Rare disease priorities in the European Parliament 2019-2024: the voice of PID patients

March 2019

Introduction

On 27 November 2018, IPOPI[1] organised its 12th PID Forum called “Rare disease priorities in the European Parliament 2019-2024: the voice of PID patients” at the European Parliament in Brussels (Belgium). The event was hosted by the Member of the European Parliament (MEP) Mr José Inácio Faria (EPP, Portugal). The meeting brought together various stakeholders such as MEPs, patient organisations, academics, healthcare professionals, industry representatives and other stakeholders. The meeting provided IPOPI with a platform to recall the work they achieved during the 8th term of the European Parliament and prepare the ground for the next five years.

This document is the result of the 12th PID Forum as the meeting saw several policymakers calling for further actions in the field of rare diseases. Through this document, IPOPI and the MEPs aim to ensure the prioritisation of care for patients with PIDs and to promote opportunities for healthcare professionals and researchers during the European Parliament’s next legislature. While awareness of PID care has significantly improved in recent years, a significant amount of work remains to be done. The EU needs to prioritise the delivery of lifesaving interventions across the region. This is especially true in the case of newborn screening for rare diseases and the promotion of equal and continuous access to immunoglobulin replacement therapies (IGs) and other therapies such haematopoietic stem cell transplantation or gene therapy for PID patients.

Call to action for political priorities 2019-2024 in the field of rare diseases

The European Union has a long legacy of caring for rare disease patients and investing in researching this field. EU interventions can substantially impact the lives of PID patients and can positively influence PID care and rare diseases care in general.

Despite the EU principle of subsidiarity in health-related matters, the EU’s added value in the field of rare diseases including PIDs cannot be underestimated. The European Reference Networks (ERNs), established in 2017, are a clear illustration of the positive strides the EU has taken on health-related matters. In addition to ERNs, EU research funding, access to

[1] IPOPI is the International Patient Organisation for Primary Immunodeficiencies. As the association of national patient organisations, it is dedicated to improving awareness, access to early diagnosis and optimal treatments for primary immunodeficiency (PID) patients worldwide.

IPOPI is a charity registered in the UK. Registration No. 1058005.

cross-border healthcare, and the mobility of healthcare professionals are further examples of how the EU can ensure high-quality PID care and its availability.

The EU significantly contributes to improving the lives of rare disease patients, and IPOPI calls on to have rare diseases maintained high on the political agenda in the next parliamentary term so as to secure continued political support for initiatives on rare diseases.

Members of the European Parliament should keep rare diseases high on their agenda through continued political support for rare disease initiatives both at European and national levels.
Patients should be actively involved in the decision-making processes, especially in the definition of legislative provisions directly affecting their daily lives.

Rare disease diagnosis and patient referral

The European Union's recent effort in improving timely access to treatment for PID patients deserve to be highlighted. However, the problem of delays in patient referrals persists to this day. Opportunities for timely interventions are regularly missed because a PID is hiding behind common symptoms. Common symptoms can be responsible for misdiagnosis and it is estimated that 70% of PID patients are undiagnosed on the global scale.

Newborn screening for rare diseases has a key role to play in enabling prompt diagnosis and timely access to life saving specialist treatments. This is why the implementation of newborn screenings programmes for rare diseases is crucial as they would allow for a prompt diagnosis and effective treatment strategies for patients with severe immunodeficiencies.

There is a demonstrated need for the European Union to put measures in place which can improve the diagnosis of PID patients. This begins with creating an EU-wide platform for the introduction of newborn screening for rare diseases. Moreover, the EU should position itself as the central point for data and information on rare diseases newborn screening practices. The pooled data could potentially be linked with the already established ERNs.

The European Union can have significant added value in encouraging uptake of the newest scientific evidence in newborn screening for rare diseases among Member States. To this end, the EU should actively support a better coordination and exchange of best practices in the field.

The development and the implementation of an overarching framework/guidelines in the field of newborn screening for rare diseases can greatly benefit patients, reducing delays in referrals to diagnosis and increasing timely access to appropriate treatment. To this end, policy-makers should encourage the introduction of a harmonised access to newborn screening in all Member States, which is crucial for PIDs and particularly for Severe

IPOPI is a charity registered in the UK. Registration No. 1058005.

Combined Immunodeficiency (SCID) as well as for numerous other rare diseases.

PIDs diagnosis in Europe is not yet optimised as the proportion of patients who receive an accurate and timely diagnosis – a crucial component for successful PID management and care – is not satisfactory. Increased awareness on PIDs on the different levels of medical education, as well as amongst those specialists that are more likely to see a patient with a PID not yet detected is key. Additionally, the ERN on immunodeficiency, autoinflammatory and autoimmune diseases (ERN-RITA) can potentially improve equal access to genetic diagnosis, a key healthcare challenge across Europe. ERNs foresee a clinical patient management system which aims to enable sharing of patient data, provide advice on diagnosis and treatment and archive patient data. To fully benefit from the ERNs there is a need to recruit more HCPs in the countries which are currently not represented; ensure use of advanced cross-border information technologies; and raise awareness on PID for better patient referral.

A standardized and uniform European-wide approach for genetic molecular diagnosis of PIDs is needed to ensure access for genetic diagnosis for all patients.

Availability of life-saving therapies for PIDs and rare disease patients

It is essential for PIDs and rare disease patients that the European Union ensures better availability of life-saving therapies for them. The need for individualised health plans for PID patients means that the availability of multiple therapies is a necessity for the effective treatment of their condition. However, inequalities in care still exist throughout Europe and need to be tackled in a centralised and effective manner. For this reason, we need to ensure the availability of an adequate framework to support the development of therapies for rare disease patients (including orphan medicinal products and plasma-derived medicinal products).

Members of the European Parliament should advocate for better and sustained access to treatment for PIDs and rare disease patients. This could be done by ensuring a favourable political framework for the availability of a wide range of products and calling for the treatments to be delivered on the basis of the medical prescription allowing these patients to have access to the most suitable treatment.

Upcoming legislative provisions on Health Technology Assessment should also take into account the particularity of various medicinal products. For PIDs, it is key to understand that immunoglobulin replacement therapies are not interchangeable medicines and that each of them will be tolerated by a patient differently. Patients with PIDs require an individualised approach and that should also be reflected in any HTA appraisal.

Risks in the supply of immunoglobulins and plasma-derived medicinal products are the major safety concern for patients with PIDs relying on a stable source of their live-saving treatments. Replacement immunoglobulin therapy is needed for the life of the patient and

IPOPI is a charity registered in the UK. Registration No. 1058005.

for most patients, especially adults, there are no alternative therapies. This therapy should be prioritised for PID patients in times of shortages. The EU should work on strategies to increase EU plasma supply and collection, have clear definitions on key concepts, and the revision of evidence-based selection and deferral of donors amongst other measures that should be patient-centred.

Access to bone marrow transplantation, gene or advanced therapies should be equal for patients with rare conditions in different Member States. A sustained and equal access to these therapies should be promoted throughout the EU.

Transition of care and elderly care

Transitioning from paediatric care to adult care has numerous implications for people with PIDs through repercussions on their personal lives which can range from physical, social, educational, and/or domestic disturbances. Frequently overlooked or mismanaged, the transition is often inadequately planned and ignores the individual needs of the patient. Proper management of this transition would be a cost-effective way of ensuring compliance with treatment in adult life.

Moreover, discrepancies amongst national health systems, in addition to the stigma of PIDs being sometimes wrongly perceived as paediatric conditions only, mean that care must address specific stages in life where support is a key aspect to ensuring the wellbeing of the patient. Identifying best practices for the transition of care and elderly care of people with a PID has a central role in improving patients' quality of life.

Policy-makers, together with patients, should advocate for the need of a defined and coordinated pathway guiding young patients toward adult services. Planned transitional care for PID patients is often overlooked but it is a cost-effective way of preventing poor compliance. This will help preventing adolescent patients being lost in the healthcare system, which will lead to poor compliance with treatment, potential irreversible organ damage, lower life expectancy, and reduced quality of life.

Policy-makers at European and national level should support the inclusion of elderly healthcare into national rare disease plans where the establishment of a framework of accessible and sustainable home care and long-term care services for elderly PID patients would play an essential role.

Policy-makers should advocate for targeted measures with regards to infrastructure, the supply of adequate health care and care services, as well as participation in economic, political and social life. Clinical guidelines should also be developed to ensure that care provided to elderly patients with PIDs is adequate to this population's needs. IPOPI's PID Principles of Care recommends such guidelines.

IPOPI is a charity registered in the UK. Registration No. 1058005.

European Reference Networks

The European Reference Networks (ERN) are a means to establish a structured and voluntary collaboration among the EU Member States in the field of healthcare. These cross-border networks bring together healthcare providers across Europe to tackle rare and complex diseases that require highly specialised healthcare knowledge. The ERNs give access to a much larger, cross-border pool of expertise and knowledge, increasing the chances of patients to receive the best advice to treat and diagnose their diseases. By collecting a large amount of patient data, the ERN will, in the longer term, facilitate large clinical studies to improve the understanding of diseases or support the development of new medicinal products.

European Reference Networks (ERNs) are virtual networks involving healthcare professionals (HCPs) across Europe. Launched in 2017, they aim to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment and concentrated knowledge and resources. Their potential to improve quality, safety, and access to specialised healthcare for patients affected by a rare disease is significant as they raise awareness of PIDs and enable better patient referrals.

Currently, certain countries are seeing the added-value of ERNs limited by the fact that there is a clear need for more HCPs. To render ERNs more effective, the EU should also ensure a widespread use of advanced cross-border information technologies.

The EU should continue supporting and funding ERNs to improve diagnosis and outcomes of rare disease patients', including those of PID patients. ERNs are the best possible tool to ensure that expertise, care and research on PIDs and other rare diseases is spread throughout Europe, including Eastern European countries.

Research funding

When developing its policy on rare diseases, the European Union considered that these diseases affect so few people that combined efforts are needed to reduce the number of people contracting the diseases, prevent new-borns and young children dying from, and preserve sufferers' quality of life and socio-economic potential. Thus, through Framework Programme 7 (FP7) and Horizon 2020, the EU has ensured that over EUR 1 billion was invested in research on rare diseases by funding more than 200 collaborative projects related to rare diseases. Patients with rare diseases, such as PID patients, are highly dependent on research and innovation which leads to the development of new medicines and treatments. Research projects supported through the EU research budget such as

IPOPI is a charity registered in the UK. Registration No. 1058005.

SCIDNET[2] or RECOMB[3] show the potential of collaboration in the development of therapies for very rare conditions, such as severe combined immunodeficiencies. The European elections, in May 2019, coincide with preparations for the next EU budget and consequently for the next Research and Innovation programme: Horizon Europe.

The EU should build on the achievements and success of Horizon 2020 and keep itself at the forefront of global research and innovation. Given that EU research funding and collaborative research networks are crucial factors in ensuring high-quality care for PIDs and rare diseases, the EU should strive to ensure rare diseases funding remains a priority for the next EU legislature. The EU should pledge to commit a significant percentage of Horizon's Europe budget to funding innovative rare diseases projects.

Education of healthcare professionals

Many PIDs are complex and require genetic testing to ensure the best possible diagnosis. In some EU countries, genetic diagnosis for PIDs is still unavailable. In other cases, the specific PID could be easily diagnosed by a PID specialist. Problems arise when general practitioners and other healthcare professionals do not suspect a PID and patients are confronted with many tests and repeated visits to specialists before getting the right diagnosis.

In order to reduce the number of undiagnosed and misdiagnosed rare disease patients policy-makers should further endorse policies raising awareness on primary immunodeficiencies.

Member States should ensure the use of advanced cross-border information technologies and raise further awareness on PID for better patient referral.

As of March 2019, the manifesto is endorsed by:

Members of the European Parliament

José Inácio Faria (EPP, Portugal)

Anna Záborská (EPP, Slovakia)

Sirpa Pietikäinen (EPP, Sweden)

Tilly Metz (Greens/EFA, Luxembourg)

[2] SCIDNET is a research project that looks into gene therapy as a curative option for over 80% of all forms of SCID in Europe. It is financed through funding from the European Union's Horizon 2020 research and innovation programme. For more information, please refer to: https://scidnet.eu/

[3] EU funded research project that looks into creating a novel treatment for one of the most common types of SCID, RAG1-SCID by performing phase 1 and phase 2 clinical trials using autologous haematopoietic stem cell–based gene therapy. RECOMB has received funding from the European Research Council (ERC) under the European Union's Horizon 2020 research and innovation programme. More information is available at: http://www.recomb.eu/project/

IPOPI is a charity registered in the UK. Registration No. 1058005.

Patient representatives

Martine Pergent	President, IPOPI
Johan Prévot	Executive Director, IPOPI
Dr Susan Walsh	Director, PID UK
Savvas Savva	Secretary, Cyprus PID Association
Christine Jeffery	Executive Officer, Australian PID association (IDFA)
Bruce Lim	President, Malaysian PID association (MyPOPI)
Jose Drabwell	Board member, IPOPI
Andrea Gressani	Vice-President, Italian PID association (AIP)
Violetta Kozhereva	Vice-Chair, Russian patient support group (OPPID)
Roberta Anido de Pena	President, Argentinian PID association (AAPIDP)
Annie Pienaar	Chair, South African PID association (PiNSA)
John Seymour	Chair, US PID association (IDF)

Medical experts

Dr Nizar Mahlaoui	Necker-Enfants Malades University Hospital, Paris (France)
Prof Martin van Hagen	Erasmus Medical Center Rotterdam (the Netherlands)
Dr Mirjam Van der Burg	Leiden University Medical Centre (the Netherlands)
Prof Surjit Singh	Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh (India)
Prof Isabelle Meyts	Catholic University of Leuven (Belgium) President, European Society for Immunodeficiency (ESID)
Prof Antonio Condino Neto	Sao Paolo University (Brasil)
Dr Olaf Neth	University Hospital Virgen del Rocio, Sevilla (Spain)
Prof Helen Chapel	University of Oxford (UK)
Prof Bobby Gaspar	University College of London (UK)
Prof Tadej Avcin	University Medical Center Ljubljana (Slovenia)
Prof Klaus Warnatz	Medical Center – University of Freiburg (Germany)

IPOPI is a charity registered in the UK. Registration No. 1058005.