



Research is hope for early diagnosis, treatment and care of Primary Immunodeficiency

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Today the Primary Immunodeficiency (PI) community joins forces with rare disease advocates in celebrating Rare Disease Day 2018 around the theme of research. On this important occasion, the World Primary Immunodeficiency Week (WPIW) outlines the importance of medical research and encourages Governments, health authorities, and investors in all countries to further support and stimulate rare disease research to change the lives of people with PI.

70-90% of people with PI worldwide remain undiagnosed. Because Primary Immunodeficiencies - which are rare, genetic diseases occurring in people with an absent or deficient immune system - often manifest themselves in the form of recurring and “common” infections in individuals, diagnosis is complex. General practitioners and healthcare professionals may face difficulties in making an early and appropriate diagnosis due to those commonalities between PI symptoms and other recurrent infections. Furthermore, lack of scientific knowledge of the diseases contribute to a delayed diagnosis.

Medical knowledge of PI has been increasing, but there is still a long way to go to fully understand these life-impairing, complex conditions. Diagnostic challenges and gaps in scientific knowledge can be tackled with research. Research means hope for an improved quality of life for the estimated 6 million people living with PI worldwide: it is key to improve the understanding of PI, leading to early testing and definitive diagnosis, but also to support the development of innovative treatments and care.

This year, World PI Week (22nd-29th April) will gather the PI community around the theme “*My future starts with early testing and diagnosis of PI*” highlighting how testing and diagnosis are essential first steps in the care pathway and are life-enhancing for patients with PI and other rare diseases.

Therefore, on Rare Disease Day, the PI community unites under the World PI Week to encourage policy-makers, health regulators and investors to: maintain and further support PI research, by exchange of scientific knowledge and good practices worldwide in the collection of data as well as by developing new, effective care models and treatments; to recognise the central role that patients play in research processes hence to fully engage them. These can be achieved by prioritising research in political agendas and funding schemes. In the context of political discussions on the financial framework for research post 2020 in the European Union (EU), the World PI Week especially invites EU decision-makers to ensure that research funding is not withdrawn nor reduced, but rather increased to meet rare disease research needs. This will ultimately benefit the population and sustain the tireless efforts of medical researchers.

PI represent more than 350 of the 6,000 recognised rare diseases, and as the significant research and medical progress of the past decades testify, research remains the backbone of any improvements in the diagnosis, treatment and care of patients worldwide.

For more information on **Rare Diseases Day 2018**: www.rarediseaseday.org

For more information on **World PI Week 2018**: www.worldpiweek.org

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