PREVENTION GENETICS part of EXACT SCIENCES

Pharming

navigateAPDS by Pharming

navigateAPDS GENETIC TESTING PROGRAM DESCRIPTION

Pharming offers sponsored genetic testing, and pre- and post-test genetic counseling for eligible individuals who may carry a pathogenic variant in one of two genes known to be associated with Activated PI3K Delta Syndrome (APDS), an underdiagnosed, progressive primary immunodeficiency.

The navigateAPDS testing program helps reduce barriers to genetic testing and increases certainty in obtaining a correct diagnosis by enabling patients suspected of having APDS, and their family members, to have access to genetic testing, as well as pre-test and post-test counseling.

ABOUT APDS

APDS is an underdiagnosed, progressive primary immunodeficiency that affects approximately 1-2 people per million.

Signs and symptoms of APDS start in childhood, and patients are vulnerable to repeat infections and autoimmune/inflammatory signs such as lymphoproliferation, splenomegaly, and even lymphoma.

APDS occurs when there is an abnormal change in either one of two specific genes, the *PIK3CD* gene or the *PIK3R1* gene, in an autosomal dominant mode of inheritance. These genes are involved in making parts of a protein that help in the growth and division of white blood cells, particularly the B-cell and T-cell lymphocytes.

Patients are often misdiagnosed with other immunodeficiencies or autoimmune disorders and have a protracted course to obtain a correct diagnosis. A definitive APDS diagnosis is made by genetic test.

For more information about APDS visit: www.allaboutapds-hcp.ca

PROGRAM ELIGIBILITY

This program is available to individuals in the US and Canada who meet **two or more** of the following bulleted criteria below:

Clinical features:

- Bronchiectasis
- Lymphadenopathy for greater than one month
- Chronic hepatomegaly or chronic splenomegaly
- Severe, persistent, or recurrent Herpesviridae infections (e.g., EBV, cytomegalovirus)
- Enteropathy
- Lymphoma at 0-25 years – meets the 2 eligibility criteria
- Lymphoma at ≥ 26 years of age – requires second eligibility criteria

Laboratory:

- Elevated levels of immunoglobulin M
- Increased number of follicular helper T cells
- Reduced number of naïve B cells

History:

- Common Variable Immune Deficiency (CVID) phenotype or direct family member with CVID phenotype
- Relative with *PIK3CD* or *PIK3R1* genotype (first or second degree) – meets the 2 eligibility criteria

PANEL INFORMATION

The **navigateAPDS** program offers testing with the choice of either the **Primary Immunodeficiency and Malignancy Predisposition Panel (680 genes)** or the **Inborn Errors of Immunity/Primary Immunodeficiency (PID) Panel (609 genes)**, which analyzes genes that are associated with inherited disorders of the immune system. Broad panel testing allows for an efficient evaluation of several potential genes based on a single clinical indication.

CLINICAL CONSULTATION SERVICES FOR HEALTHCARE PROVIDERS

PreventionGenetics' team of board-certified and experienced genetic counselors trained in medical genetics are available to assist clinicians by phone and/or email. PreventionGenetics' counselors can be reached throughout the testing process to:

- review patient cases that may benefit from the Program
- aid in explaining genetic test results
- provide result-specific background regarding variants, genes, and the condition
- identify gene-specific information including relevant literature and studies, published management guidelines if available, and patient resources

For more information or to order a test, please visit
www.preventiongenetics.com/sponsoredTesting/navigate-apds
or scan the QR code.

While third parties and commercial organizations may provide financial support for this program, tests and services are performed by PreventionGenetics. Healthcare professionals must confirm that patients meet certain criteria to use the program. Third parties and commercial organizations may receive de-identified patient data from this program, but at no time would they receive patient identifiable information. Third parties and commercial organizations may receive contact information for healthcare professionals who use this program. Genetic testing and counseling are available in the US and Canada only. Healthcare professionals and patients who participate in this program have no obligation to recommend, purchase, order, prescribe, promote, administer, use or support any other products or services from PreventionGenetics or from third parties or commercial organizations.