Using EURORDIS' Position Paper and Rare 2030 Recommendations to advocate for diagnosis, treatment and care

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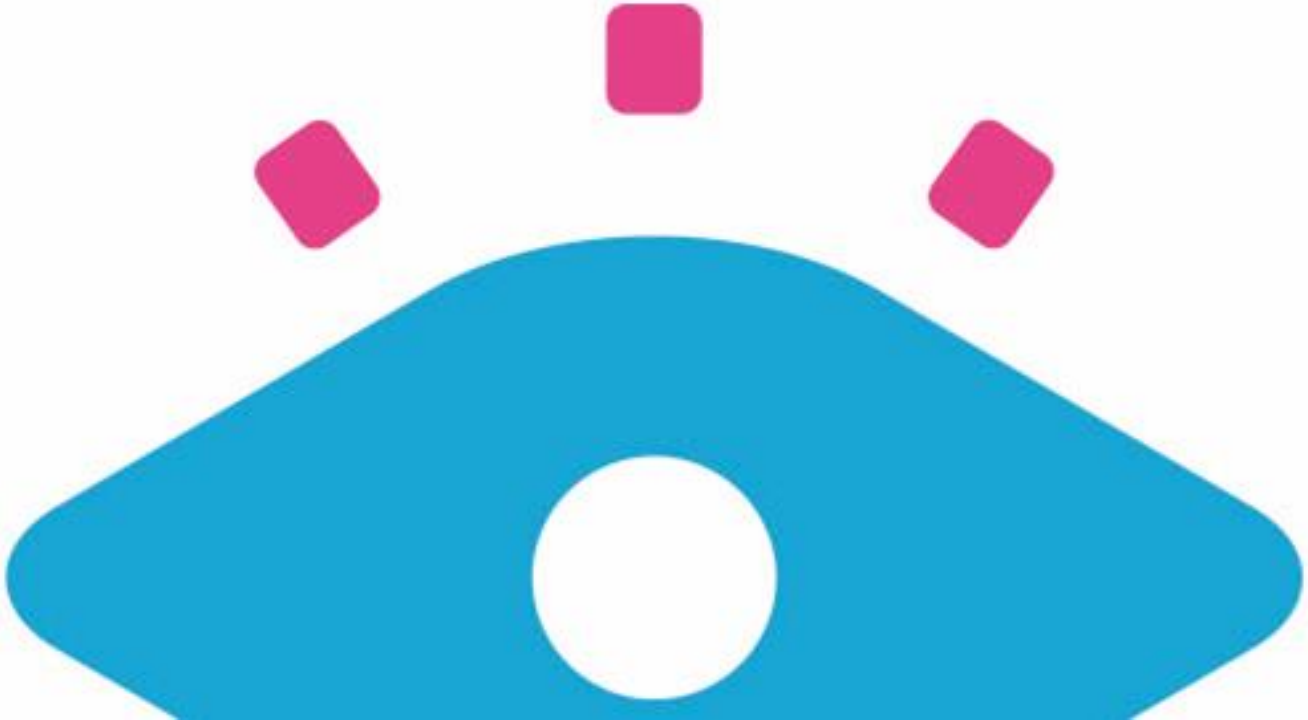
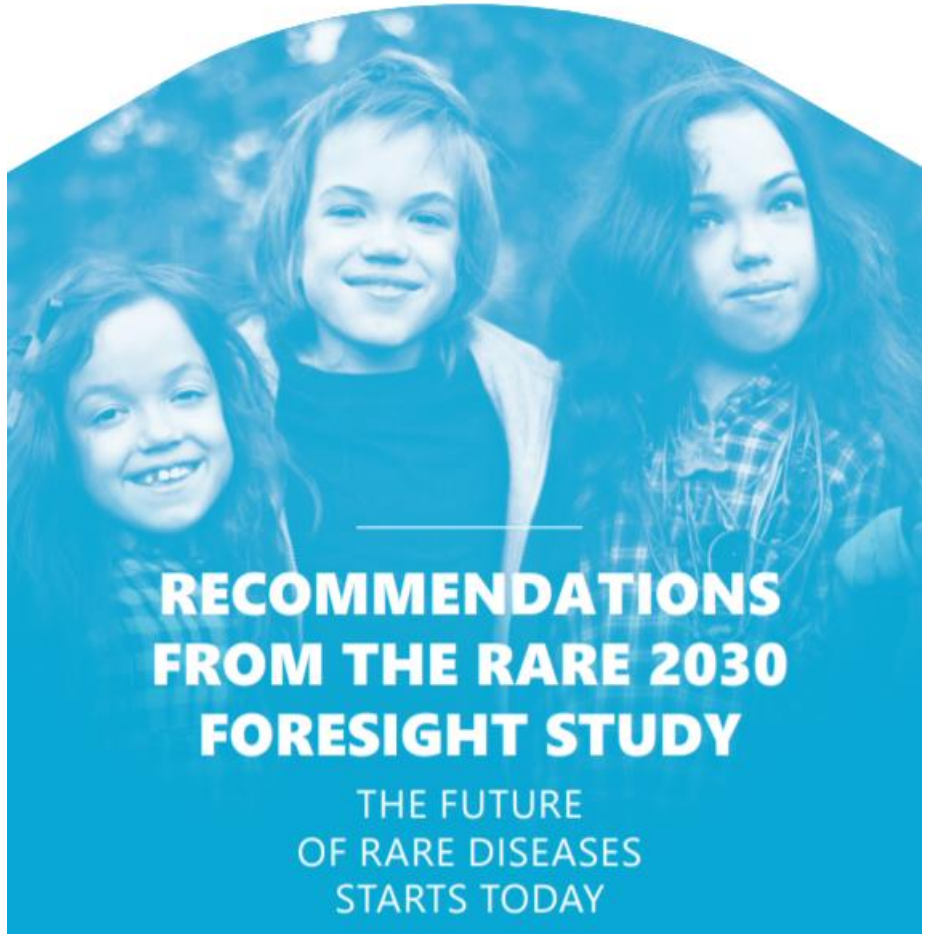
After two years of extensive consultation, the Rare 2030 Foresight study recommends a new policy framework for rare diseases …with a set of policy recommendation to reach the preferred scenario



Rare2030
Foresight in Rare Disease Policy

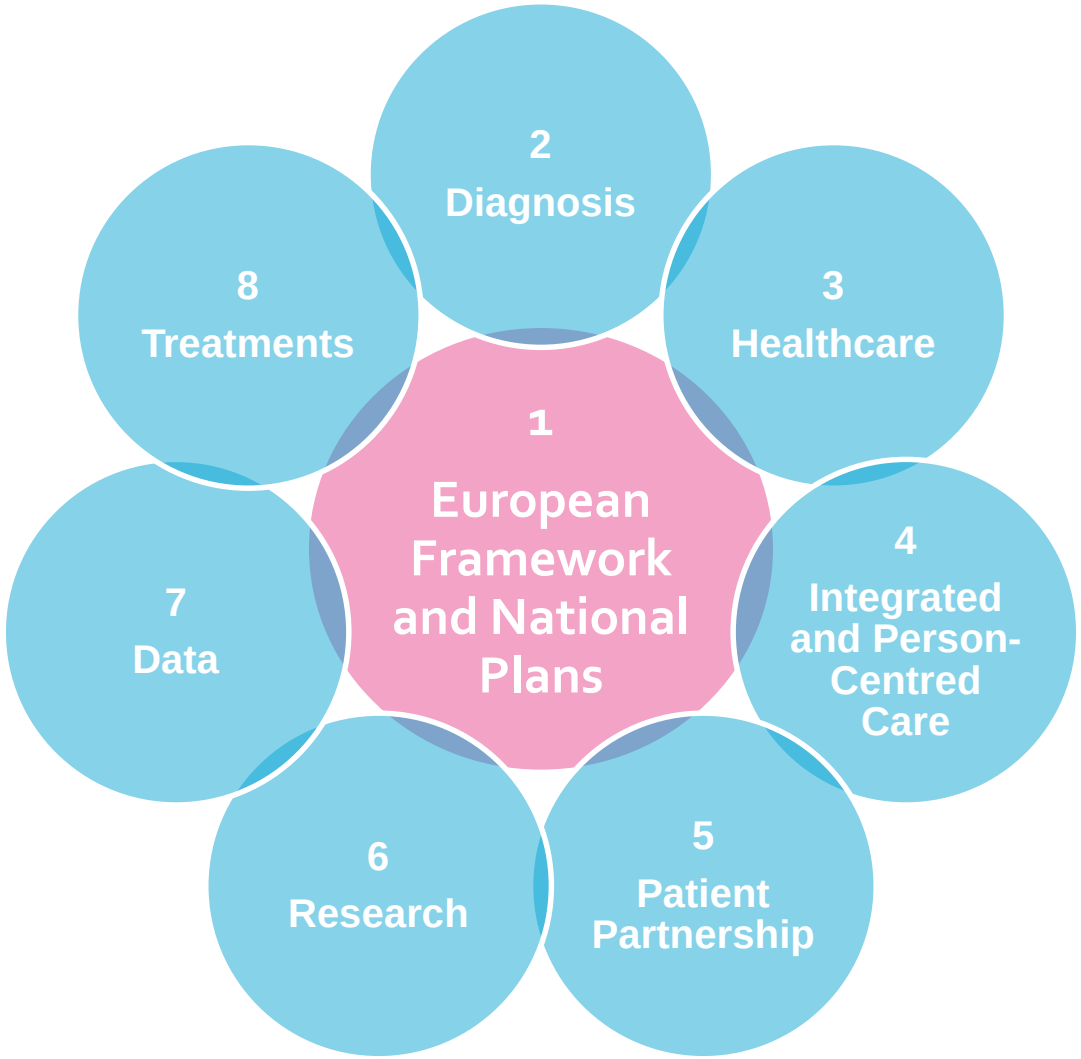


RECOMMENDATIONS FROM THE RARE 2030 FORESIGHT STUDY

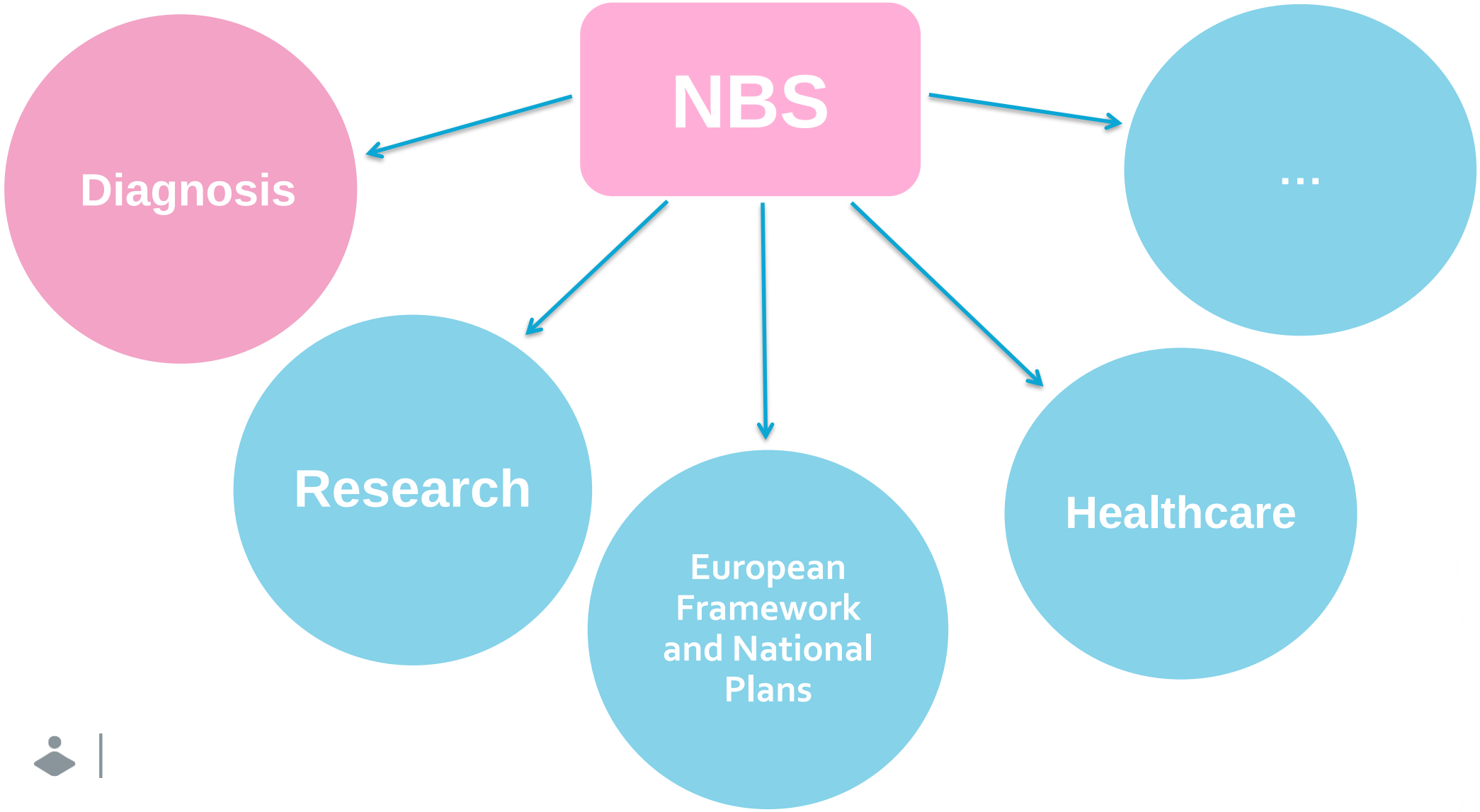
THE FUTURE
OF RARE DISEASES
STARTS TODAY

http://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

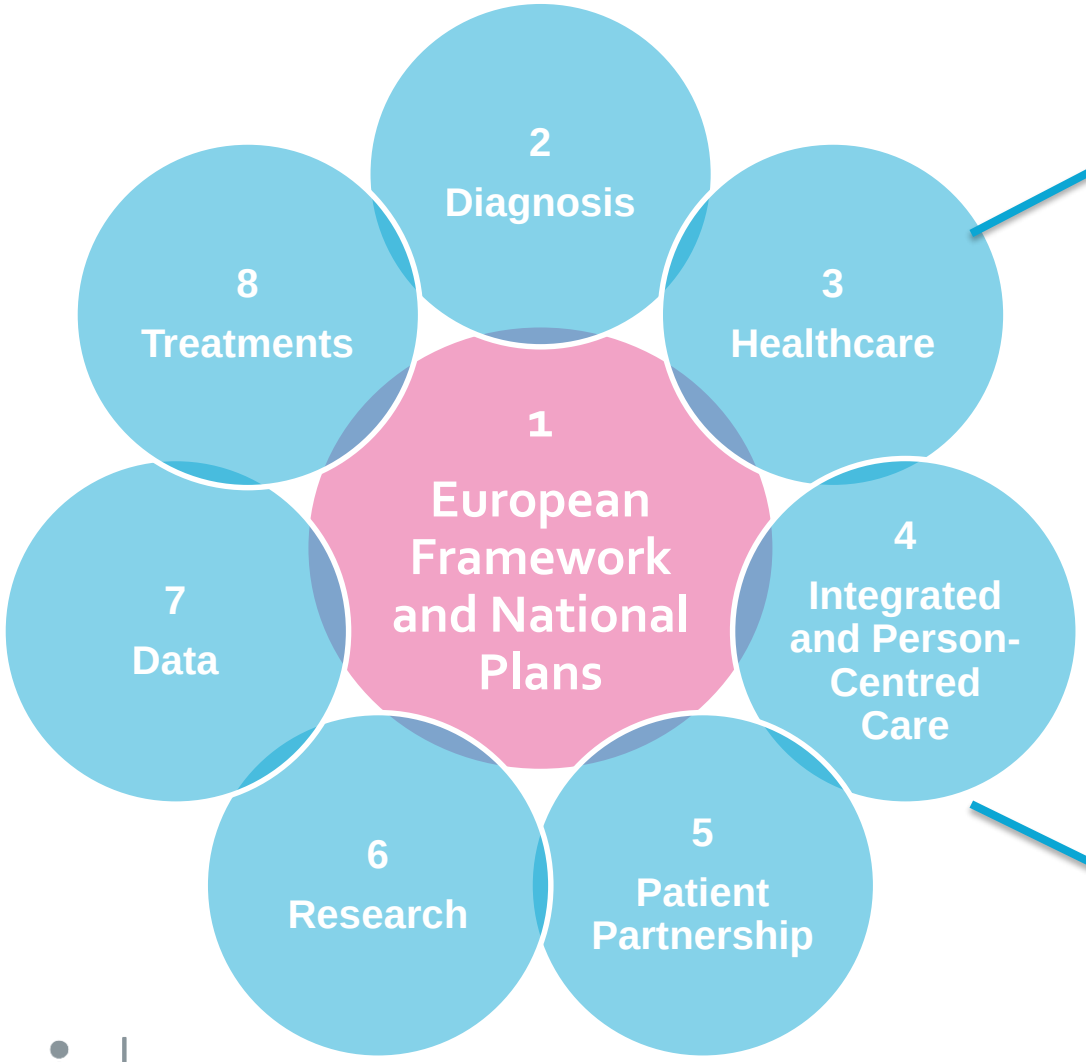
8 Interconnected Recommendations ....



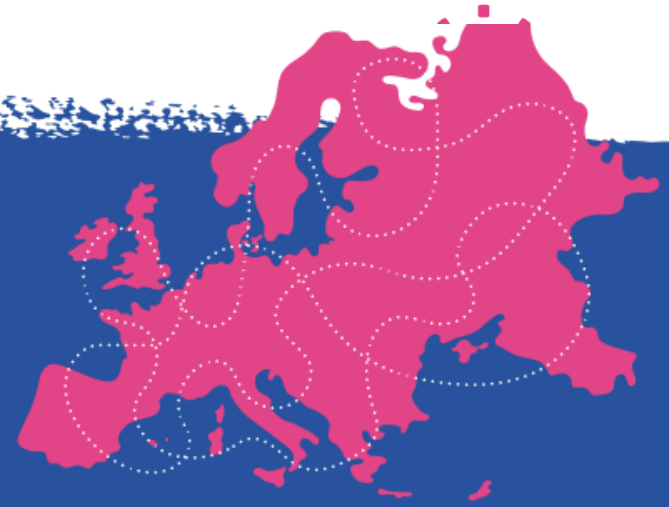
Example of interconnectivity: NBS



…with one overarching Framework



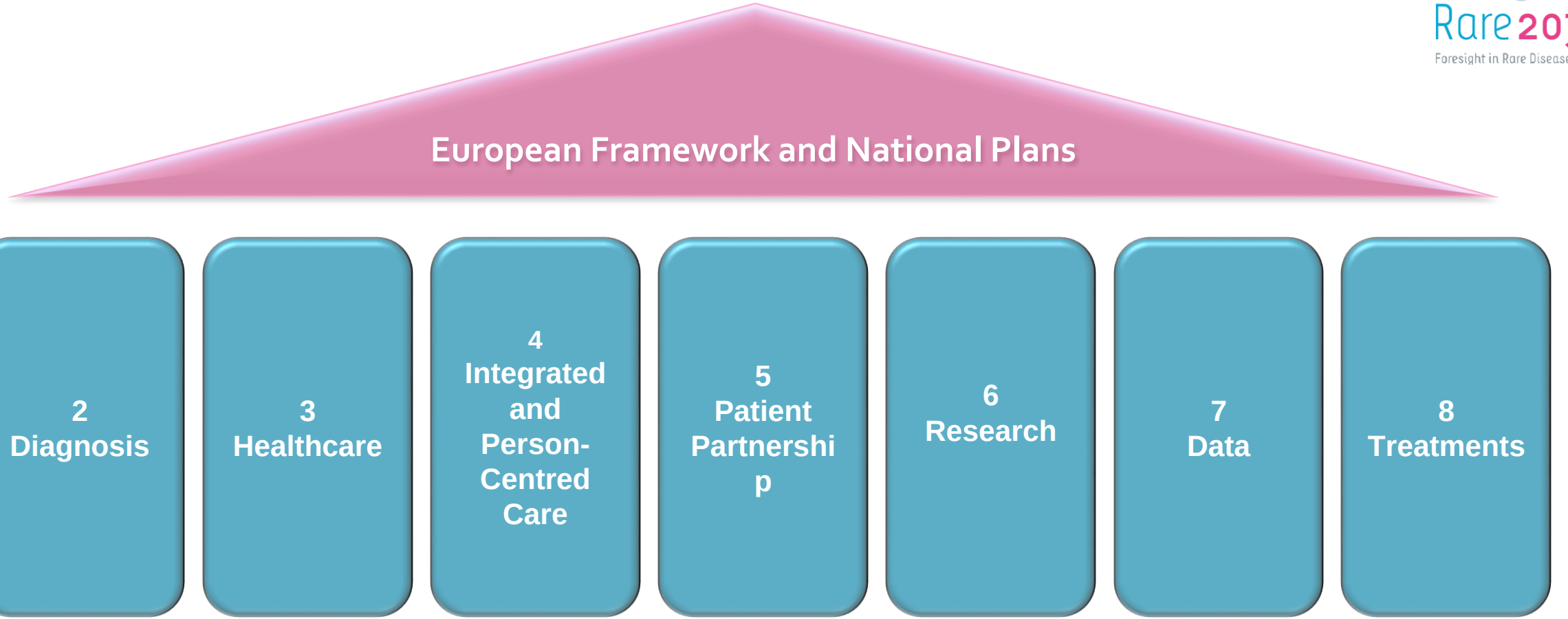
A EUROPEAN ACTION PLAN WILL...



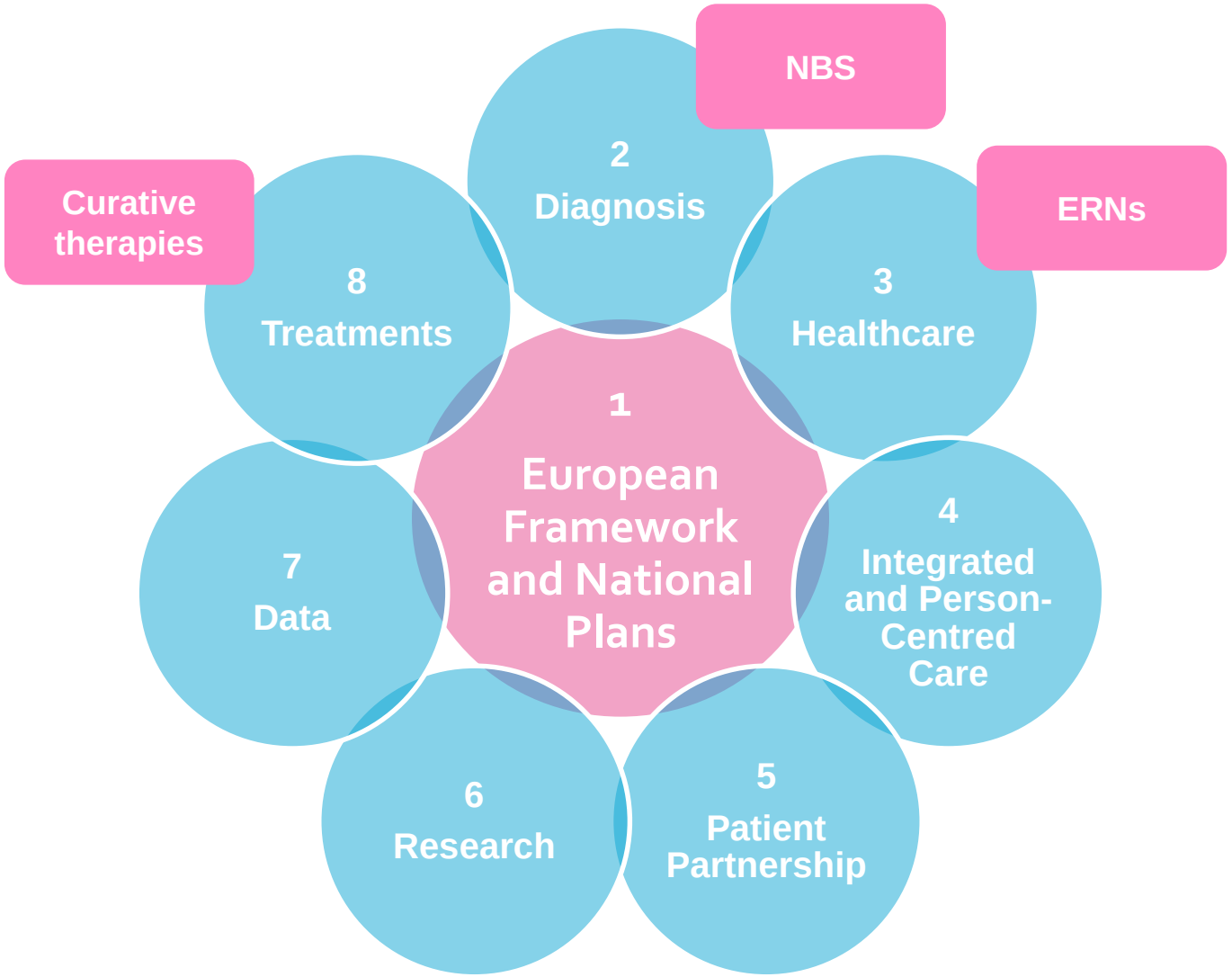
BRING TOGETHER ACTIONS, INCLUDING EXISTING LEGISLATION, ACROSS THE RARE DISEASE PATHWAY WHERE THE EU CAN ADD THE MOST VALUE UNDER ONE FRAMEWORK TO FOSTER RESEARCH AND CARE

A EUROPEAN ACTION PLAN WILL...

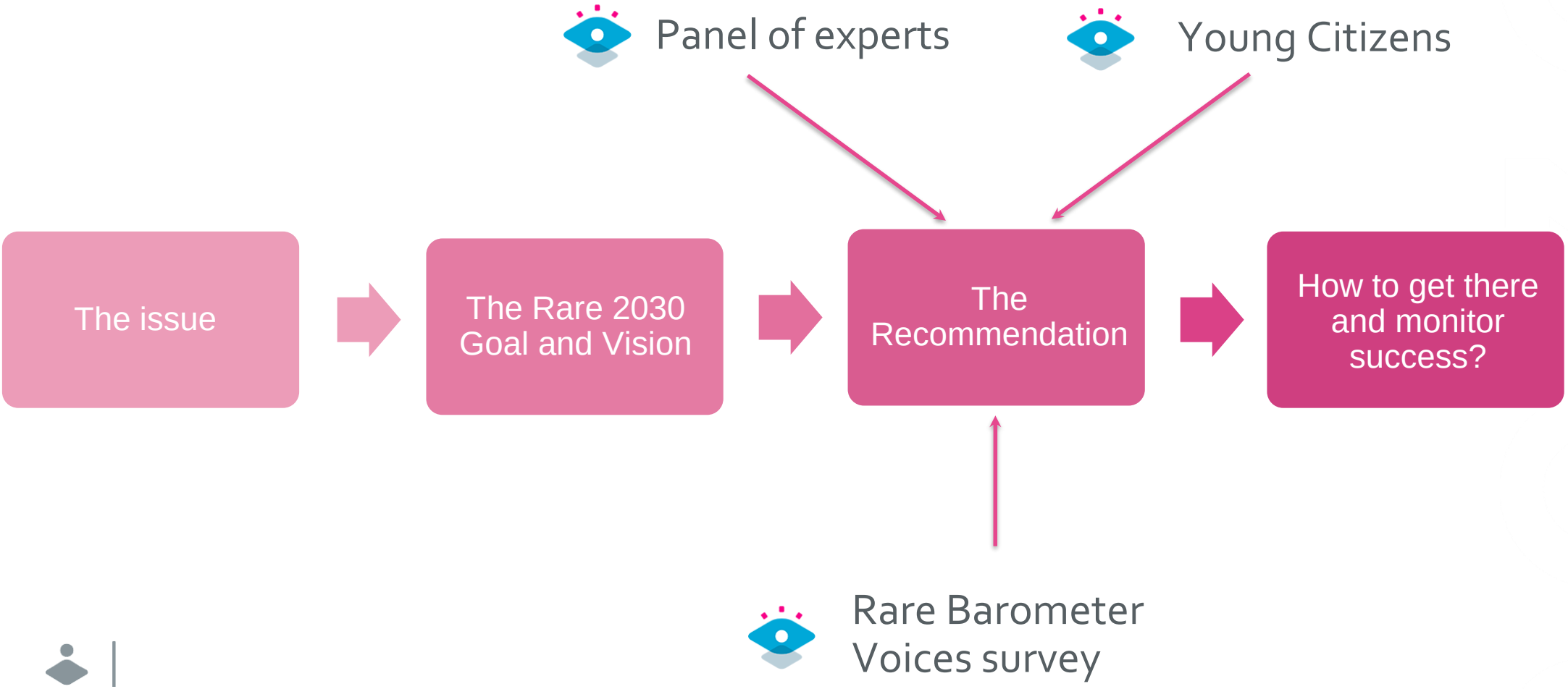
GIVE A NEW FOCUS TO NATIONAL PLANS TO ENSURE THAT SCIENTIFIC, TECHNOLOGICAL AND THERAPEUTIC ADVANCES REACH EVERY PERSON LIVING WITH A RARE DISEASE IN EUROPE


European Framework and National Plans
2 Diagnosis
3 Healthcare
4 Integrated and Person-Centred Care
5 Patient Partnership
6 Research
7 Data
8 Treatments

8 Interconnected Recommendations ....



Structure of Recommendations



RARE 2030 **GOAL** 2

All people living with a rare disease known in the medical literature will be diagnosed within six months of coming to m

All people will have access to diagnostic technologies, b programmes (including sc discrimination and regardless in Europe. All currently undiag will enter a European and gl diagnostic and resear

The search for an accurate diagnosis very often remains a diagnostic 'odyssey', for many reasons; for instance, the sheer number of conditions under the heading of 'rare diseases'; the scarcity (by definition) of patients with any single condition and the corresponding scarcity of experts acquainted with each condition; the tendency for rare diseases to manifest as complex, multisystemic conditions; the fact that existing te or implem of a diagn diagnosis) quences fo terogeneit genetic tes implement impact acc qualities f Europe.

VISION OF THE RARE DISEASE COMMU

The rare community recognises that diagnosis is the first step towards any improvements in health and well-being and envisions a future where families across Europe no longer have to endure a diagnostic odyssey to obtain this critical information. Whether it be access to a treatment or the ability to plan for their future, by 2030 all people living with a rare disease will receive the attention they need as quickly as possible - and this begins with a timely and accurate diagnosis. The community strongly supports the better application of current diagnostic testing and screening approaches (which includes low-cost but highly effective solutions such as raising awareness) as well as the use of new technologies to overcome current barriers in diagnosis equally across all countries.

RARE 2030 **RECOMMENDATION** 2

EARLIER ACCUR

The time to diagnosis should be shortened - whilst avoidin consequences - which should be achieved by better use and available diagnostic testing technologies, best practices and innovative approaches must be driven by patient-needs and qualities in access to diagnosis and ensuing care must be en standards and programmes across Europe (and beyond). A p with undiagnosed rare diseases, which demands greater and data-sharing and diagnostic platforms and infrastructures.

Specifically, to achieve earlier, faster, and more accurate diag supported:

- Promote equality in access to diagnostic opportunit sease or suspected rare disease, no matter where the
- Enable patients to navigate health systems with ease, direct route to obtaining a diagnosis, to connect wi learn how to best manage their disease and particip manner.
- Co-design - with healthcare professionals and pati ciently guide people living with a rare disease from di where possible and appropriate, to the most relevant
- Improve diagnostic expertise by fostering Europea specialised healthcare providers and by ensuring greater interoperability and standardisation of data able to support diagnostics whilst ensuring particular transnational collaboration to diagnose the most complex presentations, rarest diseases and undiagnosed cases.
- Ensure an integrated, international approach to patients with currently undiagnosable conditions, ensuring the absence of diagnosis does not preclude access to the best possible care and support
- Raise awareness on rare diseases in medical curricula and amongst all primary/ front-line health and social care professionals and specialists
- Foster broad and equitable implementation of next generation sequencing and other emerging new technologies into national healthcare systems, to facilitate and speed up access to diagnosis.

HOW TO ACHIEVE THIS?

BETTER USE AND ACCESSIBILITY OF EXISTING SOLUTIONS, WITHIN A MORE STRATEGIC AND COORDINATED DIAGNOSTICS ECOSYSTEM

Obtaining a timely and accurate diagnosis is a human right, whether there is an available medical treatment or not. The following steps should be pursued to better apply existing tools, best practices and programmes

AT THE EUROPEAN AND GLOBAL LEVELS

- A clear, systematic and European-wide (indeed sometimes global) approach to rare disease diagnostics must be ensured, founded upon the ability to guide patients towards centres of expertise or equivalent, access transnational diagnostics platforms, and capture - and systematically manage - data on patients for whom a diagnosis is not forthcoming
- Continued support must be ensured for multinational and multistakeholder research linking omics data, clinical data and biomaterials with well-defined patient cohorts and applying them in the clinic, building on the work of existing initiatives such as the European Joint Programme on Rare Diseases and Solve-RD
- Existing and future best practice guidelines to support the diagnosis of rare diseases (such as decision trees and patient pathways) should be visible and findable at the European level (via European Reference Networks ERNs and Orphanet) and should be adopted and implemented to a greater degree at the national level
- The Orphanet services pertaining to diagnostics (resources concerning the definition and inventorying of diseases, and the database on expert clinical centres and laboratories) should increasingly be co-created and co-curated together with ERNs, and should be sustained by European action
- Funding bodies in Europe and all other world regions should target diagnostics for subpopulations, indigenous people, and other culturally and linguistically diverse populations in a culturally safe manner (including populations in developing nations): this will support the genetic and phenotypic characterisation of rare disease populations to enlarge patient cohorts and advance knowledge and understanding.
- Appropriate and targeted funding should be dedicated at EU and national levels to foster research into aetiology of rare diseases with no evident underlying genetic causes
- Research should be fostered at European level to elucidate the determinants of the heterogeneity across EU Member States in terms of diagnostic performance
- Research should be fostered at the European level (inline with the Commission Expert Group on Rare Disease Recommendations on Cross-Border Genetic Testing) to conduct a cross-border health economics assessment of diagnostic and screening technologies, comparing costs and benefits relative to those currently incurred under the diagnostic 'odyssey'

Recommendation 2

Earlier, Faster and more Accurate Diagnosis

What do we recommend?

THE ISSUE

Continuous delays in diagnosis (Odyssey)

GOAL AND VISION

Diagnosis within 6 months

RECOMMENDATION

Earlier, Faster and more Accurate Diagnosis

A better use of **currently effective and available tools, new technologies and innovative approaches** driven by patient-needs. **Inequalities** in access to diagnosis and care must be **eradicated** through the **harmonisation of standards**

Recommendation 3

Access to High Quality Care

What do we recommend?

Rare 2030
Foresight in Rare Disease Policy

THE ISSUE

Lacking and scattered expertise and care

GOAL AND VISION

Right to health care, leaving no one behind

RECOMMENDATION

Ecosystem for access to high quality care

Provision **of political, financial, operational and technical support** at European, national and regional levels to establish a highly specialised healthcare ecosystem that leaves no one behind

Recommendation 8

Available, Accessible and Affordable Treatments

What do we recommend?

THE ISSUE

Significant unmet needs

GOAL AND VISION

1,000 new therapies, 3 to 5 times more affordable

RECOMMENDATION

Available, Accessible and Affordable Treatments

Establishing **streamlined regulatory, pricing and reimbursement policies**, encouraging a continuum of evidence generation and a European ecosystem that **attracts investments, fosters innovation and addresses challenges of sustainability**

From the Recommendations Rare 2030 to your Advocacy Work

Recommendations Rare 2030

Rare2030 material

Position Papers

RareBarometer studies

Tell and share a story

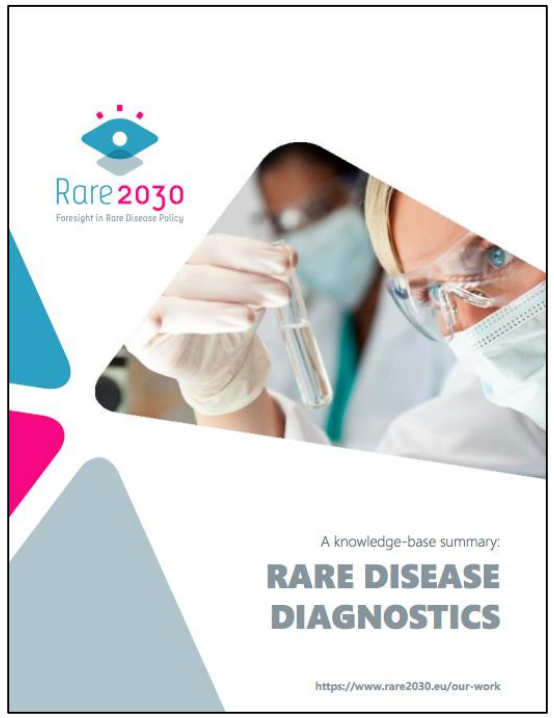
Get the level right (EU/ national)

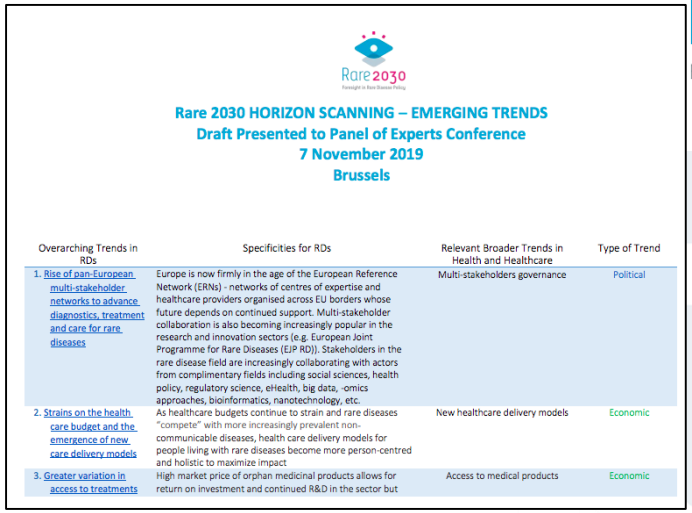
Co-create

EURORDIS.ORG

EURORDIS RARE DISEASES EUROPE

Advocacy Work – Available Tools and Materials




Rare 2030

Rare 2030 HORIZON SCANNING – EMERGING TRENDS
Draft Presented to Panel of Experts Conference
7 November 2019
Brussels

Overarching Trends in RDs	Specificities for RDs	Relevant Broader Trends in Health and Healthcare	Type of Trend
1. Rise of pan-European multi-stakeholder networks to advance diagnostics, treatment and care for rare diseases	Europe is now firmly in the age of the European Reference Network (ERNs) - networks of centres of expertise and healthcare providers organised across EU borders whose future depends on continued support. Multi-stakeholder collaboration is also becoming increasingly popular in the research and innovation sectors (e.g. European Joint Programme for Rare Diseases (EJP RD)). Stakeholders in the rare disease field are increasingly collaborating with actors from complimentary fields including social sciences, health policy, regulatory science, eHealth, big data, -omics approaches, bioinformatics, nanotechnology, etc.	Multi-stakeholders governance	Political
2. Strains on the health care budget and the emergence of new care delivery models	As healthcare budgets continue to strain and rare diseases "compete" with more increasingly prevalent non-communicable diseases, health care delivery models for people living with rare diseases become more person-centred and holistic to maximize impact	New healthcare delivery models	Economic
3. Greater variation in access to treatments	High market price of orphan medicinal products allows for return on investment and continued R&D in the sector but	Access to medical products	Economic

Rare 2030 Report

8 Knowledge Base Summaries

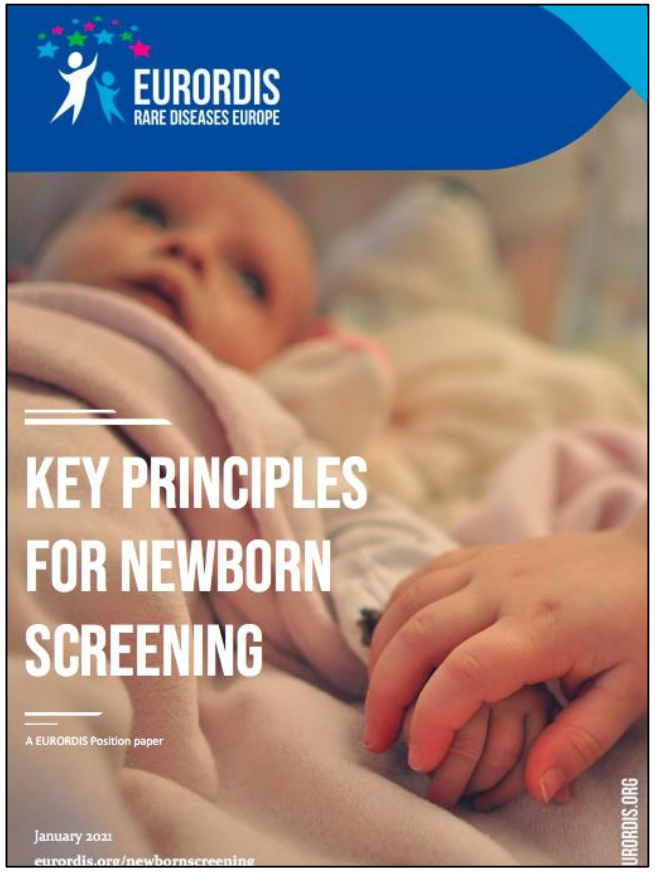
Topic-specific documents

Overview of knowledge

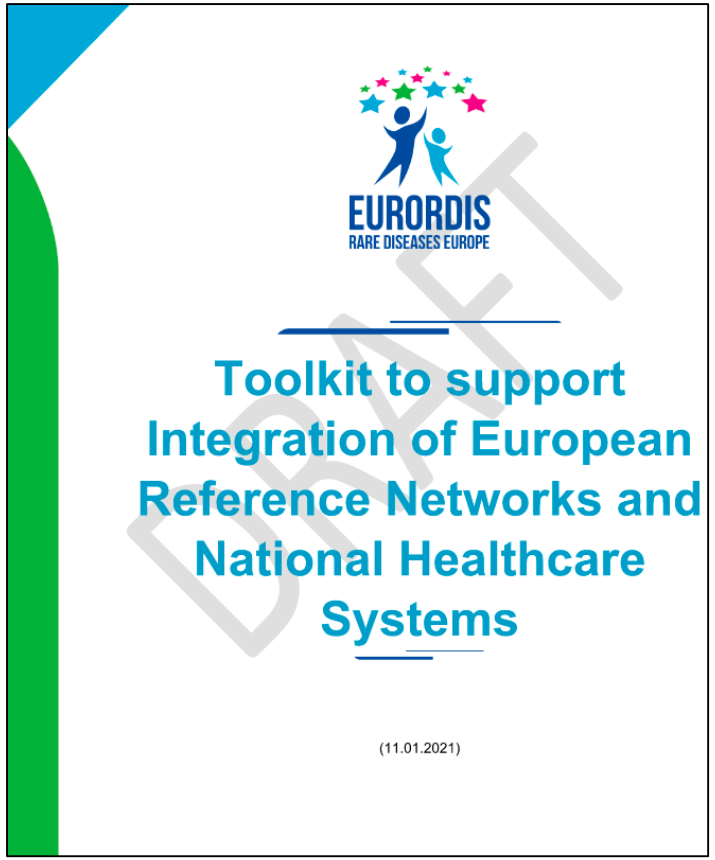
Trends

Overview of current trends in rare diseases

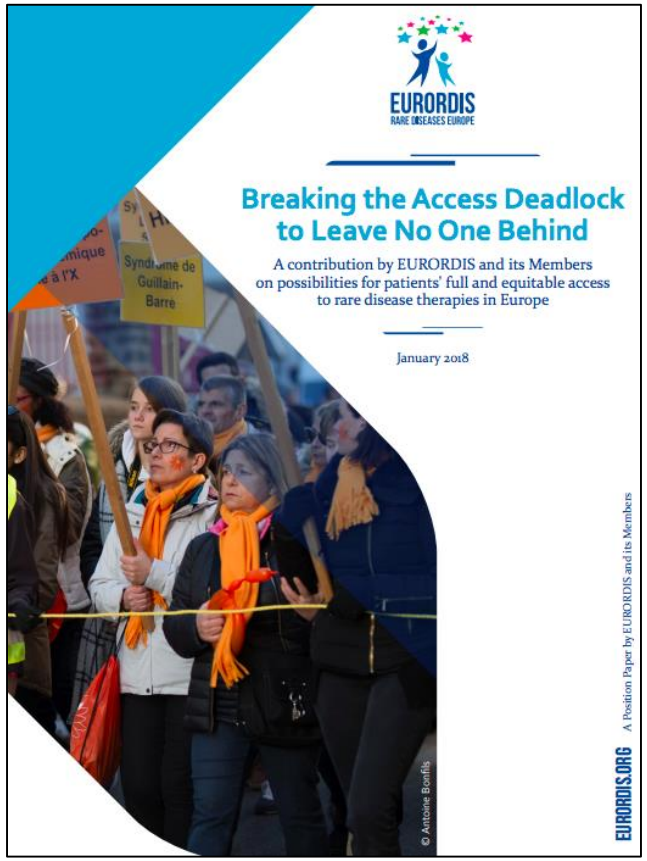
Advocacy Work – Available Tools and Materials



Position Paper



Advocacy Work – Available Tools and Materials



Position Paper

Report

Advocacy Work – Available Tools and Materials

Rare Barometer

https://www.eurordis.org/voices

- H-CARE Survey full results
- H-CARE Survey condensed results
- Your experience of COVID-19-Dashboard
- Your experience of COVID-19-Infographics
- Share and protect our health data! Full report
- Share and protect our health data!
- Survey results: Access to treatment
- Survey results: Juggling care and daily life
- Survey results: Rare disease patients' paticipation in research
- Results infographic: Rare disease patients' paticipation in research

Rare2030 RareBarometer Voices survey

ACCESS TO MORE EFFECTIVE DIAGNOSIS STRATEGIES: NEW-BORN SCREENINGS

94% of respondents support the diagnosis of rare conditions at child's birth, pushing for implementing new-born screening across Europe.

People living with a rare disease also favour the use of new technologies to diagnose rare conditions before birth: either during pregnancy (87%) and around the time of conception (80%).

TREATMENTS NEED TO BE MORE AVAILABLE, ACCESSIBLE AND AFFORDABLE FOR PEOPLE WITH RARE DISEASES

22% of people with rare diseases could not get, in 2019, the treatments they needed because it was not available where they live.

14% of people with rare diseases could not get, in 2019, the treatments they needed because the waiting list was too long.

12% of people with rare diseases could not get, in 2019, the treatments they needed because they could not pay for it.

Advocacy Work – Your experience and stories

"We tell our stories. You don't have to be an expert, you just have to be on board"

EURORDIS.ORG

EURORDIS RARE DISEASES EUROPE

Advocacy Work – getting it right

The right level

Global ?

Local ?

The right role for everyone

- ✓ Patient community
- ✓ Healthcare providers
- ✓ Researchers & scientists
- ✓ Health authorities & payers
- ✓ Regulators
- ✓ Policymakers
- ✓ Industry
- ✓ xxx

Key Messages

- Interconnected recommendations = a comprehensive tool
- Materials and experience **enable us** to do things in areas **where we want to see change**
- « A long road and we must work in concert and collaboration »
- Co-creation starts today
- Think about what you/your organisation can do

→ **2 things you/your organisation could do in the next 2 weeks**