

Policy Pledge

Access to quality care for those living with primary immunodeficiency matters: it saves lives





Access to healthcare is a fundamental human right.

Everyone living with a primary immunodeficiency (PID) worldwide is entitled to the correct diagnosis, appropriate treatment and care. It is a matter of saving lives.

Primary Immunodeficiencies are rare diseases caused by inherited defects of the immune system. PID leaves people living with the disease more prone than others to “common” infections; including: severe autoinflammation, autoimmunity, allergies, and malignancy – resulting in poor health outcomes.

There are over 450 forms of PID and an estimated 10 million people live with the disease worldwide. However, only between 10 and 30% have been diagnosed; and those that are diagnosed are not all receiving the best possible care. **Lack of access to early diagnosis and quality care can lead to repeated infections, organ damage, and even death.**

This document outlines **priorities to make access to quality care a reality for all PID patients, no matter where they live.** It unifies organizations and individuals who are committed to improving awareness and recognition of PIDs; as well as addressing current challenges in access to quality care – understanding the complex realities of healthcare systems across the world. It further reflects combined efforts to inform policy makers on issues related to PID and encourage actions that will support better diagnosis and care, for the benefit of all those living with the disease.



Early diagnosis and screening

Early diagnosis is the first step in improving quality of life for patients and is critical to ensure patients have the best outcomes for treatment and care, preventing years of complications.

- Newborn screening for severe types of PID such as Severe Combined Immunodeficiency (SCID) and X-linked agammaglobulinemia (XLA) is a life-saving and efficient method to achieve early diagnosis and where this is appropriate in the health economy it deserves the highest possible priority.
- Education about the symptoms and characteristics of all forms of PID helps to improve early diagnosis rates.
- Access to genetic testing for inheritable PIDs as well as other testing methods should be ensured worldwide along with access to trained, specialist health professionals.



Quality Care

Universal and timely access to treatment and specialist care shall be ensured worldwide; so that no matter what country a PID patient is from, they are still able to live their life to the fullest.

- A sustainable and high-quality supply of human plasma worldwide allows for the availability of safe immunoglobulin replacement therapy, which remains one of the most important treatments for a wide range of PIDs. Access should be guaranteed for all patients in need, alongside other therapies.
 - Prophylaxis treatments should be provided to all patients as they are effective for several types of PIDs and can be beneficial to patients of all ages. These treatments may act both as complements to other treatments or compensate for measures that aim to provide antibody protection when proven ineffective.
 - Patients should be guaranteed access to Hematopoietic Stem Cell Transplantation (HSCT or BMT) without delay, when it is the best suitable treatment. This requires high quality training and uniform expertise in administering transplantations to patients, as well as adequate donation systems.
 - Medical innovations such as gene therapy hold promise for a cure for some PIDs. Efforts shall be directed towards their uptake by health systems and patients' safe access to them.
 - Vaccination can protect PID patients from specific viruses and diseases as well as assist in the diagnosis of PID. Vaccines should be made available to patients where indicated and systematically to those around them.
 - Efforts to improve secondary prevention measures should be enhanced, including diagnosis of potential co-morbidities in PID patients.
- The creation and upkeep of patient registries in each country underpin all advances; allowing for further universality of care application.



Health Awareness for PID patients

Preventative medicine and education are necessary to avoid health complications and help patients live longer lives.

- Ensuring patients' access to and education about safe hygiene practices and health promotion measures, including healthy diets and regular physical activity as well as mental health support, is crucial to enhance quality of life alongside medical treatment.
- These are all part of integrated, multidisciplinary care for those with PIDs.
- Increasing access to preventative medicines and setting up adequate screening protocols for complications and co-morbidities in PID patients helps to reduce rates of relapse in treatment and poor health outcomes.



Research

Investment in medical research will always be fundamental in advancing our understanding of the immune system, progressing clinical practices and delivering advanced treatments that improve outcomes for people living with PIDs.

- Efforts to support research into and the development of gene therapies for PIDs should be pursued to ensure safe access for patients across the globe.
- COVID-19 studies have generated new findings and contributed to improving knowledge of the immune system. These advances shall be used to strengthen care for PID patients and further the understanding of PID's causes.
- Studies into the impact of COVID-19, development of symptoms, and efficacy of treatment can benefit PID patients by better informing medical professionals on the protection of those with compromised immune systems from future viruses and known infections.



Signatories

 European Federation of Immunological Societies		
 Arab Society for Primary Immunodeficiencies		




Key facts and figures



10 million
estimated people have **PID worldwide**

> 70-90%
are undiagnosed

There are **450** different forms of PIDs, with unique care requirements



Due to research support, this number is up by almost **+200** in the past 10 years




Early diagnosis and efficient treatment can drastically improve the quality of life for PID patients




If treated before **3.5 months** of age, infants with SCIDs and other PIDs experience fewer complications and higher survival

The overall survival rate of PID patients after Hematopoietic stem cell transplants (HSCT) is now over **80%**

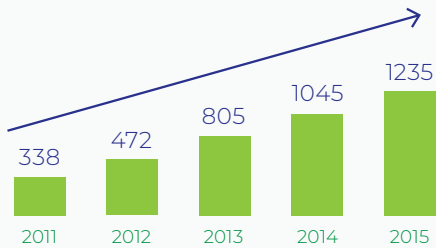
1:1200
the suspected number of people with a genetic primary immunodeficiency that significantly impacts their health



The **10** warning signs of PID have heightened awareness and offer further drive towards early diagnosis



“Celebrate these advancements with us and strive for better quality of care for PID patients every 22 to 29 April, on World PI Week!”



Signatures to the WPIW petition since 2011