The global organisation working to improve the quality of life for people with primary immunodeficiencies.

PID Leaflet

FACTS ABOUT PRIMARY IMMUNODEFICIENCIES

WWW.IPOPI.ORG

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, physicians treat the infections, with repeated prescription antibiotics, while missing the underlying cause, allowing the illnesses to recur, and leaving the patient vulnerable to vital organ damage, physical disability, and even death.

Early diagnosis and access to appropriate treatment enables people living with a PID to lead normal productive lives whilst significantly reducing healthcare expenditure. Lack of awareness of PID remains a major issue, and due to this problem the majority of patients are diagnosed too late. Unlike other rare disorders, there are many effective treatment options for PID patients, which can enable them to carry out a normal life.

PID Facts

- PID affects at least **10 million** people worldwide.
- There are currently **200** 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 health.
- It is estimated that **70–80% of PIDs remain undiagnosed**, globally.
- A simple and relatively inexpensive **blood test** Complete Blood Count (CBC) **can identify over 95% of PID patients.**

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 autoimmune or rheumatic disorder.

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

World PI Week
22nd - 29th April 2011

www.worldpiweek.org

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Diagnosis

The first step in diagnosing a Primary Immunodeficiency disease is a good evaluation. **Paediatricians and immune system specialists (i.e. immunologist)** can help with diagnosis and treatment.

Evaluation of the immune system may include:

- Detailed medical history
- Physical exam
- Blood tests
- Vaccines to test the immune response

If a PID disease is suspected, a series of blood tests and vaccines may be required. Blood tests will show if any part of the immune system is missing or not working properly.

PID Warning signs

Correct diagnosis of a PID disease begins with awareness of the **10 Warning Signs**

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

What treatments are available?

Once a diagnosis is established, much can be done for people living with PID. At a minimum, the recurring infections can be treated with low or moderate doses of appropriate antibiotics. These can help prevent permanent organ damage, thus promoting the patient's long-term survival while improving the quality of life. When appropriate, immunoglobulin therapy is used for a wide range of PID diseases. Bone marrow transplantation and gene therapy is also used for more severe disorders. Advanced treatments such as the interleukins, PEG-ADA, and gamma interferon can also help in some complex cases.

Is nutrition important?

Good nutrition gives the body the energy and the resources to fight infections. Eating well always makes good sense, and families with antibody deficiencies should take extra care to maintain a healthy and balanced diet.

Is it possible to lead a normal life?

Thanks to new therapies, greater public awareness, and better access to information, many people with PID are leading normal lives - going to school, work, playing sports, and enjoying a better quality of life. There has never been more hope for people with PID.

About IPOPI

IPOPI is the Association of national patient organisations dedicated to improving awareness, access to early diagnosis and optimal treatments for primary immunodeficiency (PID) patients worldwide. Established in 1992, IPOPI works as the global advocate for the PID patient community in cooperation with its National Member Organisations (NMOs) and other global stakeholder groups including, among others:

- ASID, African Society for Immunodeficiencies
- CIS, Clinical Immunology Society
- EFIS, European Federation of Immunological Societies
- ESID, European Society for Immunodeficiencies
- INGID, International Nursing Group for Immunodeficiencies
- JMF, Jeffrey Modell Foundation
- LASID, Latin American Society for Immunodeficiencies

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