EURORDIS
RARE DISEASES EUROPE

Patient involvement & engagement in research

17
ORGANISATIONS

136
COUNTRIES

10 PATIENT ORGANISATIONS AS FULL PARTNERS

5 EUROPEAN ORGANISATIONS

7 INTERNATIONAL ORGANISATIONS

7
YEARS

385.5M€
OVERALL BUDGET (EC, MEMBER STATES, INDUSTRY)

25
WORK PACKAGES

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ERDERA started on 1st September 2024 under the EU Research & Innovation funding programme Horizon Europe as a co-funded partnership between the European Commission, European Member States, and beyond.

PPIE, what it is?

Patient and Public Involvement and Engagement: working **with** patients and the public to shape research and engage with it.

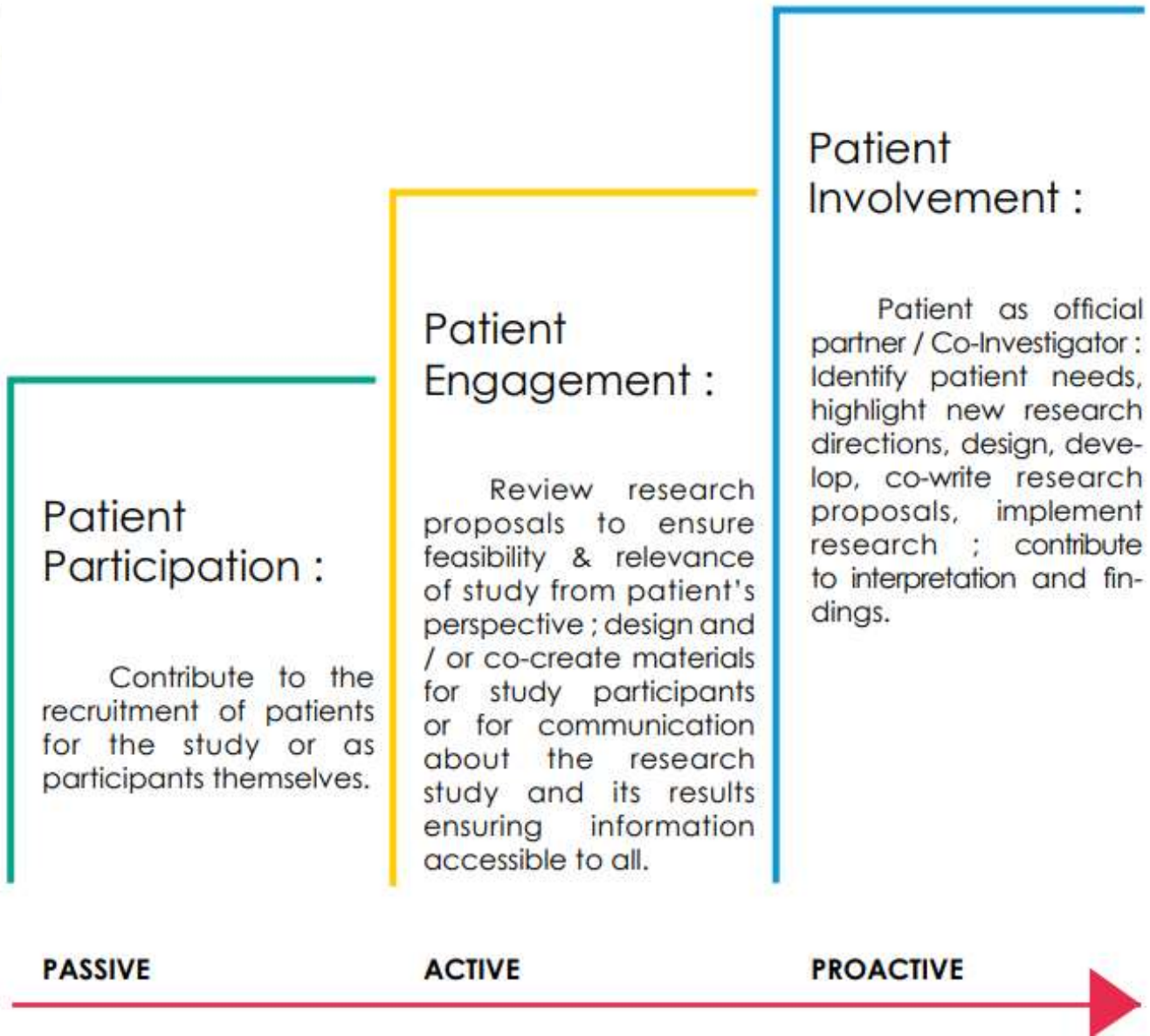
Patient and public involvement and engagement (PPIE) describes the **different ways in which members of the public can inform and shape research.**

"PPIE is different to research participation where members of the public can directly take part in a study, for example by being given a new treatment as part of a clinical trial." – University of Birmingham

*"Patient and public involvement (PPI) entails **research being carried out 'with' or 'by'** members of the public, rather than 'to', 'about' or 'for' them"*- National Institute of Health Research

PPIE is for **everyone.**

A continuum of active and meaningful partnership

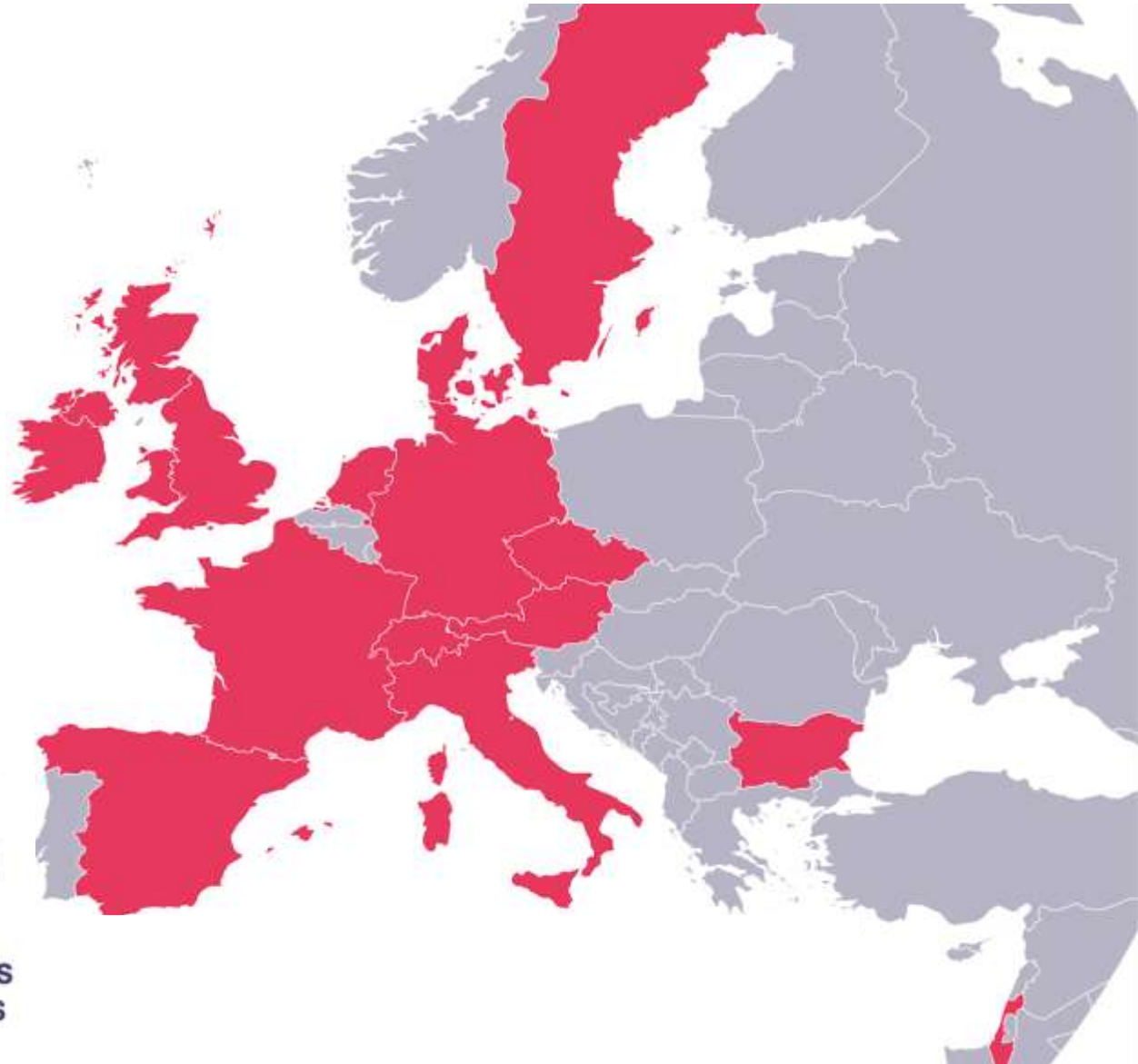


Building evidence on diagnostic needs and expectations

EURORDIS & NBS Research

SCREEN 4CARE

Accelerating Diagnosis for Rare Disease Patients Through Genetic Newborn Screening and Artificial Intelligence



START DATE
1 OCTOBER 2021

DURATION
5 YEARS

BUDGET
25 MIO €

14 COUNTRIES
35 PARTNERS

Rare Barometer Survey on Newborn Screening

CARERS STRONGLY SUPPORT NEWBORN SCREENING

8/10 **Carers** would have liked the person they care for to be diagnosed at birth

Rare on Air podcast with Iuliana Dimitriu:
Her 7-years-long odyssey for her son to have a confirmed diagnosis of Coffin-Lowry syndrome, and how she thinks that early diagnosis could have improved his health and everyday life.

eurordis.org/rare-on-air

Q: If it is or were possible, I would have liked the person I care for to be diagnosed at birth (agree + strongly agree). N=3,002

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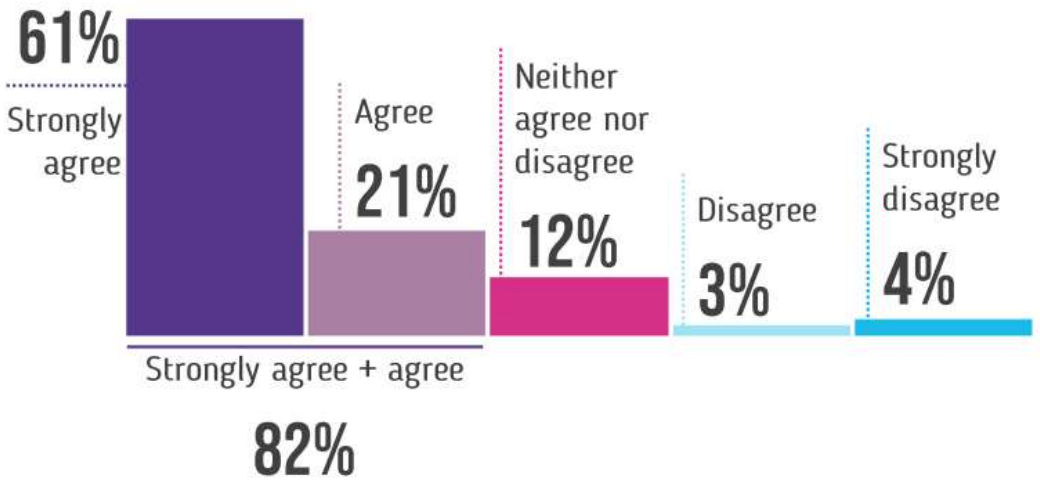
Rare Barometer Survey on Newborn Screening

PARENTS OF PEOPLE LIVING WITH A RARE DISEASE STRONGLY SUPPORT THE DIAGNOSIS OF THEIR CHILD AT BIRTH

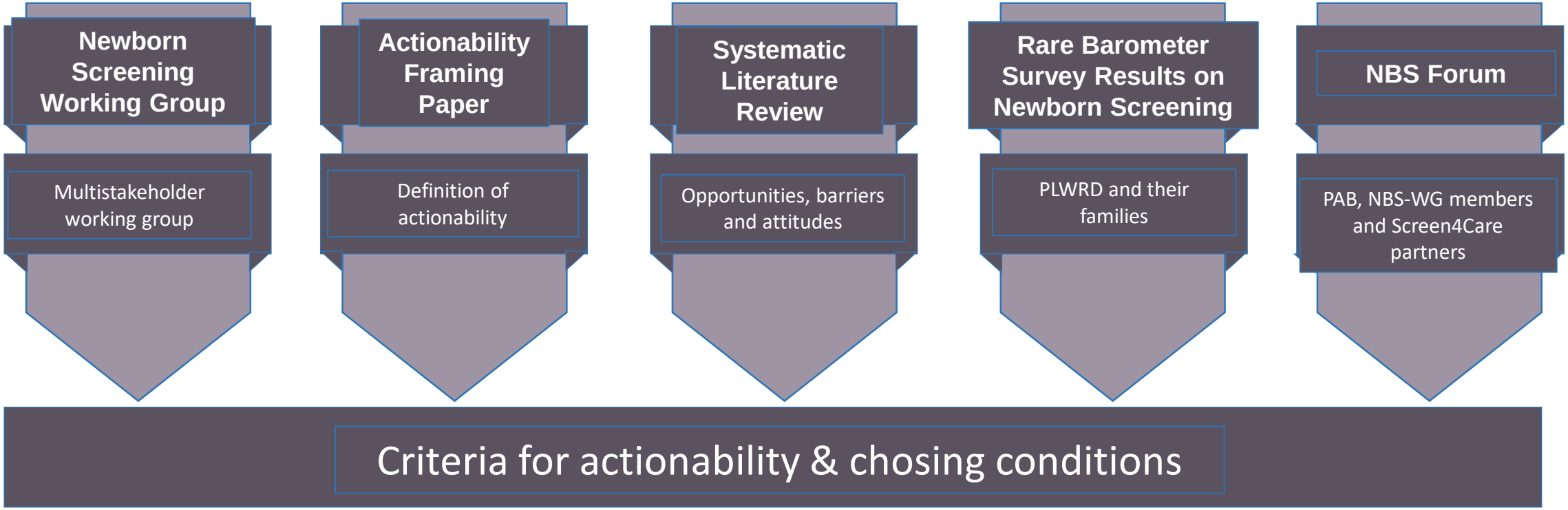
If it is or were possible, I would have liked the person I care for to be diagnosed at birth – *Only parents of people living with a rare disease; n=2,701*



Comparison: parents of people living with a rare disease were also more in favour of newborn screening than adults living with a rare disease in a study on a specific condition[3].

[3] *Boardman et al. (2019). Newborn screening for haemophilia: The views of families and adults living with haemophilia in the UK. Haemophilia.*

Developing a list of actionable condi



Areas of Actionability

Groups of actionable diseases – disease characteristics

- Availability of intervention (actions to prevent, delay or reduce the symptoms of the health condition) such as physiotherapy, symptom control (seizure control), prevention of complications leading to a positive impact on the patient´s quality of life.

Importance for reproductive choices

- Disease clinical characteristics or inheritance modes that would have an impact on reproductive choice decisions, or could lead to a complicated pregnancy management.

Availability of support

- Availability of Centers of expertise, ERNs, Patient organizations or communities, Availability of a specialised healthcare team
- Access to a tailored education plan, rehab, speech therapy or other supportive care

Research and development

- Research in advanced stages or active clinical development on the particular disease

Pharmacogenetic passport

- Would pharmacogenetic testing for a gene be beneficial?

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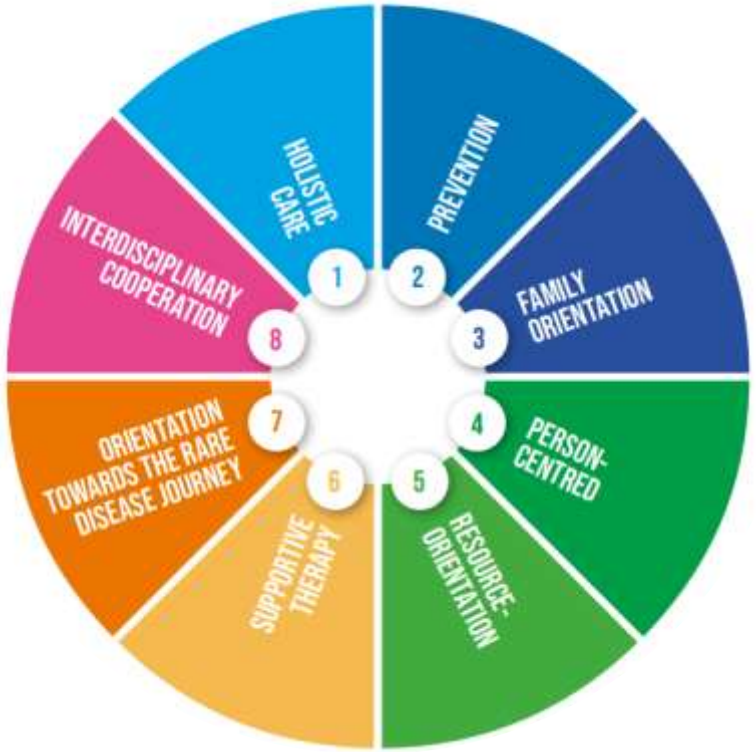
RARE DISEASES EUROPE

Mental Health & Wellbeing

Mental Health & Wellbeing

The rare disease community has identified as an **absolute priority the need to look beyond the physiological aspects of rare conditions** and to take concrete **action to address the psychological impacts** associated with these complex conditions (Rare 2030).

June 2024



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