



XVIII
IPOPI GLOBAL
PATIENTS' MEETING
an IPOPI event

#GPM2024

SUPPORTING
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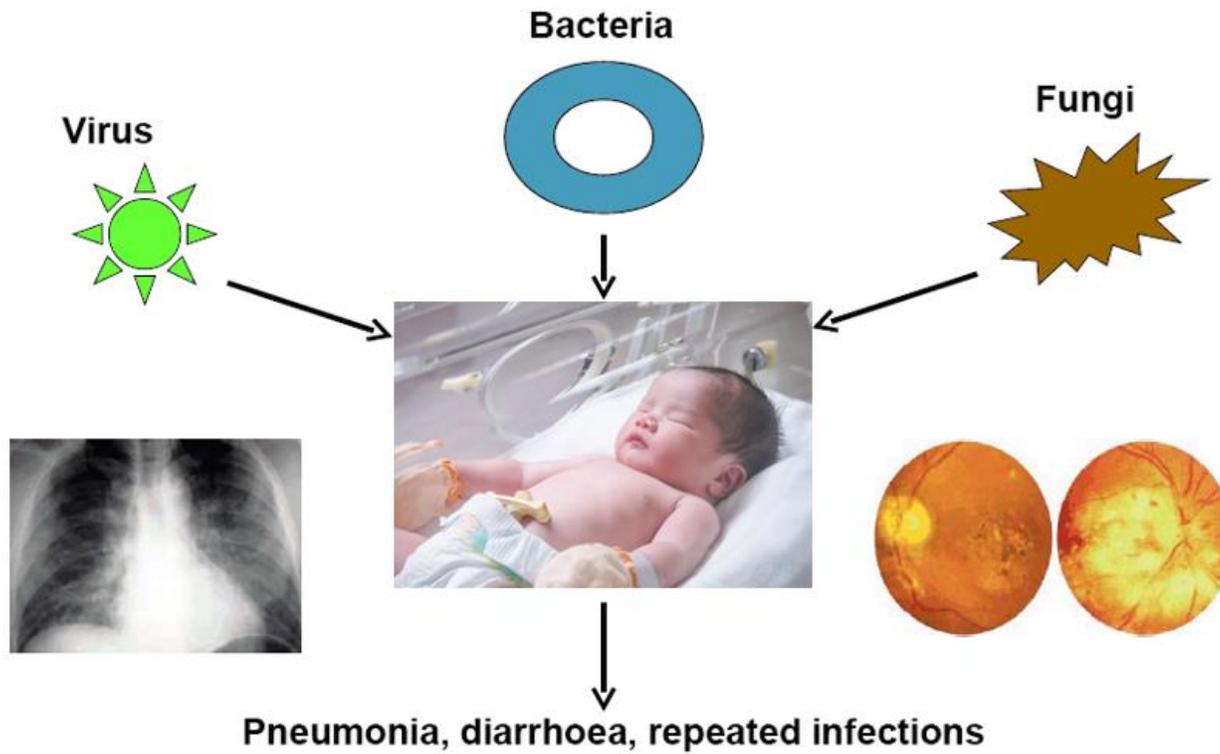


The newborn screening international landscape & how to get screening off the ground

Johan Prévot, IPOPI Executive Director

Why is NBS important to us?

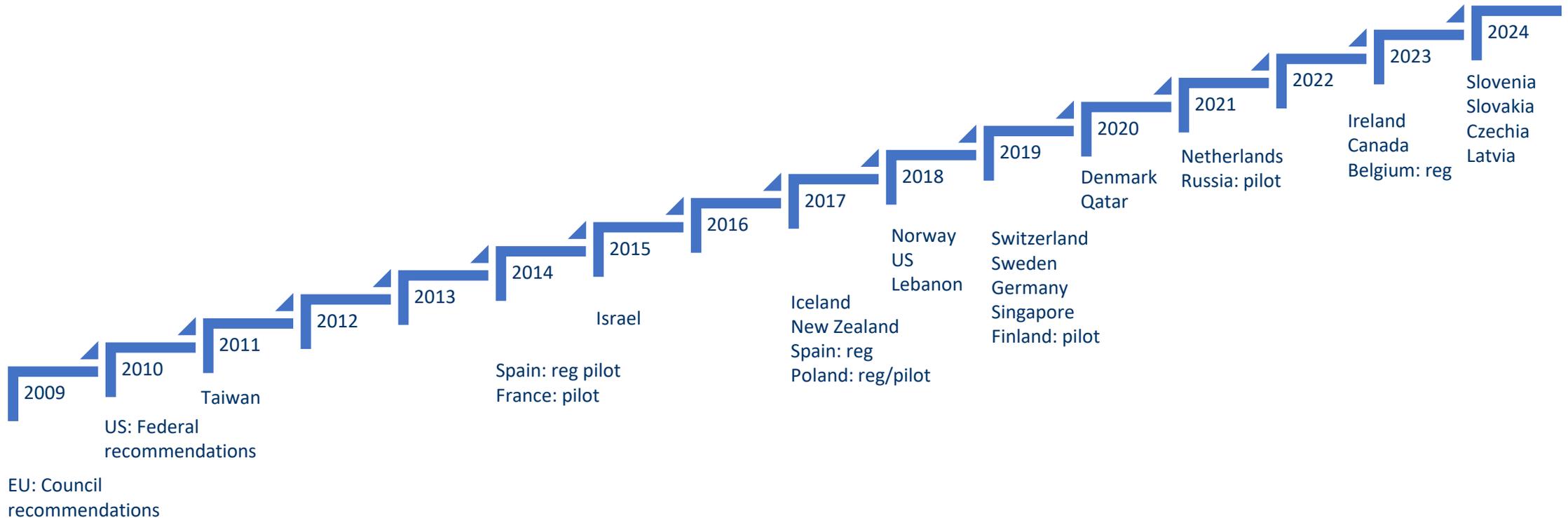
Severe Combined Immunodeficiency (SCID)



- Paediatric emergency
- Without early detection & effective intervention, the condition is associated with almost 100% mortality
- Meets all Wilson & Jungner criteria
- **Other PIDs may benefit from NBS in the future**

Implementation of SCID NBS

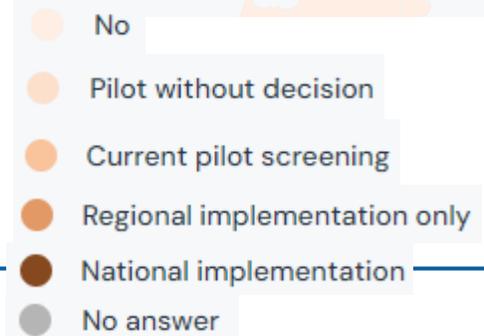
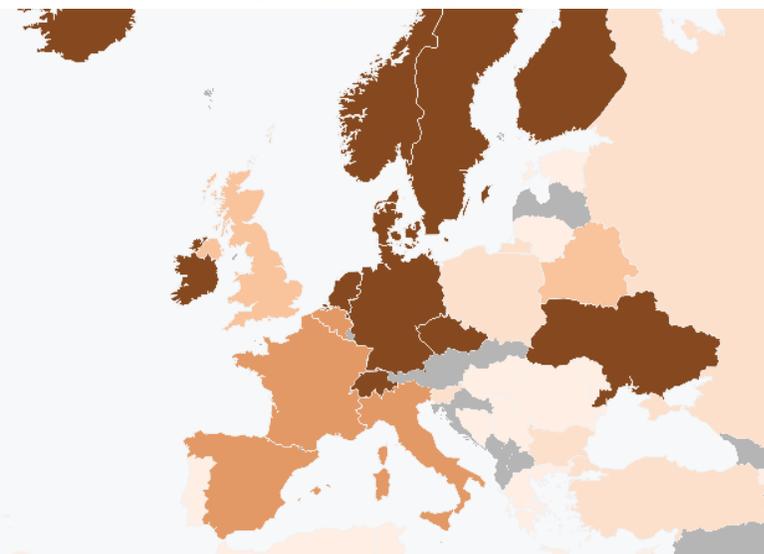
A long journey, with each new step a victory!



Regional campaign: screen4rare

an IPOPI event

- In the EU, Health is a **national competence** → countries have the last word **unless** EU can **add value**
- Co-founded by IPOPI, ISNS and ESID, **Screen4Rare** is a **multi-stakeholder** platform aiming to support & exchange knowledge and best practices on NBS for rare diseases :
 - By **partnering** with EU Commission to support member states
 - By launching the **MEP Alliance** on NBS
 - By **including NBS** in EU Presidencies' agenda
 - By **collaborating** with the ERNs
- To ensure all babies have **equitable access** to newborn screening



Source: <https://pidlifeindex.ipopi.org/#/en/principles/world-map>

S4R recent activities



Meeting with EU Commissioner, September 2023



December 2023

EN English

Home > Press corner > Screen4Rare MEP Alliance

Available languages: English

SPEECH | 6 December 2023 | Brussels | 1 min read

Commissioner Stella Kyriakides delivers a Speech via Video Message to the Screen4Rare MEP Alliance on Newborn Screening

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Related topics



April 2024



Conference Report

Newborn Screening Today and Tomorrow: A Brief Report from the International Primary Immunodeficiencies Congress

Leire Solis¹, Samya Van Coillie¹, James R. Bonham², Fabian Hauck³, Lennart Hammarström⁴, Frank J. T. Staal⁵, Bruce Lim¹, Martine Pergent¹ and Johan Prévot^{1,*}

- 1 IPOPI, BE-1050 Brussels, Belgium; leire@ipopi.org (L.S.); samya@ipopi.org (S.V.C.); bruce@ipopi.org (B.L.); martine@ipopi.org (M.P.)
- 2 International Society of Neonatal Screening, Reigerskamp 273, 3607 HP Maarssen, The Netherlands; j.bonham@nhs.net
- 3 Division of Pediatric Immunology and Rheumatology, Department of Pediatrics, Dr. von Hauner Children's Hospital, University Hospital, Ludwig-Maximilians-Universität München, DE-80337 Munich, Germany; fabian.hauck@med.uni-muenchen.de
- 4 Department of Medical Biophysics and Biochemistry, Karolinska Institutet, SE-17177 Stockholm, Sweden; lennart.hammarstrom@ki.se
- 5 Departments of Immunology and Pediatrics, Leiden University Medical School, 2300 RC Leiden, The Netherlands; f.j.t.staal@lumc.nl
- * Correspondence: johan@ipopi.org

Abstract: This article presents the report of the session on “Newborn Screening for Primary Immunodeficiencies—Now What?” organised during the International Primary Immunodeficiency Congress (IPIC) held in November 2023. This clinical conference was organised by the International Patient Organisation for Primary Immunodeficiencies (IPOPI), the global patient organisation advocating for primary immunodeficiencies (PIDs) in patients. The session aimed at exploring the advances in newborn screening (NBS) for severe combined immunodeficiency, starting with the common practice and inserting the discussion into the wider perspective of genomics whilst taking into consideration the ethical aspects of screening as well as incorporating families and the public into the discussions, so as to ensure that NBS for treatable rare disorders continues to be one of the major public health advances of the 20th century.



Citation: Solis, L.; Van Coillie, S.; Bonham, J.R.; Hauck, F.; Hammarstrom, L.; Staal, F.J.T.; Lim, B.; Pergent, M.; Prévot, J. *International Journal of Neonatal Screening* **2024**, *10*, 1–10. <https://doi.org/10.3390/ijns10010001>



Upcoming! 6 November 2024:

Screen4Rare & MEP Alliance for NBS for RD meeting at the European Parliament. Hosted by **MEP Billy Kelleher** (Renew, Ireland)

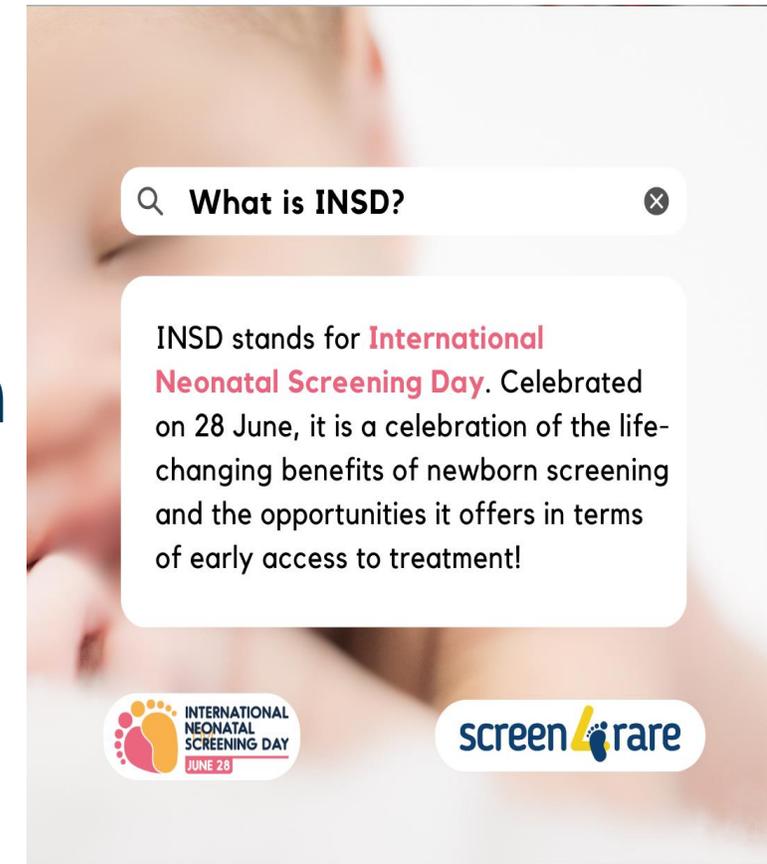
Global campaign: INSD

an IPOPI event

International Neonatal Screening Day (INSD)



Celebrating the achievements & raising awareness on the value of NBS Worldwide.



INSD – Around the world!



Happy International Neonatal Screening Day!
JUNE 28

INTERNATIONAL NEONATAL SCREENING DAY
screen4rare



Good morning and happy Europe Day!
8 June, Screen4Rare will be celebrating International Neonatal Screening Day. An initiative to save and improve the lives of newborns with treatable rare diseases worldwide.



Neonatal screening is not merely a medical test; it is a life-changing public health achievement that has the power to diagnose and treat rare diseases in their earliest stages.



"I'm proud to be part of the #InternationalNeonatalScreeningDay event. Neonatal screening is a life-changing public health achievement that has the power to diagnose and treat rare diseases in their earliest stages. If you have any questions, please feel free to contact me at [email address]."

Rare Links
in line with International Neonatal Screening Day
Diagnosis & Management of Biotinidase Deficiency

Wednesday July 24, 2024 | 11 am to 12:30 pm
Hybrid Webinar Series via CCKM & at Conference Rooms Superstars Building 2nd Floor, AKU

Dr. Ayesha Akhtar Associate Professor of Pediatrics Department of Pediatrics The Aga Khan University	Prof. Dr. Ayesha Akhtar Associate Professor of Pediatrics Department of Pediatrics The Aga Khan University	Dr. Ayesha Akhtar Associate Professor of Pediatrics Department of Pediatrics The Aga Khan University	Dr. Ayesha Akhtar Associate Professor of Pediatrics Department of Pediatrics The Aga Khan University

Learning Objectives:

1. Understand the screening and diagnosis of Biotinidase Deficiency
2. Clinical presentation and management of biotinidase deficiency in children.
3. Discuss the challenges in access to Biotinidase Deficiency

Who should attend?
All Paediatricians, Paediatricians and Nurses.

For Registration click here

CME Activity Code: GRS-PATH-136
"Rare Links: A Webinar Series 2024"
2.00



Ministers
Department of Health and Aged Care

Home Media centre Mark Butler Anika Wells Ged Kearney Emma McBride

Malarndirri McCarthy

Home > The Hon Mark Butler MP > Minister Butler's media

Ending the postcode lottery in newborn health screening

Today, on International Neonatal Screening Day, the Albanese Government is delivering on our commitment to end the postcode lottery, by investing \$39 million to achieve national consistency and expand the program.



The Hon Mark Butler MP
Minister for Health and Aged Care



2024- WORLD NEWBORN SCREENING DAY

[In honour of Dr. Robert Guthrie, Father of NBS]

Chemical Pathology Department

University of Ibadan / University College Hospital (UCH), Ibadan

Please Rest

SHINE Screening for Health, Intervention and Nurturing of Every child
with Arcadia

"Act NOW for Tomorrow's Future"

SHINE Program Launching

June 28 2024

9:30 am to 12:00 pm
L3-E-11, Level 3, HIVE 5, MRANTI Technology Park, 57000 Bukit Jalil, Kuala Lumpur

Haikal Ghezali
+6013 4186084
haikal@arcadialife-science.com

RSVP by 19 June 2024
SCAN OR CLICK HERE

YOU'RE INVITED

Importancia del tamizaje neonatal: Prevención temprana de enfermedades



IMT
Irish Medical Times

Number of diseases screened among newborns should be doubled, TDs and Senators to hear



DR ANSHU BANERJEE

Director of the Department of Maternal, Newborn, Child and Adolescent and Ageing, World Health Organization (WHO)

1:21

World Health Organization



The passage of the neonatal screening law is the culmination of many years of tireless teamwork between patients, medical professionals and lawmakers in the Dominican Republic.

This monumental step will mean a brighter future for Dominican newborns and their families, is a victory for all of Latin America and shows a path to follow for the advancement of equitable health globally.

screen4rare



UK Newborn Screening Laboratories Network

UKNSLN Annual Scientific Meeting – Friday 28th June 2024

Millennium Point, Curzon Street, Birmingham, B4 7XG

California Department of Public Health @CAPublicHealth

Today is International Neonatal Screening Day! Nearly all babies born in California are screened for many serious but treatable disorders.

To learn more about our CA Newborn Screening Program, visit: cdph.ca.gov/NBS.

#InternationalNeonatalScreeningDay

Newborn screening saves lives.

Nearly all babies born in California are screened for many serious but treatable disorders.

National campaign: Spain

an IPOPI event

Identify best strategy to get your campaign off the ground...



Asociaciones pediátricas españolas exigen la incorporación del cribado neonatal de IDCG

Los expertos de estas asociaciones aseguran la eficacia que tiene el cribado neonatal como diagnóstico rápido y replican que evitará muchas muertes.



Declaración de apoyo de IPOPI

A la implantación del cribado neonatal de IDCG en España

La Organización Internacional de Pacientes con Inmunodeficiencias Primarias (IPOPI), por sus siglas en inglés) apoya y solicita la implantación del cribado neonatal de la Inmunodeficiencia Combinada Grave (IDCG) en España.

IPOPI es la organización global que representa a los pacientes con inmunodeficiencias primarias (IDP), un amplio grupo de más de 200 enfermedades crónicas y raras en las que el sistema inmune o alguno de sus componentes, no funciona adecuadamente. Una de las formas más severas y que pone en peligro la vida de las personas que la sufren es la Inmunodeficiencia Combinada Grave (IDCG). La IDCG es una enfermedad compleja que afecta la salud de los niños. Los bebés nacidos con IDCG no tienen inmunidad celular o humoral y son incapaces de combatir infecciones graves causadas por virus, bacterias u hongos. La IDCG resulta fatal debido a una variedad de infecciones generalizadas en el primer año de vida a menos que un tratamiento específico pueda ser usado para corregir el defecto inmune subyacente. Al tratarse de la forma más severa de inmunodeficiencia primaria hereditaria, la IDCG es una emergencia pediátrica que pone en riesgo la vida del paciente.

Este estado de emergencia también ha sido puesto en evidencia por el Parlamento Europeo en una Pregunta Oral a la Comisión Europea pidiendo la elaboración de unas recomendaciones europeas sobre el cribado neonatal, incluyendo la IDCG.

2017

Initiatives in regional parliaments (Andalucía, Murcia)

2018

Clinical efficacy of SCID NBS report by the MoH



2021

HTA on SCID NBS by the Spanish MoH

2022

Scientific societies & AEDIP call for SCID NBS in the media

January 2017

Implementation of SCID NBS in Catalonia

23 October 2017

Roundtable at the Congress on SCID NBS in Spain

2018 - 2020

Initiatives in Parliament on SCID NBS, incl. in the Health Committee adopted by a large majority

2021 - 2024

Implementation of SCID NBS in some Spanish regions

April 2024

MoH announcement: Q2 2024 evaluation of SCID NBS



INMUNODEFICIENCIAS • Uno de cada 40.000 niños nace en España con el 'síndrome del niño burbuja'

El día que Vera rompió su burbuja

#GPM2024

BEATRIZ G. PORTALATÍN • 11 JUN. 2018 | 08:34

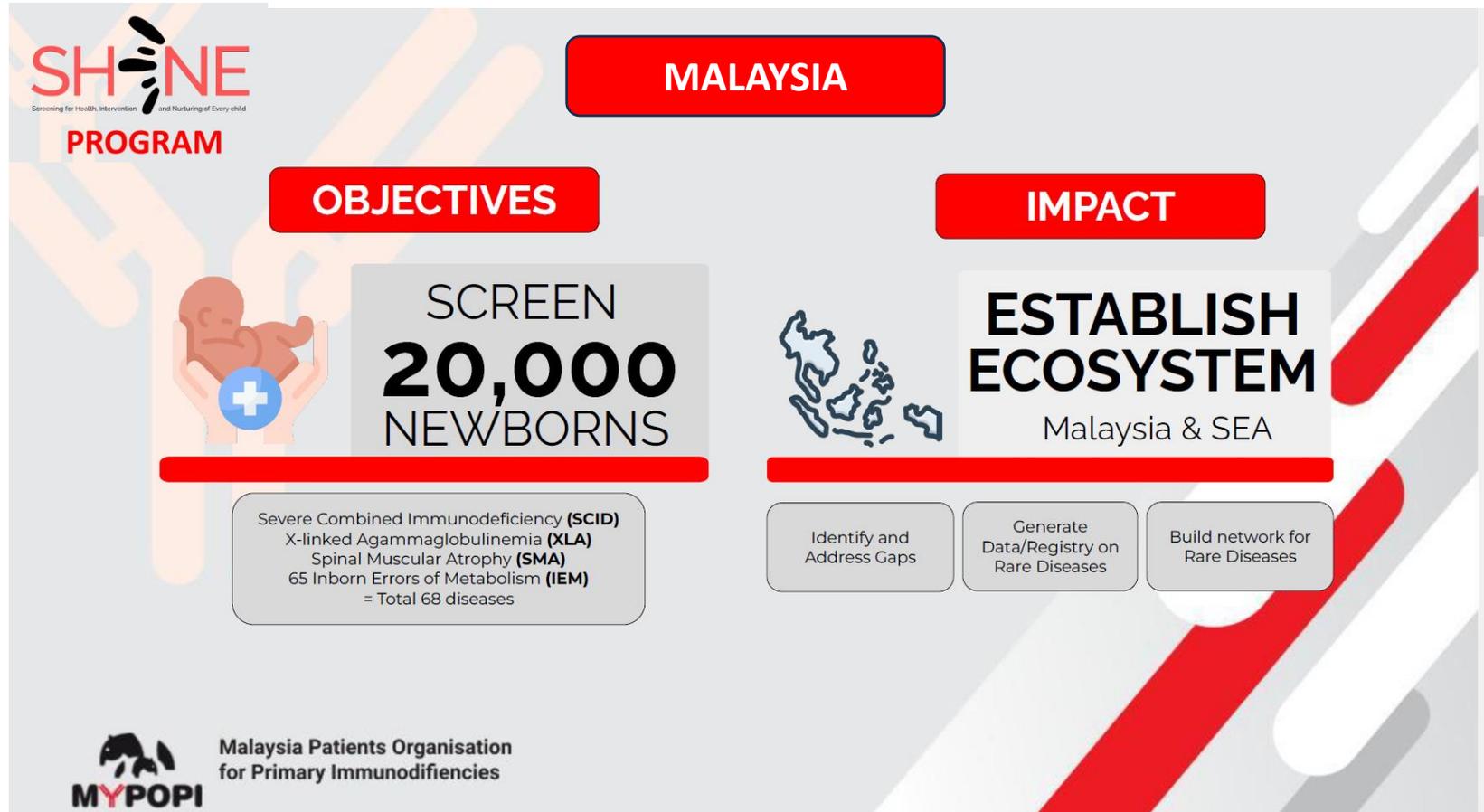
EL MUNDO

IPOPI.org

National campaign: Malaysia

an IPOPI event

Identify best strategy to get your campaign off the ground...



Getting a national campaign off the ground

TAKE HOME MESSAGES

- Takes time, patience...
- Understand your regional & national NBS environment (Status of your programme? other screened conditions? Federal or regional scope? Rare diseases plan? HTA aspects? Is treatment available? Other neighbouring countries are screening?)
- Identify your stakeholders and who can support / partner with you (doctors, screening experts, other patient groups, industry?...)
- Choose your angle & message based on your national environment (SCID focused or other focus with SCID as case study example i.e. Malaysia?)
- INSD - an opportunity to build momentum, making NBS best practise become common practise
- We are here to help!!