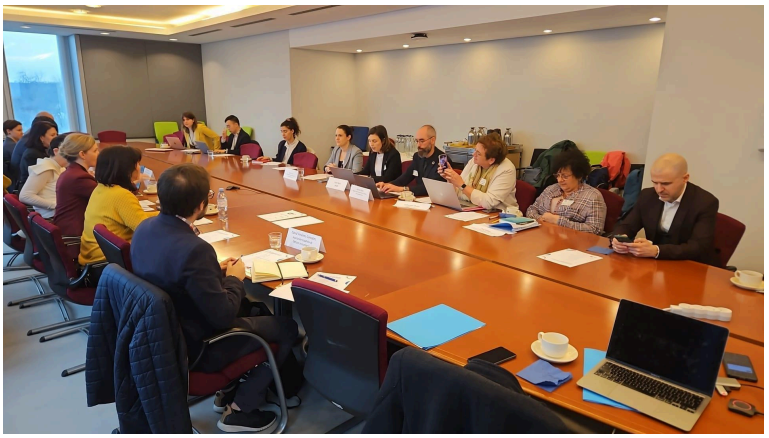
Event Report



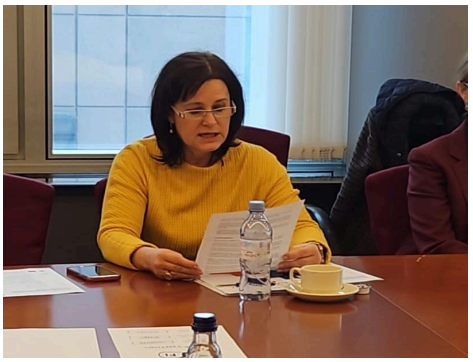
PID Forum - Empowering the PID Community Through Rare Disease Policies 2024-2029

21 March 2024

On 21 March 2024, the International Patient Organisation for Primary Immunodeficiencies (IPOPI) organised a PID Forum titled "*Empowering the PID Community Through Rare Disease Policies 2024-2029*". The event was hosted by **Member of the European Parliament (MEP) Radka Maxová** (S&D, Czechia) in the European Parliament.

Opening the event, moderator **Leire Solís,** health policy and advocacy senior manager at IPOPI, underscored the significance of the upcoming European elections in shaping policy-making for the next five years, particularly in the realm of rare diseases. She underlined IPOPI's intention to publish a 2024-2029 Manifesto that would include the demands of patients with primary immunodeficiencies (PIDs) as they relate to unmet needs, access to diagnosis, and the affordability of treatment and care.

Welcome address



In her opening remarks, **MEP Radka Maxová (S&D, Czechia)** highlighted the strides made by the EU in addressing rare disease challenges and cross-border healthcare challenges in recent years. She noted that a number of files, such as the Pharmaceutical Package, the European Health Data Space (EHDS) and the Substances of Human Origin (SoHO) regulation, have brought rare diseases to the forefront of the EU health agenda. However, despite these advancements, MEP Maxová stressed that issues

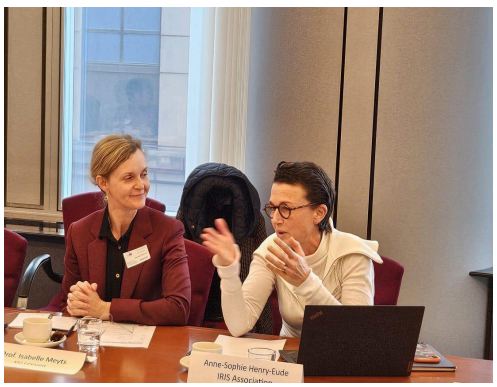
persist for rare disease patients, such as unequal access to medicines and affordability of treatments, medicine shortages and the lack of incentives to encourage the development of new medicine. She also called for continued efforts in research, cross-border collaboration, and patient-centred policy making to advance the European Health Union. MEP Maxová concluded with hopes that the efforts to date will serve as a basis for a broader conversation on rare diseases and PIDs.

Setting the scene

Ms Solís provided an introduction to PIDs and some of the challenges faced by patients with PIDs in getting a diagnosis, living with the disease and ageing with it. Ms Solís began by highlighting the delays European PID patients have to overcome to receive a diagnosis in Europe and highlighted variations in diagnostic delays across EU countries for the same PID. She highlighted that newborn screening can be a critical step to shorten diagnostic delays. It has the potential to significantly improve patient outcomes through early detection of the most severe types of PIDs such as severe combined immunodeficiency (SCID).

Ms Solís further addressed the profound impact of day-to-day challenges confronting patients after their diagnosis, including navigating appointments with multiple medical specialists, coping with medication shortages, and enduring lengthy travels for treatment. Ms Solís also spoke about ageing with a PID and what it means to grow older with a chronic and rare disease, the potential co-morbidities and subsequent treatment and care needed, or the disability status needed in some cases.

The EU's impact on rare disease patients and PID communities

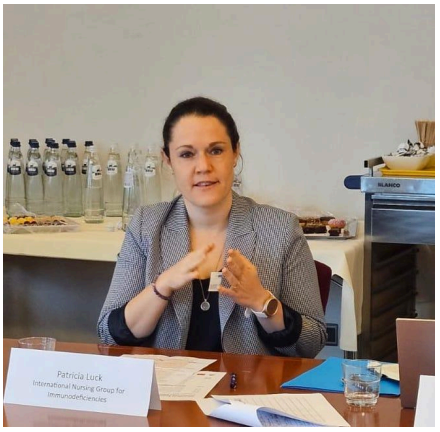


Ms Anne-Sophie Henry-Eude, a patient member of the French PID patient organisation IRIS, kick-started the panel discussion by sharing her own experience of living with a PID. She shared some insights into her diagnostic journey and the time it took her to receive a diagnosis as an adult, an arduous process of around 5 years.

Ms Henry-Eude emphasised that PID patients want to live as normal a life as possible, but that the realities of having a PID mean that it took her a year to identify where her therapy was available and the correct specialists in order to relocate to another country.

When asked about her wishes for the upcoming mandate, Ms Henry-Eude called for:

1. The swift implementation of the Substances of Human Origin (SoHO) regulation and more incentives for plasma donors.
2. The creation of a user-friendly list monitoring shortages. She also expressed her satisfaction with the extended mandate of the EMA.
3. The development of a list of available therapies in EU countries to improve patient access to vital information.
4. More support for PID patients who wish to study and work abroad.

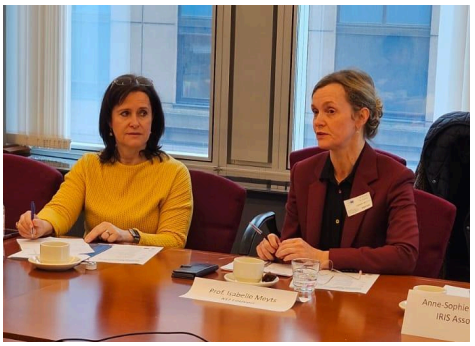


Ms Patricia Luck, Vice President of the International Nursing Group for Immunodeficiencies (INGID) spoke about her experience working as an advanced practice nurse in Immunology at University Children's Hospital Zurich. She spoke about her work in supporting the parents of children with a PID diagnosis.

To assist parents of babies with PID (B- and T-cell deficiencies) diagnosed through Newborn Screening (NBS), she launched a project focused on providing parents with a psycho-educational intervention supporting them in the first three months after diagnosis. The intervention provides a range of information (practical tips, illness knowledge and background information) as well as individual counselling sessions supporting them in their family management. Expanding on points raised by Ms Henry-Eude, Ms Luck specifically underscored the many practical hurdles confronting patients and their families in their daily lives, encompassing symptom management, medical appointment coordination, and the financial strains borne by families.

With this in mind, Ms Luck called for:

1. Greater financial support for families
2. Reducing the administrative burden on PID patients and healthcare professionals.
3. Greater investment in research, particularly in the PID and rare disease fields for all healthcare professions and encouraging personal and public involvement (PPI).



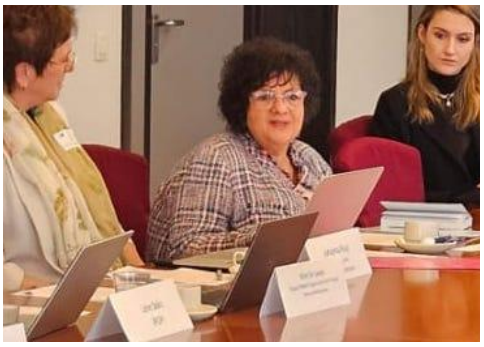
Building on this, **Professor Isabelle Meyts**, PID Physician Paediatrician at University Hospitals Leuven shared her perspective on the advancements in PID care over the past five years.

In particular, she noted: 1) the key role that European Reference Networks (ERNs) have played in deepening the understanding of rare diseases, like ERN RITA for PIDs and other immunological disorders; 2) the genetic revolution and the importance of finding a way to navigate genetic testing by favouring research; 3) access to treatment, including the availability and affordability of therapies, as

well as tackling inequities in the access to them; 4) mental health and disability status as two important aspects to be taken into consideration for PIDs.

Speaking about what she would like to see in the future, Professor Meyts called for:

1. Supporting rare disease research and integrating PIDs and rare diseases into the medical curriculum to ensure a broader understanding of these conditions and better patient support.
2. Streamlining and improving the use of data.
3. Increasing access to genetic diagnosis.
4. Treatment availability and affordability to enhance equity of access.

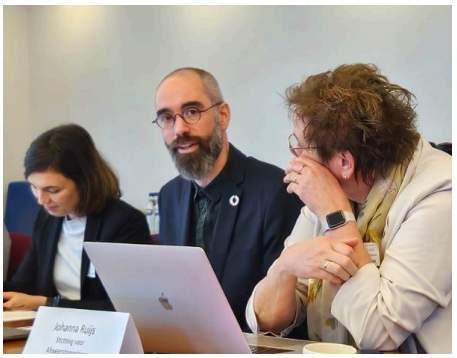


Dr Eva Varga, Vice President of the Hungarian Organization for Patients with Immunodeficiencies, shared insights into her experience of being both a PID patient and a general practitioner. She highlighted that in Hungary, around 600 patients benefit from regular immunoglobulin replacement therapies across 14 licensed centres. Previously, patients in Hungary faced shortages of immunoglobulin, but the national health insurance system has introduced an itemised financing approach for plasma-derived medicines, enhancing accessibility for PID patients.

Dr Varga also outlined notable achievements in Hungary, including the establishment of a national registry for PIDs and the involvement of healthcare units in the European Reference Network (ERN). Despite these advancements, she acknowledged enduring challenges in disease diagnostics and screening, particularly regarding conditions like SCID, the lack of representation of ERN RITA centre for PIDs in Hungary or the absence of coverage for the equipment needed for subcutaneous immunoglobulin replacement therapies.

In terms of future areas to work, Dr Varga highlighted the need for:

1. Easier (or more widespread) access to subcutaneous immunoglobulin replacement therapy
2. Accelerated diagnosis through genetic testing
3. SCID newborn screening in Hungary
4. Recognition of a Hungarian centre as an ERN RITA reference centre



Mr Wim De Geest, Chairman of the Belgian Patient Organisation for Primary Immunodeficiencies (BePOPI) focused on the experiences of PID patients and their families in Belgium, drawing a line between the complexities of the country's regional levels with the approach to public health at a European level. In particular, he referenced a story of a mother and the obstacles and turmoil she encountered when trying to access treatment for her two children with a PID. Mr De Geest echoed similar sentiments made by the

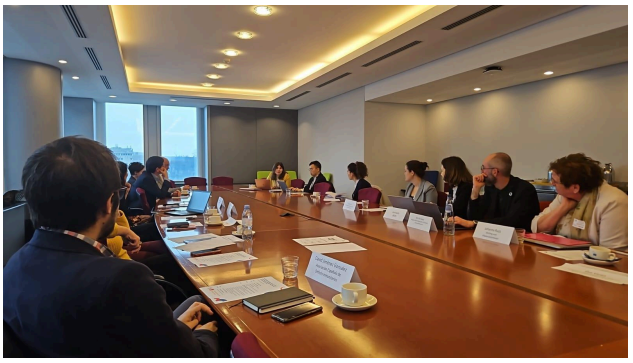
previous speakers, noting that medicine shortages are a major challenge for PID patients in Belgium. He also drew upon his own experience of being a father of a child with a PID and highlighted how these medicine shortages resulted in long hospital stays and long school absences.

Mr De Geest highlighted recent strides made in Belgium, such as the addition of SCID to neonatal screening programmes by the Flemish government. However, he also underscored how the ongoing complexities inherent in Belgium's multi-tiered governmental structure have added further complications to PID care in the country.

In terms of areas for improvement, Mr De Geest highlighted:

1. The development of an emergency plan for patients with PIDs in need of their therapy
2. Mental health support for patients with PIDs and their families
3. The establishment of a “care trail” for patients with PIDs (similar to other, more prevalent, diseases).

Open Floor Discussion



Mr David Jiménez González, board member of the Spanish PID patient organisation, Asociación Española de Déficit Inmunitarios Primarios (AEDIP) echoed the sentiments expressed during the panel discussion and underscored some of the positive impacts that the EU has brought to PID patients, such as the pharmaceutical package. More specifically, Mr Jiménez González called for more equality in diagnosis, treatment and care for patients in Spain and also across the EU.

Ms Kersti Urbala, President of the Estonian Patient Society for Primary Immunodeficiencies called for the implementation of SCID newborn screening in Estonia and also a swift access to early diagnosis of PIDs.

Ms José Verstegen, President of the Dutch PID patient organisation, Stichting voor Afweerstoornissen (SAS), spoke about the improvements in PID research in the Netherlands. She also highlighted some of the challenges facing PID patients, such as a lack of medical personnel equipped to care for PID patients and the shortage of immunoglobulin treatments for patients.

Ms Linda Engel, a member of the German PID patient organisation, Deutsche Selbsthilfe Angeborene Immunodefekte (DSAI), shared her own experience of living with a PID. She compared her experience to Ms Henry-Eude and the challenges of moving abroad as a PID patient. More specifically, Ms Engel

called for swifter diagnosis of patients with PIDs and for patient-centred care to ensure the successful integration of the treatment into a patient's daily life.

Closing Statements

In her concluding remarks, **MEP Radka Maxová** expressed gratitude to all participants for their valuable contributions, emphasising that the dialogue initiated during the PID Forum would surely extend beyond its confines. She strongly encouraged patient representatives to actively advocate for the establishment of a European Parliament Committee on Health in the upcoming mandate. Maxová highlighted the potential of such a committee to catalyse ongoing efforts in addressing some of the challenges associated with rare diseases that were outlined throughout the event.

Closing the PID Forum, Ms Solís thanked the event's sponsors as well as the representatives from the European Parliament, the medical sector, and patient organisations. She highlighted the insightful perspectives shared during the event which will inform the development of IPOPI's 2024-2029 Manifesto. She also emphasised the importance of this document in fostering ongoing collaboration and advocacy for a comprehensive rare disease strategy that could focus on supporting individuals with PIDs.