

All about APDS A predominately inherited condition

Family Health Tree

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APDS, activated PI3K delta syndrome (previously known as PASLI disease) is an underrecognized and progressive primary immunodeficiency, first characterized in 2013. It can be difficult to diagnose, with a reported median diagnostic delay of 7 years, and is caused by genetic variants in either one of two identified genes known as *PIK3CD* or *PIK3R1*, which are vital to the development and function of immune cells in the body.

APDS is a genetic condition with an autosomal dominant inheritance pattern. This means that only one parent needs to have the altered gene for APDS to be inherited. Simply put, people with APDS have a 50% chance of passing it to their children.

People with APDS suffer from a wide variety of symptoms that may include:

- Ear, Sinus, and Respiratory Tract Infections
- Chronic Cough
- Enlarged Tonsils, Lymph Nodes or Spleen
- Nodules in the Airway or Digestive Tract
- Herpes Infections
- Gastrointestinal (Digestive) Tract Issues
- Autoimmune and Autoinflammatory Disorders such as Rheumatoid arthritis, Lupus and Celiac disease.
- Low Numbers of Blood Cells
- Developmental Delay
- Lymphoma

Discover Your Family's Health History

The onset of APDS symptoms often start very early in a child's life and continue into adulthood. APDS symptoms evolve over time and may be underdiagnosed. Earlier diagnosis may spare patients from irreversible damage such as bronchiectasis, lymphoma, or early mortality.

People with APDS may have very different symptoms that vary in severity. These differences are seen even within the same family, which emphasizes the need for genetic testing even among family members.

Use the worksheet on the following page to fill in your family's health history. It may help you track your family's history of the rare primary immunodeficiency, APDS.

If, like many people, you are still confirming your Primary Immunodeficiency diagnosis, talk to your doctor about your eligibility for a sponsored genetic test at navigateAPDS.com. If you have APDS, encourage your family members to talk to their doctor about getting a genetic test.



For information about our sponsored genetic testing program visit:

navigateAPDS.com

Available in the US and Canada only.

COMPLETE YOUR FAMILY HEALTH TREE ON THE NEXT TWO PAGES >

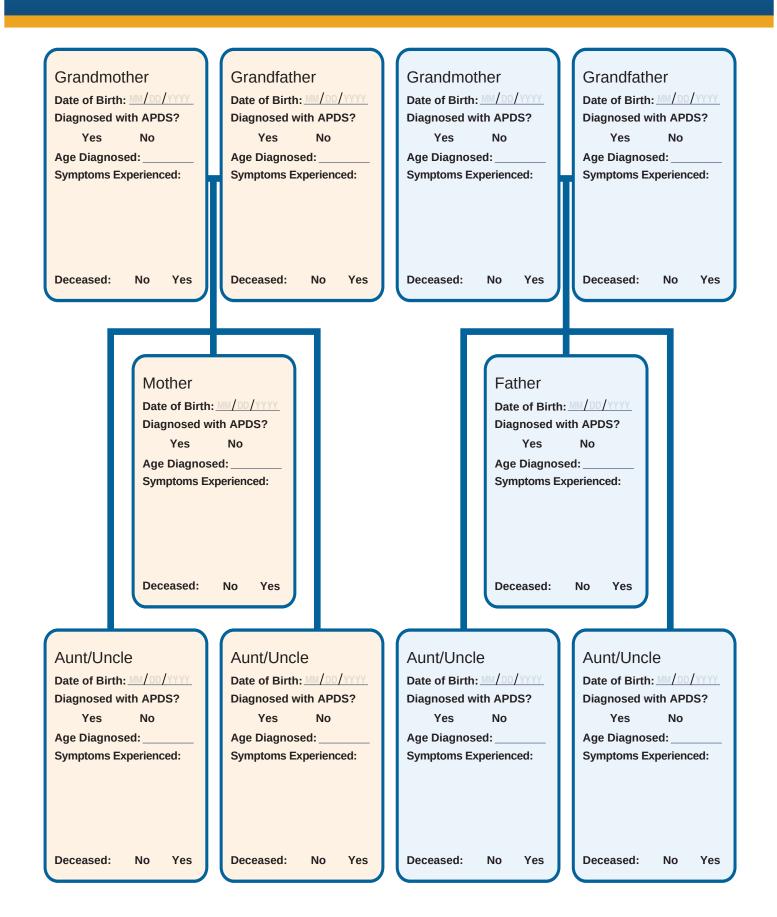


People with APDS experience a wide variety of symptoms making it difficult to diagnose.

Even if you don't yet have an APDS diagnosis, write down all the symptoms experienced by you or your family members in the health summary below.

Symptoms may include, but should not be limited to:

- Ear, Sinus, and Respiratory Tract Infections Autoimmune and Autoinflammatory Chronic Cough
- Enlarged Tonsils, Lymph Nodes or Spleen
- Nodules in the Airway or Digestive Tract
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Sister/Brother Date of Birth: ////////////////////////////////////	Sister/Brother Date of Birth: <u>MM/DD/YYYY</u> Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	You Date of Birth: // / /////// Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Spouse Date of Birth: <u>M/DD/YYYY</u> Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:
Deceased: No Yes	Deceased: No Yes		Deceased: No Yes
	Child	Child	Child
	Date of Birth: MM/DD/YYYY Diagnosed with APDS?	Date of Birth: <u>MM/DD/YYYY</u> Diagnosed with APDS?	Date of Birth: MM/DD/YYYY Diagnosed with APDS?
	Yes No	Yes No	Yes No
	Age Diagnosed: Symptoms Experienced:	Age Diagnosed: Symptoms Experienced:	Age Diagnosed: Symptoms Experienced:
	Deceased: No Yes	Deceased: No Yes	Deceased: No Yes

Additional information or list other members of your family with symptoms here:

No data input into document is retained by Pharming or held on the website.