

All about APDS

A predominately inherited condition

Family Health Tree







All about APDS Family Health Tree

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, Puerto Rico 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.

Deceased:

No

Yes

Deceased:

No

Yes

Deceased:

Yes

Deceased:

Yes

Symptoms may include, but should not be limited to:

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

Grandmother Date of Birth: M/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced: Deceased: No Yes	Grandfather Date of Birth: M/DD/YVYV Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced: Deceased: No Yes	Grandmother Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced: Deceased: No Yes	Grandfather Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced: Deceased: No Yes
Mother Date of Birth Diagnosed of Yes Age Diagnose Symptoms E	No sed:	Father Date of Birth Diagnosed w Yes Age Diagnos Symptoms E	No sed:
Aunt/Uncle Date of Birth: M/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Aunt/Uncle Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Aunt/Uncle Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Aunt/Uncle Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:

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- Gastrointestinal (Digestive) Tract Issues
- Disorders such as Rheumatoid arthritis, Lupus and Celiac disease.

 • Low Numbers of Blood Cells
- Developmental Delay
- Lymphoma

Sister/Brother Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Sister/Brother Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	You Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Spouse Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:
Deceased: No Yes	Deceased: No Yes		Deceased: No Yes
	Child Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Child Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:	Child Date of Birth: MM/DD/YYYY Diagnosed with APDS? Yes No Age Diagnosed: Symptoms Experienced:
	Deceased: No Yes	Deceased: No Yes	Deceased: No Yes
Additional information o	r list other members of your fa	ımily with symptoms here:	