RARE DISEASE DAY 2020
POLICY BRIEFING
FROM PLEDGE TO ACTION

European Parliament, Brussels

Room 3H1

18Th of February, 10h00 – 11h00

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RARE DISEASE DAY®

Our agenda for today

Welcome and opening

What are rare diseases? What is the network, rationale, main purpose

What topics will be working on? Where does EURORDIS have expertise in to offer?

How can you help concretely?

Q&A/Closing

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WHAT ARE RARE DISEASES?

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There are

6,000+ IDENTIFIED RARE DISEASES

Article 'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', European Journal of Human Genetics (2019)

#RareDiseaseDay

29 FEBRUARY 2020

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72% OF RARE DISEASES ARE GENETIC

whilst others are the result of **infections** (bacterial or viral), **allergies** and **environmental causes** or are **rare cancers**.

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70% OF GENETIC RARE DISEASES start in CHILDHOOD

Article 'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', European Journal of Human Genetics (2019)

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300 MILLION PEOPLE WORLDWIDE

living with a rare disease

Article 'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', European Journal of Human Genetics (2019)

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Rare diseases currently affect **5%** OF THE WORLDWIDE **POPULATION**

Article 'Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database', European Journal of Human Genetics (2019)

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Collectively the number of people living with a rare disease is equivalent to the population of the

WORLD'S 3RD LARGEST COUNTRY

300 million people worldwide are living with a rare disease

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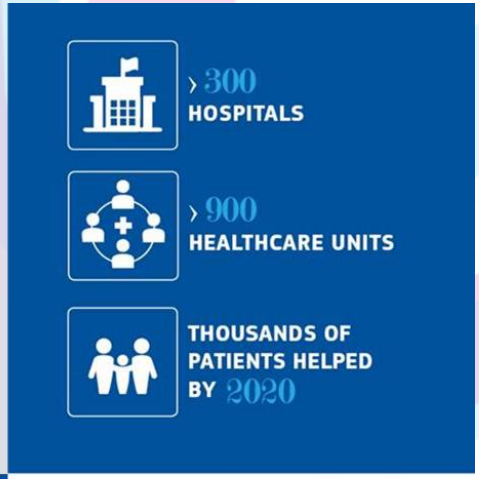
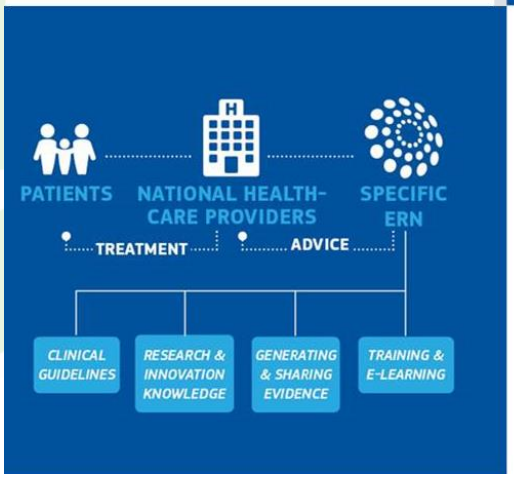
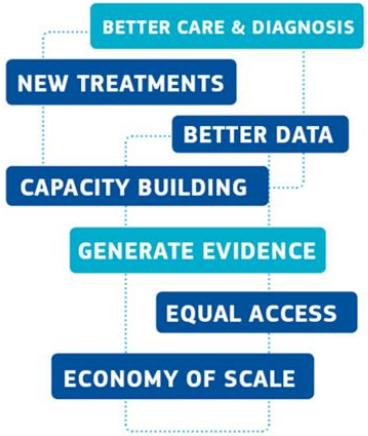
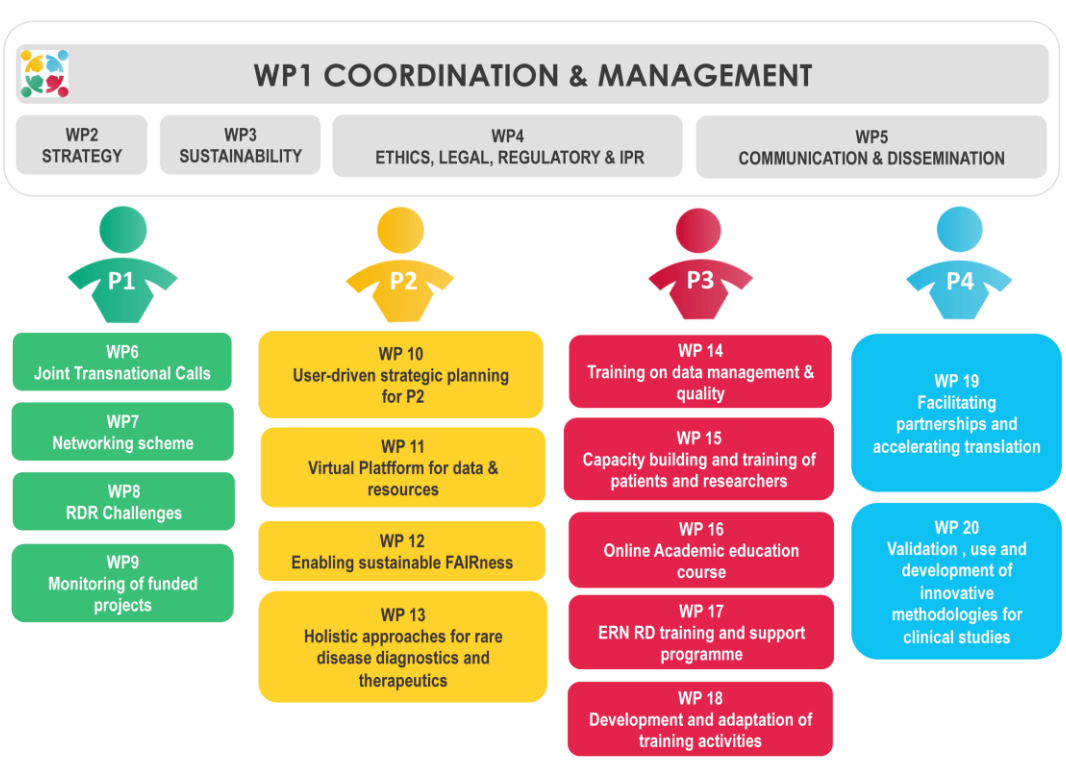
The EU added value (1): Pooling knowledge to improve diagnosis







The EU added value (2): partnership for research



EUROPEAN JOINT PROGRAMME RARE DISEASES

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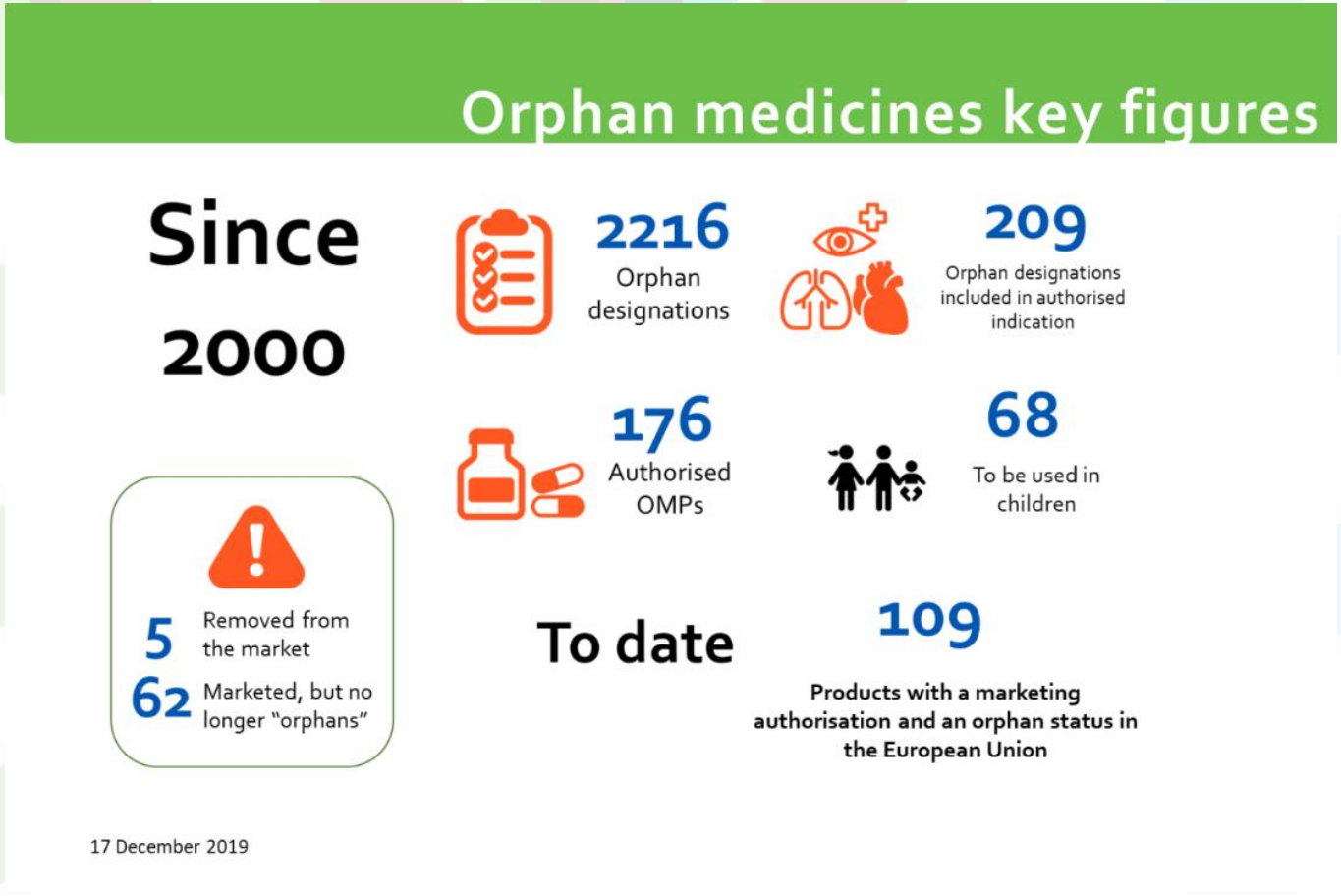
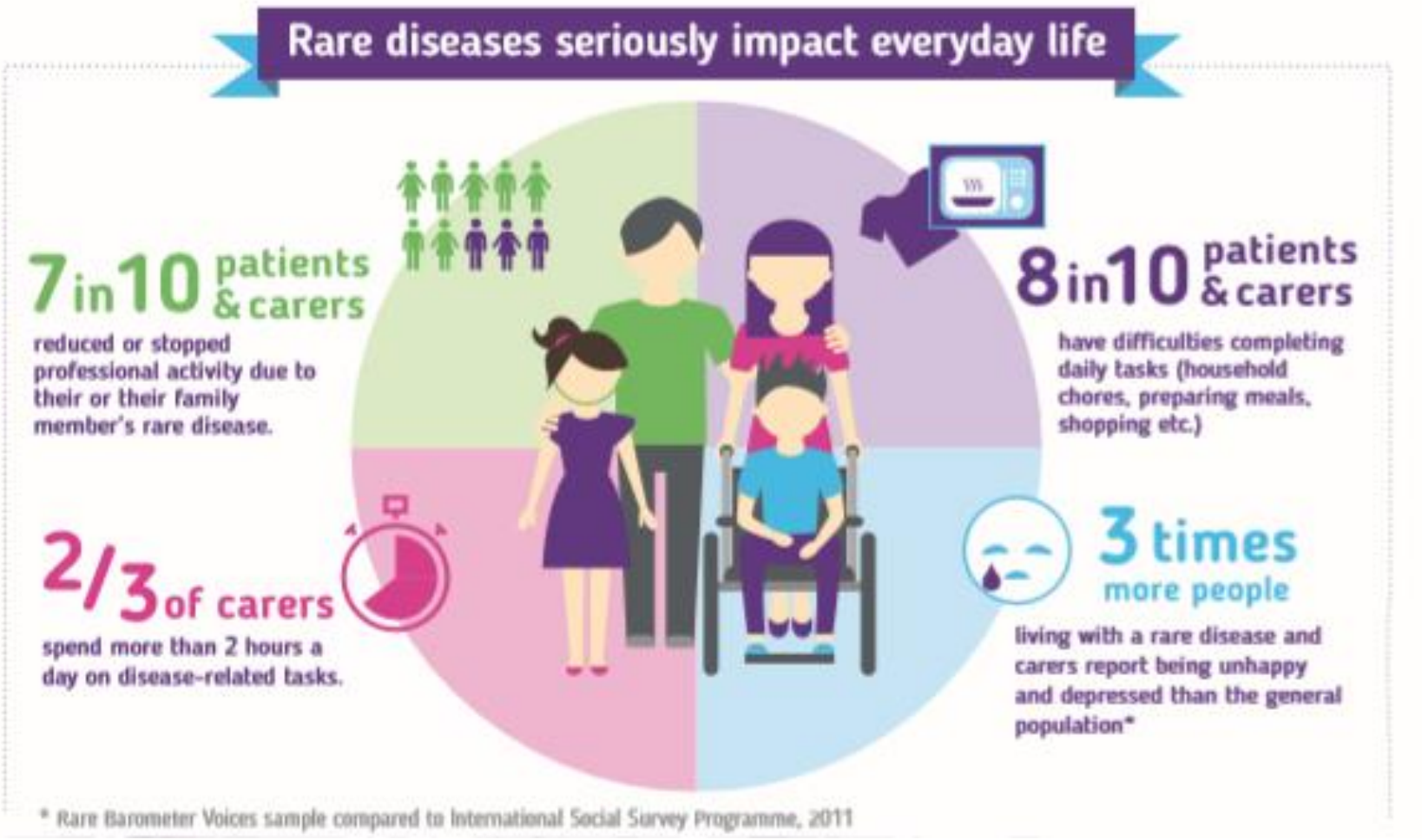
The EU added value (3): therapies development





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An insight into the participation of rare disease patients in research

30 million people are living with a rare disease in Europe and 300 million worldwide

No cure for the vast majority of diseases and few treatments available

3213 rare disease patients and their families responded to the survey, conducted in 23 languages across 42 countries worldwide

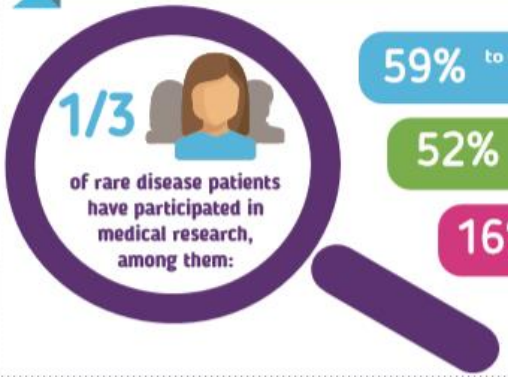
Key findings of a Rare Barometer Voices survey on the participation of rare disease patients in research:

Top 3 obstacles to rare disease research

Top 3 priorities for patients

- Develop therapies
- Improve diagnosis
- Identify mechanisms & causes of rare diseases

What type of research do patients participate in?



- **59%** to develop treatments & therapies
- **52%** in research on quality of life
- **16%** to develop genetic therapis

What matters to patients?

Rare Barometer Voices is a EURORDIS-Rare Diseases Europe online survey initiative. It brings together over 8,000 patients, carers and family members to make the voice of the rare disease community stronger.

Thank you to all Rare Barometer Voices participants and partners! To read the full report, register to participate in future surveys or for more information, visit: eurordis.org/voices

Rare disease patients' experience of treatments

69% have already experienced a treatment

31% have never experienced any treatment

Only **5 %** have already experienced a **curative** treatment

3% a treatment to **prevent** the disease

31% a treatment to **slow down** the disease

62% a symptomatic treatment

During the past 12 months…

Rare disease patients compared with the **General public**

Were unable to get their medical treatment because they could not pay for it

12% +6 6%

Were unable to get their medical treatment because it was not available where they live

22% +5 7%

Were unable to get their medical treatment because the waiting list was too long

14% +5 9%

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WHAT IS THE NETWORK?

Why a network of Parliamentary Advocates for Rare Diseases?

- **European Union added-value of action on rare diseases**
  - ✓ Need of action beyond national borders
- **Breakthrough legislation driven mostly by the EU institutions & implemented at national level**
  - ✓ Need for collaboration at EU level & between the EU and national level
- **Create a pool of advocates amongst policy makers**
  - ✓ Need to raise awareness amongst decision-makers
  - ✓ Strong signal of Parliamentarians commitment & relevance of rare diseases

Core missions of the Parliamentary Advocates for Rare Diseases

1. Explore and discuss specific challenges faced by people living with a rare disease
2. Shape political input for future legislation and programmes in a new framework for Rare Diseases
3. Work to make rare diseases an integral part of EU, national and regional programmes in health, research, social affairs and other relevant policies

From Pledge to Action

1

ENABLES ALL PEOPLE LIVING WITH A RARE DISEASE TO RECEIVE AN ACCURATE DIAGNOSIS AND APPROPRIATE CARE WITHIN ONE YEAR OF COMING TO MEDICAL ATTENTION

2

SUPPORTS HOLISTIC CARE AND SOCIAL SYSTEMS THAT ARE INCLUSIVE OF PEOPLE LIVING WITH A RARE DISEASE, THROUGHOUT THEIR LIVES

3

SEIZES OPPORTUNITIES IN SCIENCE AND INNOVATION THAT EMBODY HOPE FOR PEOPLE LIVING WITH RARE DISEASES AND THEIR CARERS AND CAN CHANGE THEIR LIVES

PEOPLE WITH RARE DISEASES AND THEIR REPRESENTATIVES MUST BE ENGAGED IN ALL FORA WHERE DECISIONS THAT AFFECT THEIR LIVES ARE MADE.

On access to diagnosis and care

1. **Reframe the current European strategy tacking rare diseases** to adequately address the needs of the rare disease community

2. Advancing the work that has been initiated during the previous legislature to support **EU-wide coordination on HTA**

3. Scrutinise activity on the implementation of **the Directive on Patients' Rights in Cross-Border Healthcare** in line with report of European Court of Auditors

Support holistic care

1. Ensure that in the MMF 2021 – 2027 and successive annual budget and related programme, **adequate and streamlined funding is appropriated for activities of the European Reference Networks (ERNs)**

2. Promote the **implementation of the recommendations on ERN integration in national healthcare system**s;

3. Ensure that measures to achieve holistic care for people living with rare diseases and their families, such as access to social services, disability assessment for rare diseases are streamlined in the upcoming **European Disability Strategy**

Foster research

1. Promote the importance of **fostering research in the area of rare cancers** in the context of the Commission's Beating Cancer Plan.

2. Support the rare disease patient community in getting engaged in the upcoming discussions around the **Pharmaceutical Strategy**, including the **legislation on medicines for special populations** (Regulation on Orphan Medicinal Products and on Paediatrics)

3. **Support rare diseases research** by ensuring adequate support to a successful Partnership like the European Joint Programme on Rare Diseases.

Other actions that need support

Organise a European Parliament (EP) event in the context of the RareImpact project that addresses the difficulties of patients accessing advanced therapies;

Promote and organise the final meeting of the Rare 2030 pilot project, promoted by the European Parliament and co-funded by the European Commission.

Encourage patient organisations to participate in European policy-making by supporting 2021 Rare Disease Week, either by meeting your constituents during this week and/or organise a bigger meeting in the EP.

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EURORDIS EXPERTISE

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Our mission

EURORDIS works across borders and diseases to improve the lives of people living with a rare disease

869

Member patient organisations

71 countries (28 EU countries)

44 National Alliances of rare disease patient organisations

Founded in

1997

Outreach to over

2,500

patient groups

72 European Federations for specific rare diseases

40+

Staff members with offices in Paris, Brussels and Barcelona

Over

440

volunteers

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Advocating, empowering & engaging patients in the areas of:

Research

Rare disease policy

Diagnosis

Social care

Treatments

Healthcare

EURORDIS volunteers at the heart of policy making

440+ **volunteers involved in EURORDIS activities**

Drug Information, Transparency and Access Task Force (DITA)
focusses on the areas of product information, transparency of the regulatory process and access to medicines

30 + ePAG Advocates who are members of the EURORDIS **ePAG Steering Committee** active across 24 European Reference Networks

Rare Disease Day Working Group
Co-creating the global campaign

HTA Task Force
to facilitate the participation of patients in health technology assessment activities

340+ **RareConnect moderators** animate the RareConnect communities

SPAG (Social Policy Action Group) advocating for holistic & integrated care

Therapeutic Action Group (TAG), composed of EURORDIS volunteers in the scientific committees and working party at the **European Medicines Agency** (EMA)

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HOW CAN YOU HELP CONCRETELY?

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