About primary immunodeficiency

- Primary immunodeficiencies are rare diseases which occur when a person's immune system is absent or does not function properly. When a defect in the immune system is inherited (carried through the genes), it is called primary immunodeficiency. There are over 380 forms of primary Immunodeficiency (PI or PID), ranging widely in severity.
- Primary Immunodeficiency often presents in the form of "common" infections, sometimes leading physicians to treat the infections while missing the underlying cause, allowing the infections to reoccur, and leaving the patient vulnerable to vital organ damage, physical disability, and even death.

Early testing and diagnosis

- It is estimated that 6 million people worldwide are living with primary Immunodeficiency
- 70-90% of people with primary immunodeficiencies are still undiagnosed worldwide
- A number of different signs and symptoms can help detect primary immunodeficiencies as they occur commonly in many types. The most common are severe, persistent and/or recurring infections such as ear, sinus, or skin infections, as well as inflammation in the lungs, liver and intestines. Although it varies from individual to individual and from disorder to disorder, those signs can enable doctors to refer the patient for further medical examinations. It is also important that family carers and the public are aware of symptoms to visit an immunologist in case of suspicion.
- The first step in diagnosing primary immunodeficiencies consists of a detailed evaluation of the immune system through several types of testing including blood and/or vaccine tests, reviewing medical and family history, physical examination. The initial testing is usually performed via blood testing, followed by further tests of the immune system. Genetic testing is often needed to identify the particular form of primary immunodeficiency and the appropriate care pathway, however it is not yet available in many countries.
- For severe forms of primary immunodeficiency, newborn screening is a lifesaving, cost effective method to drive early diagnosis and save lives. Routine newborn screening programmes for severe primary immunodeficiency (e.g., SCID and XLA) should be widely implemented in public healthcare settings in all countries.

Care & treatment

Patient access to care and appropriate treatment helps to prevent serious and life-threatening complications in people with primary immunodeficiency

Treatment options: Treatment for primary immunodeficiencies depends on the type of disease and other factors. Several treatment options exist, which include immunoglobulin (IG) therapies, haematopoietic stem cell transplantation (HSCT or BMT), gene therapy, granulocyte-colony stimulating factor (G-CSF), and other advanced treatments such as anti-fungals, prophylactic antibiotics, interleukins, Gamma interferon or PEG adenosine deaminase, among others. Immunoglobulin therapy is the accepted protocol for a wide range of primary immunodeficiencies. Most patients should receive immunoglobulin replacement therapy on a regular basis to prevent further recurring infections. This treatment prevents permanent damage to the lungs, organs and bronchial tubes, therefore promoting the patient's long-term survival while improving the quality of life.

About Immunoglobulins (IG):

- IGs are made from human plasma: as plasma derived medicinal products, plasma and blood donations are crucial to produce immunoglobulin therapies. Patients rely on the commitment of plasma donors.
- How IGs work: IGs replace the protective antibodies that are missing in the immune system, without which the patients would be prone to recurring and severe infections. They can be administered either intravenously or subcutaneously.
- IGs are not generic medicines: No single immunoglobulin therapy or delivery method is suitable for all individuals. IGs are included in the World Health Organisation (WHO) Lists of Essential Medicines for both children and adults. Countries should provide access to the broadest spectrum of immunoglobulin therapies as possible.

Quality of life

- Early diagnosis and appropriate treatment can prevent complications and have considerable impact on the lives of people with primary immunodeficiencies and their families.
- As a result of new therapies, greater public awareness, and better access to information, many patients are leading more normal and productive lives - going to school, work, playing sports, enjoying social activities and a better quality of life.
- Patients may have difficulties coping with the many medical appointments, treatment regime and personal life as well as the psychological aspects of living with a chronic disease.
- Raising awareness among the patients' social environment, general public, medical professionals and policy makers helps improve understanding of the disease in support of people with primary immunodeficiency.