WHY EUROPE'S ACTION PLAN FOR RARE DISEASES IS CRITICAL FOR OUR COMMUNITY

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Overview

- Why is it needed?
- Why now?
- How can this be achieved?
- What could this look like?

Why is a European Action Plan on Rare Diseases critical for our community?

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RARE DISEASES EUROPE

Rare diseases

All together, an estimated

30

MILLION PEOPLE

are living with a rare disease in

48

COUNTRIES

in Europe

Each one affects fewer than

1 IN

2000

PEOPLE

Affects

4%

of the population in the course of the lives (3,5% to 5,9%)

OVER

6000

distinct rare diseases

Onset of

70%

of rare diseases is in

CHILDHOOD

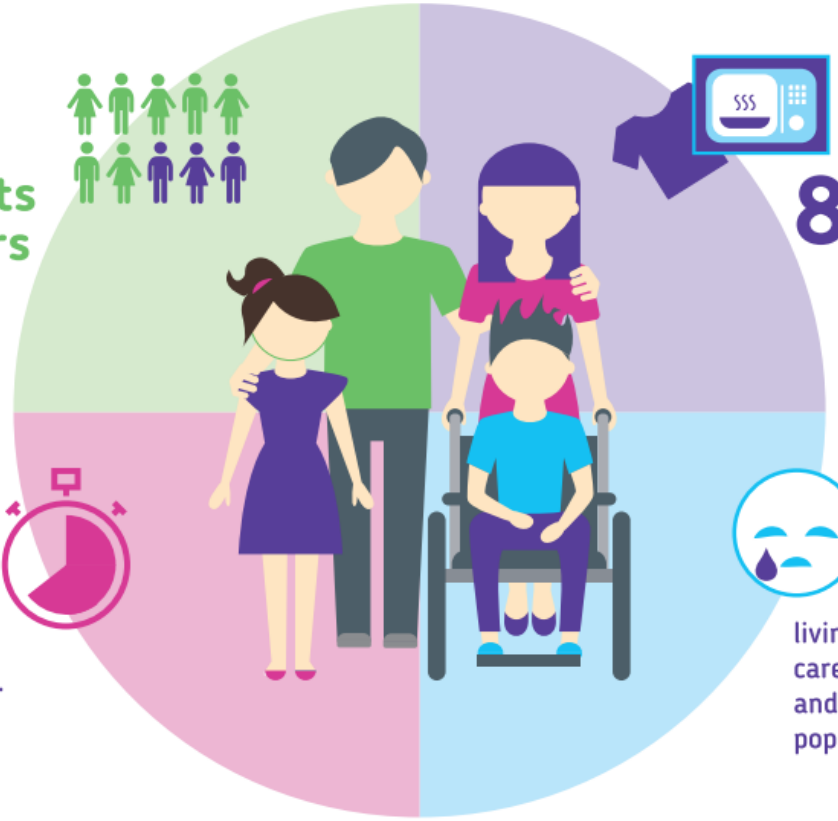
72%

of rare diseases are genetic

28%

are non-genetic, including rare cancers, rare infections, and health hazards

Rare diseases seriously impact everyday life



7 in 10 patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.

8 in 10 patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)

2/3 of carers

spend more than 2 hours a day on disease-related tasks.

3 times more people

living with a rare disease and carers report being unhappy and depressed than the general population*

* Rare Barometer Voices sample compared to International Social Survey Programme, 2011

Vast unmet needs

- Still, long road to diagnosis
  - 5 years to diagnosis
- Still, lack of treatment options… or access to treatments when they are available
  - Only 6% of rare diseases have a treatment
  - 22% of people with rare diseases could not get, in 2019, the treatments they needed because it was not available where they live.
- Lack of coordination between health and social care
- Still stigmatised in society, poorly integrated in society, far from optimal well-being

Why is European action needed?

"To create support centers in Greece equipped with the whole team that every rare disease needs, so that no time is spent looking for the right scientist for our children. Personally, it took me 13 years to find specialists to give a quality of life to my child."

— Magda, Greece

"I have a progressive genetic disease. My wish is to improve the treatment options and care, especially the emotional stability and self-esteem of those affected, even if the disease cannot be cured."

— Maria, Austria

"No treatment specialists, no government help. We don't have financial aid. It shouldn't be like that. Why are we required to be treated at work as healthy when we are disabled? No understanding, no empathy."

— Natalia, Poland

Progress has been made thanks to…

- Empowered patient communities
- Legislation to incentivise investment in rare disease research
- A European mandate to Member States
- National Plans and Strategies setting national priorities
- Disease registries
- Centres of Expertise and European Reference Networks
- European research programmes

2000 OMP Regulation
2006 Paediatric Use Regulation
2007 Advanced Therapies Regulation

2008 Council Recommendation
2009 Commission Communication

Cross border healthcare Directive

Why now?

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New challenges… and new opportunities

- Science & Knowledge
- Diagnostic tools
- Advanced therapies
- Data & Digital transformation
- Social and Economic impact

The rare disease story in Europe so far

Rare 2030 Foresight Study

The main recommendation called for a renewed European policy framework on rare diseases by 2023, to connect the dots between different policy areas and different countries in Europe

2000
EU Regulation on Orphan Medicinal Products

incentivised investment in rare disease research

2006
EU Regulation on Paediatric Use of Medicines

2007
EU Regulation on Advanced Therapies Regulation

2008
The first Rare Disease Day

global solidarity for rare diseases

2008–2009
European Commission's Communication and EU Council Recommendation on rare diseases

rare diseases recognised as a public health priority; Member States recommended to introduce a national plan on rare diseases

2011
EU Cross-Border Healthcare Directive

European Reference Networks have a foundation

2017
Launch of 24 European Reference Networks

2019
European Court of Auditors report on EU actions for cross-border healthcare

calling for a review of rare disease strategies

Feb 2021
Rare 2030 Foresight Study Recommendations published

Dec 2021
UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families

End of a policy cycle

- 14 years since the last "rare disease strategy" – actions from 2008/2009 legislations complete or evolved (Commission Communication 2008 and Council Recommendation 2009)
  - No longer the existence of an EU expert group
  - Only 11 active National Plans and Strategies currently
- Different areas of legislation under review or development (OMP, Pharma Pack, CBHC, EHDS, Cancer Plan, Mental Plan)

A call from across the community

- The 2000 rare diseases patient organisations across Europe, all national alliances, all European federations
- All stakeholders: patients & families, clinicians (eg ERNs), researchers, pharma & biotech, national policy makers
- European Court of Auditors' Report
- European Parliament debate, 45 MEPs call upon the Commissioner for Health, mention in resolutions on building back better
- 50+ partners of the European Conference on Rare Diseases July 2022
- European Economic and Social Committee opinion on rare diseases
- Member State support through the Slovenian, French and Czech EU Council Presidencies

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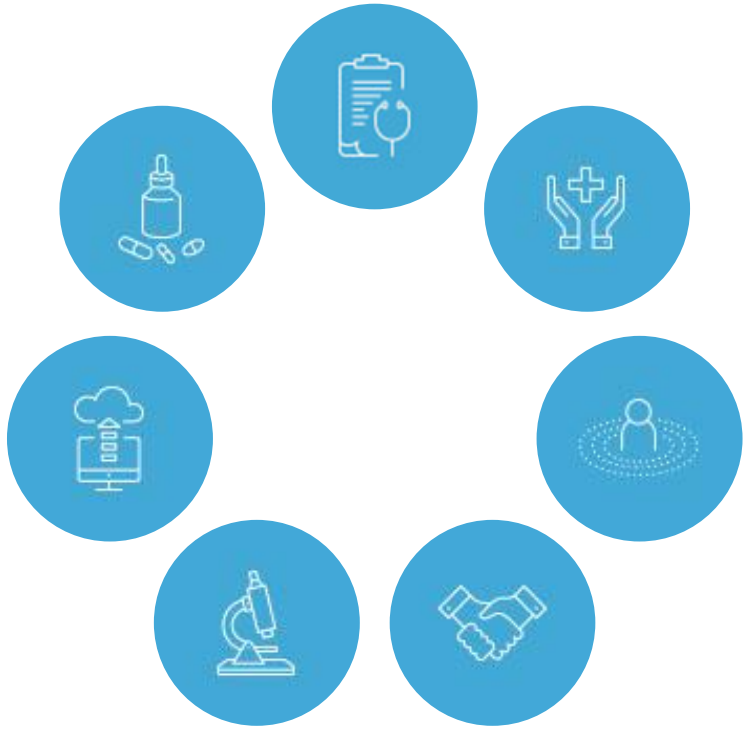
How should this be done?

A comprehensive, integrated, goals based framework

- Bridging the gaps between different legislative pieces on data, research, treatment, healthcare, social care
- Bridging the gaps between national and EU initiatives
- A holistic and life-long view
- Measurable goals to set a common direction
- Upstream clinical research… using data…connecting with patient advocates… linking downstream with access to treatments… at specialist centres



Goal based strategy

8 Rare 2030 Recommendations

1. European/national plans and strategies
2. Diagnosis
3. Access to care
4. Person-centred care
5. Patient Partnerships
6. Research
7. Data
8. Treatment

Europe's Action Plan

GOAL 1: Ensuring healthy lives and promoting well-being

GOAL 2: Reducing inequalities

GOAL 3: Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation

4 SDGs

SDG3: Ensure healthy lives and promote well-being for all at all ages

SDG 9: Build resilient infrastructure, promote inclusive and sustainable industrialisation and foster innovation

SDG10: Reduce inequalities within/among countries

SDG17: Revitalise the global partnership for sustainable development

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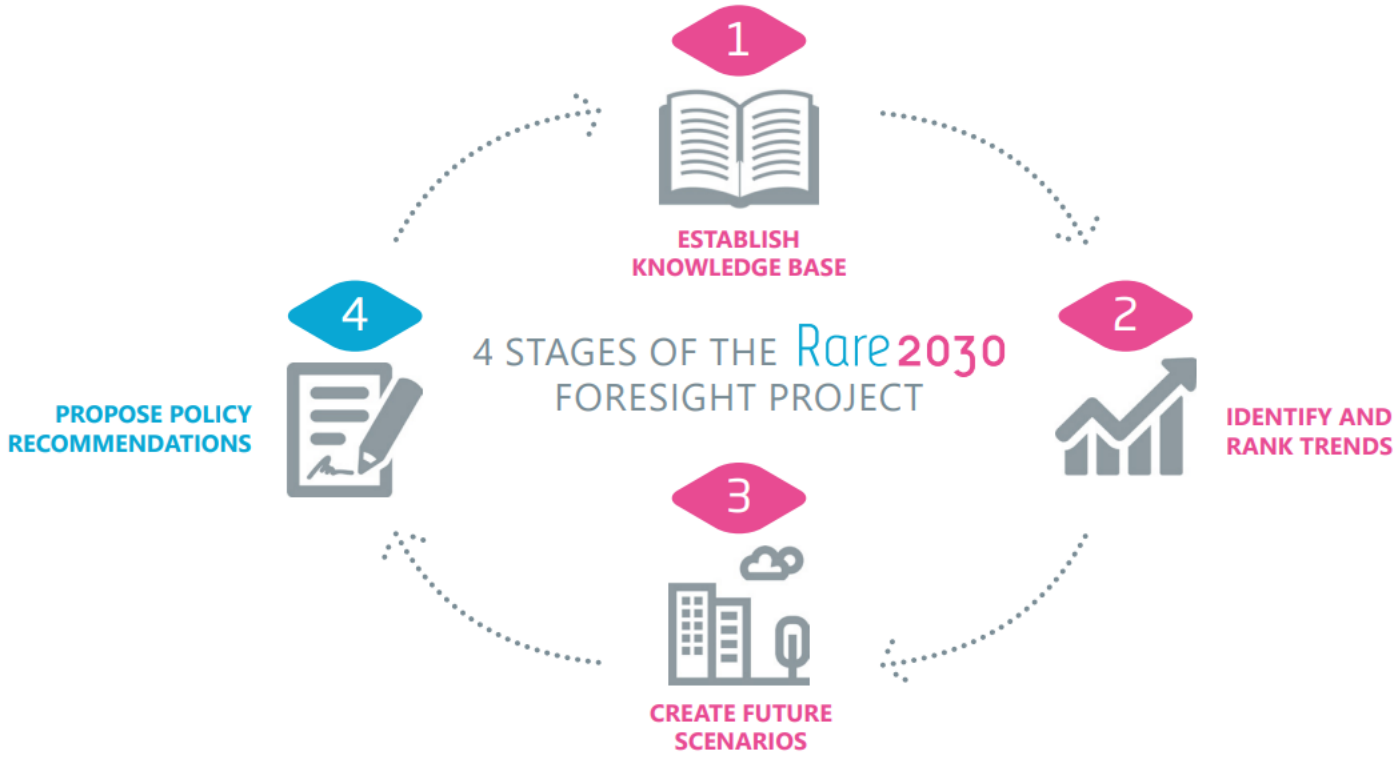
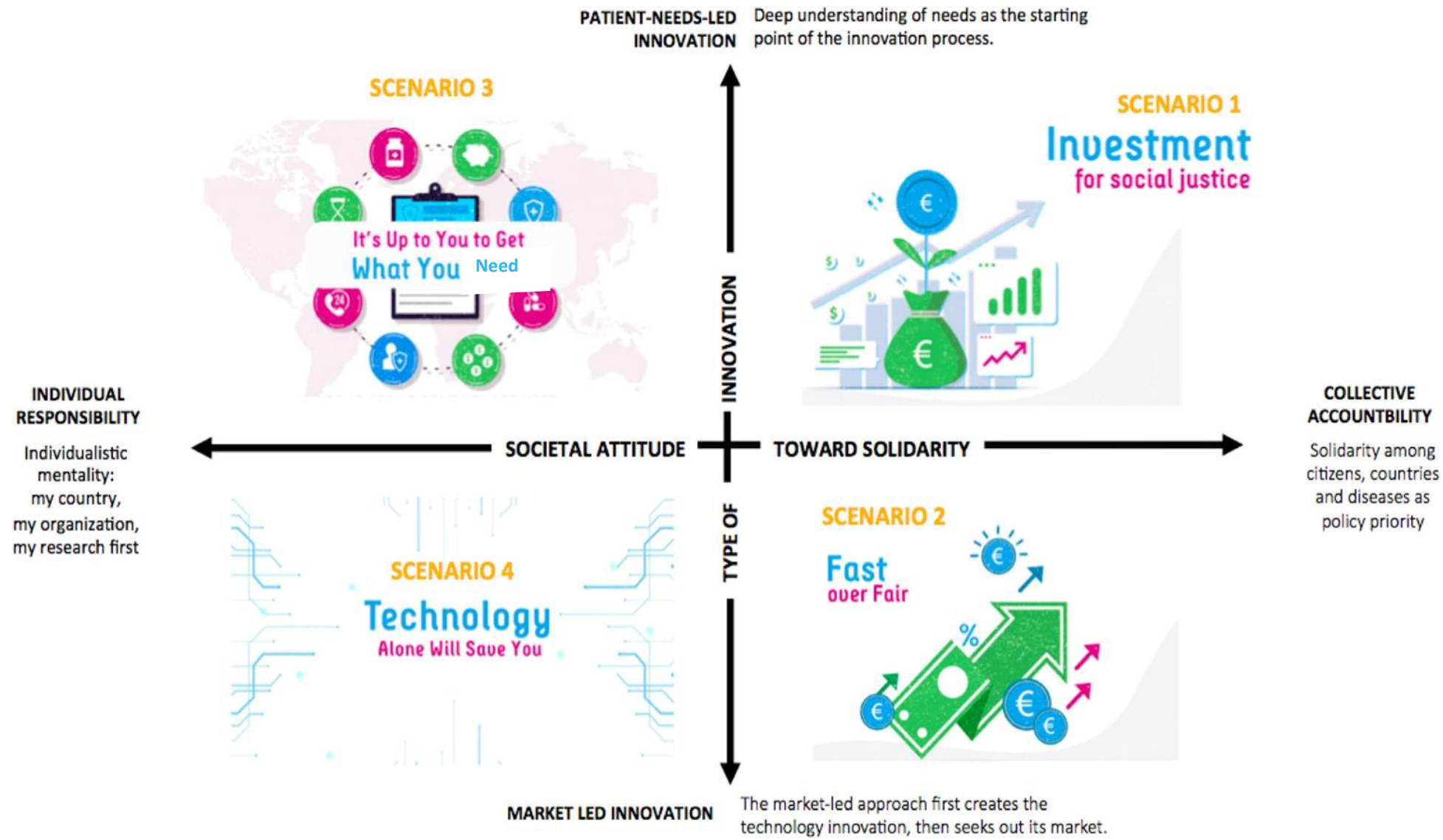
What should this look like?

Rare 2030 Foresight Study:
A roadmap for rare disease policy to 2030

- Two year large scale Foresight Study (2019-2021)
- Over 250+ expert stakeholders from 38 countries
- Input of Young Citizens panel
- European Parliament initiated, European Commission funded
- Participatory and iterative methodology




PATIENT-NEEDS-LED INNOVATION
Deep understanding of needs as the starting point of the innovation process.
SCENARIO 3
It's Up to You to Get What You Need
SCENARIO 1
Investment
for social justice
INDIVIDUAL RESPONSIBILITY
Individualistic mentality: my country, my organization, my research first
INNOVATION
SOCIETAL ATTITUDE
TOWARD SOLIDARITY
COLLECTIVE ACCOUNTBILITY
Solidarity among citizens, countries and diseases as policy priority
TYPE OF
SCENARIO 4
Technology
Alone Will Save You
SCENARIO 2
Fast
over Fair
MARKET LED INNOVATION
The market-led approach first creates the technology innovation, then seeks out its market.

Rare 2030 Foresight Study:
A roadmap for rare disease policy to 2030

- **Eight detailed policy recommendations** across diagnosis, access to healthcare, integrated care, partnerships with patients, research, data and treatments

- Central recommendation for a **new European Policy Framework** driven by the needs of people living with a rare disease, to guide the implementation of consistent national plans and strategies

Recommendation 1: Long-term, integrated European and national plans and strategies

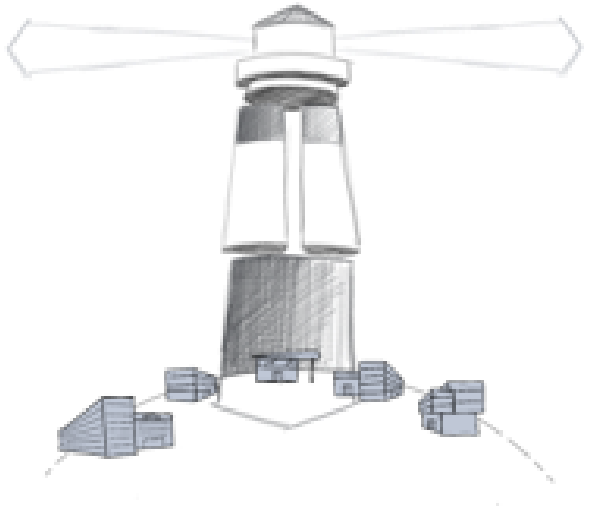
- Align with other **European and national strategies** (e.g. cancer, data, research, access, social rights)
- Support a **harmonisation of the definition of rare diseases and rare cancers** in European and national plans, strategies and policies
- Align with the **international objectives** established in Sustainable Development Goals, Universal Healthcare and other UN system policies relevant to rare diseases.
- Place a new focus **on EU level monitoring of rare disease diagnostics, treatment, care, research, and holistic wellbeing**, with countries encouraged to collect and pool such data to publically and transparently illuminate the status quo and enable benchmarking
- **European and national plans and strategies should be sustained on a long-term basis**, with adequate funding, and should be monitored by the appropriate authorities and key opinion leaders in the field
- Suitable **forum should be created or designated to advance multistakeholder policy-oriented debate on rare diseases**, enabling the identification of good practices and support for implementation to suit national realities

What can we be doing now?

- Leverage opportunities and infrastructures that exist
  - Revision of legislation
  - Joint Action on Integration of ERNs
  - EU4Health
  - Horizon 2030
- Prepare the ground for the strategy
  - Baseline metrics for measurable goals
  - Establish and support a drafting group to design the new Commission Communication on a European Action Plan on Rare Diseases and Council Recommendation

Flagships that will address unmet needs and innovate

- Joint Action on Newborn Screening
- European Comprehensive Care Centres for ultra rare and complex diseases
- EU Fund to generate real world evidence



How would European action help?

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— Magda, Greece

ERNs better integrated into national healthcare systems to improve access to specialist

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— Maria, Austria

Holistic approach to managing symptoms and mental health

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— Natalia, Poland

Improved link with social care