REPORT

IPOPI 12th PID Forum

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

27 November 2018, **European Parliament, Brussels (Belgium)**

Hosted by MEP José Inácio Faria (EPP, Portugal)

Introduction

On Tuesday 27th November 2018, IPOPI organised its 12th PID Forum entitled “Rare Disease Priorities in the European Parliament 2019-2024: the voice of PID patients” at the European Parliament in Brussels. The event was kindly hosted by MEP José Inácio Faria, a long-standing and dedicated political supporter of the PID community for the past years.

The meeting represented an opportunity for Members of the European Parliament (MEPs), patient organisations, academics, healthcare professionals and other stakeholders to identify and discuss the challenges facing rare disease and PID patients and how these issues can be addressed in the next 5 years. At the heart of the discussions were crucial topics such as the need for newborn screening for severe forms of PIDs and other rare diseases, the need for improved diagnosis and availability of life-saving therapies for PID patients, and funding for rare diseases.

The Forum attracted a great attention from the European Parliament with six additional MEPs participating: Dr Anna Záborská (EPP, Slovakia), Sirpa Pietikäinen (EPP, Finland), Norica Nicolai (ALDE, Romania), Demetris Papadakis (ALDE, Cyprus), Takis Hadjigeorgiou (GUE/NGL, Cyprus) and Tilly Metz (Greens/EFA, Luxembourg). MEP José Inácio Faria called on fellow MEPs to keep rare diseases high on the political agenda and to endorse and act upon the actionable and concrete political recommendations which will be developed based on the Forum’s discussions.

Opening remarks

Mr José Inácio Faria MEP opened the 12th PID Forum. Due to an urgent mission in Georgia, he joined the Forum via a video intervention. MEP Faria pointed out the remarkable work achieved by IPOPI during their national campaigns for newborn screening programmes for Severe Combined Immunodeficiencies (SCID). Mr Faria called upon his colleagues to support him in a joint effort to help IPOPI achieve their goal regarding newborn screening which can help to secure a better life, or even a cure, for thousands of infants. He noted that IPOPI’s campaigns should serve as a reminder that it is vital to integrate newborn screening programmes nationally. Moreover, Mr. Faria emphasised that the fight against rare diseases should continue beyond the 8th term of the European Parliament and that the EU should place itself at the forefront of global research and innovation for the years to come.

Ms Sirpa Pietikäinen MEP highlighted the EU's added value in the field of rare diseases. She re-confirmed her political support towards rare disease patients and noted her commitment to improving the outcomes for patients. Ms. Pietikainen's intervention focused on three crucial areas for patients: (1) diagnosis, (2) treatment and (3) medicines' availability. She underlined the need for early and timely diagnosis, specifically in the field of rare diseases. She also called for the establishment of a European framework to fund and distribute orphan medicinal products. Finally, she touched upon the need for adequate prevention in the field of rare diseases and the need to protect patients' personal data.

Dr Anna Záborská MEP started her opening remarks by noting the essential need for rare disease patients and PID patients to have access to life-saving therapies. She noted that while some of the shortcomings in the field of rare diseases, such as coordination and exchange of best practices may be addressed at the EU level, Members States should not forget that they hold the primary responsibility for tackling these shortcomings. Dr Záborská underlined that very often rare diseases are left out of insurance reimbursement lists which is ultimately detrimental for patients. Moreover, the variety of healthcare system management throughout the EU leads to inequalities in care and called to tackle this challenge in a centralised and effective manner. Dr Záborská recalled the need for an adequate framework to support the development of therapies for rare disease patients. Finally, Dr Záborská pledged to support upcoming initiatives aimed at benefiting the lives of patients living with PIDs.

Rare diseases: what can be improved in the next 5 Years

Mr Johan Prévot, IPOPI Executive Director, set the scene of the 12th PID Forum by providing the audience with an overview of the milestones achieved at EU level in the field of rare diseases. These milestones included the establishment of the European Reference Networks and the Cross-Border Healthcare Directive. Mr Prévot pointed out that the implementation of newborn screening programmes for rare diseases still constitutes a major missing piece in the EU's efforts to improve patient outcomes. He recalled that the 2009 European Council recommendations on action

in the field of rare diseases emphasised this as well by noting that "*it is of utmost importance to ensure an active contribution of the Member States to the elaboration of some of the common instruments foreseen in the Commission communication on rare diseases …, especially on diagnostics and medical care and European guidelines on population screening*".[1] Mr Prévot went on to identify the changing landscape of rare diseases at European level as an opportunity towards a harmonised approach on newborn screening for rare diseases. He further noted the challenges of PID patients in terms of shortages of immunoglobulin. He recalled that the 2nd European Association of Hospital Pharmacists report has listed immunoglobulins as the 3rd most frequently referred medicines experiencing shortages. Mr. Prévot agreed with Dr Záborská on the need for the EU to keep supporting the promotion of scientific research initiatives within the framework of the Horizon Europe and with Ms. Pietikäinen on the protection of patient personal data. He stressed the need to not de-prioritise rare diseases on the EU agenda and to not look at what the EU has achieved on rare diseases in the last 15 years as an accomplished job well done but as a solid foundation for further much needed EU actions on this topic.

Newborn screening for severe forms of PIDs and other rare diseases

Gerard Loeber, from the International Society for Neonatal Screening (ISNS), started his intervention by giving an overarching presentation on the situation of newborn screening for rare diseases. He pointed out that newborn screening has been mainly considered as a national level programme. This has led to large differences among programmes and thus to a situation of inequality of acquiring good health across Europe. He urged European policy-makers to work together and promote a harmonised approach at EU level on newborn screening for rare diseases. Mr. Loeber praised the several EU actions that followed the 2008 Public Consultation on rare diseases and emphasised that initiatives for pooling and sharing data have prompted increased scientific research and further technological developments in the field of rare disease.

[1] https://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

Improving diagnosis for rare diseases

Dr Mirjam van der Burg, from Leiden University Medical Hospital (Netherlands), elaborated on the need for an improved framework for better diagnoses on rare diseases. Dr van der Burg emphasised the importance of the Human Phenotype Ontology (HPO) classification for each disease as well as the standardised laboratorial diagnosis and genetic testing for PIDs. She further noted that neonatal screening specifically for SCID can improve survival outcomes. While some countries in Europe have either implemented SCID screening (i.e. Norway) or are working on pilot projects (e.g. the Netherlands, UK, France, Spain, Italy, Germany, Denmark), there is still more work to be done and a harmonised approach would ultimately prove beneficial for patients.

Availability of life-saving therapies: patient's perspectives on areas of improvement

This panel was devoted to the availability of life-saving therapies. It brought together the testimonies of three patient representatives. Their presentations provided the audience with the reality PID patients are facing.

Ms Martine Pergent, IPOPI President and Board member of IRIS (French patient organisation for PIDs), provided an overview of the current availability of life-saving therapies in France. Ms Pergent reflected on the management and consumption of immunoglobulin (IG) by presenting the discrepancies in the supply and demand of plasma and blood products in the different French regions. She noted the recurrent tensions on Prescription Drug Monitoring Programs and the impact on the health and life of patients relying on the plasma supply chain. Moreover, Ms Pergent highlighted the preliminary conclusions of the pilot study jointly launched by IRIS-PERMEDES-CEREDIH, "PRIODIP", aimed at carrying out an exhaustive inventory of patients with PIDs and their needs of intravenous and subcutaneous immunoglobulins. As a result of the study, Ms Pergent noted the difficulties in describing exhaustively the patient-pathway and the challenges pharmacists confront in arbitrating between priority indications and emergency situations for patients. Furthermore, Ms Pergent spoke about the preparation of the law on bioethics. The French organisation representing patients with PIDs, IRIS, has already submitted their views on the necessity for sustainable access to a large range of safe IG, allowing a choice between biological medicines, tolerance and quality of life.

Finally, Ms Pergent highlighted some areas for improvement such as the consideration of PDMPs as global medicinal products rather than merely national ones; the support for vaccination programmes, and the consideration of ethical issues through the patient's perspective.

Dr Susan Walsh, Director of PID UK, conveyed the important testimonies of PID patients facing the challenges with immunoglobulin in the UK. The presentation started by noting that 2850 patients are receiving immunoglobulin out of the estimated 5,000 patients affected by PID in the UK. Dr Walsh emphasised the supply insufficiency and instability affecting the lives of patients in the UK. Dr Walsh also indicated that Brexit is likely to bring additional concerns and challenges to these patients not only in terms of possible shortages, but also regarding the expectation of companies moving away from the UK market. Lastly, she noted that PID UK are constantly keeping the community informed; calling for increase in number of commissioned products; opposing the government's planned surcharge on plasma products; and working with the plasma protein therapeutics association's taskforce to improve government's awareness of the plasma market and asking them to review the collection of UK plasma.

Mr Savvas Savva, Secretary of Cyprus Primary Immunodeficiencies and Friends, provided the participants with an overview of the challenges faced by PID patients in Cyprus. One of the main challenges for these patients is the lack of national registry for PIDs and the scarcity of educated healthcare professionals in the field. Mr Savva emphasised the overall unawareness of the disease among the local medical community and the inexistence of monitoring systems on the availability of medicinal products for patients in need of immunoglobulin. Mr Savva envisaged that a bigger place for rare diseases in the European budget as well as a bigger allocation of the government budget to health will certainly make a difference to the lives of PID patients.

Discussion

Ms Norica Nicolai MEP noted the need for real and political support for rare disease patients. She highlighted the challenges of diagnosis and treatment for PID patients and affirmed her support to IPOPI and the rare disease community.

During the discussion section, it emerged that in Germany, the treatment of adult PID patients is a major concern since the waiting list to see a specialist can sometimes reach several months. The scarcity of plasma supply also impacts negatively patient outcomes. In this sense, **Ms Claudine Deckers,** from the Belgian PID patient organisations, expressed hopes that the current on-going campaign in Belgium on plasma donation may attract new donors and incentivise existing ones to donate on a more regular basis.

Silke Mader, from the European Foundation for the Care of Newborn Infants, called on policy makers to urgently consider the needs of newborns suffering from rare diseases and to politically endorse newborn health programmes. She further recalled the existing inequalities of diagnosis and treatment around the EU and called for the address of this challenge.

Mr Takis Hadjigeorgiou MEP praised the work developed by the Cyprus Primary Immunodeficiency and Friends. Mr Hadjigeorgiou communicated his wishes for seeing more comprehensive actions in the future where patient organisations for rare diseases should combine forces with other interest groups in the European Parliament such as that on Rheumatic and Musculoskeletal Diseases (RMDs) with the aim of increasing awareness on these conditions.

Ms Tilly Metz MEP proposed a global and centralised approach to patients, with standardisation of diagnosis. She further supported the idea of data sharing across Europe on rare diseases. Ms Metz expressed great concern with regard to shortages of immunoglobulin for PID patients. She gave her support to the PID community and emphasised the need for newborn screening for rare diseases.

Concluding remarks

Ms Martine Pergent concluded the meeting by noting that the discussions should echo within the European institutions. She thanked all participants and noted that tangible recommendations have emerged from the Forum. As per the call from MEP Faria, Ms Pergent announced that these recommendations will be developed and shared with policy-makers for their political endorsement.

Overarching policy recommendations stemming from the discussion

- Rare diseases should be kept high on the political agenda through continued political support for rare disease initiatives both at European and regional levels.

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

- Policy-makers should advocate for better access to treatment for PIDs and rare disease patients by ensuring a favourable political framework for the availability of a wide range of products, allowing the most suitable treatment for these patients.

- 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.

- Based on the ERNs' performance and identified need, the EU should continue funding ERNs to improve diagnosis and outcomes of rare disease patients', including those of PID patients.

- The EU should build on the achievements and success of Horizon 2020 and keep itself at the forefront of global research and innovation.

- Policy-makers should further endorse policies raising awareness on primary immunodeficiencies.

List of participants

European Parliament

- Anna Záborská MEP (EPP, Slovakia)
- Demetris Papadakis MEP (ALDE, Cyprus)
- José Inácio Faria MEP (EPP, Portugal) (video intervention)
- Norica Nicolai MEP (ALDE, Romania)
- Sirpa Pietikäinen MEP (EPP, Finland)
- Takis Hadjigeorgiou MEP (GUE/NGL, Cyprus)
- Tilly Metz MEP (Greens/EFA, Luxembourg)
- Alexandru Serpe, office of MEP Norica Nicolai
- Andreia da Silva, office of MEP José Inácio Faria
- Luis Baiao, office of MEP José Inácio Faria

External Participants

- Cesar Rubio, Grifols
- Claudine Deckers, BOPPI (IPOPI's Belgian national member organisation)
- Esperanza Guisado, Grifols
- Frank Willersinn, Alpha-1 Plus
- Gerard Loeber, International Society for Neonatal Screening (ISNS)
- Joelle Khraiche, CSL Behring
- Johan Prévot, IPOPI
- José Verstegen, Stichting voor Afweerstoornissen (IPOPI's Dutch national member organisation)
- Karl Petrovsky, Plasma Protein Therapeutics Association (PPTA)
- Kit Greenop, RPP Group
- Leire Solis, IPOPI
- Martine Pergent, IPOPI
- Mirjam van der Burg, Leiden Hospital University
- Neele Jongen, RPP Group
- Paul Strengers, International Plasma Fractionation Association (IPFA)
- Ricardo Arieira, RPP Group
- Savvas Savva, Cyprus Primary Immunodeficiencies and Friends (IPOPI's Cypriot national member organisation)
- Silke Mader, European Foundation for the Care of Newborn Infants
- Steffen Ball, DSAI (IPOPI's German national member organisation)
- Susan Walsh, PID UK (IPOPI's UK national member organisation)
- Yordan Aleksandrov, RPP Group