

## REPORT

### IPOPI 13th PID Forum

#### Newborn screening for rare diseases.

#### A PID perspective.



**Wednesday 4th December 2019 – 15:00-17:00**

**Room A5G305, European Parliament, Brussels (Belgium)**

## Introduction

On Wednesday, 4th December 2019, IPOPI organised its 13th PID Forum entitled “Newborn Screening for Rare Diseases. A PID Perspective” at the European Parliament in Brussels (Belgium). The event was co-hosted by the following Members of the European Parliament (MEP): Manuel Pizarro (Social and Democrats, Portugal), Tilly Metz (Greens/EFA, Luxembourg), Sirpa Pietikäinen (European People’s Party, Finland) and Irena Joveva (Renew Europe, Slovenia). Our co-hosts represented a mixture of long-standing and dedicated political supporters of the primary immunodeficiencies (PID) community and MEPs who have taken office in May 2019 and are dedicated to improving the lives of rare disease and PID patients.

Also welcomed at the Forum was MEP Tudor Ciuhodaru (S&D, Romania) in addition to a wide range of stakeholders, from patient group representatives, healthcare professionals and representatives of the European Parliament who used this time to elaborate on the advancement of newborn screening for rare disease in their countries and support the Call to Action on newborn screening for rare diseases jointly supported by IPOPI, the International Society for Neonatal Screening (ISNS) and the European Society for Immunodeficiencies (ESID).

Throughout the event, our co-hosts called on fellow MEPs to work together to:

- Develop and implement overarching guidelines in the field of newborn screening for rare diseases;
- Develop an EU-wide platform on newborn screening for rare diseases and pool from the expertise of recognised societies working in the field; and
- Cooperate to position the EU as the central point for data collection.

## Opening Remarks



**Dr Manuel Pizarro** MEP opened the 13<sup>th</sup> PID Forum. MEP Pizarro welcomed attendees and stated that he was delighted to co-host the event with so many of his colleagues from various political parties.

MEP Pizarro began his address by stating that he treated primary immunodeficiency patients in the past and was only too aware of the challenges posed by not diagnosing conditions such as severe combined immunodeficiencies (SCID) early enough. He deplored that in his home country of Portugal, the average delay for patients getting a PID diagnosis is seven years. Despite being a frontrunner in the number of rare conditions screened at birth – with more than 20 conditions being screened – Portugal does not screen for SCID and is still far away from the 80 screenable conditions. MEP Pizarro offered his heartfelt support for the IPOPI-ISNS-ESID Call to Action and outlined his belief that the European Parliament offers one of the most effective political venues to discuss these issues. He called on the Council of the European Union and its upcoming Presidencies to consider concrete activities on newborn screening in their political programmes.

**Ms Tilly Metz** MEP is a supporter of IPOPI and praised their continued efforts towards implementing the considerations contained within the [Political Manifesto on the EU’s Political Priorities for 2019-2024](#) which she endorsed following the 12<sup>th</sup> PID Forum.

Ms Metz’s intervention focused on the complexity of gathering data at national level for Member States like Luxembourg. Although the newborn screening programme in her country had recently expanded to include cystic fibrosis, she noted that with no centrally located point to share existing screening practices, uptake is not as fast as it could. As such, European children continue not to be screened for diseases which could be cured with a prompt diagnosis at birth. She concluded by welcoming the call to action which was being launched and that she looked forward to implementing initiatives at the European level which can overcome the challenges rare disease patient face every day.





**Ms Irena Joveva** MEP highlighted that despite the national newborn screening programme beginning in 1979, Slovenia is currently only screening for two diseases, one of the lowest figures in the EU.

In her mind, Slovenia's situation and the fact that other Member States screen for nearly 30 conditions perfectly illustrates how countries lagging behind could benefit from the sharing of data, best practices and guidelines on this topic.

She expressed her formal support of the Call to Action and called on the approaching Slovenian Presidency of the Council of the EU from July 2021 to December 2021 to place newborn screening for rare diseases within their priorities in order to ensure that rare diseases patients and especially the lives of newborn babies can be improved and saved.

**Ms Sirpa Pietikäinen** MEP re-confirmed her political support towards rare disease patients and IPOPI's activities. She said newborn screening for rare diseases has a key role to play in enabling prompt diagnosis and timely access to life saving specialist treatments.

She highlighted that in Finland, several hospitals are running pilot programmes for SCID and pointed out that other Member States, if they desire to do so, should be able to consult and be aware of the results and outcomes from these pilots. Consequently, she stated the EU should actively support a better coordination and exchange of best practices in the field. She concluded by saying that newborn screening for rare diseases is no longer a topic which can be neglected and called on the European Parliament to support and engage in a concrete dialogue with the European Commission and Member States to progress towards EU action on this topic.



**Dr Tudor Ciuhodaru** MEP expressed his delight at joining IPOPI for the 13<sup>th</sup> PID Forum on newborn screening for rare diseases. He stated that, as a healthcare professional and as a politician, he has long stood by the belief that health should be considered a human right for everyone.

He spoke of his ambitions within the European Parliament and his hope to work on topics which would positively impact the lives of European patients for the duration of his mandate. He therefore strongly supported the Call to Action and pledged to support upcoming initiatives aimed at benefiting the lives of patients living with PIDs.

## Setting the Scene

**Mrs Martine Pergent**, IPOPI President, set the scene for the 13<sup>th</sup> PID Forum by providing the audience with a presentation of IPOPI and its activities in the field of newborn screening for rare diseases. She then followed by saying that cooperation on newborn screening for rare diseases is the missing piece in EU policy on rare diseases. Not only is it cost-effective, most notably through the reduction in ongoing care, but it also plays a critical role improving healthcare outcomes for rare diseases and mortality for rare diseases.

She highlighted SCID as a prime illustration of the importance of newborn screening. SCID is a rare, in most cases fatal syndrome of diverse genetic cause in which extreme susceptibility to serious infections that can result in permanent organ damage or failure or even death unless definitive treatment can be used in time to correct the underlying immune defect. Early diagnosis, before the infant has had a chance to develop any infections, is extremely valuable since a transplant of bone marrow or blood or gene therapy given in the first 3 months of life have a 96% success rate.

IPOPI has been active at the European level for a number of years and Mrs Pergent noted that in 2013, some Member States showed a distinct lack of interest in a coordinated approach to newborn screening for rare diseases.

However, by supporting a number of national initiatives, IPOPI helped place newborn screening as a public health priority in a number of EU Member States. This led to many Member States becoming more inclined towards promoting an EU approach to newborn screening. IPOPI thus renewed its efforts at the European level – in collaboration with the International Society for Neonatal Screening and the European Society for Immunodeficiency – to engage with the European Parliament, the European Commission and Member States.

Before giving the floor to the first panel she called on the EU to tackle the major discrepancies between Member States on newborn screening in order to ensure that babies who are born in the EU have the same opportunities to be diagnosed early and access the lifesaving care they need. She concluded that this could be achieved through (1) the development and implementation of overarching guidelines in the field of newborn screening for rare diseases, (2) launching an EU-wide expert committee on newborn screening for rare diseases to pool from the expertise of recognised societies working in the field and (3) Encouraging the exchange of best practices that would support Member States' efforts in the development of newborn screening practices.

### The Aspects of Newborn Screening



**Dr Peter Schielen**, International Society for Neonatal Screening (ISNS), provided a historical overview of neonatal screening in Europe and an indication of the differences in neonatal screening between European countries which he used to underline the vision of his organisation and how it has come to align with the goals of IPOPI and ESID.

He pointed out that the list of diseases which could be screened for at birth is growing incessantly. Indeed, only three diseases were being screened in the early 70s and 80s, but innovative laboratory techniques have facilitated the expansion of potential conditions to screen for as well as the establishment of screening principles by Maxwell Wilson and Gunnar Jungner.

Despite positive advancements in medical terms, discrepancies between EU Member States regarding the number of diseases screened still exist and we should eradicate these differences. Concluding his presentation, Dr Schielen called for added collaboration at the EU level and awareness raising campaigns in addition to asking for MEPs to support the IPOPI-ISNS-ESID Call to Action.



**Dr Nizar Mahlaoui**, Necker-Enfants Malades Hospital, and Chair of IPOPI's Medical Advisory Panel, said that an estimated 6 to 8% of the total European population suffer from one of the currently identified eight-nine thousand rare diseases which amounts to more than 30 million people. Patients with rare diseases, he said, often harbour multiple disorders simultaneously which have a high impact on their daily lives. Subsequently, they need different kinds of care. However, Dr Mahlaoui said that there was a critical lack of essential knowledge on rare diseases across the board from healthcare providers to governments and local authorities.

Dr Mahlaoui specifically mentioned the case of SCID as a disease which would benefit from newborn screening as the sooner diagnosis came, the better the outcome for the patient, including in the long term as they could be cured from their condition and no longer be patients. He stated there is strong evidence that SCID fulfils internationally established criteria for newborn screening and that it meets the World Health Organization's (WHO) criteria.



### The Impacts of Newborn Screening

**Ms Stela Andreea Pirvu**, Romanian Association for Patients with Primary Immunodeficiencies (ARPID), shared her and her son's story preceding and following his diagnosis with Complete DiGeorge Syndrome, a rare and severe type of primary immunodeficiency.

She spoke of the challenges her family faced as her son, Daniel, spent many months in the hospital with a number of symptoms which doctors did not immediately link to a PID condition. Daniel is the only Complete DiGeorge patient from Romania who survived long enough to receive diagnosis and treatment, but some families were not so lucky.

Early identification can provide an immediate lifesaving treatment and consequently lead to avoiding an inefficient, costly and dangerous diagnostic odyssey.

Because of late diagnosis, PID patients are vulnerable to infections, spend more time in the hospital and are more likely to succumb to their conditions. This is why, she concluded, newborn screening and early identification is crucial. The EU should tackle newborn screening for rare diseases in order to ensure Daniel's story is not repeated over and over.

**Mr Alberto Casaca**, Associação Portuguesa de Doentes com Imunodeficiências Primárias (APDIP), conveyed the challenges PID patients face in Portugal, but also how lucky they are to have dedicated centres in Lisbon, Porto, Coimbra and Funchal and a large number of health professionals experienced in primary immunodeficiencies.



He stated that his diagnosis as a PID patient saved his life but, like many patients in Portugal, it came late. The average delay for diagnosis is seven years in Portugal, he was diagnosed when he was seventeen. Delays in diagnoses is one of the main challenges facing PID patients in Portugal and he stated that an EU approach to newborn screening could serve to drastically reduce the delays for conditions such as SCID.

Although newborn screening for SCID has not been adopted in Portugal yet, he was delighted to relay the information that a hospital in Porto will start a pilot project on newborn screening for SCID in 2020. Obtaining a diagnosis, he said, is critical for PID patient as they can finally start being treated for their disease instead of their symptoms.

### Open Floor for Discussion

This session was devoted to giving the floor to national member organisations of IPOPI and other stakeholders who together lent to their voice to the actions of IPOPI, ESID and the ISNS to advocate for the Call to Action which was launched during the Forum.

**Prof Isabelle Meyts**, President of ESID, kickstarted this discussion by emphasising she was delighted to be partnering with likeminded organisations such as IPOPI and ISNS in promoting an EU approach to implementing newborn screening for rare diseases. By outlining the importance of sharing best practices and data, she expressed her hope that the 13<sup>th</sup> PID Forum would help launch a political discussion at the European level on the issue.



**Prof. Filomeen Haerynck**, President of Symposium of the Belgian Primary Immune Deficiency Group (BPIDG), provided a brief overview of the countries in which pilot projects on SCID neonatal screening have been implemented or are planned and provided a glimpse into the TREC pilot study taking place at Ghent University Hospital, the hospital in which she works. In anticipation of the implementation of SCID newborn screening programme in Flanders, this pilot study aims to compare both screening methods in order to select the most suitable one for patients.

**Mr Alain Delfante**, Belgian Organisation for Patients with Primary Immunodeficiencies (BOPPI), called on policy makers to urgently consider the need to reduce the overall cost of newborn screening and eagerly supported the call to action.

**Mr Sergio Zeno Vincentini**, Associazione immunodeficienze Primitive (AIP), highlighted the need to eradicate the differences inside and outside Member States. He said EU guidelines for newborn screening were a critical necessity for the EU and endorsed the call to action.

**Mr Gabriel Bodor**, Slovakian Association of Patients with Primary Immunodeficiency, conveyed that their organisation is delighted to support the call to action on newborn screening for rare diseases as PID patients would



strongly benefit from the EU tackling this issue, he volunteered to raise awareness of the call to action at national level.

Just like Mr Casaca, **Ms José Vertegen-Ruijs**, Stichting voor AfweerStoornissen (SAS), also took the opportunity of the event to reveal the development of a pilot project for SCID in the Netherlands which should begin in 2020. Likewise, she strongly supported the call to action and hope to see many policymakers support it.

Additionally, a number of stakeholders noted the need for political support for newborn screening for rare diseases. Dr Mahlaoui made it known that in France, the DEPISTREC study which ran a pilot project on SCID since 2012 now stands at the health technology assessment phase and that a positive decision on the expansion of the newborn screening programme could be hoped for in 2020.

### Wrap-up from IPOPI & ISNS & ESID

Following the interventions of the various stakeholders within the room, **Mr Johan Prevot**, IPOPI Executive Director summarised the day's discussions and was joined by supporting remarks from Dr Peter Schielen, ISNS, and Prof Isabelle Meyts, ESID. The policymakers in attendance were thanked for their invaluable support to this joint campaign and positive stance on health-related issues.

The sheer number of people represented by Members of the European Parliament is a clear demonstration of the importance of the European Union and the added value it can bring in the field of rare diseases. Extrapolating best practices from Member States has also played a crucial role and it is undoubtedly clear that this extrapolation has improved the lives of rare disease patients. It is evident, however, that the EU cooperation on Newborn Screening for Rare Diseases is still an underdeveloped area and should seek to follow the lead of the United States.



With some Member States screening for many diseases and others lagging behind, it was pointed out by Mr Prevot that it is important to complement what has already been achieved in the field of rare diseases. This is why cooperating to position the EU as the central point for data collection and information on rare diseases newborn screening practices and encouraging the exchange of best practices that would support Member States' efforts in the development of newborn screening practices is critical.

It was highlighted that, ultimately, the development and implementation of overarching guidelines in the field of newborn screening for rare diseases will assist Member States make better-informed assessments help them prepare their healthcare systems for the implementation of a newborn screening for a specific rare condition.

The European Commission Steering Group on Health Promotion and Prevention (SGPP) is taking a new role in prevention affairs and it was underlined that the creation of a European newborn screening standing committee could add significant value in encouraging uptake of the newest scientific evidence.

### Concluding remarks

Mrs Martine Pergent concluded the meeting by thanking every participant and the MEPs in attendance for endorsing the call to action as we look to improve conditions for newborn screening for rare diseases. Before giving the floor to MEP Pizarro for the final remarks, Mrs Pergent proposed to have him sign the Call to Action which he did. He was joined by MEP Ciuhodaru who likewise signed the call to action.

MEP Pizarro likewise thanked everyone for the productive discussions and for the insights which were provided from a number of Member States. He concluded that Europe has the right tools at its disposal to encourage further cooperation in the field of newborn screening for rare diseases. He expressed his personal willingness to support EU action on newborn screening and assured he would cooperate with IPOPI, the ISNS and ESID again in the future.



## List of participants

### European Parliament

- Manuel Pizarro, MEP (S&D, Portugal)
- Tilly Metz, MEP (Greens/EFA, Luxembourg)
- Irena Joveva, MEP (RE, Slovenia)
- Sirpa Pietikäinen, MEP (EPP, Finland)
- Tudor Ciuhodaru (S&D, Romania)
- Eduardo José Valentim Dos Santos Leal, Office of MEP Manuel Pizarro

### External Participants

- Margarida Albuquerque Arenga, Portuguese Permanent Representation to the European Union
- Yordan Aleksandrov, Rhode Public Policy (RPP) Group
- Gabriel Bodor, Zdruzenie pacientov s primarnou imunodeficiencious, o.z.
- Lucia Bodor, Zdruzenie pacientov s primarnou imunodeficiencious, o.z.
- Malicorne Buysse, UZ Gent
- Alberto Casaca, Associação Portuguesa de Doentes com Imunodeficiências Primárias (APDIP)
- Alain Delfante, Belgian Organisation for Patients with Primary Immunodeficiencies (BOPPI)
- Nicole Grillet, Belgian Organisation for Patients with Primary Immunodeficiencies (BOPPI)
- Prof. Dr Filomeen Haerynck, Belgian PID Group / Centre for Primary Immunodeficiency Ghent
- Joelle Khraiche, CSL Behring
- Ava Lloyd, FTI Consulting
- Nizar Mahlaoui, Necker-Enfants Malades Hospital, and Chair of IPOPI's Medical Advisory Panel
- Isabelle Meyts, UZ Leuven – European Society for Immunodeficiencies (ESID)
- Aline Mola, Rhode Public Policy (RPP) Group
- Julia Nordin, International Patient Organisation for Primary Immunodeficiencies (IPOPI)
- Elliot O'Farrell, Rhode Public Policy (RPP) Group
- Yvonne Okx-van der Linde, Stichting voor AfweerStoornissen (SAS)
- Martine Pergent, International Patient Organisation for Primary Immunodeficiencies (IPOPI)
- Karl Petrovsky, Plasma Protein Therapeutics Association (PPTA) Global
- Jan Philippé, UZ Gent
- Stela Andreea Pirvu, Romanian Association for Patients with Primary Immunodeficiencies (ARPID)
- Johan Prevot, International Patient Organisation for Primary Immunodeficiencies (IPOPI)
- Françoise Rossi, International Plasma and Fractionation Association (IPFA)
- César Rubio, Grifols
- Peter Schielen, National Institute of Public Health and the Environment
- Leire Solis, International Patient Organisation for Primary Immunodeficiencies (IPOPI)
- Maartin Van Baelen, Plasma Protein Therapeutics Association (PPTA) Global
- José Vertegen-Ruijs, Stichting voor AfweerStoornissen (SAS)
- Sergio Zeno Vincentini, Associazione immunodeficienze primitive onlus
- Charles Waller, Rhode Public Policy (RPP) Group
- Dr Frank Willersinn, Alpha-1 Plus Belgium