Empowering The PID Community Through Rare Disease Policies 2024-2029

Primary immunodeficiencies (PIDs) are a large and growing group of over 485 different chronic and genetic defects caused when some components of the immune system (mainly cells and proteins) are lacking or do not work properly.[1] People with these conditions are highly susceptible to infections, inflammation, autoimmunity, granulomas, severe allergies, and malignancies. Despite their rarity, PIDs collectively affect millions worldwide, significantly impacting the lives of patients and their families.

The International Patient Organisation for Primary Immunodeficiencies (IPOPI) calls on the European Union's (EU) policymakers to take steps to accelerate early diagnosis, patient access to safe and affordable medicinal products, innovative treatments and accessible care, including cross-border treatment when needed. Increasing awareness and funding for rare diseases can also support a more holistic approach to the non-medical needs of PID patients and improve quality of life and outcomes.

Ensuring fast and accurate diagnoses

Diagnosis of PIDs and rare diseases in general remains a challenge in many countries, no matter how developed they are. PID patients across the EU are often prone to illness, disability, organ damage and poor quality of life because of the consequences of lengthy diagnostic delays and treatment that is focused on symptoms.

We must increase awareness of PIDs amongst healthcare professionals, facilitate affordable access to diagnosis and work towards the integration of genetic testing into healthcare systems. By supporting patients and their families throughout the process, we can ensure they are appropriately cared for, irrespective of the rarity of their condition.

Newborn screening offers a unique opportunity to expedite diagnosis by allowing for early detection of some treatable rare diseases that are severe and asymptomatic at birth. EU Member States have different approaches to newborn screening, which have led to wide discrepancies in the evaluation of new techniques and the range of diseases screened at national and even regional levels.

Severe combined immunodeficiency (SCID) is an ideal candidate for newborn screening as infants who are diagnosed and receive treatment for this life-threatening PID before 16 weeks have a 96% survival rate.[2] However, less than ten countries in the EU currently screen for the condition nationally.[3]

EU policymakers should take decisive action to increase equity of access to newborn screening in the EU by supporting the development of comprehensive guidelines on newborn screening for rare diseases and act as a focal point for education and data sharing. The European Reference Networks (ERNs) are perfectly positioned to act as key figures in this process as their expertise can support the growth and ensure data collection and information on newborn screening practices is in line with scientific advancements.[4]

[1] Human Inborn Errors of Immunity, https://bit.ly/4cxLg4T
[2] The Value of SCID Newborn Screening, https://bit.ly/3TSfGHW
[3] Neonatal Screening in Europe Revisited: An ISNS Perspective, https://bit.ly/3JdBsjj
[4] Screen4Rare's Call to Action, https://screen4rare.org/calltoaction/

From digital data collection to Artificial Intelligence (AI), the digitalisation of healthcare has also created new pathways to timely diagnosis for rare disease patients. While IPOPI welcomes innovation, EU policymakers must ensure that it benefits patients. Digital tools should offer user-friendly interfaces that benefit data gathering and research. Moreover, the implementation of the **European Health Data Space (EHDS)**, for instance, should be accompanied by communication efforts that educate patients about how their data is being used and why.

Optimising access to medicines and treatments

Sustainable access to appropriate treatment and care is fundamental in the effective management of chronic conditions like PIDs. From anti-infectious to immunoglobulin replacement therapy or curative therapies, a range of treatments can treat PIDs, alleviate associated symptoms and improve quality of life.

Patients with PIDs need to have access to a **wide range of therapies**. This would improve patient adherence, management of their disease, and quality of life. Patients in need should be able to benefit from therapies wherever they are located and without incurring additional costs. This includes improving access to different types of immunoglobulin therapies, either intravenously or subcutaneously, and those involving a curative treatment such as hematopoietic stem cell transplantation, regenerative therapies or gene therapies.

A 2022 IPOPI survey found that two-thirds of surveyed patients in 15 EU countries had experienced a **shortage of immunoglobulin** in the past year. In 7 out of 15 countries, the shortage lasted more than 7 months and many patients were forced to adapt their treatment.[5] The EU is currently reviewing the legislation regulating blood and plasma, so that more can be collected for the development of live-saving therapies such as immunoglobulin therapies. In the coming years, national policy makers will need to work hard to promote other means to incentivise the collection of plasma at the national level if the EU is to better manage systemic shortages of these substance

Other PID treatments, such as anti-infectious and some biological medicines, face shortages, some even recurrent ones, due to a multi-faceted array of reasons.. We welcome the EU's intention to address these shortages, as per the proposal for a Pharmaceutical package or the extended mandate of the European Medicines Agency (EMA).

Finally, the EU's right to free movement means EU citizens are entitled to travel, work and live in another EU country.[6] However, PID patients in need of regular treatment can not readily enjoy this privilege. For them, long journeys abroad pose significant challenges as they require organisation, planning and management of their medical care. The EU should work towards ensuring EU citizens living with rare chronic illnesses have equal access to the right to free movement by supporting them in their efforts to work, travel and live in another EU country without undue burdens.

Promoting a qualitative, holistic approach to patient management and care

No patient should be left behind or feel like they are. Involving rare disease patient organisations in political discussions and public forums is a crucial pathway to educating decision-makers on the specifics of their conditions and ensuring the unmet needs are addressed.

[5] IPOPI PID Forum, https://bit.ly/49hxxMB
[6] European Commission, https://bit.ly/3vNDiE7

Patients require support from the moment they seek a diagnosis. Developing a patient-friendly system that helps patients overcome the hurdles of their condition would go a long way to ensuring that their quality of life is not severely impacted by the care they need. The EU and national decision-makers should therefore seek to create a "care trail" that ensures continuity of care for PID patients and facilitates the navigation of healthcare systems.

In the upcoming mandate, the EU must also prioritise continued investment in public health and rare diseases through EU4Health. Increased access to funds can help overcome systemic cross-border disparities and ensure a qualitative and holistic approach to rare disease and PID care that empowers patients to lead fulfilling lives.

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Many persons with PIDs fit the definition of the UN Convention on the Rights of Persons with Disabilities. Physically impaired patients should be recognised as such and receive the necessary financial and practical support to improve their quality of life.[7] Likewise, financial and practical support must be extended to mental health care. Rare diseases patients are disproportionately affected by mental conditions but the area is often overlooked and underfunded.[8]

Making rare diseases an EU priority for 2024-2029

Navigating the complexities of rare diseases demands concerted efforts from all stakeholders. As we embark on the 2024-2029 mandate, EU policymakers have a unique opportunity to enact meaningful change for PID patients by forging a path towards solutions that were previously deemed unattainable.

The EU must continue to support the development of new medicines and advanced treatments to prevent it from lagging behind in terms of medicine development and availability, especially important in the case of some gene therapies, and mitigate upcoming problems like antimicrobial resistance.

Prioritising patient needs, fostering cross-border collaboration and leveraging digitalisation are just some of the pathways to reduce the burden of many rare diseases. That is why IPOPI calls for the establishment of a permanent European Parliamentary Committee for Health for the 2024-2029 mandate. It would be the ideal setting for policymakers looking to ensure the PID community is empowered by proactive and future-proof legislation that tackles unmet needs in the areas of diagnosis, access to treatment options and care.

Together, we can build a future where the EU can make a tangible difference in the lives of every PID patient.

[7] IPOPI's Disability Statement, https://ipopi.org/disability-statement/
[8] Psychological distress of adult patients consulting a center for rare and undiagnosed diseases: a cross-sectional study, https://bit.ly/3xbco9F