

All about  
**APDS**

# APDS Glossary

Not an actual patient

## Know the terms associated with activated PI3K delta syndrome (APDS)

### WHAT IS APDS?

APDS is one of 400+ primary immunodeficiencies (PIs). PIs are rare genetic disorders that prevent the immune system from functioning properly. APDS may cause a wide variety of symptoms throughout the body, and **can only be definitively diagnosed through genetic testing**.

You will likely encounter various acronyms and terms specific to your APDS diagnosis, and some may be unfamiliar to you. Inside this booklet is a collection of terms you may hear in discussions with your healthcare team or encounter in your personal research. We hope that this collection will be a useful reference as you learn more about APDS.

# APDS Symptom Terms

APDS can cause signs and symptoms, such as:

**ANGIOEDEMA:** A disorder that causes swelling beneath your skin that may occur anywhere on the body. It can be dangerous if it occurs in the throat or tongue, as it makes breathing difficult.

**ATOPIC DERMATITIS:** Also known as eczema, it is an allergic condition that makes your skin dry and itchy. This rash can occur anywhere on the body.

**BRONCHIECTASIS:** A condition in which the bronchial tubes in the lungs widen and thicken. This makes them stiff and causes difficulty breathing. The condition also makes it easier for bacteria to accumulate, which can result in frequent infections and permanent damage to the lungs. Bronchiectasis can also cause coughing and chest pain.

**CHRONIC URTICARIA:** An allergic condition that causes red, itchy hives (welts) to appear on your skin. Because it is a chronic condition, it can occur repeatedly over a long period of time.

**CYTOPENIAS:** Reduced numbers of blood cells in the body. There are multiple types of cytopenias. Anemia is a low number of red blood cells, while leukopenia and neutropenia are reduced white blood cells.

**ENTEROPATHY:** Damage, irritation, or swelling in the small intestine that can cause diarrhea, abdominal pain, and malabsorption (difficulty digesting or absorbing nutrients from food).

**LYMPHADENOPATHY:** Swelling of the lymph nodes as a result of infections from bacteria or viruses, or in some cases resulting from cancer.

**LYMPHOMA:** A type of cancer of the immune system that occurs when B or T cells change and grow out of control (see next page for B and T cell definitions).

**NODULAR LYMPHOID HYPERPLASIA:** The occurrence of small nodules (bumps or swellings) in the intestinal tract, containing large numbers of white blood cells. This can lead to enteropathy (see definition above).

**ORGANOMEGALY:** The abnormal enlargement of an organ. In APDS, enlargement of the liver (hepatomegaly) or spleen (splenomegaly) can occur and may require surgical intervention.

# Terms for Understanding APDS

**ANTIBODY:** A specially shaped protein made by B cells (see definition below) of the immune system in response to an infection. The special shape allows antibodies to bind with foreign microbes (like bacteria and viruses) in a way that neutralizes these invaders. Antibodies are an essential part of fighting infection.

**AUTOSOMAL DOMINANT DISEASES:** Genetic conditions in which, if one parent is affected, there is a 50% chance that their child will inherit the disease. APDS is an autosomal dominant disease.

**B AND T CELLS:** Types of white blood cells that play important roles in an immune response.

**B CELLS:** Produced in the bone marrow (*think "B" for bone*), they produce antibodies and can activate other immune cells.

**T CELLS:** Originating from the thymus (*think "T" for thymus*), T cells can kill infected host cells and activate other immune cells.

**COMMON VARIABLE IMMUNODEFICIENCY DEFICIENCY (CVID):** A PI where people have low levels of antibodies in their blood, similar to APDS. It is one of the most common PIs and is usually diagnosed in adults. Many people with APDS are often initially diagnosed with CVID.

**IMMUNE DEFICIENCY:** A condition that makes the body less able to fight off infections. APDS is a disease that leads to immune deficiency and disruption to the functioning of the immune system.

**IMMUNE DYSREGULATION:** Breakdown in control of the immune system, which can lead to a range of symptoms throughout the body.

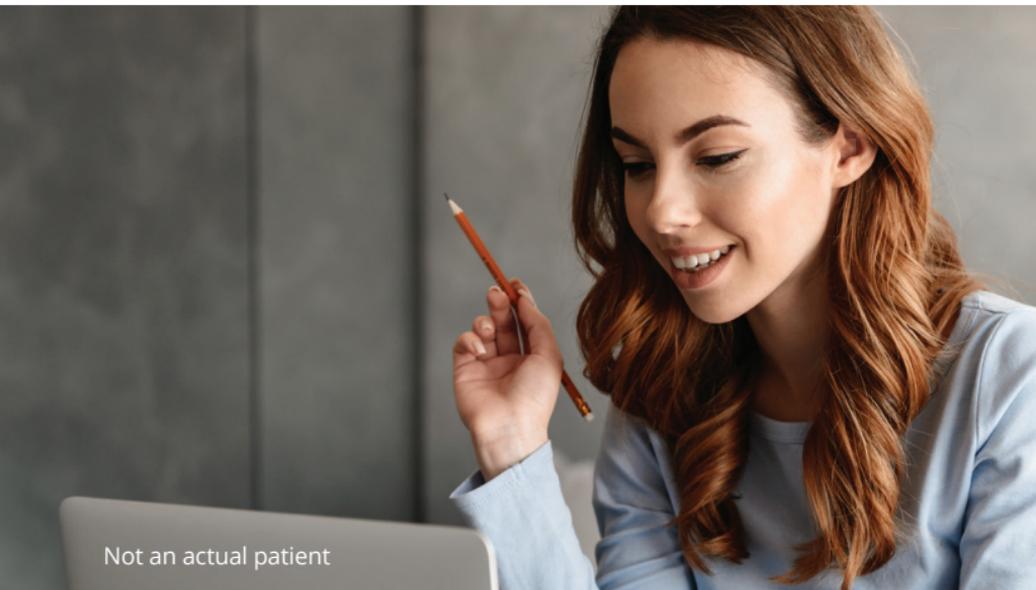
**PASLI:** A previous name for APDS. PASLI is an abbreviation for *P110 delta-activating mutation causing senescent T cells, lymphadenopathy, and immunodeficiency*.

**PI3K DELTA:** A complex of 2 proteins that controls how B and T cells develop and function. In APDS, PI3K delta is more active than it should be, leading to problems with B and T cells.

# Terms for Understanding APDS (continued)

**PIK3CD AND PIK3R1 GENES:** These genes provide the instructions for making PI3K delta, which regulates how B and T cells develop and work. When these genes are abnormal, the immune system cannot function correctly. Variations in either of these genes can cause APDS.

**PRIMARY IMMUNODEFICIENCIES (PIs):** A group of rare genetic disorders that impair the immune system. Without a functional immune response, people with PIs may experience frequent infections and other symptoms such as swollen lymph nodes/spleen or autoimmunity. "Primary" means that the disorder is genetic, so the immune impairments are present at birth.



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**Knowledge is power when living with APDS.** For more information and to sign up for regular updates, visit [www.AllaboutAPDS.com](http://www.AllaboutAPDS.com). Also consider sharing this glossary with your support system or anyone else affected by APDS or other PIs.

