

# Key Messages for Key Audiences

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## I. Key messages

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### Facts about PI:

- PI affects at least 10 million people worldwide<sup>1</sup>.
- While each of the more than 200 identified PI conditions<sup>2</sup> may be rare, taken together, they are more common than childhood leukaemia and lymphoma combined and has four times the incidence of cystic fibrosis.
- Around 70–80% of PI remain undiagnosed<sup>2</sup>.
- Current diagnosis levels suggest that around 1 in 8-10,000 people have a genetic primary immunodeficiency that significantly affects their health<sup>2</sup>
- A simple and relatively inexpensive blood test CBC (Complete Blood Count) can identify over 95%<sup>2</sup> of the patients.
- Lack of awareness on PI remains a major issue, and due to this deficit, the majority of patients are diagnosed too late.
- Unlike many rare diseases, there are many effective treatment options for PI patients, which can enable them to carry out a normal life. PI patients must therefore be diagnosed early and informed about the most adequate treatment for their particular condition.
- None of the available therapies can reverse the damage of late diagnosis, which is why awareness and education is paramount across the World.

### The importance of early detections:

- The PI Community recommend that all patients affected by two or more of the “10 Warning Signs” should be tested for PI. These signs are:
  1. Four or more new ear infections within 1 year
  2. Two or more serious sinus infections within 1 year
  3. Two or more months on antibiotic 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 fungal infection on skin
  8. Need for intravenous antibiotics to clear infections
  9. Two or more deep-seated infections including septicemia
  10. A family history of PI
- Detecting the disease early can save lives and avoid unnecessary vaccinations that can cause serious illnesses and even death for undiagnosed PI patients.
- Detecting the disease early can save patients from lifelong disabilities like hearing loss or lung scarring.

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

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



- Detecting the disease early can significantly improve the patient’s quality of life, by reducing the usually numerous visits to the doctor and stays in hospitals. If treated early, the patient can get back to school or work.
- Detecting the disease early has huge benefits for the patients, their family and society as a whole

### The cost of late diagnosis:

- PI are the reservoirs of severe infections – bacterial, viral, and fungal and can therefore present a serious threat to schools, day care centres, the workplace and even hospitals.
- Studies show that one hospitalisation with pneumonia is more costly to the healthcare system than one year of appropriate treatment with immunoglobulins.
- A peer-reviewed study documenting the financial impact associated with early diagnosis and management of PI in the United States found that each undiagnosed patient with an underlying PI costs the healthcare system an average of 75,488 Euro [\$102,552] annually, while after diagnosis and treatment costs to the healthcare system averaged 16,633 Euro [\$22,610] annually; yielding an average savings of 58,792 Euro [\$79,942] per patient annually.

### The major problem - lack of awareness of PI:

- There is a lack of awareness of PI across the World. A concerted effort at both national, regional, European and International level needs to be undertaken in order to raise awareness of PI amongst the general public, primary caregivers (GPs and nurses), specialists and government officials in terms of:
  - Disease recognition
  - Recognition of PI symptoms and their association with these diseases
  - Impact on quality of life
  - Early detection methods
  - Availability and efficacy of treatment options
  - Cost of the disease

### Steps governments need to take to understand, appropriately diagnose and manage PI in their respective countries:

- Raise awareness on Primary Immunodeficiencies
- Educate health professionals and promote exchange of expertise
- Promote early diagnosis and screening
- Gather knowledge and data (e.g. epidemiological studies, creation of registries, Centres of Reference,..)
- Ensure adequate and easy access to treatment

## II. Key Audiences

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- Government/Health Authorities/Ministers of Health
- WHO / UNICEF and Other International health-related organisations
- Medical Professionals (GPs, nurses, related specialists)
- General Public



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- Existing PI Patients
- Schools
- International and local media
- Industry to promote further research and involvement
- The media / celebrities –to multiply the message
- Medical Press