

APDS Diagnosis: Family Discussions



Once you or someone in your family has been diagnosed with APDS, which is a genetic disorder, you may have questions about whether or not someone else in your family could also have APDS.

What does this mean for my family?



Who should be tested?



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 on to their children.

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

Chronic cough

Ear, sinus, and respiratory tract infections

Nodules in the airway or digestive tract

Low numbers of blood cells

Developmental delay

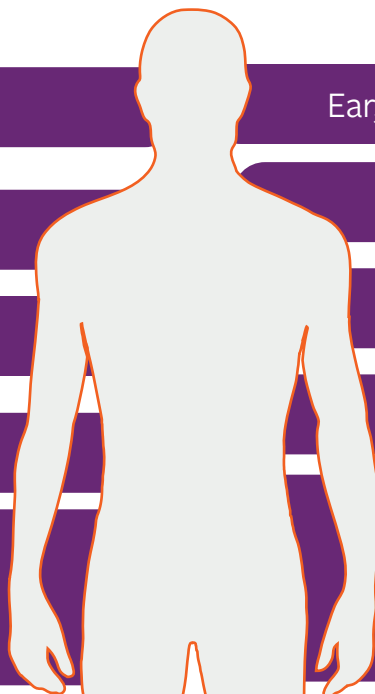
Lymphoma

Gastrointestinal (digestive) tract issues

Enlarged tonsils, lymph nodes, or spleen

Autoimmune and autoinflammatory disorders such as rheumatoid arthritis, lupus, and celiac disease

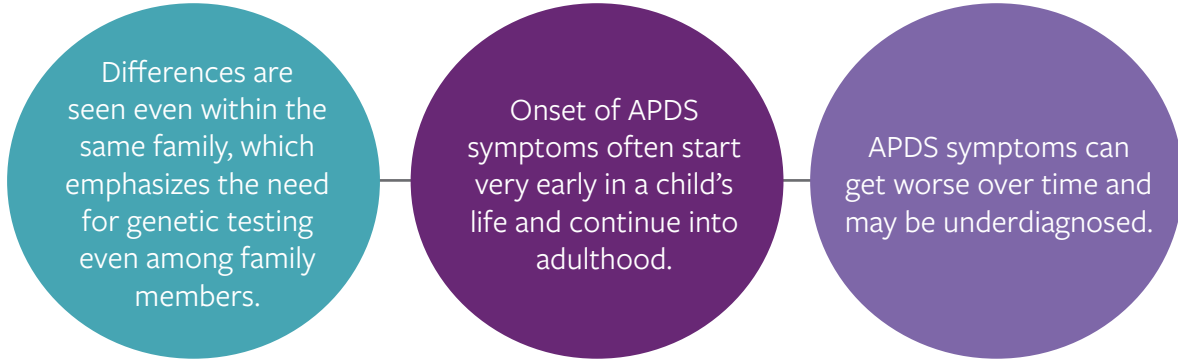
Herpes virus infections, including Epstein-Barr virus (EBV), cytomegalovirus (CMV), herpes simplex, and varicella zoster (chicken pox or shingles)



Family Mapping

Once someone in your family has a diagnosis, it is important to recognize and evaluate the symptoms that other family members may be experiencing as well. Immediate family members, such as parents, siblings, and children may need to be genetically tested, but this may also apply to extended family members such as aunts, uncles, grandparents, and cousins.

People with APDS may have very different symptoms that vary in severity



Ask your APDS Clinical Educator (ACE) about the “Family Health Tree.” They can also help you fill it out and work through the next steps for discussing genetic testing with your family.

Be Prepared in an Emergency

The emergency leaflet summarizes key information that can be used in the ER, with a school nurse, or with any medical provider who is not familiar with APDS.

