About the Immune Deficiency Foundation & Primary Immunodeficiency Diseases

Founded in 1980, the Immune Deficiency Foundation (IDF) is the national nonprofit organization dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases (PI) through advocacy, education, and research.

There are approximately 250,000 people who are diagnosed with PI in the U.S. These individuals often find it difficult to receive specialized healthcare and proper diagnosis and treatment. Individuals affected by PI also experience difficulties financing their healthcare, finding educational materials on the disease and locating others with whom to share their experiences. IDF helps individuals overcome these difficulties so they can live healthy and productive lives. The constant presence of IDF assures people with PI, their families and their healthcare providers that there is a place to turn for help.

Frequently Asked Questions about Primary Immunodeficiency Diseases

What is a primary immunodeficiency disease?
Primary immunodeficiency diseases, or PI, are a group of more than 350 rare, chronic disorders in which part of the body's immune system is missing or functions improperly. These problems lead to an increased susceptibility to infection.

Who does primary immunodeficiency affect?
According to IDF, there are approximately 250,000 people with PI in the U.S. Approximately 1 in 1,200 persons have a diagnosed PI. Thousands more go undetected. While not contagious, these diseases are caused by hereditary or genetic defects, and although some disorders present at birth or in early childhood, the disorders can affect anyone, regardless of age, gender, or ethnicity.

How is primary immunodeficiency diagnosed?
Medical and family history, physical exam, blood and immunoglobulin level tests and vaccines to test the immune response may be included in the diagnosis process. IDF estimates that the average length of time between onset of symptoms and diagnosis is between nine and 15 years. Fifty percent of those patients are 18+ years of age when diagnosed.

What are the symptoms of primary immunodeficiency?
You should be suspicious if you have an infection that 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 your family have a similar susceptibility to infection

If any of these describe your infection, ask your physician to check for the possibility of a PI. People with PI are more susceptible to infections and health problems that lead to serious and debilitating diseases. It is critical to get an early diagnosis and proper medical care.

How is primary immunodeficiency treated?
Treatment for primary immunodeficiency depends on the specific type of disorder. Immunoglobulin (Ig) replacement therapy is the treatment for people with certain types of antibody deficiencies. For other types, treatments may include one or more of the following: gene therapy, hematopoietic stem cell transplantation (bone marrow transplantation), interferon gamma, and/or enzyme replacement therapy. Prophylactic antibiotics and/or antifungals are also used by themselves or in combination with other therapies.

Contact IDF for more information
800-296-4433
www.primaryimmune.org
Preserve Protections Established by the Affordable Care Act for People with Primary Immunodeficiency Diseases

Individuals and families impacted by primary immunodeficiency diseases (PI) and other rare, chronic conditions rely on the patient protections established by the Affordable Care Act (ACA) to ensure they have continued access to the treatments they require in order to live a healthy and productive life.

- **Continue Prohibition on Exclusion for Pre-Existing Conditions**
  The ACA forbids health insurers from denying coverage or charging patients more based on pre-existing conditions. PI is a group of more than 350 genetic disorders of the immune system. Individuals with PI are extremely susceptible to infections and illnesses that, without proper treatment, could lead to serious and debilitating health conditions. The pre-existing condition protections included in the ACA ensure all people with PI have access to adequate health coverage. Any reversal of this policy would restrict access to care and endanger lives. Proposals that claim to offer such protections but are less comprehensive are equally problematic because access to inadequate coverage will not address the treatment needs of the PI community.

- **Prohibit Annual or Lifetime Caps on Coverage**
  The only treatment option available to approximately 70% of people with PI who have antibody deficiency is immunoglobulin replacement (Ig) therapy, a blood plasma product that helps prevent severe infections. Ig replacement therapy requires lifelong infusions and costs on average $7,500 to $10,000 per month. When access to this therapy is limited, many people with PI will experience frequent, severe and potentially life-threatening infections. The prohibition of annual and lifetime health coverage limits helps patients who use Ig therapy and who, without such protections, would exceed annual or lifetime caps imposed by health plans and would lose their insurance.

- **Maintain Ability for Dependents to Remain on Parents Plans Until Age 26**
  Coverage of the young adult population drastically reduced the uninsured population and especially helped those with chronic conditions. Maintaining coverage for these chronic conditions prevent these young people from being a greater burden on the healthcare system. Young people with PI who rely on Ig replacement therapy need continuous coverage to afford their treatment.

- **Limit Maximum Out-of-pocket Expenses**
  People with PI who require Ig replacement therapies and other expensive therapies to treat their conditions need predictable and affordable cost-sharing in order to manage their complex conditions. Predictable and limited out-of-pocket expenses are necessary to ensure access and protect the health of people who rely on lifesaving treatments.

- **Ensure New Plans Protect Patients**
  The legality of association health plans, short term limited duration plans, and certain Medicaid waiver restrictions is being heard and appealed in the courts. Plans or eligibility criteria that do not offer patient protections and limit access to needed treatments, including Ig replacement therapy and gene therapy, endanger the health of individuals with severe chronic conditions. Congress must exercise oversight to ensure adequate access protections for these extremely vulnerable populations.

IDF urges members of Congress to support policies that preserve these patient protections
Support the Newborn Screening Saves Lives Reauthorization Act of 2019 (HR2507)

The Newborn Screening Save Lives Reauthorization Act reauthorizes critical federal programs that provide assistance to states to improve and expand their newborn screening programs; support parent and provider education; and ensure laboratory quality and surveillance for newborn screening.

Background

- In 2008, Congress passed the original Newborn Screening Saves Lives Act, which established guidelines and helped facilitate comprehensive newborn screening in every state. Prior to passage of this act, newborn screening tests varied greatly from state to state.
- In 2007, only 10 states and Washington, DC required screening for all disorders on the Recommended Uniform Screening Panel (RUSP). Today, 49 states and Washington, DC require screening for 31 of the 35 core conditions.
- In 2010 when Severe Combined Immunodeficiency (SCID), a severe form of primary immunodeficiency diseases (PI), was added to the RUSP, there were only 4 states that required screening for SCID. As of December 2018, all 50 states screen for SCID.
- While all states are now screening for SCID, there are still barriers to full implementation for screening and treatment, and there is a need for parent and provider education to ensure babies with SCID are able to thrive.

Key Bill Provisions

- Increases funding for Health Resources and Services Administration (HRSA) and Centers for Disease Control and Prevention (CDC) grants to expand state screening programs, educate parents and healthcare providers, and improve follow-up care for infants with a condition detected through newborn screening.
- Renews the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, which provides states with a RUSP to help ensure every infant is screened for conditions that have a known treatment.
- Reauthorizes HRSA’s Clearinghouse for Newborn Screening Information, the Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs), and the CDC Newborn Screening Quality Assurance Program, supporting test accuracy and ensuring programs keep pace with the rapid improvements in newborn screening.
- Continues funding for research through the National Institutes of Health Hunter Kelly Newborn Screening Program, expanding permitted activities under the program to include promising new research and further enables private/public partnerships to support pilot research and technology studies.
- Commissions an expert report on the modernization of newborn screening to, among other things, identify factors that impact the decision to add new conditions to the RUSP and to identify barriers to state implementation.

Impact on the Primary Immunodeficiency Disease (PI) Community

- SCID Compass, an educational program of the Immune Deficiency Foundation is funded by HRSA through the Newborn Screening Saves Lives Reauthorization Act of 2014. This program seeks to improve outcomes for infants with SCID by enhancing access to educational resources, providing linkages to critical services and developing mechanisms for follow-up. Reauthorization of this act will ensure this vital program continues.
- With SCID as the first condition to be added onto the initial RUSP, the PI community was a pioneer in advocating for screening to ensure infants born with serious conditions are identified and treated quickly. The federal funds provided to states through the Newborn Screening Saves Lives Act ensured all states are now screening for SCID, and support for its reauthorization in 2019 will ensure screening continues and any existing barriers are addressed.
- Families impacted by SCID will continue to benefit from the reauthorization through programs focused on long-term surveillance of individuals identified at birth to assess patient progress and outcomes to inform future treatments.
- With over 350 types of PI, there may be opportunities in the future to identify and treat other conditions early through newborn screening to ensure children with PI are able to thrive. This act supports research and pilot studies that will help the PI community realize that possibility.