The global organisation working to improve the quality of life for people with primary immunodeficiencies.

IPOPI EU POLICY FORUM

NEW BORN SCREENING FOR SEVERE COMBINED IMMUNODEFICIENCIES

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Introduction

- IPOPI, the Association of national patient organisations dedicated to improving awareness, access to early diagnosis and optimal treatments for PID patients worldwide
- Work with policy makers to address patients needs
- Dialogue with other stakeholders to design best approach

Introduction

- Thanks to Mrs. Willmott, MEP.
- Thanks to participants and experts who accepted to join and contribute to the elaboration of recommendations
- Thanks to sponsors supporting this IPOPI initiative

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We are stronger if we work together!

Introduction

- *Primary Immunodeficiencies* (PID) is a group of +/- 200 rare disorders that can affect anyone regardless of age or gender
- PID occur in persons born with failed immune systems
- Prevalence is difficult to establish: PID are massively UNDER DIAGNOSED in most countries

Consequences are dramatic for patients as symptoms of their condition are treated rather than their cause – even when treatments or cures exist!

Early Diagnosis – a chronic issue with PID

- Early diagnosis of all PIDs has been identified as public health priority in various EU initiatives

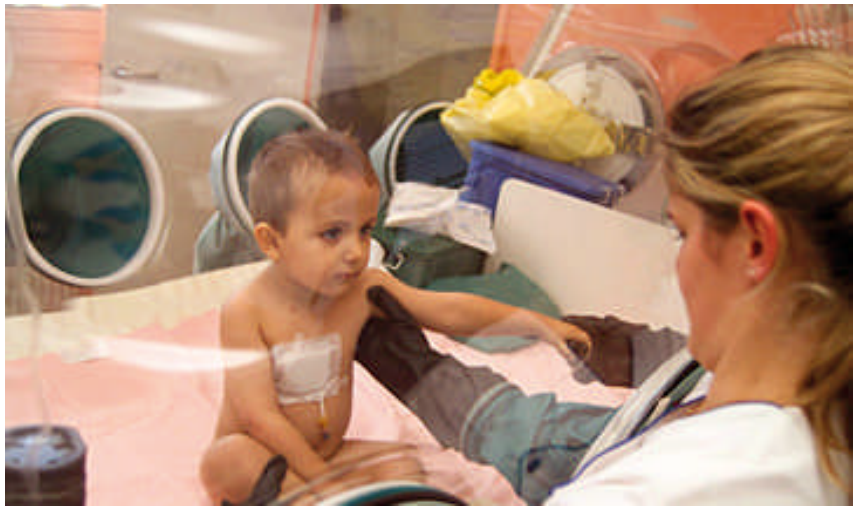
European Primary Immunodeficiencies Consensus Conference
19 – 20 June 2006
Paul-Ehrlich-Institut, Langen, Germany

Recommendations of the PID Expert group chaired by Jorgo Chatzimarkakis MEP

- Early diagnosis saves lives
- Significantly reduces the costs of treating symptoms (rather than the causing defect)
- Allow patients to receive effective treatment live a productive life; working and contributing to society

Severe Combined Immunodeficiency (SCID)

- (SCID) is one of the most serious PID – known to the public as the “bubble boy disease”, it occurs in 1 out of 50,000-100,000 newborn



- No immune protection against viruses, bacteria and fungi
- Exposed to serious and life threatening infections, which are the main cause for early death
- Without early diagnosis and proper treatment babies will die before their first birthday.

Need for Screening

- SCID is not apparent at birth
- Babies screened and diagnosed at birth have a MUCH HIGHER chance to be treated successfully
- Cure affecting the outcome is available
- Effective & easy to use tests are available

= meets the WHO newborn screening criteria!

Screening for SCID is a

Paediatric Emergency

The end of an exception?

- SCID newborn screening is subject to nation-wide implementation in the USA
- In the EU, SCID newborn screening is not implemented, even though the tools are available.
- Successful EU initiatives show added value of a coordinated EU action to address rare diseases.
- Communication 679 of the Commission and Council recommendations on Rare Diseases confirmed the importance of supporting the evaluation of neonatal screening strategies in Europe.

Countries / Disorder / prevalence[i]	France[ii]	Germany[iii]	Sweden[iv]	United Kingdom[v] England (E)[vi] Northern Ireland (NI); Scotland (S); Wales (W)
Phenylketonuria *(1-9/100000)*	Mandatory	Mandatory [vii]	Mandatory	Recommended: E/ NI /S/W
Cystic fibrosis *(1/8000-10000)*	Mandatory	Recommended		Recommended: E/ NI/S/W
Hypothyroidism (incl. congenital hypothyroidism) *(1/3000)*	Mandatory	Mandatory [viii]	Mandatory	Recommended: E/ NI/S/W
Congenital adrenal hyperplasia (including AGS) *(1-5/10000)*	Mandatory	Mandatory	Mandatory	
Sickle cell disease *(1/4000)*	Only on targeted population			Recommended: E/ NI/S
Galactosemia *(1/35000)*		Mandatory [ix]	Mandatory	
MCADD (Medium Chain Acyl-CoA Dehydrogenase Deficiency) *(1-5/10000)*		Mandatory		Recommended: E/ NI/S
Biotinidase Deficiency *(1/70000)*		Mandatory	Mandatory	
Homocystinuria *(1/1000000)*				Recommended: NI
Tyrosinaemia *(1/1500000)*				Recommended: NI
Duchenne Muscular Dystrophy (boys only) *(1/3,3000)*				Recommended: W
Maple syrup urine disease (MSUD) *(1-5/10000)*		Mandatory		
Long-chain 3-OH-acyl-CoA dehydrogenase deficiency (LCHAD) *(1-9/100000)*		Mandatory		
Very long-chain acyl		Mandatory		

Conclusion – We need to take action!

- SCID is a paediatric emergency - Introduce SCID in the obligatory list of diseases newborns are screened for
- Early diagnosis (within the first 3.5 months of life) allows access to a timely treatment that greatly improve the chances of survival of a SCID patient.
- Prioritise early diagnosis of PID to save lives and reduce costs of treatment and care
- Ensure appropriate information of healthcare professionals to facilitate diagnosis
- The European Commission should ensure that future policy initiatives, including a possible proposal for a Council Recommendation on Newborn screening, will include SCID in the list of diseases newborns are screened for on a routine basis.

THANK YOU!