



# IPOPI 4<sup>TH</sup> REGIONAL ASIAN PID MEETING

19-20 NOVEMBER 2022  
KUALA LUMPUR, MALAYSIA

an **IPOPI** event

**CARE FOR RARE**  
GLOBAL ALLIANCE EDUCATIONAL SEMINAR



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# **Newborn screening for rare diseases:**

**Real life experiences and future opportunities from the  
Screen4Rare & Malaysia experience**

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RESEARCH

Open Access



# The PID Life Index: an interactive tool to measure the status of the PID healthcare environment in any given country

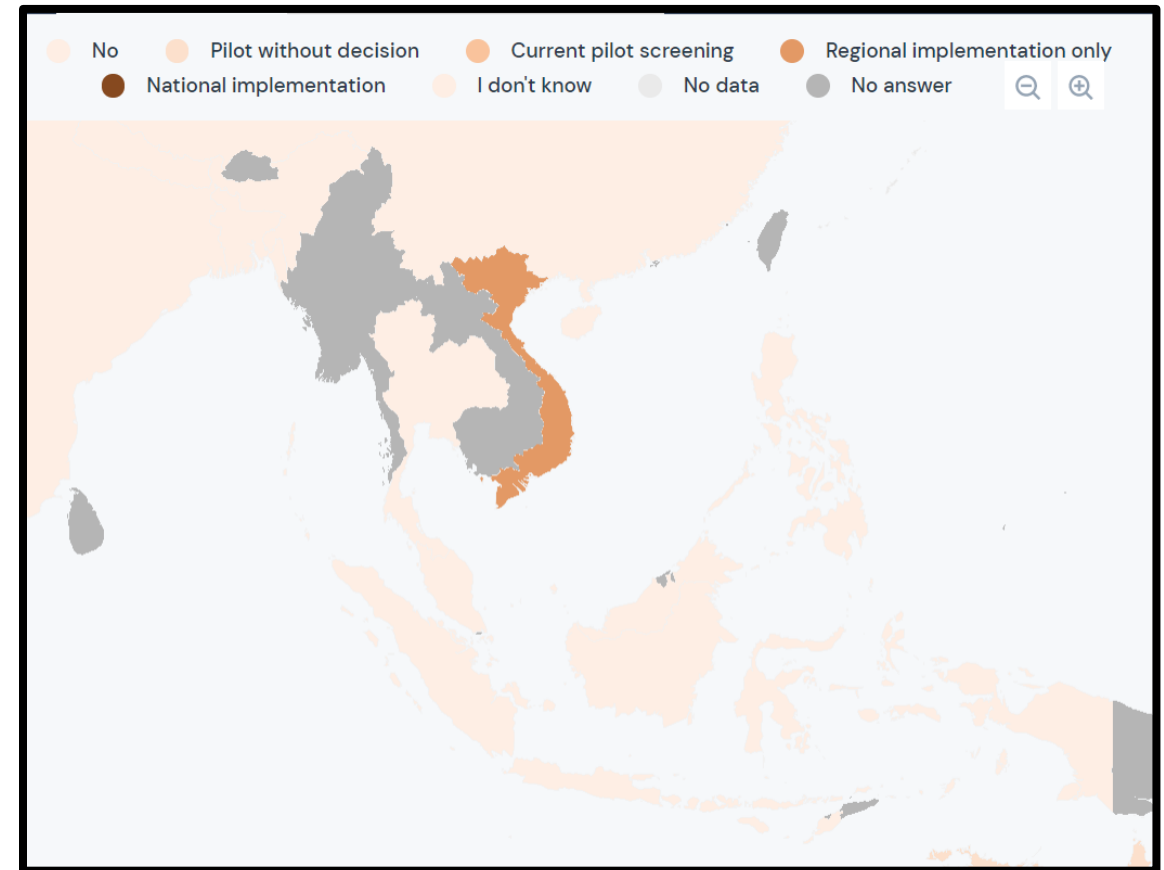
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# PID Diagnosis: Newborn screening for SCID



## Newborn screening for SCID

Identifies babies born with a SCID before the onset of an infection, so as to significantly reduce mortality and morbidity in affected patients





# Newborn Screening for SCID in Malaysia: The Initial Phase

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2014

Interest of starting Newborn  
Screening for SCID  
Conflict due to curative  
treatment



2015

Exploration of technology  
Engagement with the  
diagnostic provider



2016

Feasibility & Framework  
Program withheld

# Newborn Screening for SCID in Malaysia: The Incubation Phase

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Enhancing ecosystem: Strengthening expertise in curative management



Establishing Database : real-national data of SCID

Observation: Experiences from other countries

Capacity Building: Learning and understanding the technology

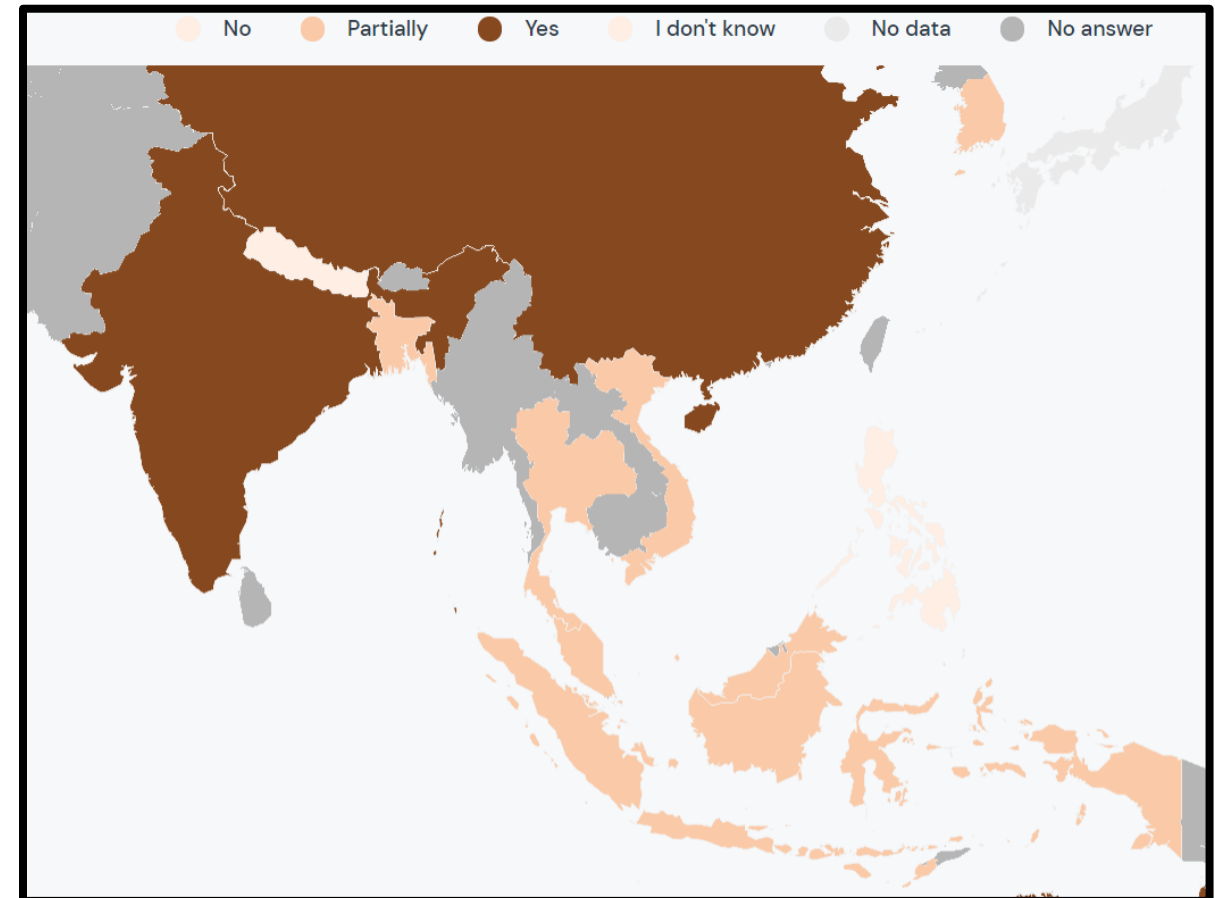
Pondering : The need, the feasibility in Malaysia

# PID Diagnosis: Genetic Diagnosis availability



## Genetic Diagnosis

Aims at identifying the mutations in one gene leading to a particular PID. Particularly helpful for those PIDs whose exact genetic defect is known.



# PID Treatment:

## Curative treatment (BMT/ HSCT)



### **Bone marrow transplantation or haematopoietic progenitor cell transplantation**

Specialised treatments in which HSC of an HLA compatible donor are provided to patients with usually severe PIDs (SCID, severe CGD) when the immune cells are missing or malfunctioning.

- **Gene Therapy**
- **Thymic Transplant**





# **Newborn Screening for SCID in Malaysia: Re-visiting and Re-strategizing**

## Determining the needs:

Survey, Pilot Project, Expert Discussion

## Evaluating the technology :

Pilot project

## Partners Engagement :

IPOPI, Other NGO, MOH, MOHE

## Advocacy:

WPIW campaign, Whitepaper,

## Feasibility and the cost-benefit analyses :

Framework and Pilot evaluation

# Conclusions

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## ❖ NBS for SCID in developing nation

- ❖ Understanding why, when, and how
- ❖ The pre-requisite ecosystem

## ❖ Importance of understanding local context

- ❖ Knowing the disease burden and management
- ❖ Identifying the stakeholders and the game changer
- ❖ Framing a practical roadmap