



Using Sponsored Genetic Counseling to Boost Provider Engagement for Rare Disease Molecular Diagnosis


Program Launched to Increase APDS Awareness

Activated PI3K Delta Syndrome (APDS) is a rare, progressive primary immunodeficiency, characterized by recurrent infections and increased risk for lymphoma. It shares many features with other immune disorders making identifying APDS as the specific diagnosis difficult. And since APDS has only recently been fully characterized, knowledge of the disease, even among medical professionals, may be limited.¹


Genetic testing is needed to precisely diagnose APDS, enabling clinicians to recommend targeted treatment options or clinical trial opportunities. But testing costs and a shortage of genetic specialists to support both patients and providers can be barriers to testing.

Pharming, a biopharmaceutical company dedicated to transforming the lives of patients with rare, debilitating and life-threatening diseases, took action to elevate awareness of APDS and increase access to genetic testing by launching the *navigateAPDS* Program. This program offers no-charge genetic testing and counseling for eligible individuals from the U.S. and Canada who have features that could be consistent with APDS, providing them with the opportunity to determine a molecular diagnosis that could lead to more precise treatment options.

Program Goals



Increase **AWARENESS** of APDS



Increase **ACCESS** to genetic services

Genetic Counseling Service Fosters Provider Participation

Patients with phenotypes consistent with APDS can be seen by a variety of specialists, from asthma and immunology to gastroenterology and hematology. These providers have varying degrees of genetics expertise themselves and may have limited access to genetic specialists, resulting in hesitance to order genetic testing.

To aid providers in delivering optimal care and increase program participation, Pharming partnered with Genome Medical to provide genetic counseling services to patients participating in *navigateAPDS*.

“Offering genetic counseling as part of the *navigateAPDS* program has made providers more comfortable with ordering genetic testing and has been an invaluable resource for patients.”

NAMI PARK, PHARM D
DIRECTOR OF MEDICAL AFFAIRS AT PHARMING

GENETIC COUNSELING'S VALUE TO PHARMING	
Pre-Test:	Helps patients understand the disease and genetic testing so they feel comfortable participating in the program.
Post-Test:	Reviews complex test results and potential next steps to support patients and providers with follow-up care.

Over 420 different providers have referred their patients for genetic counseling as part of the program, demonstrating the need for this support.

Participating physicians noted they also appreciate the ability to provide a full service offering to patients who are interested in testing, made possible with the inclusion of genetic counseling.

Program Increases Access to Genetic Testing

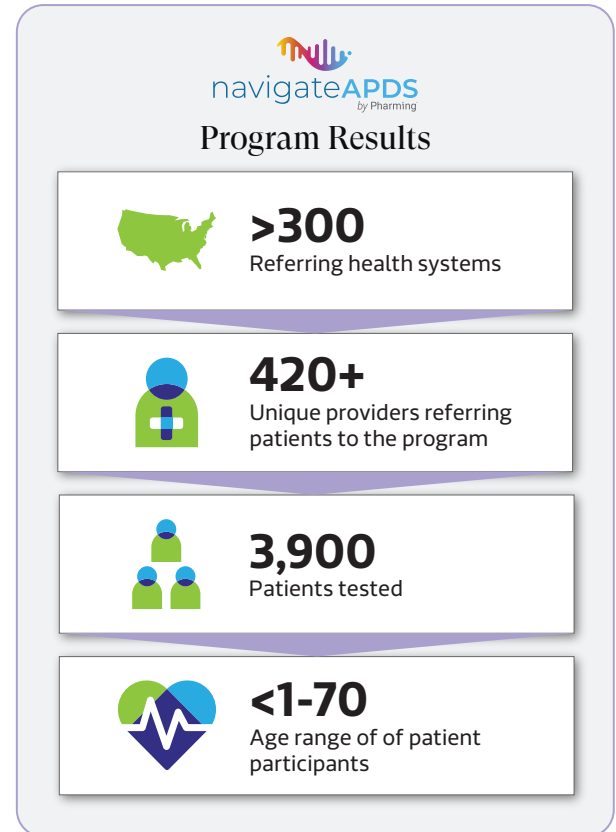
With Pharming's field team increasing provider awareness of navigateAPDS, the program grew quickly and Genome Medical scaled to meet the increased volume, from delivering 5 genetic counseling sessions per quarter to over 340.

The navigateAPDS program has grown beyond academic medical centers to expand equitable access for patients at community hospitals and clinics across the country that may not have genetic counseling resources. Patients can continue to receive care from their local provider, while easily completing their telehealth genetic counseling appointment within a week.

“The genetic counseling service is increasing access by allowing patients to get care closer to home and reducing time to testing.”

HEATHER MCLAUGHLIN, PHD, FACMG, NYCOQ, CGMB
DIRECTOR MOLECULAR DIAGNOSTICS AT PHARMING

Over 3,900 individuals from all 50 U.S. states have participated in the navigateAPDS program, from infants to adults in their 60s and 70s. The program is helping these patients find the answers and the support they need.



Supporting Diagnosis and Follow-up Care

As part of the genetic counseling service, patients and providers receive a visit summary explaining test results and what they may mean as well as resources tailored to the individual results. This is provided not only for patients with a positive APDS molecular diagnosis, but also for other findings that may be uncovered such as carrier status for recessive disorders or variants of uncertain significance (VUS). Equipped with this information, patients and providers can make more informed care management and treatment decisions.

In addition, providers participating in the program have access to educational materials that help them identify symptoms and learn what treatment options and clinical trials may be beneficial to their patients, such as Pharming's APDS clinical trials.

When asked what the program means for Pharming, Heather McLaughlin commented, “navigateAPDS has allowed Pharming to increase awareness of APDS among clinicians, identify patients eligible for enrollment in clinical trials related to APDS and gain a better understanding of the genetic landscape of the *PIK3CD* and *PIK3R1* genes.”

“I am extremely grateful for my genetic counselor's expertise and for the navigateAPDS testing program in getting this important information to me.”

NAVIGATEAPDS PROGRAM PARTICIPANT

¹ <https://allaboutapds-hcp.com>