3rd PID FORUM on National Plans for Rare Diseases: Centres of reference and patients' registries
Hosted by Mrs. Nessa Childers MEP, 26 September 2012
Report

INTERNATIONAL PATIENT ORGANISATION FOR PRIMARY IMMUNODEFICIENCIES
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Recommendations

1. Member States should fully develop their national plans/strategies by the end of 2013, and ensure that patients with rare diseases have access to high-quality care.
2. It is not sufficient to adopt a national plan: the adoption must be fully implemented and the plan should have access to sufficient funding to develop the envisioned activities.
3. Patients should be consulted and taken into consideration whenever Member States or the European Union develop policies that may affect their lives.
4. The heterogeneity between national plans demonstrates the need for the EU to play a coordinating role and tackle inequalities in the access to treatment across the Union.
5. Policymakers should ensure that national plans/strategies fully comply with the Council Recommendations, especially when supporting the creation or development of centres of reference and patient registries.
6. Patient registries and national centres of reference should be included when Member States are developing their National Plans for Rare Diseases to encourage research and improve diagnosis and information to patients.
7. PIDs are an important group of rare diseases and as such should be taken into consideration by national policy makers when developing the national plans for rare diseases.
8. Patient registries and centres of reference should be recognised and financially supported both nationally and at EU level, for the value they can add to patients' lives.
9. National and European policymakers should support the development of PID centres of reference so as to ensure high quality information to patients and the care they need to live normal lives.
10. Reference centres for PID should be encouraged as a means of tackling under diagnosis, and improving knowledge among the medical community about the condition.
11. Member States should set up national centres of reference for PID to improve the information to patients and training of medical professionals and to support the enrolment of patients into PID registries.
12. Member States should set up PID patient registries where there are none, and support the development of the ones already established.
13. The EU should help Member States with less experience in managing PID to invest in appropriate frameworks for care for patients leaving with a PID.

Introduction

On Wednesday 26th September 2012, Mrs. Nessa Childers MEP (S&D, Ireland) hosted IPOPI's third Primary Immunodeficiency Forum at the European Parliament in Brussels, Belgium. The meeting which was organised in collaboration with the International Patient Organisation for Primary Immunodeficiencies (IPOPI) focussed on national plans for rare diseases, centres of reference and patient registries.

Experts representing patients, academics, physicians and industry discussed with Members of the European Parliament the benefits afforded to patients by national plans for rare diseases and the need for Member States to fully implement and finance their national plans including the setting up of patient registries and designate centres of reference. All participants in the meeting agreed that Member States should involve patients and their representatives when developing and implementing national plans.

Summary of discussions

Mrs. Childers initiated the meeting by highlighting how patients with PIDs, as for the rest of rare diseases, would benefit from actions taken both at Member States and European level to ensure that they are provided with adequate information, treatment and care. Amongst the European initiatives, the Council of Ministers adopted in June 2009 a Recommendation on an action in the field of rare diseases. This Council Recommendation called for Member States to develop and implement their national plans or strategies by the end of 2013. Centres of reference on rare diseases were described as key elements to increasing the knowledge of rare diseases, to enable faster diagnosis and to ensure that patients receive the best treatment available. Mrs. Childers furthermore highlighted the importance of creating and coordinating European registries, as these are a prerequisite for research into treatment and diagnostics and the development of knowledge about rare diseases such as PID, disorders that today remain under diagnosed.

Mr. Johan Prevot, Executive Director of IPOPI, briefly described how patients affected with PIDs are often treated for the symptoms of their condition, rather than for the disease itself. As it was explained, this is due to the lack of awareness and knowledge about this group of around 250 different disorders. Mr. Prevot highlighted that only 8 Member States out of 27 have adopted their national plan for rare diseases and out of 8, only 3 had fully or partially implemented them. Member States should therefore ensure that they meet the Council deadline of end of 2013 to develop their plans. It was also argued that the adoption of such strategies was not sufficient and that the plans

should be fully implemented and provided with sufficient funding. In that sense, an essential element in the plans would be the inclusion of screening programmes for curable and treatable rare diseases, such as Severe Combined Immunodeficiency. All participants agreed that during the development of the national plans on rare diseases, PID patients should be consulted in the development of the national plan.

Dr. Nizar Mahlaoui, Manager of the French Centre de Référence Déficits Immunitaires Héréditaires (CEREDIH), provided an overview of the work of the CEREDIH in the management and knowledge development about PIDs. Dr. Mahlaoui explained that the network of centres of reference in France has been key to registering patients to determine the prevalence and ensure that patients have access to the best available treatment. Throughout his presentation, Dr. Mahlaoui showed how patients with missed or a delayed diagnosis of PIDs would go under unnecessary suffering and depend heavily on healthcare resources. Centres of reference, such as the CEREDIH, benefit patients by collecting a full range of expertise and deliver the best and most appropriate care for a specific disease or group of disorders. In this sense, centres of reference for PIDs provide patients with access to treatment, training for physicians to identify and diagnose PIDs and support spreading best practices regarding the treatment of the conditions.

Dr. Gerhard Kindle, from the Centre of Chronic Immunodeficiency of the University Hospital of Freiburg and Head of the ESID Online Database, presented the European Society for Immunodeficiencies (ESID) registry: its potentialities and future perspectives. The ESID registry is a database combining information on PIDs from different European registries covering more than 16,100 patients. The aim of such registry is to improve knowledge about the conditions, facilitate research and help doctors answer the questions they and their patients might have when faced with a PID diagnosis. Given the great utility of patient registries for PIDs for better managing the diseases, it is required that Member States keep on developing national PID registries.

Representing the European Commission, Dr. Karl Freese, principal administrator of the Health Information Unit, presented an overview of the European legislative and non-legislative initiatives in the field of rare diseases. Dr. Freese explained that currently it is a Member State competency to develop the national plans for rare diseases. In this respect, patients and patient representatives are the best placed to call for adequate development and implementation of the plans. MEPs were highlighted as key stakeholders to help convey patients' messages. Newborn

screening is also being supported at European level, with the objective of evaluating neonatal screening strategies for rare diseases in Member States. It was also mentioned that the European Union Committee of Experts on Rare Diseases is currently analysing a report on the status of screening practices in the EU, which will result in recommendations being publish in 2013.

After the presentations there was a lively discussion on several very important topics. On the subject of screening and early diagnosis, Dr. Gaspar, from the Institute of Child's Health (UCL) in London, explained the importance for newborn screening for the most severe forms of PIDs, such as Severe Combined Immunodeficiencies (SCID). In the case of SCID, a timely diagnosis would allow the patient to access a treatment that would cure him, while late detection greatly reduces the chances of survival. Dr. Keogan, from Beaumont Hospital in Dublin, highlighted that for PID the right treatment means improved functionality when applied early enough, while late diagnosis means frequent hospitalisation and disruption of work and education as a consequence for both patient and parents. Mrs. Childers agreed and reminded the participants that if the patients were left untreated, they could suffer chronic damages that would not allow them to fully contribute to society.

On the topic of the implementation of national plans for rare diseases, Johan Prevot asked for mechanisms to remind Member States about their obligation. Mr. Freese responded that it was a national competence but that MEPs and national members of parliament could be very good allies to help them raising pressure for implementation. Mrs. Rooney, P.R.O. of the Irish Primary Immune Deficiency Organisation (IPIA), explained that indeed any support patients could receive from policy makers in ensuring the development of better diagnosis and treatment for rare diseases in their countries would be welcomed. Mrs. Rooney explained the difficulties that patients go through for getting an adequate diagnosis and that sometimes, treatments were not accessible enough for patients. To this Mrs. Childers responded by volunteering her support to raise awareness and push for the inclusion of best practices and timely implementation of the Irish National Plan for Rare Diseases.

A set of key recommendations identified during the meeting was produced and endorsed by Mrs Childers. These recommendations will be particularly helpful to patient organisations and other key PID stakeholders in their discussion around the implementation of rare diseases national plans and the importance of carving out Primary Immunodeficiencies as an important group of rare diseases deserving specific attention and action.

List of participants

Ms. Nessa Childers, Member of the European Parliament, host of the PID Forum

Ms. Phil Prendergast, Member of the European Parliament

Ms. Marian Harkin, Member of the European Parliament

Ms. Emer Costello, Member of the European Parliament

Ms. Emily Hunter, office of Mrs. Glenis Willmott, Member of the European Parliament

Mr. Mark Taylor, office of Mrs. Mairead McGuinness, Member of the European Parliament

Ms. Stanislava Benova, office of Dr. Miroslav Mikolášik Member of the European Parliament

Dr. Karl Freese, European Commission

Dr. Gerhard Kindle, ESID/University of Freiburg

Dr. Nizar Mahlaoui, CEREDIH (Le Centre de Référence Déficits Immunitaires Héréditaires)

Dr. Hubert Baburaj Gaspar, UCL Institute of Child Health

Dr. Mary Keogan, Beaumont Hospital

Ms. Yvonne Rooney, IPIA

Mr. Johan Prevot, IPOPI

Ms. Martine Pergent, IPOPI

Ms. Sónia Pereira de Figueiredo, IPOPI

Mr. Johan Eerens, European Haemophilia Consortium

Ms. Isabel Henkel, Grifols

Mr. Rüdiger Gatermann, CSL Behring

Mr. Charles Waller, PPTA

Ms. Laura Savini, PPTA

Mr. Sebastian Rohde, Rohde Public Policy

Ms. Leire Solis Garate, Rohde Public Policy

Mr. Rune Orloff Pedersen, Rohde Public Policy

Mr. James Kennedy, Rohde Public Policy