



## **About the Immune Deficiency Foundation**

The Immune Deficiency Foundation (IDF) improves the diagnosis, treatment, and quality of life of people affected by primary immunodeficiency through fostering a community empowered by advocacy, education, and research.

Primary immunodeficiencies (PI) are a group of more than 400 rare, chronic disorders in which part of the body's immune system is missing or functions improperly. PI is caused by genetic defects in immune response pathways and can affect anyone, regardless of age or gender.

PI results in high susceptibility to infections. Illnesses such as repeated bouts of pneumonia, persistent skin abscesses, and sepsis, or blood infections, are common and can lead to permanent organ damage. Antibiotics often have little effect on the infections long-term. The infections can become chronic and develop into life-threatening conditions requiring hospitalization. PI is also linked to autoimmune disorders and increased risk of certain forms of cancer.

There are approximately 250,000 people who are diagnosed with PI in the U.S., and thousands more go undetected. Individuals affected by PI often find it difficult to receive proper diagnosis, treatment, and specialized healthcare. They experience difficulties financing healthcare, finding educational materials, and locating others who share their experiences.

As a national non-profit located in Towson, Md., IDF provides accurate and timely information for individuals and families living with PI and offers valuable resources and support.

IDF operations are carried out by staff, medical professionals, and volunteers. Through this network, IDF conducts research on various PIs, provides education seminars and support groups, coordinates nationwide fundraising activities, influences healthcare policy, develops PI specific programs, assists with navigating health insurance, provides general information about managing care, and offers annual conferences for the PI community.

**Contact IDF for more information**  
**800-296-4433 | [www.primaryimmune.org](http://www.primaryimmune.org)**

# Primary Immunodeficiencies: Frequently Asked Questions

## What is a primary immunodeficiency?

A primary immunodeficiency is one of more than 400 rare, chronic disorders in which part of the body's immune system is missing or functions improperly. A person with PI has one or more genetic mutations in cells or antibodies related to the immune system which causes the normal protective immune response to fail when confronted with bacteria, viruses, or fungi. The person with PI experiences increased susceptibility to infection.

## Who does a primary immunodeficiency affect?

PI can affect anyone, regardless of age, gender or ethnicity and onset of symptoms may occur in childhood or adulthood. According to the Immune Deficiency Foundation, there are approximately 250,000 — or 1 in 1,200 people — diagnosed with a primary immunodeficiency in the U.S. Thousands more go undetected.

## How is a primary immunodeficiency diagnosed?

Evaluations for PI may include review of medical and family history, physical exam, laboratory bloodwork to examine immune system functions, skin tests, and exome sequencing, which examines genetic causes for disorders. PI is rare and often missed as a diagnosis by primary care physicians; the immune system is a very complicated function of the body. The Immune Deficiency Foundation estimates that the average length of time between onset of symptoms and diagnosis is between nine and 15 years. Half of patients diagnosed are 18 or older.

## What are the symptoms of a primary immunodeficiency?

A person with PI develops frequent and persistent infections in the sinuses, the throat, the ears, the lungs, the brain or spinal cord, the blood, the urinary tract, the intestinal tract, and on the skin. The infections can lead to debilitating illnesses that must be treated with intravenous antibiotics and/or hospitalization, and some infections are fatal.

An infection that may be a PI is:

- **Severe** – requires hospitalization or intravenous antibiotics
- **Persistent** – won't completely clear up or clears slowly
- **Unusual** – caused by an uncommon organism
- **Recurrent** – keeps coming back
- **Runs in the family** – others in a family have a similar susceptibility to infection

A person experiencing this type of infection should request testing for a primary immunodeficiency.

## How is a primary immunodeficiency treated?

Regular immunoglobulin (Ig) replacement therapy, a blood product administered intravenously or subcutaneously, is the primary treatment for antibody deficient patients. Gamma interferon injections can be used to treat a PI called chronic granulomatous disease (CGD). Sometimes, antibiotics are used to treat persons with PI. For more severe PI disorders, bone marrow transplantation and clinical trial gene therapy are options. PI is a lifelong condition and cannot be cured, only treated. It is imperative to pursue continual medical follow-up.