fighting infections.[1]

Why is it called WHIM syndrome?

WHIM syndrome is named after four manifestations[1]:

- **W**arts
- **H**ypogammaglobulinemia (low antibody levels)
- **I**nfections (frequent bacterial and viral infections)
- **M**yelokathexis (retention of infection-fighting white blood cells in bone marrow)

You could have some of these, all of these, and even other potential symptoms or consequences that are not in the name of the disease.[1,2]



Talk to your doctor about your symptoms and ask if it could be WHIM syndrome.

How WHIM syndrome may affect daily life

Living with WHIM is unpredictable. Uncertainty about how you will feel the next day, making time for doctor's appointments, or even scheduling treatments can be very disruptive to daily living.

"I never know from day to day if I'm going to feel okay tomorrow because there isn't really a predictor but it's not a constant. It's always going to be this way."
– person living with WHIM

"We work day to day ... we don't plan long term ... we never know how things are going to look so we're always kind of on edge."
– caregiver of a person with WHIM

How is WHIM syndrome diagnosed?

A person with WHIM may experience many different symptoms which can make it difficult to diagnose. The following methods can be used to diagnose WHIM syndrome[1]:

Clinical evaluation
Physical exam and evaluation of clinical symptoms

Detailed patient history
Evaluation of medical and family history of symptoms and prior diagnoses

Bloodwork
Blood test to determine white blood cell (WBC) counts

Bone marrow biopsy
Removal and examination of bone marrow tissue

Genetic testing
Lab tests to look for variants in genes such as *CXCR4*

Has anyone in your family been diagnosed with or experienced the symptoms of WHIM syndrome?

WHIM syndrome may run in families. Examining your family history and taking a genetic test can help identify other family members who may be at risk.[1]

Scan here
to see if you or your family member is eligible for genetic testing.

SCAN ME

People with WHIM syndrome may get a diagnosis like chronic neutropenia or primary immunodeficiency (PID).[1]

An accurate WHIM syndrome diagnosis can help you and your doctor to appropriately manage your care and minimize potential long-term complications.[1]

We are here to help.

SCAN ME

At X4 Pharmaceuticals, **Nurse Educators** serve as key points of contact for people with WHIM syndrome.*

Scan here
to connect with an X4 Nurse Educator to learn more about WHIM syndrome and your diagnostic options.

Access additional resources to learn more about WHIM syndrome:

X4 Pharmaceuticals:
WHIMSyndrome.com

Immune Deficiency Foundation:
primaryimmune.org/disease/WHIM-syndrome

National Organization for Rare Disorders:
rarediseases.org/rare-diseases/WHIM-syndrome

**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, please contact your healthcare provider.*

References: 1. Geier CB, Ellison M, Cruz R, et al. Disease progression of WHIM syndrome in an international cohort of 66 pediatric and adult patients. *J Clin Immunol.* 2022;42(8):1748-1765. **2.** Data on file. X4 Pharmaceuticals. 2023.

X4 PHARMACEUTICALS