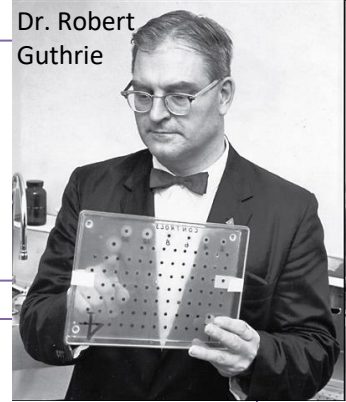


Regional Diagnosis and Clinical Challenges

Dr Nizar MAHLOUI (Past ESID Registry Chair Elect 2014-2018, Chair of the IPOPI MAP), adapted from the Slidedeck prepared by **Prof. Isabelle MEYTS** (ESID President) and **Prof. Fabio CANDOTTI** (ESID President-Elect)



Dr. Robert Guthrie



Newborn screening for SCID

ESID position:

- **Early detection** is the cornerstone of optimized management
- Recognize the value of **equitable access worldwide**
- Support efforts of political and awareness campaigns of **Screen4Rare**





Newborn screening for SCID: path ahead —ESiD IEWP to develop and publish guidelines

Guidelines for countries performing screening:

- how to manage babies awaiting treatment (at home? hospital?)
- links with reference centers

When/How to transplant?

- Role of conditioning
- Role of donor type





Clinical benefits of ESID-Registry

Publications: 21 articles in the past 5 years



- Clinical criteria
- Presenting symptoms
- Disease(Group) specific (AT, sIgM, UnPAD, XIAP, APDS...)
- Comorbidity (GLILD)
- Burden of disease/Burden of treatment (CVID)/LongTerm Outcomes
- Database & comparisons with other registries — international collab.

Pharma interest:

- Pharming — APDS Level3
- Takeda — ESID Registry / unPAD study
- Chiesi — ADA1 def.



Regional good news



European
Commission

Funding & tender opportunities

Single Electronic Data Interchange Area (SEDIA)

Development of new effective therapies for rare diseases

HORIZON-HLTH-2022-DISEASE-06-04-two-stage

Call for proposal

Grant

Programme	Horizon Europe (HORIZON)	Status	Closed
Type of action	HORIZON Research and Innovation Actions	Deadline model	two-stage
Opening date	06 October 2021	First stage deadline	01 February 2022 17:00:00 Brussels time
		Second stage deadline	06 September 2022 17:00:00 Brussels time

Budget: 60,000,000 €



Emerging concerns

- Orchard Therapeutics Press Release March 2022:
“...intends to **discontinue its investment in ... rare primary immune deficiencies** ...Wiskott-Aldrich syndrome (WAS), ...X-linked chronic granulomatous disease (X-CGD) and ... adenosine deaminase severe combined immunodeficiency (ADA-SCID).”
 - This includes Strimvelis® a gammaretroviral vector-based gene therapy approved in Europe
- Follows **Bluebird Bio leaving the European market**
 - making gene therapy for hemoglobinopathies unavailable



Opportunities

- **NATIONAL PATIENT ORGANIZATIONS REINFORCEMENT – PATIENT EMPOWERMENT**
 - ESID to work hand in hand with patient organizations to implement screening, to establish guidelines
 - IPOPI
 - Creation and support of strong national and international patient organizations > lobbying at the government level
 - ERNs could play an important role
- **Focus on national registries reporting to regional registries**
 - e.g. CEREDIH - ESID Registry > moving politics by numbers – no room for ignoring