

Facts About PID for the Medical Profession

We would like to thank World PI Week for the use of their materials in compiling the IPOPI PID factsheets.

What is PID?

Primary Immunodeficiencies (PID's) are hereditary and genetic defects in the immune system that cause increased susceptibility to a wide range of infections. PID often presents itself in the form of "ordinary" infections which do not respond to treatment. Too often infections are treated while missing the underlying cause, allowing the illnesses to recur, and leaving the patient vulnerable to vital organ damage, physical disability, and even death.

Unlike many rare diseases, effective treatment options for PID patients are available, which can enable them to carry out a normal life. PID patients must therefore be diagnosed early and informed about the most adequate treatment for their particular condition. Unfortunately, none of the available therapies can reverse the damage of late diagnosis.

PID Facts

- PID affects at least **10 million** people worldwide¹
- There are currently **200²** identified PID conditions.
- Can affect **anyone**, regardless of age and sex
- **Vary in severity** depending on whether one or several parts of the immune system are affected
- Current diagnosis levels suggest that around **1 in 8-10,000** people have a genetic primary immunodeficiency that significantly affects their health³.
- It is estimated that **70–80%⁴ of PIDs remain undiagnosed**, globally.
- A simple and relatively inexpensive **blood test** Complete Blood Count (CBC) **can identify over 95% of PID patients⁵**
- Unlike many other **rare disorders**, PIDs can be **successfully treated**.

Please insert any data/challenges which you identified in your country

What are the symptoms?

There are a number of different signs and symptoms of a Primary Immunodeficiency disease. PID patients have an increased susceptibility to recurrent and persistent infections. These vary in severity from ear and sinus infections, to lung infections, meningitis and septicaemia, amongst others. Although less common, PID patients may have abscesses of their internal organs, such as the liver, or infections of their blood. There are certain infections caused by germs that only afflict PID patients, therefore the type of infection itself may serve as a red flag for PID disease. Patients with PID may also present with a variety of autoimmune or rheumatologic problems. Gastrointestinal (digestive) problems may also occur. In some cases this can be the result of an infection of the intestines in others, they may be a reflection of an

¹ U.S. Department of Health and Human Services. National Institutes of Health. *PAR-08-206: Investigations on Primary Immunodeficiency Diseases*, 2009.

² Primary Immunodeficiencies: A Field in Its Infancy, Casanova, J.-L. Abel, L., AMERICAN ASSOCIATION FOR THE ADVANCEMENT OF SCIENCE, 2007

³ ibid

⁴ ibid

⁵ ibid

autoimmune or rheumatic disorder.

PID is treatable

- Certain patients can be treated with immunoglobulin (IG) therapies, which replace or supplement the ineffective antibodies that the patient fails to produce.
- These complex medicines are derived from donated human plasma. The therapy is either infused into the vein (intravenously) or into the layer of fat just below the skin (subcutaneously).
- Early diagnosis and access to appropriate treatment make it possible for PID patients to live a normal and productive life.
- Other treatments such as antivirals, antibiotics and antifungals, granulocyte colony stimulating factor can be utilised to back up first-line treatment such as immunoglobulin replacement therapy. The imperative for a patient is that their medical support system finds the combination of treatments that are most effective for their condition.
- In the most severe cases, bone marrow or stem cell replacement may be appropriate.

Warning Signs

The PID Community recommend that all patients affected by two or more of the “**10 Warning Signs**” should be tested for PID. These signs are:

1. Eight or more new ear infections within 1 year
2. [Two or more serious sinus infections within 1 year](#)
3. Two or more months on antibiotics with little effect
4. [Two or more pneumonias within 1 year](#)
5. Failure of an infant to gain weight or grow normally
6. [Recurrent, deep skin or organ abscesses](#)
7. Persistent thrush in mouth or elsewhere on skin, after age 1
8. [Need for intravenous antibiotics to clear infections](#)
9. Two or more deep-seated infections
10. [A family history of primary immunodeficiency](#)

Raising awareness in doctor surgeries and in clinics

All medical professionals are encouraged to learn more about PID, to recognise PID symptoms and educate other primary caregivers in their surgeries and clinics. The following are some actions that can be taken by medical professionals:

- There are several posters available that can be posted on notice boards. Various other material that explain PID are available on the IPOPI and World PI Week websites⁶;
- Monitor and educate patients and warn them of implications of going untreated;
- Get in contact with your local immunodeficiency society to find out how you can get involved in their initiatives;
- Review the guidelines on how to detect primary immunodeficiency diseases and how to

⁶ www.ipopi.org; www.worldpiweek.com

implement a screening program.

What can be done at a national level?

It is important that the medical community work and support governments to take the necessary steps to appropriately diagnose and manage PID with a focus on the following actions:

1. Raise awareness of primary immunodeficiencies
2. Educate healthcare professionals and promote exchange of expertise
3. Focus on early diagnosis and screening
4. Ensure access to treatment
5. Organise data collection

1. Raise awareness of primary immunodeficiencies

Implement targeted campaigns to increase recognition of PID among medical students, family doctors, the medical profession and nurses, with the objective of increasing understanding of the disease and available testing methods and treatments.

2. Educate healthcare professionals and promote exchange of expertise

Work together with government and medical specialists organisations such as: AAAI, EFIS, ESID, INGID, IUIS, and other national or regional professional organisations for nurses and doctors involved in the management of PID to:

- provide standards for basic and applied immunology training, with emphasis on PID, in the education programmes for general practitioners and nurses;
- integrate basic and applied immunology teaching into training for general paediatric internal medicine, rheumatology, respiratory and gastro-entriology medicine, and infectious diseases;
- develop cross country/regional initiatives to allow the exchange of experience and education, including capacity to network and funding of visits by immunology centres representatives from other regions/countries.

3. Focus on Early Diagnosis and Screening

- Develop clinical diagnostic protocols to reliably identify all forms of PID;
- Promote the development and encourage the use of simple diagnostic tests for PID at local level;
- Implement a system for recognition of symptomatic patients and ensure appropriate immunologic and genetic laboratory tests are available at national level via centres of excellence;
- Promote newborn screening programmes to ensure early detection.

4. ***Ensure access to treatment***

- Ensure satisfactory access to treatment for people with PID as it has been shown to prevent recurring infections, improve social well-being and reduce the burden of the disease for the person with a PID and their family;
- Ensure that safe immunoglobulin treatments are available to all patients who require antibody replacement;
- Recognise the social needs of PID patients and their family and facilitate their access to adequate support services such as physiotherapy;
- Ensure the reimbursement of orphan drugs according to their medical efficacy for the patient and not according to the price of the product.

5. ***Organise data collection***

- Set up a national PID patient registry. Work on maintaining consistency with other national registries in order to facilitate international collaboration. Models of national registries are available on www.esid.org;
- Participate in epidemiological studies to assess prevalence and incidence of PID in the population;
- Encourage the creation of and input into international registries which will enable future diagnostic processes by identifying: pattern of clinical presentation of these diseases; natural history of the various PID (morbidity, mortality, complications); relationships between clinical disease patterns and genetic backgrounds;
- Encourage transnational cooperative research and multinational clinical trials;
- Encourage the establishment of Centres of Reference to promote best practice in terms of disease classification, treatment outcome measures and cost assessments of treatments. The French PID study group: CEREDIH, which has been funded through the French National Rare Diseases Plan, is an excellent model of how Rare Disease programmes can benefit the PID community.