

No-cost genetic testing for WHIM syndrome

About the program

Probably Genetic's mission is to help the 200 million undiagnosed rare disease patients worldwide access affordable genetic testing. To further that goal, the company has recently joined efforts with X4 Pharmaceuticals to provide PID panel testing to people who fit the symptomatic profile of WHIM syndrome—a rare genetic immunodeficiency caused by mutations in the CXCR4 gene—at no cost to them.

How it works

Patients are first evaluated through an online symptom checker developed by Probably Genetic to determine if they are eligible. Once their information is reviewed by Probably Genetic, eligible patients will receive a genetic sequencing kit by mail and can obtain their clinical lab report from a genetic counselor without ever leaving the house.

We need your help

To ensure the program is as successful as possible, Probably Genetic encourages both diagnosed and undiagnosed people experiencing symptoms of immunodeficiency or WHIM syndrome to complete the symptom checker. If you already have a confirmed diagnosis, your symptom checker response will be used for research to improve the performance of the symptom checker. This helps other undiagnosed people access no-cost genetic testing. If you are undiagnosed, you may be able to benefit from no-cost genetic testing and counselling.

For more information, see the landing page: https://www.probablygenetic.com/immuno For questions, please reach out to cheyanne@probablygenetic.com.