1st IPOPI PID FORUM
SCID Newborn Screening
15 June 2011
Hosted by Mrs. Glenis Willmott, MEP

Report

Introduction

On Wednesday 15 June 2011, Mrs. Willmott MEP (S&D, United Kingdom) hosted the first Primary Immunodeficiency Forum organised by the International Patient Organisation for Primary Immunodefiencies (IPOPI) at the European Parliament in Brussels, Belgium. The meeting focused on newborn screening for Severe Combined Immunodeficiency (SCID), a life-threatening rare disorder causing the body's immune system not to function properly. A group of experts representing patients, academics, physicians, industry and policy makers discussed the issues facing SCID patients and their families and agreed upon a set of key-recommendations to help the European Institutions and Member States in understanding the best way forward.

This report presents the concrete recommendations for policy action and summarises the key points that were addressed by the Forum.

Summary of discussions

Mrs. Glenis Willmott MEP opened the PID Forum by noting that the European Union has successfully developed policies and legislation on rare diseases since the regulation 141/2000 on Orphan Medicinal Products[1], addressing the needs of patients who were too often neglected. The European Parliament is committed to patients suffering from

[1] OJ L 18, 22.1.2000 p.1-5

rare conditions, as recent legislation such as Directive 2011/24/EU on Patients' rights in cross-border healthcare[2], illustrates.

Mrs. Willmott reminded that the Commission and the Council of the European Union confirmed the importance of supporting the evaluation of neonatal screening strategies in Europe in the Council recommendation on a European Action in the field of Rare Diseases[3]. In this sense, Mrs. Willmott called for the EU to continue supporting citizens suffering from rare diseases.

Participants noted that SCID is a treatable and curable disease, only when the treatment is given in the first few months, but that the lack of systematic SCID screening in Member States impedes newborns to be diagnosed and treated in a timely fashion. As a result, many of them do not reach their first birthday. The fact that patients suffering from a rare condition cannot access the existing treatments and care they need undermines EU and national efforts to tackle rare diseases.

Participants considered that the European Commission should address the issue of SCID Newborn screening in its future policy initiatives, including the possible proposal for a Council Recommendation on newborn screening: the Second Public health Programme actually supported the creation of an EU Network of Experts on Newborn screening and an evaluation of current neonatal screening strategies for rare diseases – this opportunity should consider SCID, a curable rare disease, and provide Member States with the evidence on which to base their political decision.

The majority of participants stressed that timely SCID newborn screening (within the first 3.5 months of life) would increase the chance to perform a successful medical intervention - while reducing significantly the costs and the need of subsequent treatments.

Mr. Johan Prévot, representing IPOPI, presented some of the reasons why SCID newborn screening was necessary: SCID is a life threatening condition that affects 1 out of 50,000-100,000 newborn babies, but it can be cured and lives can be saved if patients are diagnosed and treated early enough. If they are left untreated, on the other hand, babies die before their first birthday. The group agreed that the EU could contribute to saving lives by calling for the inclusion of SCID in the list of diseases newborns are screened for on a routine basis, and that Member States should consider SCID as a paediatric emergency. All participants agreed that the EU must continue to encourage Member States in better diagnosing and treating patients by inter allia encouraging the

[2] OJ L 88, 4.4.2011, 45-65
[3] OJ C 151, 3.7.2009, 7-10

exchange of data and best practices in regard.

The second speaker, Professor Gaspar, Professor of Paediatrics and Immunology at the University College of London, focussed on the scientific aspect of SCID newborn screening and argued that the earlier a newborn suffering from SCID gets treatment, the higher are the chances of a positive health outcome – 90% of patients receiving timely diagnosis and treatment will survive – but if patients fail to be diagnosed on time, the urgent transplant procedure usually arrives too late and may be no longer effective.

Mrs. Jose Drabwell, representing IPOPI, provided the perspective of patients and families affected by SCID. A PID patient herself, Mrs. Drabwell had to struggle more than 20 years before receiving an accurate diagnosis. Her condition was not as fast progressing as SCID, but for several decades she had to struggle with health issues including severe infections necessitating prolonged hospital stays before she received a correct diagnosis. For years, she could not live a normal life as she was sick most of the time. Repeatedly, Mrs. Drabwell pointed at the absurdity of patients who are not given the opportunity to live normal lives and contribute to society: “save our lives - we want to be nett contributors, not nett recipients!”

Mr. Lennart Hammarström, Professor of Clinical Immunology at the Karolinska Institutet, announced that in the USA, 2.5 million children will be screened for SCID as SCID newborn screening will be generalised in that country. In the EU – such measure could be quickly implemented as all the tools already exist and the procedures are well known. Pilot projects implementing systematic SCID newborn screening have already started in Germany and Sweden, as Dr. Borte noted when presenting a pilot project in Lower Saxony. Dr. Audrain completed the picture by indicating that a multicentre study in hospitals will be developed in France. Such research is to analyse the feasibility of the diagnosis and to consider whether the cost of newborn screening is higher than the costs of early and late transplantation. The ultimate goal is to provide policy makers and Key Opinion Leaders with economic data supporting the need of SCID screening in newborns. Dr. Borte pointed out the value of such data, as in the EU some insurers would require economic prove of the importance of such screening campaigns before proceeding to do the reimbursement. Experts were confident that systematic SCID newborn screening would be implemented, eventually – but that the EU could help saving time – and hence many lives by facilitating this change.

Professor Gaspar pointed out that currently children are dying from SCID but the exact figures are not available, as too many go undiagnosed yet - which led the group to considering that it would be necessary to appropriately raise awareness among PID patient organisations, healthcare professionals and industry.

Mrs. Willmott and the expert group agreed to take action and to envisage how to accelerate the implementation process of SCID newborn screening in the European Union. It was agreed that this was a paediatric emergency that necessitates immediate action. The sooner this can be done, the more lives will be saved. As a direct outcome of the meeting a set of EU recommendations was endorsed.

EU Recommendations

See attached

List of participants

Mrs. Glenis Willmott, Member of the European Parliament, host of the PID Forum

Dr. Marie Audrain, CHU Nantes

Pr. Lennart Hammarstrom, Karolinska Institutet

Pr. Bobby Gaspar, University College London

Dr. Stephan Borte, University of Leipzig

Mrs. Katarzyna Witkos, Baxter

Mr. Klaus Hoerauf, Baxter

Mr. Ruediger Gatermann, CSL Behring

Dr. Katrin Presser, Talecris

Mr. Johan Prevot, IPOPI

Ms. Laura Savini, PPTA

Mr. Robert Perry, IPFA

Mrs. Jose Drabwell-Wuite, IPOPI

Mr. Sebastian Rohde, Rohde Public Policy

Mr. Giovanni Asta, Rohde Public Policy

Ms. Leire Solis Garate, Rohde Public Policy