PATH4WARD:
A NO-COST GENETIC TESTING PROGRAM FOR PEOPLE WITH PRIMARY IMMUNODEFICIENCIES

PATH4WARD helps confirm diagnosis of primary immunodeficiencies (PIDs) with neutropenia, including WHIM syndrome

What was the PATH4WARD report about?

PATH4WARD is a no-charge genetic testing program. This report explains how doctors used PATH4WARD to help people with suspected congenital neutropenia, including WHIM (Warts, Hypogammaglobulinemia, Infections, Myelokathexis) syndrome. Genetic testing can help people get diagnosed faster, which can help them identify therapeutic options to manage symptoms and potentially improve their quality of life.

The report included test results of eligible people from July 2019 to September 2020. People were eligible if they had **both** symptoms of neutropenia **and** an absolute neutrophil count (ANC) of 500/µL or less. PATH4WARD looked for gene mutations and variants using a 23-gene severe chronic neutropenia (SCN) panel and a 207-gene primary immunodeficiency (PID) panel.

What was the goal of the report?

Researchers wanted to describe how PATH4WARD was used to test people with suspected congenital neutropenia, including WHIM syndrome. The report was presented at the 2021 Virtual Clinical Immunology Society Annual Meeting.

What did we learn and what were the results of the report?

Researchers learned that PATH4WARD is a valuable tool for early genetic testing of people with suspected PIDs with neutropenia, including WHIM syndrome.

156 doctors used PATH4WARD. 52.5 percent were specialists in pediatric hematology/oncology. Specialists in adult hematology followed at 14.6 percent.

271 people who met the criteria and 6 family members were tested through PATH4WARD between July 2019 and September 2020.

8.9 percent of people tested were found to carry a mutation known to cause a PID.

Some people had mutations in the *CXCR4* gene, including mutations known to cause WHIM syndrome.

The program identified several variants of unknown significance (VUS). A VUS means it is not known if the mutation causes disease or not.

Based on the results from the report, PATH4WARD will now expand to include people with an ANC of 750/µL or less, and the testing panel was expanded to 407 genes to help detect more PIDs.

How can families find more information about PATH4WARD?

To learn more about genetic testing and this no-cost program, visit **IDYourPID.com/Resources.**

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