

There is so much to know about activated PI3K delta syndrome (APDS) and a lot of information to absorb. APDS is a rare primary immunodeficiency (PI). PIs are a group of genetic disorders that can impair the body's immune system. **APDS occurs when there are variations in one of two genes: *PIK3CD* or *PIK3R1*.** These genes are essential to the production and function of immune cells in your body.

In the weeks and months ahead, you may have questions about life with APDS. This guide can help equip you with some of the knowledge you need to feel more confident and informed. **Please keep in mind, your doctor and care team are always your best resource for specific questions about your diagnosis.** This is a general reference for you and your support system.

1 **QUESTION:** What are the various signs and symptoms of APDS?

ANSWER: People with APDS can experience a wide range of symptoms as a result of their impaired immune system. Symptoms can begin at birth, but some people may not experience symptoms until later in life. Some of the symptoms of APDS are listed below, but other symptoms may occur that are not listed:

- Recurrent, severe infections (such as pneumonia, sinus, and ear infections)
- Coughing phlegm (mucus), wheezing, difficulty breathing, or chest pain
- Swollen lymph nodes
- Enlarged spleen or liver
- Chronic herpesvirus infections
- Lymphoma
- Digestive issues (diarrhea, cramping, blood in stool, or malabsorption)
- Developmental delay
- Autoimmune or autoinflammatory conditions, such as anemia

These symptoms can even vary between family members who have APDS.

Because APDS is inherited in most cases, genetic testing should be considered for family members of people who have been diagnosed.

2 **QUESTION:** If I have APDS, does that mean my children or other people in my family could have it too?

ANSWER: APDS is an autosomal dominant genetic disease, meaning if one parent has APDS, there is a 50% chance that their child will also have APDS. **This makes genetic testing critical for other family members and for family planning.**



3 **QUESTION:** How can APDS impact my life?

ANSWER: People living with APDS are more likely to experience fatigue, anxiety, and depression, as well as hospitalizations and social, school, and work absences. You, your caregivers, and your loved ones can find resources to support your journey on the Resources page of www.AllaboutAPDS.com, along with a list of support organizations that you may find useful.

4 **QUESTION:** Are there better ways to manage my condition?

ANSWER: While there are currently no therapies that directly address the underlying cause of APDS, there are therapies that can help in symptom management. Any decisions about treatment strategies should be made between you and your doctor.

You may see multiple doctors as part of your healthcare team, and there can be a lot to keep track of. Take notes on the therapies you are prescribed to manage your symptoms, and share those notes with each doctor during your appointments. Tracking how your symptoms are progressing and communicating your current treatment plan among your different doctors can help your healthcare team work more efficiently.



We encourage you to ask your doctor questions about how to move forward after receiving your diagnosis, such as:

- How can we manage my symptoms now that the underlying cause of my illness has been identified?
- Will there be certain activities I will have to discontinue or modify due to my disease?
- How often should I come in for checkups?
- Which specialists should I be seeing, and will you be working with them to manage my symptoms?
- Are there any groups you can connect me with for social support?
- Which of my family members should also undergo genetic testing?

Before your next doctor visit, we encourage you to reach out to support organizations that could fit your needs in this difficult time. You can find a list of some support groups at www.AllaboutAPDS.com/useful-links.

If you know any other people living with APDS, please share this resource and encourage them to visit AllaboutAPDS.com for more information, access to additional resources, and to register for updates.