

Setting the scene on primary immunodeficiencies (PIDs)

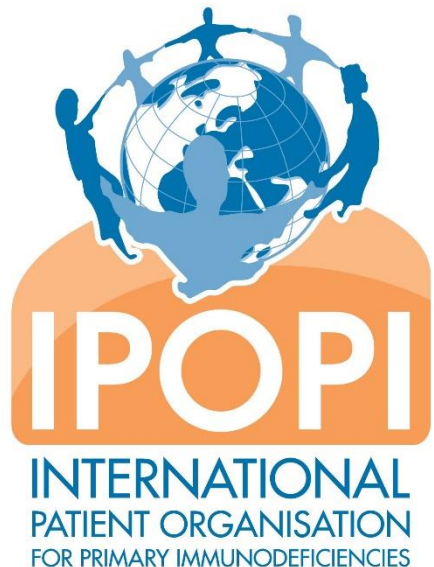
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IPOPI PID Forum

Empowering PID patients through rare disease policies 2009-2024

21 March 2024, European Parliament, Brussels

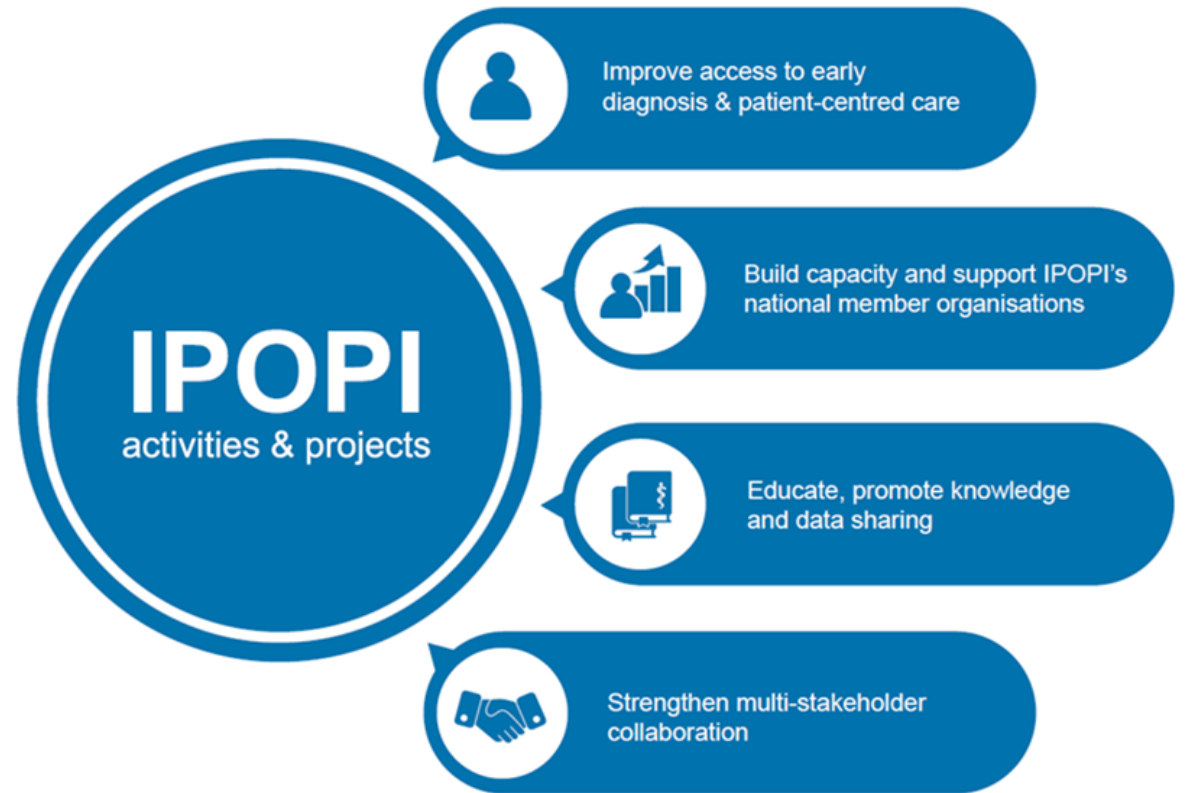


Introduction to IPOPI

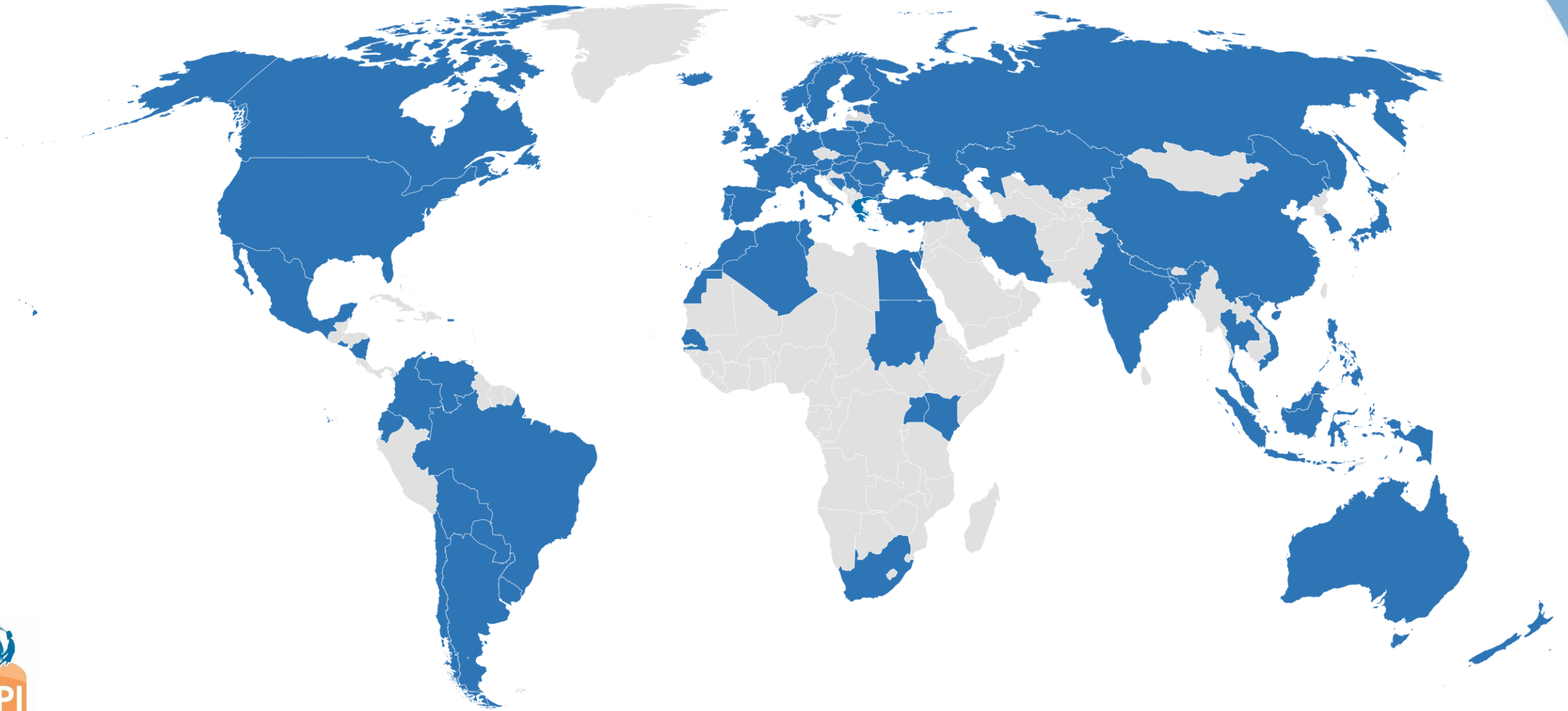
The association of national patient organisations dedicated to improving:

- Awareness
- Access to early diagnosis
- Access to care

For patients living with primary immunodeficiencies (PIDs) worldwide

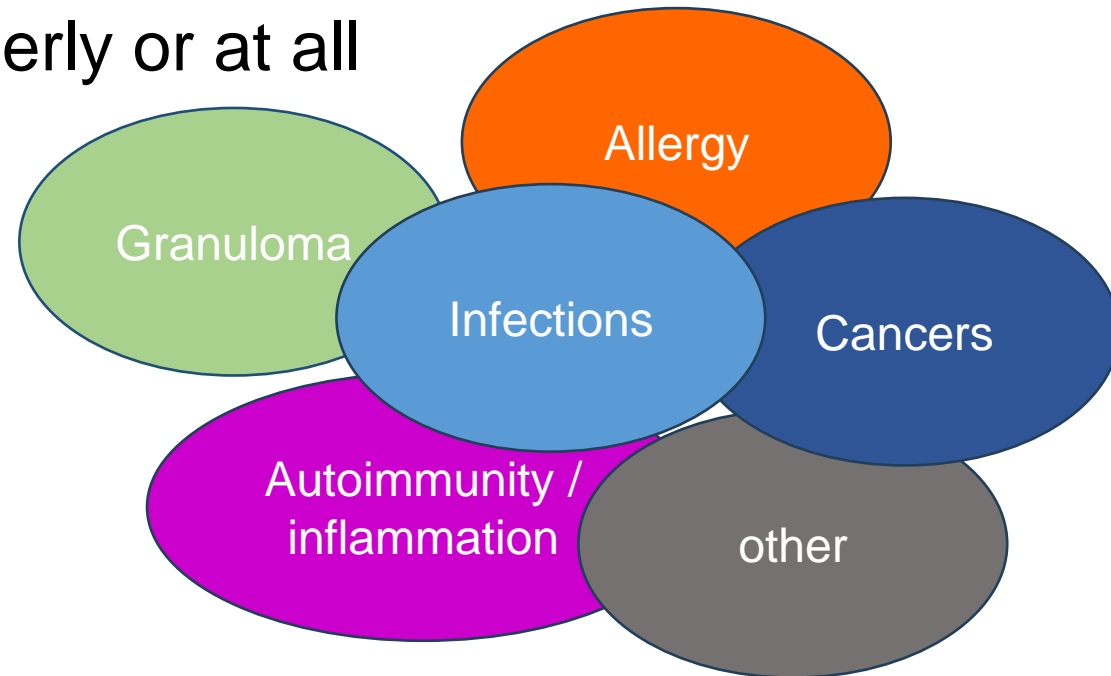


IPOPI members



What are Primary Immunodeficiencies (PIDs)?

- 485+ different genetic rare and chronic diseases
- The immune system does not work properly or at all
- Affect children and adults
- Clinical presentations are variable
- Patients with PIDs can have:
 - Increased susceptibility to infections
 - Autoimmunity
 - Inflammation
 - Lymphoproliferation
 - Allergy
 - Malignancy
 - ...



What are the treatment options for patients with PIDs?

- Anti-infectious therapies
- Vaccines
- Immunoglobulin replacement therapies
- Biological and targeted therapies
- Curative therapies
 - Hematopoietic stem cell transplantation / bone marrow transplantation
 - Gene therapy
 - thymic transplant / cultured thymic tissues

Becoming a patient...

- An individual can be diagnosed with a PID at any stage of his/her life.
- Diagnosis is key to avoid long-term damage... but hard to get sometimes → diagnostic delay / diagnostic odyssey

Country	Reported period (years)	Number of patients	Age at time of analysis	Age at onset	Age at diagnosis	Diagnostic delay	% of patients diagnosed as adults	References
Denmark	1994–2013	179	50.1 ± 17.0	29 (IQR; 3–87)	40 (IQR; 29–56) min 4; max 87	7 (IQR; 3–17)	–	Westh et al. (9)
Germany	2012–2017	728	40 (3–88)	–	Max 79	Mean: 7.35 median: 3	6.9%	El-Helou et al. (1)
Italy	1985–2015	75	50.08 ± 15.81; Median: 49	32 [17.82]*	40 [16.01]*	7 (IQR; 3–13)	–	Graziano et al. (10)
Poland	2017	77	39.19 ± 13.61	22.16 ± 14.32	32.29 ± 14.9	10.13 ± 10.53	76.6%	Wiesik-Szewczyk et al. (11)
Switzerland	2008–2014	98	–	–	–	Median: 5.95	87.5%	Marschall et al. (12)
United Kingdom (2008**)	2012–2017	1,404	–	–	–	4 (IQR; 1–10) 4 (0–69)	–	Shillitoe et al. (3)
Europe (23 countries)	2004–2014	2,700	–	18 (0–81) 22.4 ± 19.0	31 (4–89)	4 (0–69) 8.8 ± 11.4	69.5%	Odoletkova et al. (13)
Europe (16 countries)	2004–2012	2,212	–	–	–	4.1 (IQR; 1–11.8)	86.7%	Gathmann et al. (8)

If not otherwise indicated, data are presented as median (minimum-maximum) or median (interquartile range—IQR) or mean ± SD.

*Median [SD].

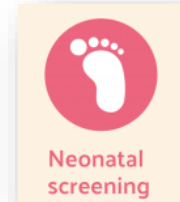
**United Kingdom Primary Immunodeficiency (UKPID) registry exists from 2008.



Summary of most relevant CVID epidemiological studies in selected countries (2020)

Becoming a patient...

- Newborn screening for some PIDs is possible: SCID can be treated and (sometimes) cured!
- “Simple” + genetic tests are needed for specific diagnosis.
- Multiple specialists are often needed in order to fully grasp the implications of the specific PID.



The fourth WHO model list of essential in vitro diagnostics (EDL 4)



Being a patient...

- Getting to terms with a chronic medical condition.
 - Learning & understanding the condition.
 - You are no longer like anyone else, certain limitations / precautions are needed.
- Getting to terms with the treatment.
 - Learning about the (complex) treatment (s).
 - Treatment is often burdensome, not so easy to handle, side effects, needs to be planned, etc.
 - Insecurity of supply with some therapies (anti-infectious, immunoglobulins, biologicals...)
 - Need to travel to receive it.

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Key words: Primary immunodeficiency; immunoglobulin treatment; qualitative research; thematic analysis.

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It's long-term, well it's for life basically: Understanding and exploring the burden of immunoglobulin treatment in patients with primary immunodeficiency disorders

Georgina L Jones,¹ Eva Brown Hajdukova,² Esmeeta Hanna,³ Rosie Duncan,⁴ Brendan Gough,¹ Jane Hughes,⁴ Debbie Hughes,⁴ Fran Ashworth,⁵ Johan Prevost,⁶ Jose Drabwell,¹ Leire Solis,⁴ Nizar Mahlaoui,⁷ Anna Shrimpton¹ on behalf of the International IgBoT study team

Norditropin (somatropin) supply shortage
First published: 05/12/2023
Last updated: 05/12/2023
Reference Number: EMA/514482/2023

English (EN) (147.45 KB - PDF)

Amoxicillin and amoxicillin/clavulanic acid supply shortage
First published: 27/01/2023
Last updated: 19/07/2023
Reference Number: EMA/27040/2023 Rev.1

English (EN) (174.5 KB - PDF) [View](#)

Source: EMA shortages catalogue



Source:

<https://www.ema.europa.eu/en/events/hma-ema-multi-stakeholder-workshop-shortages>

Being a patient...

- Getting used to a treating physician and the rest of specialists.
 - Managing appointments, several hospitals/ departments, ...
- Changing daily routine – precaution, treatment, travel to hospitals, work/school balance, family life, disease itself, travelling ... on top of “normal life”.
- Mental support is in many cases needed.



Ageing as a patient...

- Evolution of the PID over the years.
 - What is known/ felt by the patient & doctor
 - Unknown as little data
- Co-morbidities and subsequent treatment & care needed.
- Disability status needed in some cases.
- Work / family/ social adjustments.



New statement: A PID causing a heavy burden of disease must always be recognised as a disability

Key take aways

- PIDs are a large group of chronic and rare disorders that affect the individuals in a different manner.
- There is no one-size-fits-all approach for the individual patients.
- There are many aspects of patients' lives that can be profoundly impacted by EU policy and legislation.
- We look forward to continuing our collaboration with EU policy makers to jointly improve the lives of patients with PIDs.

Thank you for your attention



AN IPOPI EVENT

PID FORUM

Empowering The PID Community Through Rare Disease Policies 2024-2029

Hosted by MEP Radka Maxová
(S&D, Czechia)

MARCH 21 @ 09:00 - 11:00

ASP
5G305

