



EP PerMed
European Partnership
for **Personalised Medicine**

ICPerMed
INTERNATIONAL CONSORTIUM

ICPerMed & EP PerMed Conference on Personalised Medicine Research
Day 2, 27 November 2025

SESSION 3
Panelists' Introduction

Martina Cornel

President of the European Society of Human Genetics



The contribution of Medical Genetics to Personalised Medicine

Martina Cornel, Professor of Community Genetics & Public Health Genomics

EP PerMed & IC PerMed conference, Prague, 27th November 2025





Neonatal screening

- Public Health Intervention
- Offered to all newborn infants
- Goal: to identify infants with conditions for which effective therapy is available
- Early treatment can prevent or ameliorate the disease, so that affected children can live healthier lives
- Mainly genetic conditions (PKU, CF, MCADD), also hypothyroidism
- **More and more conditions can be treated and screened**





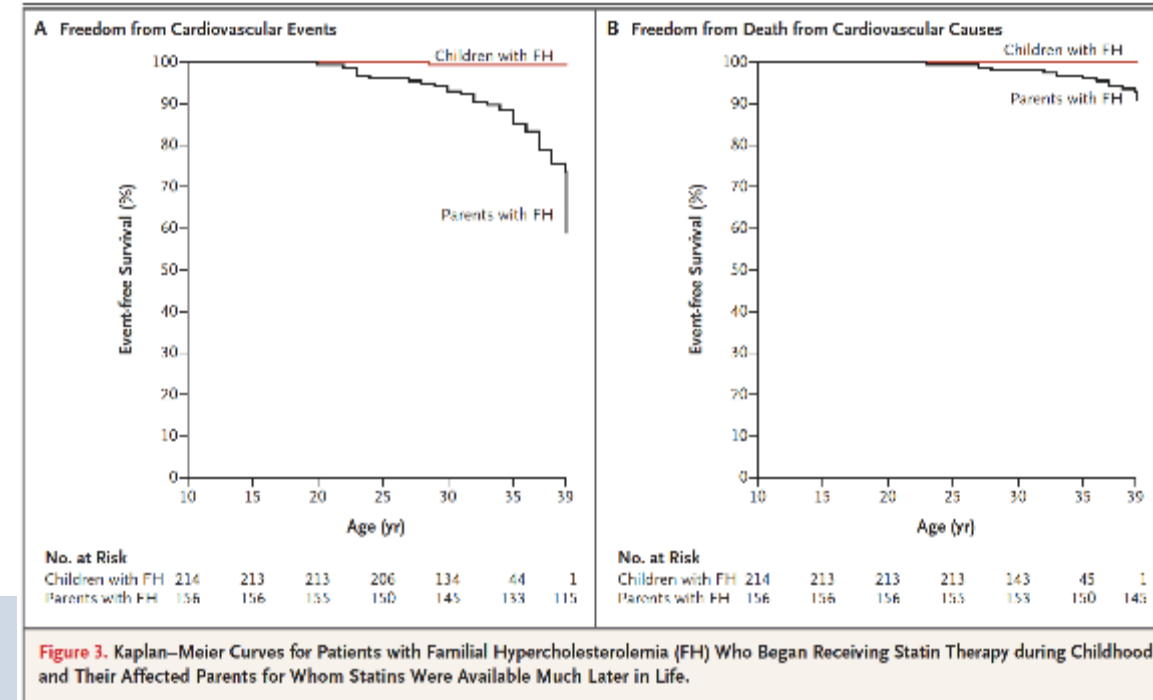
Cascade testing in a programmatic approach

- Where scientific evidence is sufficient
- Actively invite still healthy relatives at risk

ORIGINAL ARTICLE

20-Year Follow-up of Statins in Children with Familial Hypercholesterolemia

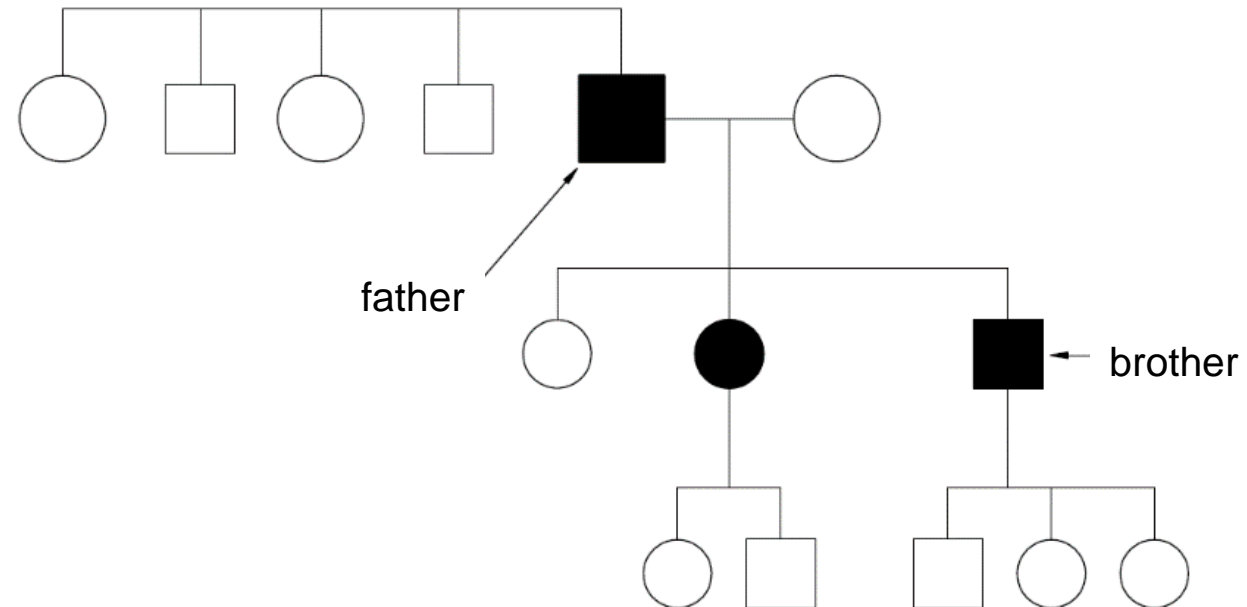
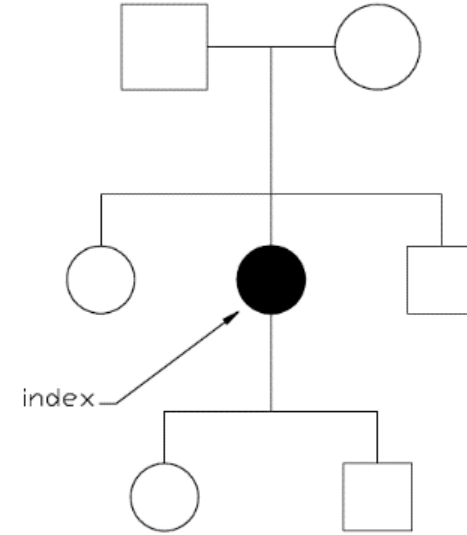
Ilse K. Lurink, M.D., Albert Wiegman, M.D., Ph.D.,
D. Meeke Kusters, M.D., Ph.D., Michel H. Hof, Ph.D.,
Jaap W. Groothoff, M.D., Ph.D., Eric de Groot, M.D., Ph.D.,
John J.P. Kastelein, M.D., Ph.D., and Barbara A. Hutten, Ph.D.





Monogenic subtypes of common conditions

- BRCA in breast cancer
- Lynch syndrome in colon cancer
- FH in cardiovascular disease
- DNA test in relatives at 50% risk
- If positive test result:
- Preventive interventions





Future of personalized prevention //

Pharmacogenomics

- Where evidence is available, implementation needs a coordinated program
 - Clopidogrel/ after stroke/ not effective with certain variants of CYP2C19
 - Fluoropyrimidine chemotherapy only after DPYD testing
 - Impact to assess
 - Deaths due to fluoropyrimidines
 - Recurrence of stroke after clopidogrel treatment
- Organize
 - Alerts in ordering in EHR
- Training, briefing, workshops, data collection
- Monitor and adjust



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Markus Kalliola

The Finnish Innovation Fund Sitra





Introduction to EHDS and TEHDAS2 joint action

Markus Kalliola, Programme Director, The
Finnish Innovation Fund Sitra

EHDS

- 1** Chapter 1: General provisions (Art. 1–2)
- 2** Chapter 2: Primary use of electronic health data (Art. 3–24)
- 3** Chapter 3: EHR systems and wellness applications (Art. 25–32)
- 4** Chapter 4: Secondary use of electronic health data (Art. 50–81)
- 5** Chapter 5: Additional actions (Art. 82–91)
- 6** Chapter 6: European governance and coordination (Art. 92–96)
- 7–9** Chapter 7-9: Delegation, Miscellaneous, Deferred application (Art. 97–105)

EHDS timeline

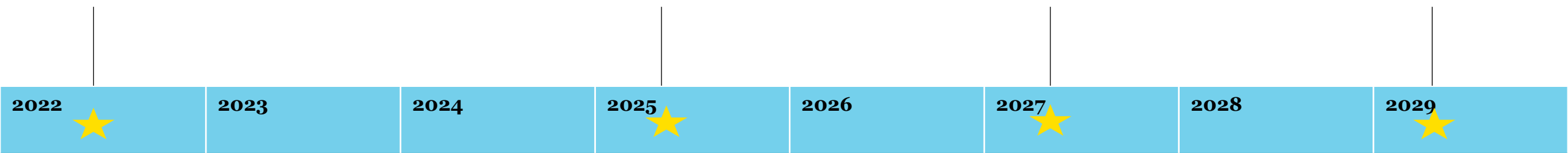
EHDS comitology 06/2025 ->

EHDS proposal
05/2022

EHDS entered into
force 03/2025

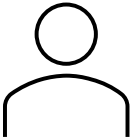
EHDS becomes
applicable 03/2027

EHDS secondary
use starts 03/2029



TEHDAS

TEHDAS2



You are here



EXPECTED RESULTS

Guidelines and technical specifications for data users, data holders and health data access bodies



EXPECTED IMPACTS

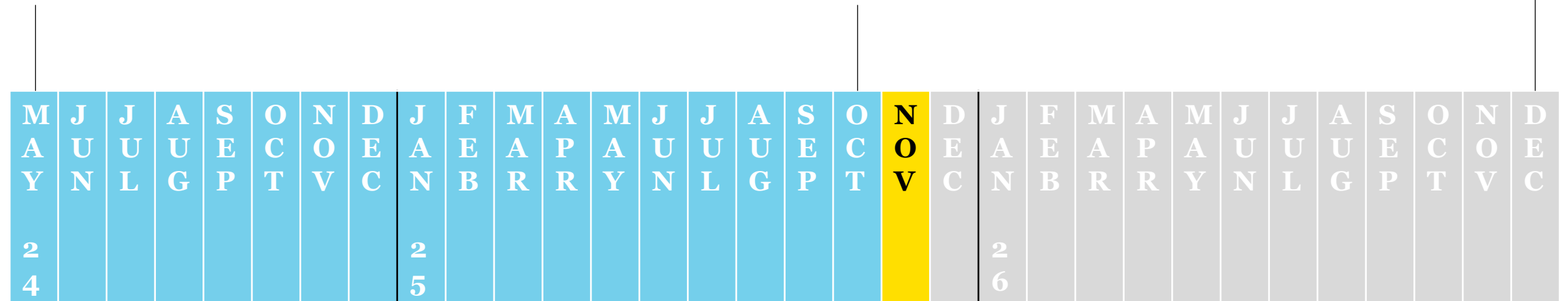
Better preparedness for implementing EHDS and better coordination on policies and less fragmentation on practices for secondary use of health data



TEHDAS2 timeline

May 2024

TEHDAS2 starts

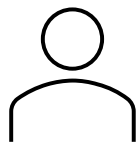


January 2025

1st stakeholder forum &
4 documents in public consultation

October–November 2025

2nd stakeholder forum &
11 documents in public consultation


You are here

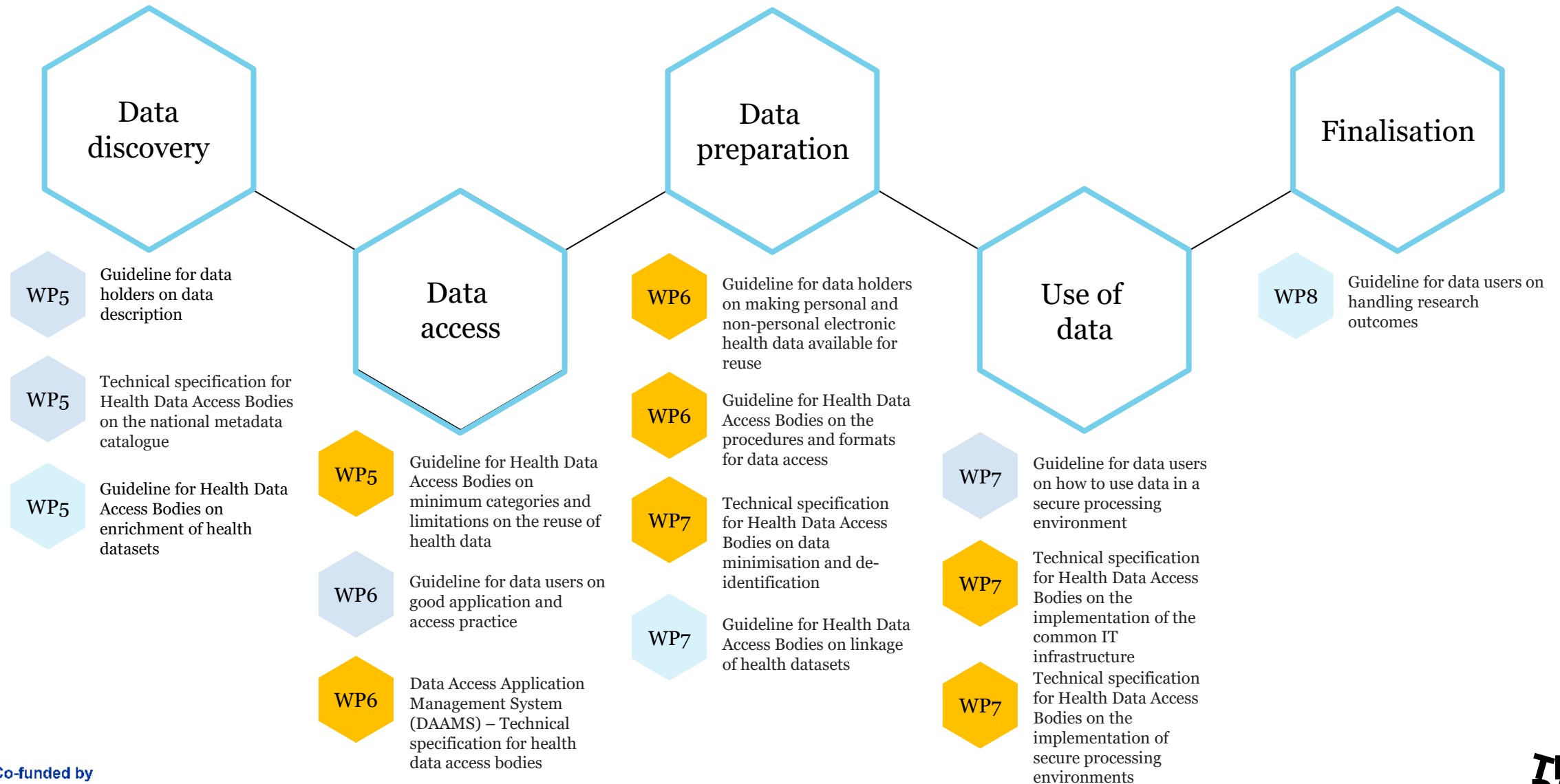
December 2026

TEHDAS2 ends

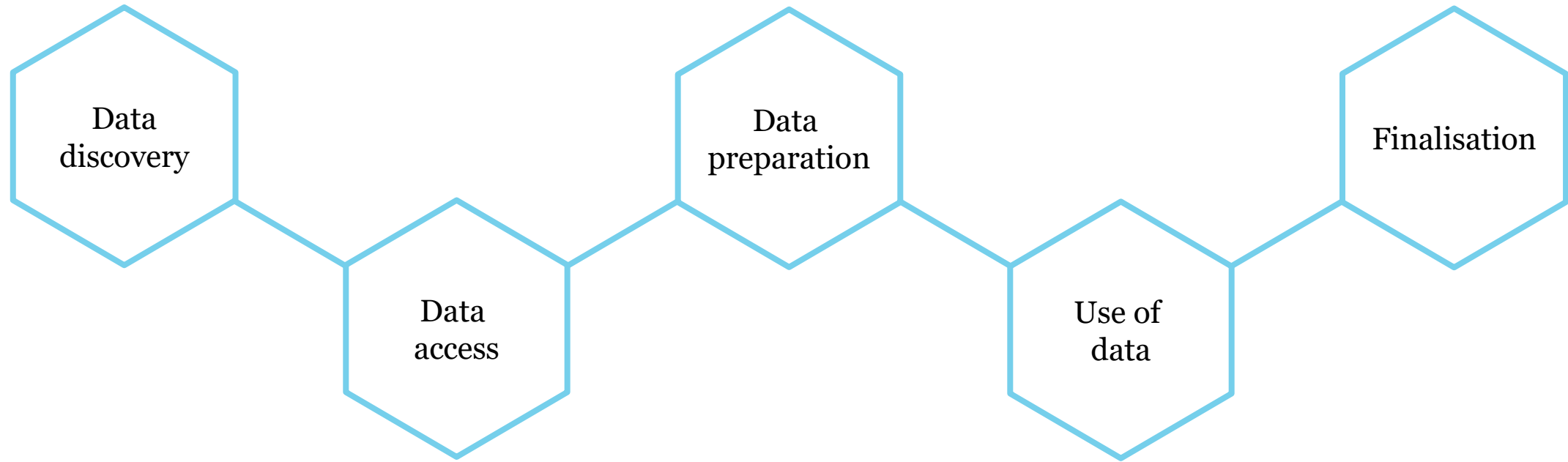
April 2026

3rd stakeholder forum &
7 documents in public consultation
(May–June)

EHDS User journey and TEHDAS2 work



and even more work...



WP4 Guideline for Health Data Access Bodies on fees and penalties for non-compliance regulated to the EHDS regulation

WP4 Guideline for Health Data Access Bodies on collaboration with other parties

WP4 Guideline for Health Data Access Bodies on international and third country access and transfer of electronic health data

WP8 Guideline for Health Data Access Bodies on the obligation of notifying the natural person on a significant finding from the secondary use of health data

WP8 Guideline for Health Data Access Bodies and trusted data holders on opt-out from the secondary use of health data

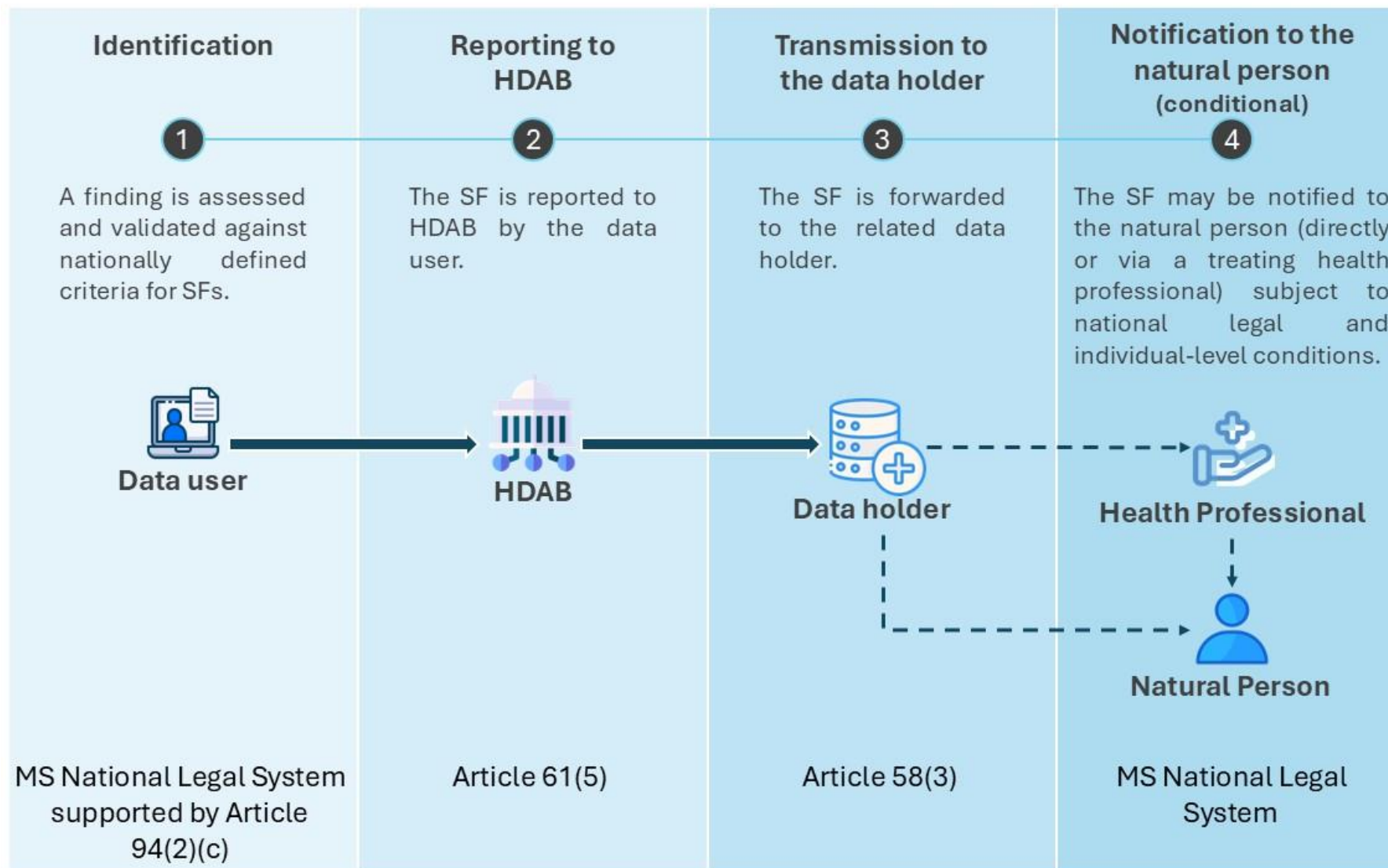
WP8 Guideline for Health Data Access Bodies on informing natural persons about the use of health data – "Citizen Information Point"

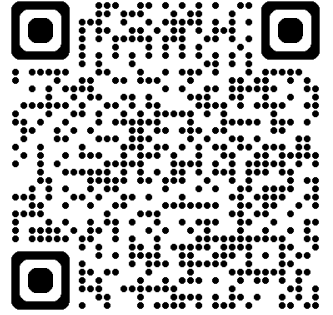


Draft guideline on notifying natural persons of significant findings from the secondary use of health data

- Explaining the meaning of Significant Findings (clarifying the term, presents legal framework, and give some typical examples)
- Outlines the journey of Significant Findings, with highlight on potential crucial points
- Focusing on the tasks of the HDABs, but not with just the responsibilities, trying to give advices on further opportunities on support the procedure.

Main elements of the draft guideline





Have your say!

Participate in the public consultation

PUBLIC CONSULTATION

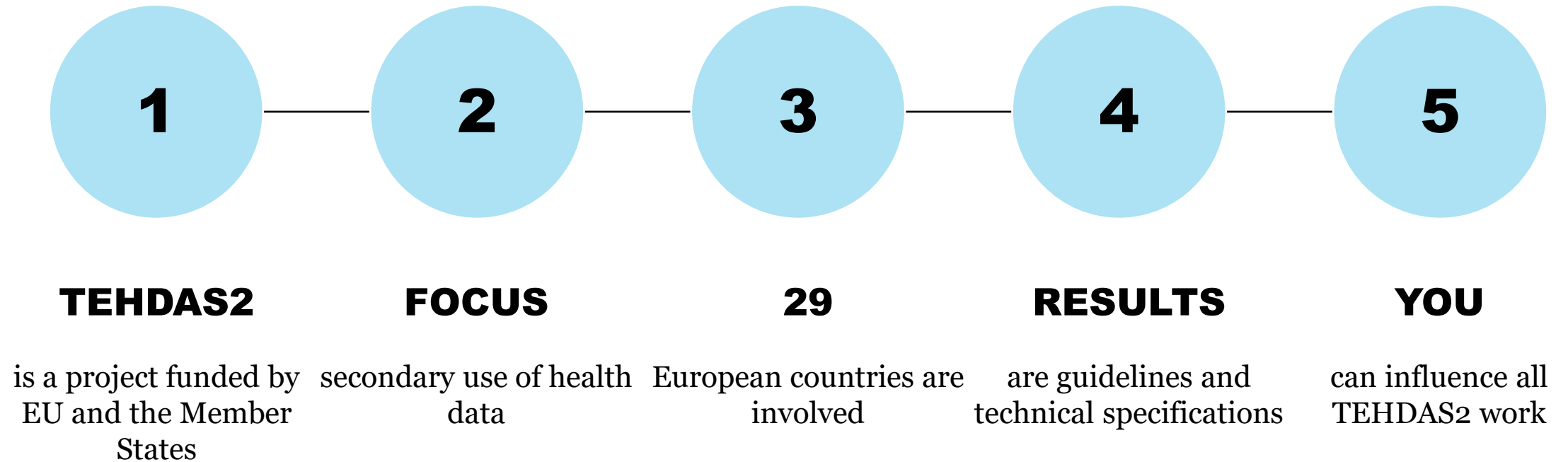
We would like to invite all stakeholders to share their views about how the **national HDABs can best help the journey** of Significant Findings! It is important that these findings reach the patients as quickly and securely as possible!

And please share your thoughts about the **definition of the Significant Findings!**

- How clear is this definition?
- Is it comprehensive enough?

The project team has tried to cover all professions and approaches, but every new perspective is valuable!

Summary



Follow us!

JOIN OUR NEWSLETTER

The next stakeholder forum will be hosted in Cyprus in April 2026.

LinkedIn: @tehdas2 joint action

bluesky: @tehdas

tehdas.eu



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Jillian Hastings Ward

Patient Advocate



10 years is a long time...

a patient/carer perspective of
personalised medicine

Jillian Hastings Ward MBE

ICPerMed & EP PerMed Conference

Prague, 26-27 November 2025

From newborn to patient



From patient to rare diagnosis



CureGRIN
Seeking treatments and cures for
GRIA, GRID, GRIK & GRIN Disorder

From diagnosis to... where?

Diagnoses

- patient-led communities
- natural history studies (and registries)
- therapeutic target identification
- trial therapies
- treatments..?



Evolving role of patient advocacy

- Explosion of diagnostic capacity
 - explosion of small support groups
 - explosion of demand for therapies
 - **Upskilling** self and community, to keep pace with technical developments
 - Getting involved in **clinical trial** design and implementation
 - Pushing for **policy** recognition and **regulatory** change
 - But also: coping with **expectation management**, **resource scarcity**, risk of **burnout**...
- Investment in major biobanking / data initiatives
 - rich opportunities for patient voices to contribute, for example:
 - Shared **decision** making or **advisory** roles on **use of data**
 - **Communications** with participants / public
 - And ideally, advisory roles in rollout of genomic **medicine** services...

Thank you

...let's keep going, together!

jillian.hastingsward@gri-uk.org