



PI and APDS **—what you need to know**

Patient guide and resources for activated PI3K-delta syndrome (APDS)

Look inside to learn more about APDS, how it is a genetic condition that may affect your children, siblings, aunts, uncles, or other relatives, and how genetic testing can help those who have gone undiagnosed or underdiagnosed for years.

APDS is a rare, progressive, underrecognized primary immunodeficiency (PI)



A PI is a genetic condition in which the body's immune system does not work correctly. There are over 450 types of PIs



APDS is a PI that can lead to a variety of symptoms like infections, autoimmune conditions, and lymphoma.



APDS is with you from birth—although symptoms may not appear until later in life.



The median time to an APDS diagnosis is 7 years, because the wide range of symptoms makes it difficult for doctors to always recognize it.



The earlier a genetic test is done, the sooner a definitive diagnosis may be made.

Symptoms can begin at any age and can vary from person to person

Pneumonia, sinus, and ear infections
(96%-100%) Very common

Developmental and growth delay
(10%-31%) Less common

Swollen lymph nodes, enlarged spleen, or enlarged liver
(71%-89%) Very common

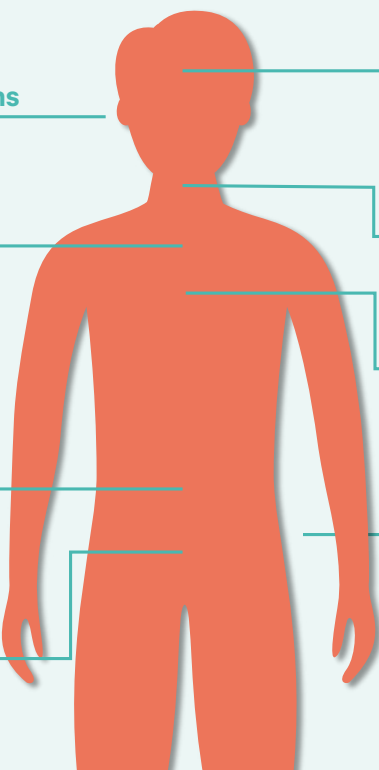
Bronchiectasis
(18%-60%) Common

Lymphoma
(12%-25%) Less common

Digestive issues
(25%-51%) Common

Autoimmune or autoinflammatory conditions
(17%-42%) Less common

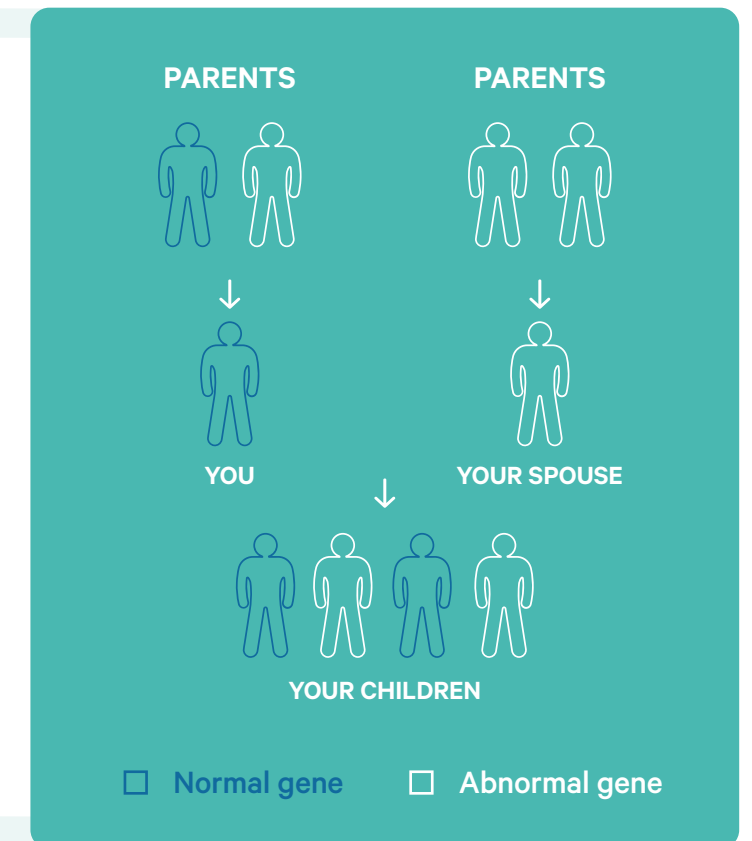
Chronic herpes virus infections
(36%-49%) Common



APDS can be passed down from generation to generation

APDS is an autosomal dominant disease, which means if either parent has APDS, there's a **50% chance each child will inherit the disease**.

The genetic variants causing APDS can also occur randomly, without the child inheriting one from a parent (that is called a de novo variant).



Relatives of those with APDS are also at risk

If you have been diagnosed with APDS, it is possible that other relatives also have the same genetic variants that cause the disease. This may include:

- Children
- Siblings
- Aunts
- Uncles
- Cousins
- Nieces
- Nephews

Here's a quick Q&A to get you prepared for genetic testing

APDS is an inherited genetic condition. If you are diagnosed with APDS, there's a good chance that other family members may also have APDS, even if they don't seem to have any symptoms or have different symptoms from you. If you think you or your family member may have APDS, talk to your doctor to see if you are eligible for a sponsored genetic test. Here's a quick Q&A to get you prepared for genetic testing.

Genetic testing sounds complicated. I don't even know where to start. Are there any risks associated with genetic testing? How is my genetic information protected?

Genetic testing can be ordered by any healthcare provider, but is most often ordered by a specialist such as a geneticist, immunologist, or oncologist. You can also self-refer to an independent genetics telehealth practice, such as Genome Medical.

How will genetic testing benefit me?

A **positive** genetic test result may lead to prevention or treatment options that have potential to improve your health, detect complications early, or prevent future complications from occurring at all.*

A **negative** result may provide peace of mind or reduce the need for future screenings or tests.

*Not all genetic conditions have prevention or screening recommendations available.

If I have a genetic condition, is there anything I can do about it? If not, I don't think I want to know.

Receiving a diagnosis through genetic testing can empower you to take action to reduce the impact of the condition, prevent future complications, or in some cases treat symptoms you didn't realize you had. As the old saying goes, **knowledge is power.**

A genetic test is performed using a standard blood draw, a saliva sample, or a cheek swab. Like any standard blood test, a genetic test is considered to have very little physical risk. Genetic test results are specifically named as protected health information and are covered by the Health Insurance Portability and Accountability Act.

This means your test results cannot be shared or accessed beyond the ordering provider's records without your express permission.

Who will see my test results?

Genetic test results are shared with the ordering physician and genetic counselor, who explain the results and suggest next steps. All records are confidential. Results are not shared directly with insurers, but may be shared by the patient or provider if chosen.

Can I be discriminated against based on my genetic test results?

In the United States, the Genetic Information Nondiscrimination Act:

- Helps protect people with diagnosed gene changes
- Makes it illegal for insurance companies to deny coverage or determine cost of coverage based on genetic testing results

Get the conversation started with your doctor

Genetic testing could provide answers for you or any family members who have gone undiagnosed or misdiagnosed for years. As you consider whether you or your relatives may be living with symptoms of APDS, these questions can encourage a productive conversation with your doctor about genetic testing.

Ask:

- If someone in my family is diagnosed with APDS, what should I do?

- How does the genetic testing process work?

- Who else in my family should consider a genetic test?

- I haven't experienced any symptoms, but my relative is living with APDS. Do I need a genetic test?

Do your relatives have unexplained symptoms? A genetic test can help

Talk with your family about APDS today

- APDS is a rare, progressive, underrecognized PI
 - PIs prevent the immune system from operating correctly
- It is possible that the relatives of a person diagnosed with APDS have the same genetic variants that cause the disease
- Genetic testing could provide answers for any family members who have gone undiagnosed or misdiagnosed for years
 - Prior to 2017, there was no genetic test for APDS. Anyone tested before 2017 should talk to their doctor about getting tested again

Get to know APDS on an even deeper level

Get help locating a physician and explore more information and resources about APDS symptoms, genetic testing, and treatment options on the All About APDS website.

All about
APDS.COM



Scan the QR code to visit the **All About APDS** website for more details.