Shaping the Future of Research: Patients as Equal Partners

14 May 2025

PID FORUM

Shaping the Future of Research: Patients as Equal Partners

AN IPOPI EVENT

Hosted by:
MEP Romana Jerković
(S&D, Croatia)
MEP Nicolás González Casares
(S&D, Spain)

14 May 2025
10.00 - 12.00
EUROPEAN PARLIAMENT
SPINELLI 1G369

EVENT PROGRAMME

10:30	*Welcome Address*
10:40	*Setting the Scene*
10:50	*EU Rare Disease Research: ERDERA Perspective*
11:00	*Perspectives on Patient Involvement in EU Research*
11:30	*Questions & Answers*
11:45	*Key Takeaways & Policy Recommendations*
11:50	*Closing Statements*

INTERNATIONAL
PATIENT ORGANISATION
FOR PRIMARY IMMUNODEFICIENCIES

WiFi: *(to be inserted)*

Follow us on social media: *@ipopi_info*

INTERNATIONAL
PATIENT ORGANISATION
FOR PRIMARY IMMUNODEFICIENCIES

Welcome Address

MEP Romana Jerković

(S&D, Croatia)

MEP Nicolás González Casares

(S&D, Spain)

Setting the Scene

Johan Prévot

Executive Director, IPOPI

Understanding the potential & challenges of participation in EU research

An international patient federation’s perspective

Johan Prevot

Executive Director, IPOPI

IPOPI PID Forum

Shaping the future of research. Patients as equal partners

14 May 2025, European Parliament, Brussels

IPOPI
INTERNATIONAL
PATIENT ORGANISATION
FOR PRIMARY IMMUNODEFICIENCIES

Introduction to IPOPI

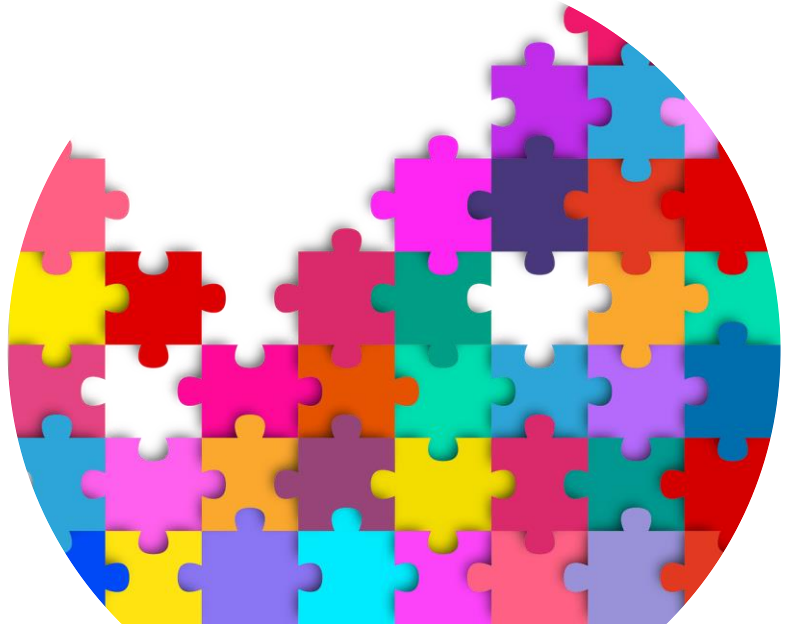
What is IPOPI?

A non-profit international organisation

- The leading advocate for primary immunodeficiency (PID) patients worldwide
- Working in collaboration with patients, doctors, politicians, regulators, pharmaceutical industry and other relevant stakeholders.

Our mission

To improve awareness, access to early diagnosis and optimal treatments for primary immunodeficiency patients worldwide through global collaboration.

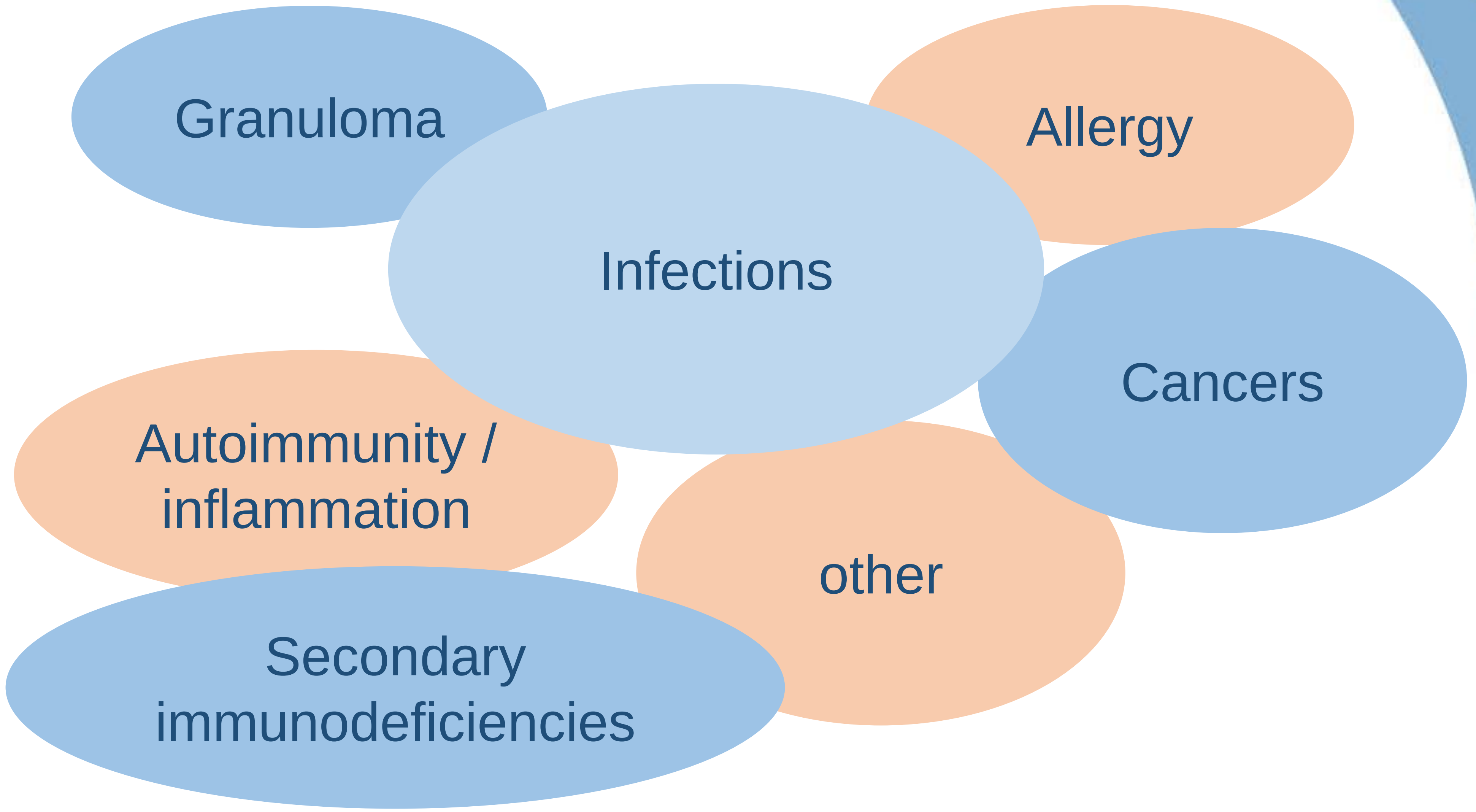


Primary Immunodeficiencies (PIDs) & associated conditions

Also called Inborn Errors of Immunity (IEI)

- 555+ different genetic rare and chronic diseases where the immune system does not function properly or at all (1)
- Affect children and adults
- Clinical presentations are variable
- PIDs are no longer defined by tendency for infections alone

  PID patients with non-infectious complications
  are increasingly recognised
  with features of immune dysregulation
- Life-impairing and life-threatening lifelong conditions
- **With associated conditions**


Granuloma

Allergy

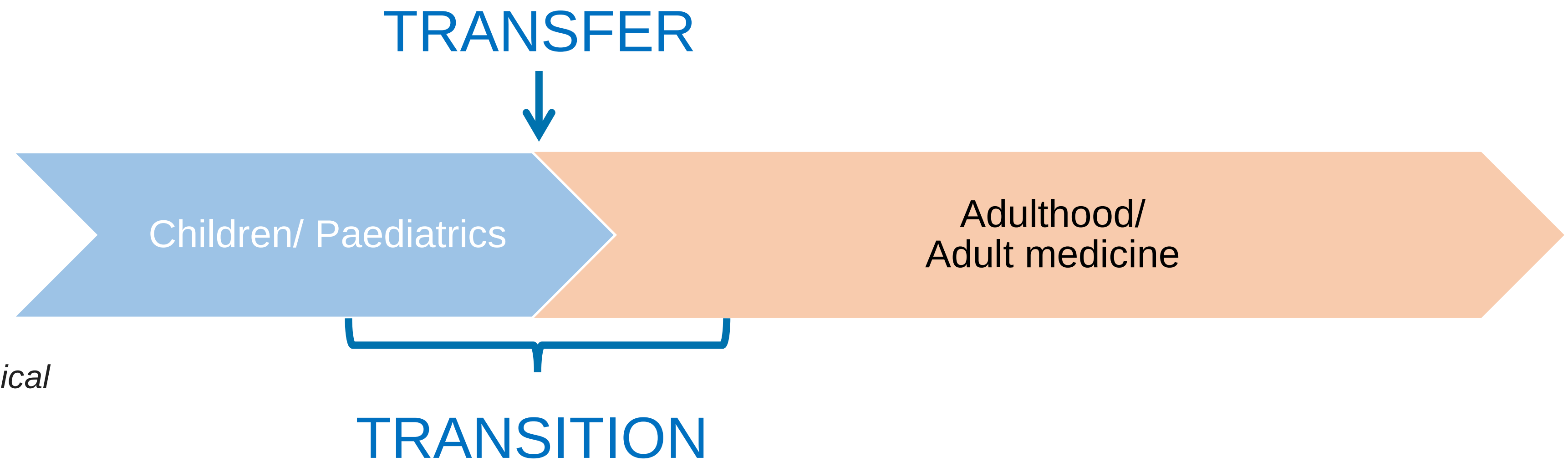
Infections

Cancers

Autoimmunity / inflammation

other

Secondary immunodeficiencies


TRANSFER

Children/ Paediatrics

Adulthood/
Adult medicine

TRANSITION

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(1) Human inborn errors of immunity:2024 update on the classification from the International Union of Immunological Societies Expert Committee. Journal of Human Immunity. April 2025. DOI:10.70962/jhi.20250003

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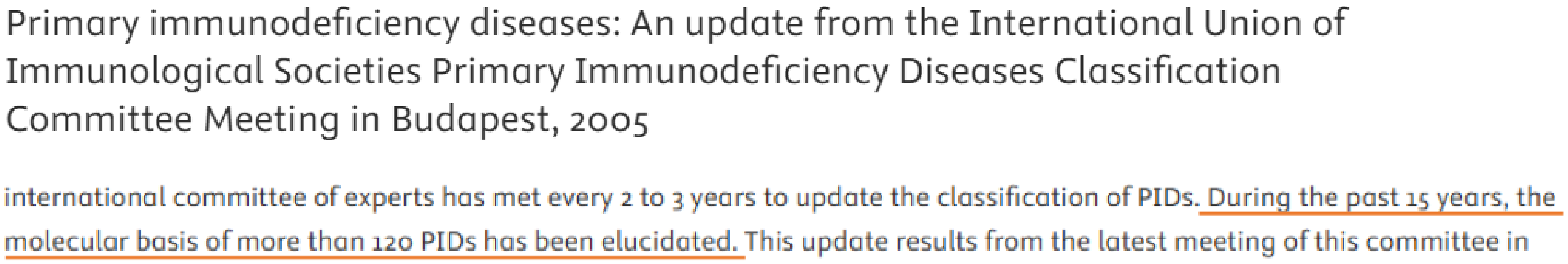

Primary immunodeficiency diseases: An update from the International Union of Immunological Societies Primary Immunodeficiency Diseases Classification Committee Meeting in Budapest, 2005

international committee of experts has met every 2 to 3 years to update the classification of PIDs. During the past 15 years, the molecular basis of more than 120 PIDs has been elucidated. This update results from the latest meeting of this committee in

→ Fast evolving field that requires constant scientific research to better understand conditions, but also diagnosis, treatments and care for the patients.

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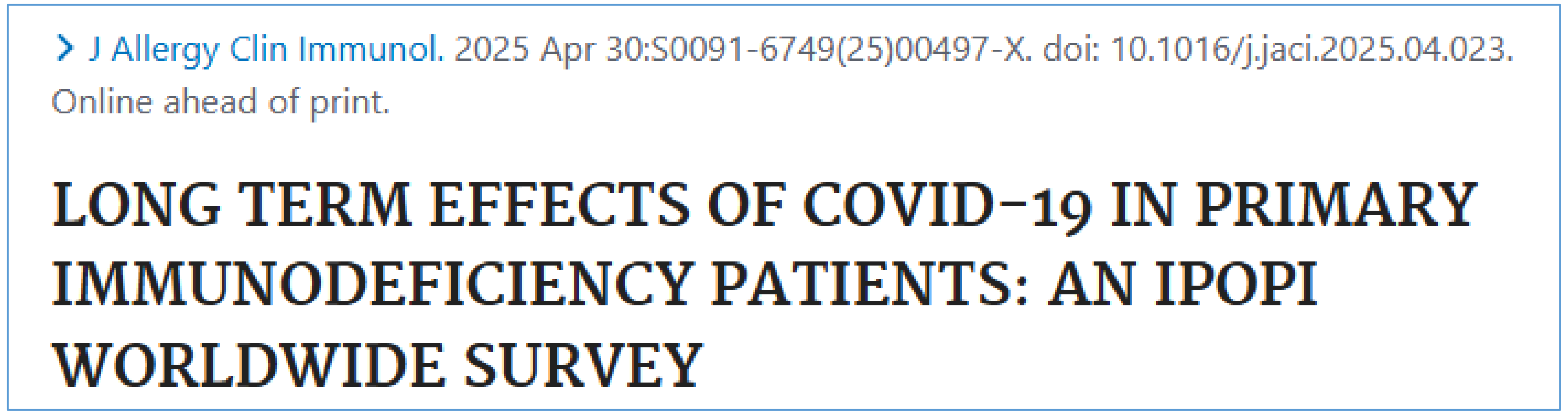
(1) Human inborn errors of immunity:2024 update on the classification from the International Union of Immunological Societies Expert Committee. Journal of Human Immunity. April 2025. DOI:[10.70962/jhi.20250003](10.70962/jhi.20250003)

IPOPI’s experience in research projects

SCIDNET

Recomb
gene therapy

3D-GATA2

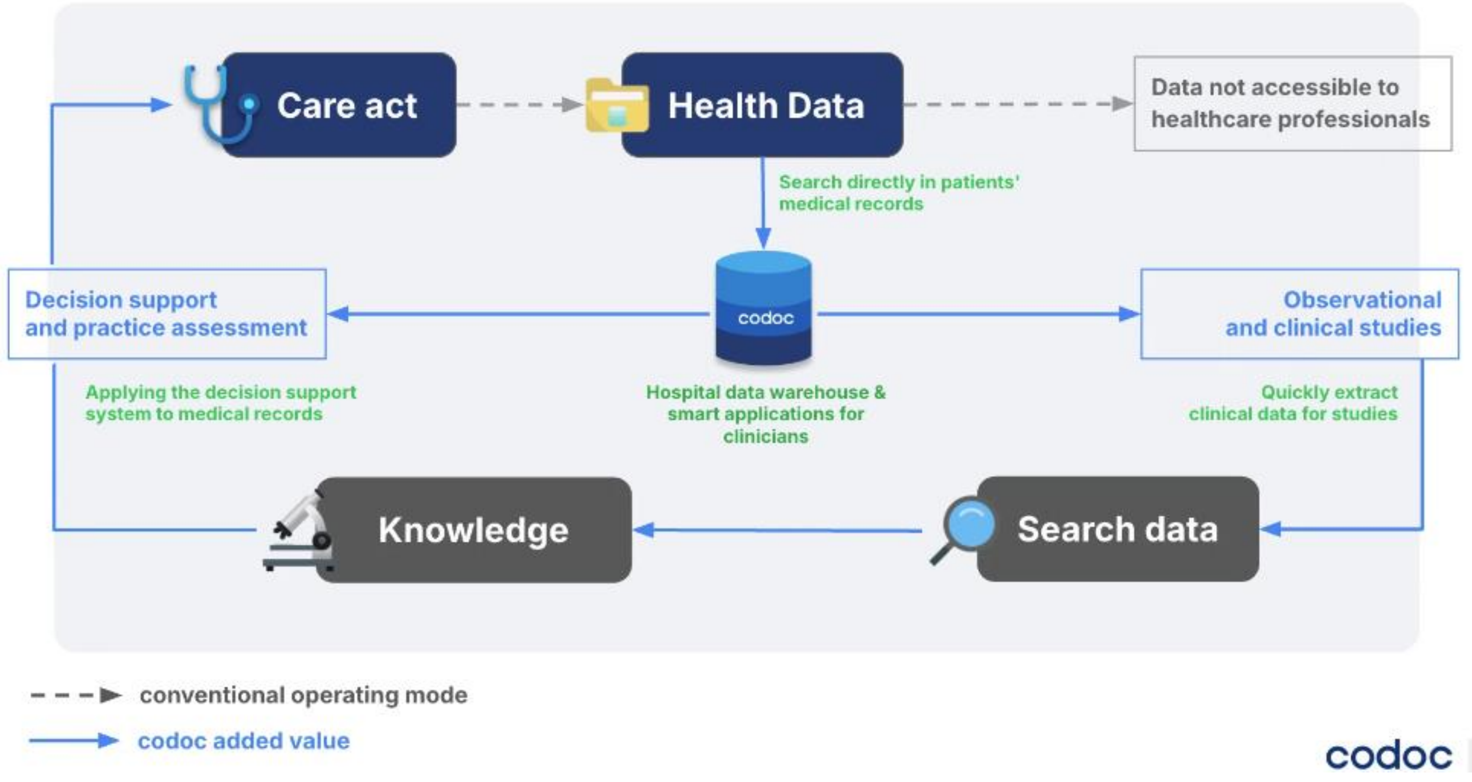


> J Allergy Clin Immunol. 2025 Apr 30:S0091-6749(25)00497-X. doi: 10.1016/j.jaci.2025.04.023. Online ahead of print.

LONG TERM EFFECTS OF COVID-19 IN PRIMARY IMMUNODEFICIENCY PATIENTS: AN IPOPI WORLDWIDE SURVEY

RareFindAI

Search & learning health systems



Care act

Health Data

Data not accessible to healthcare professionals

Search directly in patients' medical records

codoc

Decision support and practice assessment

Applying the decision support system to medical records

Hospital data warehouse & smart applications for clinicians

Observational and clinical studies

Quickly extract clinical data for studies

Knowledge

Search data

- - -► conventional operating mode

——► codoc added value

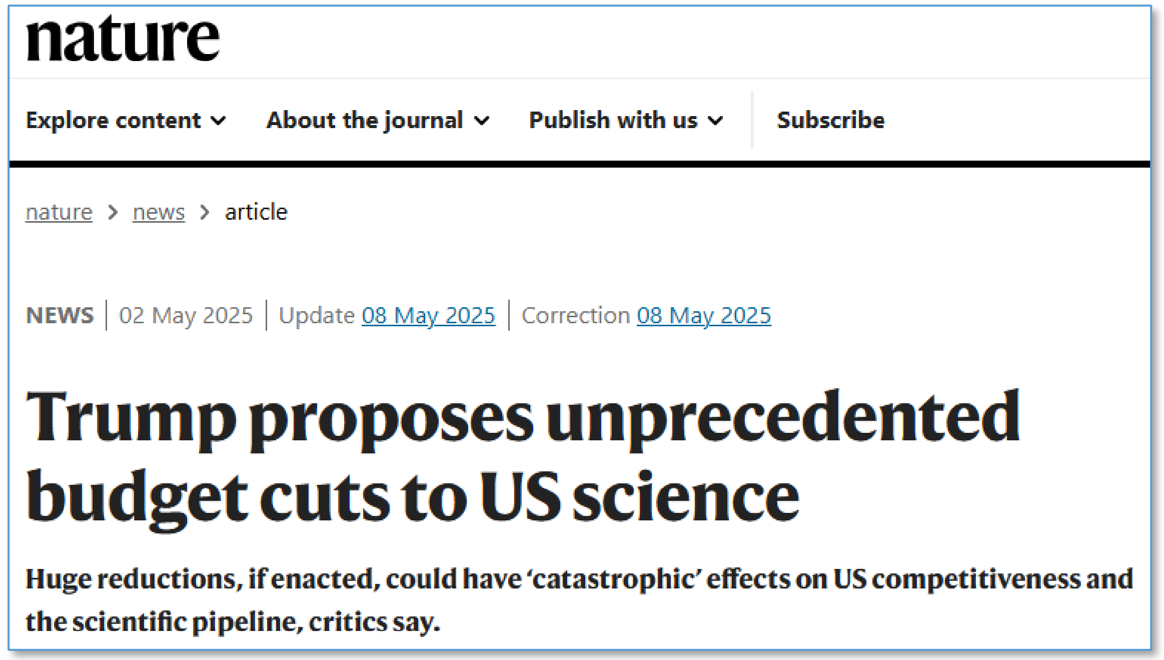
codoc

EU’s opportunity to lead on research & innovation


European Commission

President von der Leyen puts science centre stage with “Choose Europe” Initiative

https://ec.europa.eu/commission/presscorner/detail/en/ac_25_1137



NEWS | 02 May 2025 | Update 08 May 2025 | Correction 08 May 2025

Trump proposes unprecedented budget cuts to US science

Huge reductions, if enacted, could have ‘catastrophic’ effects on US competitiveness and the scientific pipeline, critics say.

https://www.nature.com/articles/d41586-025-01397-1


Choose Europe: EU launches bold plan to attract global talent for research in Europe

https://www.innovationnewsnetwork.com/eu-launches-bold-plan-to-attract-global-talent-for-research-in-europe/57757/


Research Letter

May 8, 2025

Characterization of Research Grant Terminations at the National Institutes of Health

Michael Liu, MPhil[1]; Kushal T. Kadakia, MSc[1]; Vishal R. Patel, MD, MPH[1,2]; et al

Author Affiliations

JAMA. Published online May 8, 2025. doi:10.1001/jama.2025.7707

https://jamanetwork.com/journals/jama/article-abstract/2833880


HHS Scraps Advisory Committee on Newborn Screening

— Cut comes as new conditions could have been added to a national screening panel

https://www.medpagetoday.com/special-reports/features/115070

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The PID field is one of the main drivers of scientific innovation

- With new genes being discovered every month in the PID field, new therapeutic pathways are being identified
- Development of innovative therapies to answer specific needs of different forms of PIDs/rare Diseases emanate from these discoveries
- These studies and others on rare diseases are crucial for identifying cellular and molecular pathways that are associated with the development of other more common diseases
- Patients and patient organisations are key players who contribute to ensuring research can most effectively tackle current unmet needs

Added value of patient organisations

- Expertise on the condition
- Expertise on the condition-related aspects: diagnosis, treatment, care, etc
- Patient-reported outcomes
- Input on trials design
- Regulations and medicine evaluation
- Health technology assessment of medicines & screening
- AI applied to concrete projects: diagnosis, treatment, care…

So what’s next?

- The EU wants to be a pole of attraction for research & innovation
- PID and RD research are innovation drivers
- Research & innovation without the patient voice are pointless
- Supranational federations representing specific rare diseases are a key player for many research & innovative projects
- Let’s make sure that the EU & EU research projects enable a meaningful participation and appropriately recognize the value of rare disease patient federations

Thank you for your attention

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EU Rare Disease Research: ERDERA Perspective

Daria Julkowska, Coordinator, European Rare Diseases Research Alliance (ERDERA)

ERDERA

European **Rare Diseases**
Research Alliance

Making Europe a world-leader in rare diseases research and innovation

Daria Julkowska, PhD
ERDERA Scientific Coordinator
INSERM, FRANCE

Follow us

Co-funded by the European Union

ERDERA has received funding from the European Union’s Horizon Europe research and innovation programme under grant agreement N°101156595.

Towards a comprehensive RD Research Ecosystem

2011-
Strategy
International Rare Diseases Research Consortium (IRDiRC), EC, Member States

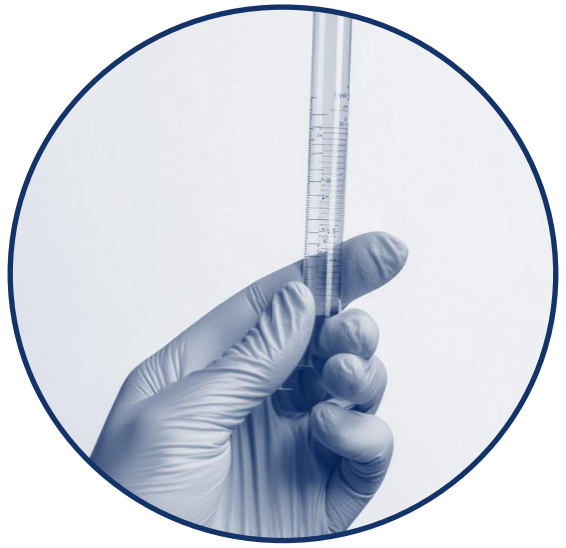
2006-
Funding
ERA-Net Rare (2006-2018) + EC + industry

1997-
Infrastructures
Orphanet, RD Connect, ERDRI, Solve-RD, EATRIS, ECRIN, BBMRI, etc.

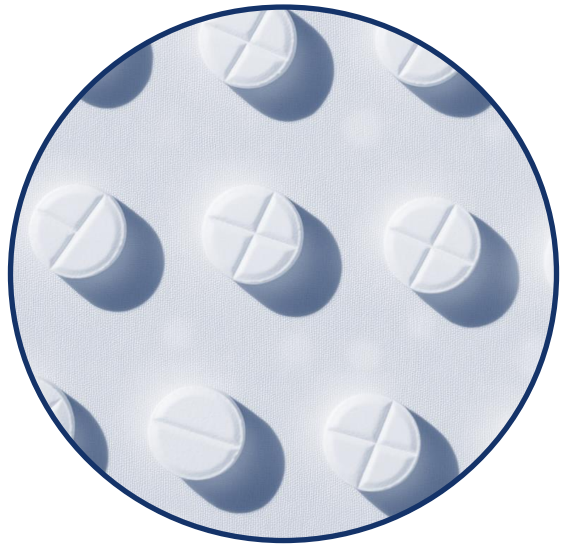
1997-
Patient needs
EURORDIS

2017-
Healthcare+
European Reference Networks (ERN)

Improving the health and well-being of 30 million people living with a rare disease by making Europe a world-leader in RD research and innovation.



Diagnosis established or enrolment in systematic research in average within 6 months after coming to medical attention



New effective therapies approved in Europe and beyond, the majority of which addressing diseases without approved options

Better understanding of the impact of rare diseases on patients, families and society to improve quality of life

Our alliance includes

+170

Organisations

Funders

Research performing organisations

Patients’ organisations

Research infrastructures

Private for-profit partners (industry & SME)

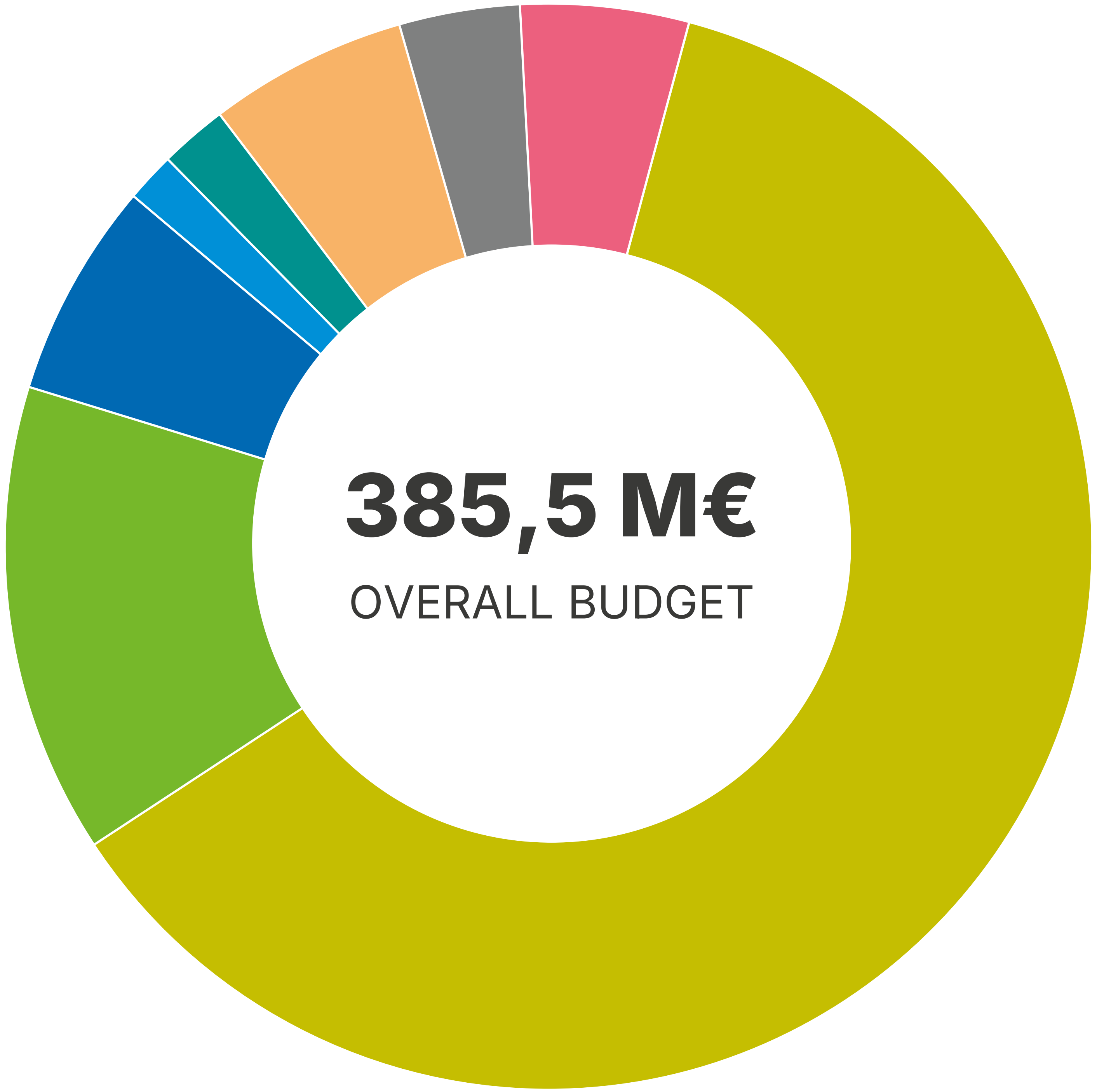
Other entities (universities, hospitals, non-profits, public administrations)

37

Countries

In Europe and beyond

Total Budget* per Work Stream



385,5 M€

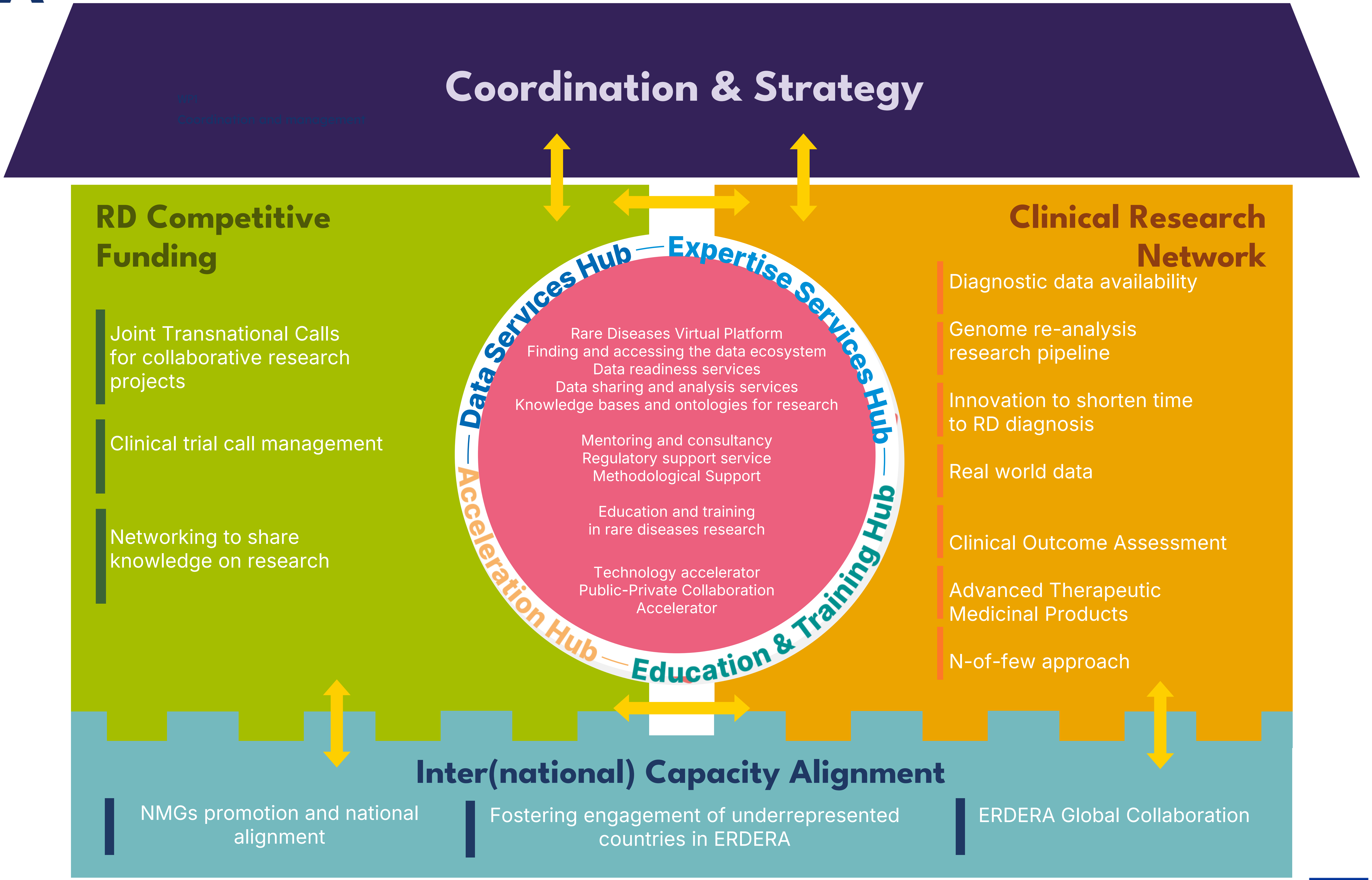
OVERALL BUDGET

- ● **244,886.552 €**
  RD Research Funding
  Joint Transnational Calls, Networking Support Scheme, and Clinical Trials funding
- ● **18,609.020 €**
  Coordination & Strategy
- ● **13,886,916 €**
  Inter(national) Capacity Alignment
- ● **23,408.401 €**
  Acceleration Hub
- ● **8,643,325 €**
  Education & Training Hub
- ● **5,335.450 €**
  Expertise Services Hub
- ● **20,125.025 €**
  Data Services Hub
- ● **50,734.575 €**
  Clinical Research Network

*estimated for **7 years**, includes: associated partners, in-kind and in-cash contributions, and 150 M€ of EC contribution

European Rare Diseases Research Alliance

ERDERA
European Rare Diseases
Research Alliance



Coordination & Strategy

WP1
Coordination and management

RD Competitive Funding

- Joint Transnational Calls for collaborative research projects
- Clinical trial call management
- Networking to share knowledge on research

Data Services Hub — Expertise Services Hub — Education & Training Hub — Acceleration Hub

Rare Diseases Virtual Platform
Finding and accessing the data ecosystem
Data readiness services
Data sharing and analysis services
Knowledge bases and ontologies for research

Mentoring and consultancy
Regulatory support service
Methodological Support

Education and training
in rare diseases research

Technology accelerator
Public-Private Collaboration
Accelerator

Clinical Research Network

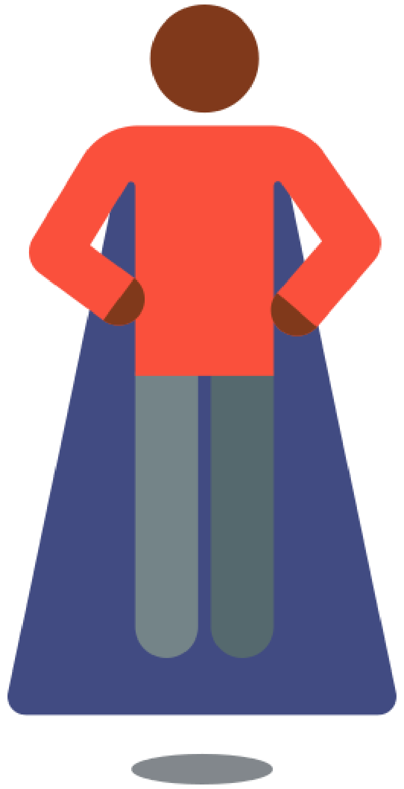
- Diagnostic data availability
- Genome re-analysis research pipeline
- Innovation to shorten time to RD diagnosis
- Real world data
- Clinical Outcome Assessment
- Advanced Therapeutic Medicinal Products
- N-of-few approach

Inter(national) Capacity Alignment

- NMGs promotion and national alignment
- Fostering engagement of underrepresented countries in ERDERA
- ERDERA Global Collaboration

Co-funded by
the European Union

ERDERA
European Rare Diseases
Research Alliance



Your voice/opinion/contribution via ERDERA:

- **Governing, executive and advisory** bodies
- **Patient & Public Engagement and Involvement** (PPIE) Group
- Direct involvement **as patient expert**
- **Open consultation**/survey

Your benefit via:

- Participation/coordination of a **Networking project**/event
- Participation/partnership in **multinational research project**
- Participation in **offered education & training**
- Participation/coordination of a **National Mirror Group**
- **Accelerated access to expertise**/advice

Co-funded by
the European Union

SHORT GUIDE
ON PATIENT PARTNERSHIPS
IN RARE DISEASE
RESEARCH PROJECTS

BASIC
PRE-CLINICAL
TRANSLATIONAL & SOCIAL

Written by the members
of the working group PENREP*
Guide first
published in July 2020
on www.ejprarediseases.org

* **P**atient **En**gagement in
Biomedical **Re**search **P**rojects.

EUROPEAN JOINT PROGRAMME
RARE DISEASES

SINCE 2015 WE IMPROVE INVOLVEMENT OF PATIENTS IN RESEARCH

- As potential funding partners: by offering them top-level, objective submission & evaluation scheme (E-Rare-3 & beyond)

- In our funding opportunities: by involving them in the topic definition and evaluation (EJP RD and beyond)

- As partners in patient-driven research projects: by supporting (funding) their participation *(In the Joint Transnational Call 2019, 36% and In 2021 we reached* ***100%*** *of funded projects involved PAOs)*

https://developpement.ejprarediseases.com/wp-content/uploads/2021/03/SHORT-GUIDE-ON-PATIENT-PARTNERSHIPS-IN-RARE-DISEASE-RESEARCH-PROJECTS.pdf

Remaining Challenges

- Mutual trust: same goal but different approaches leading to overlaps and potential dispersion of scattered funds
- Education: bi-directional(researcher-patient) training on what are partnerships in research → learning language of the other stakeholder
- Encompassing all forms of involvement in research & its objective evaluation
- Definition(s) of patient advocacy organization --> have a joint agreement to ensure that PAO involvement is fair and transparent
- Definition of partner in research --> ensure that PAOs are considered at the same level as other research partners in projects → status of PAOs as part of eligibility criteria by national funding bodies

Perspectives in Patient Involvement in EU Research

- ***Prof. Dr. Filomeen Haerynck**, Pediatric immunologist, UZ Gent*
- ***Daria Julkowska**, Coordinator, European Rare Diseases Research Alliance (ERDERA)*
- ***Christina Kyriakopoulou,** Scientific Policy Officer, DG RTD.D.2, European Commission*
- ***Johan Prévot**, Executive Director, IPOPI*
- ***Prof. Dr. Frank Staal**, Recombination Coordinator, Leiden University Medical Center*

Leiden University
Medical Center

Patient advocacy as integral part of research into novel therapies

Brussels, PID forum
May 2025

Recomb
gene therapy

Frank Staal

Immunology Department

LEIDEN UNIVERSITY MEDICAL CENTER (LUMC), NETHERLANDS

Curative stem cell-based gene therapies: key role for patient advocacy

IPOPI has played a key role as partner in 3 of my projects:
SCID-Net, RECOMB and CURE4 LIFE

- Dissemination
- Creating awareness
- Design of new trials, including PROMs (patient reported outcomes, but which?)
- Patient inclusion
- The pan European and global perspective

Five patients successfully treated for RAG1-SCID

[redacted]

[redacted]

[redacted]

En dit is nr 4 uit Wroclaw

[redacted]

IHB, LUMC
Laura Garcia Perez
Amiet Chhatta
Anna-Sophia Wiekmeijer
Martijn Cordes
Marja van Eggermond
Karin Pike-Overzet
Lieke van Roon
Sandra Vloemans
Willem Fibbe
Jaap-Jan Zwaginga
Anita Ordas
Kirsten Cante
Stefanie Klaver-Flores
Jacques van Dongen


Pediatrics, LUMC
Arjen Lankester
Robbert Bredius
Dagmar Berghuis
Mirjam van den Burg

IPOPI
Johan Prevot
Leire Solis
Lucia Mamede


Open Floor Discussion

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Key Takeaways & Policy Recommendations

Closing Statements

MEP Romana Jerković

(S&D, Croatia)

MEP Nicolás González Casares

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THANK YOU FOR ATTENDING THE PID FORUM!

Stay tuned for more...

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