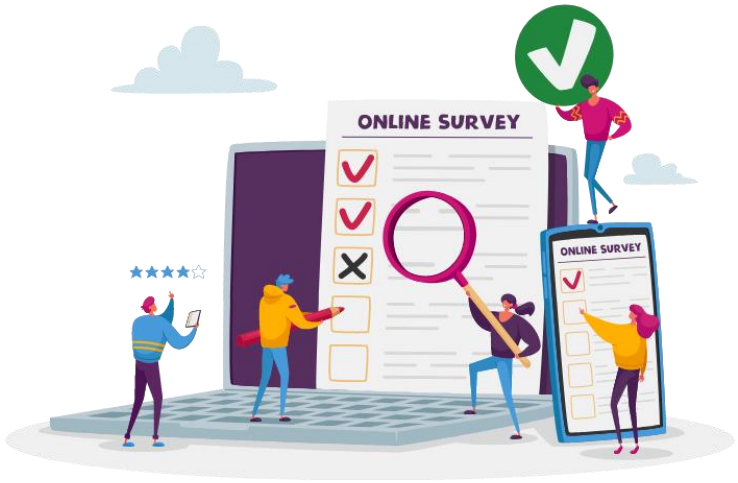
RARE BAROMETER UPDATE

CNA monthly call

8 February 2024



SURVEY RESULTS

Diagnosis
Newborn Screening

DIAGNOSIS SURVEY RESULTS

December – article submitted to the European Journal of Human Genetics

Under review

Time to diagnosis and determinants of diagnosis delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey

Dubief Jessie, Faye Fatoumata, Anido de Peña Roberta, Bellagambi Simona, Escati Peñaloza Luciana, Hunter Amy, Jensen Lene, Oosterwijk Cor, Schoeters Eva, de Vicente Daniel, Crocione Claudia, Faivre Laurence, Michael Wilbur, Yann Le Cam

NAs and EFs with 300+ respondents invited as co-authors
NAs and EFs with 100+ respondents cited in the 'acknowledgments' section.

April – Expected publication date

May – Wide communication on survey results, along with ECRD

NAs and EFs involved in communication from April to build a momentum for ECRD

NBS SURVEY RESULTS - POSTPONED TO APRIL

November 2023

Rare Barometer contribution for the Screen4Care project: proposed list of criteria and diseases for actionability.

March 2024

Discussing publications (report and factsheet) with CNA, CEF and NBS working group.

October 2023

Preliminary results discussed with CNA and CEF > **ask for the recording and ppt!**

ERTC: presentation of preliminary results

rare.barometer@eurordis.org

December-February 2024

Writing results: comparison with surveys/studies on the general population and on rare diseases, to consolidate our advocacy.

Send us any useful reference from your country (even if not in English)!

rare.barometer@eurordis.org

April 2024

Publication of results

DASHBOARD

Preliminary results

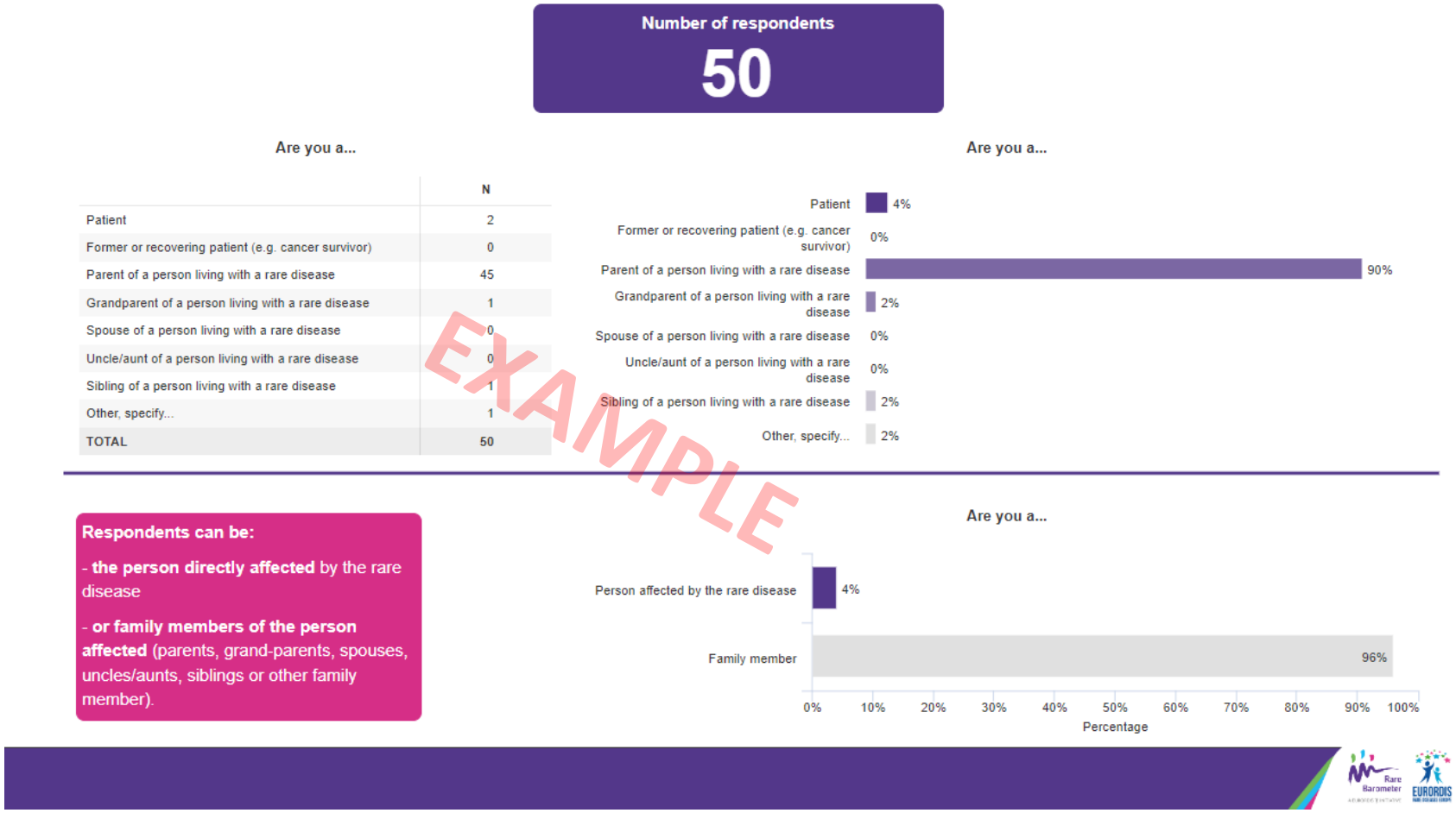
Already available

Descriptive statistics (number and percentage of respondents to each question of the survey).

Consolidated results

Available in April

Descriptive statistics + relevant crossings (to compare with results from the Rare Barometer report).



FACTSHEET

Format: 4-pager, similar to ‘The future of rare disease’ (below)

EURORDIS RARE DISEASES EUROPE · Rare 2030 Foresight in Rare Disease Policy · Rare Barometer A EURORDIS INITIATIVE

THE FUTURE OF RARE DISEASES: LEAVING NO ONE BEHIND!

Key findings from a survey on the opinion of people living with rare diseases on policies that may impact their lives

June 2021

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

Although great progress has been made over the last decade to improve the lives of people living with rare diseases, there remains a high level of unmet needs. The Rare 2030 survey aimed to collect rare disease patients' perspectives on the future of rare diseases to contribute to shaping the Rare 2030 recommendations. The survey shows that a holistic approach to rare diseases is needed, including research, diagnosis, treatment and care, and capitalising on new technological opportunities. Such policies need to be integrated in a new European framework on rare diseases by 2023 to guide National Plans for Rare Diseases and ensure that no one living with a rare disease is left behind.

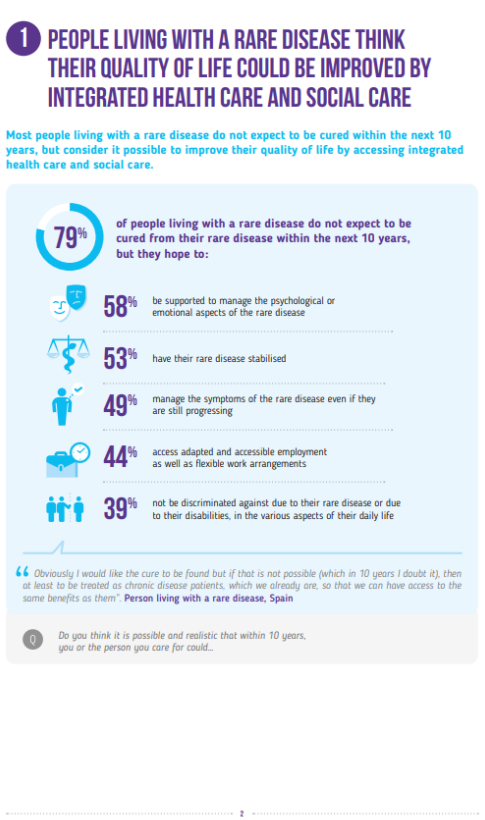
EUROPEAN RESULTS OF THE RARE 2030 SURVEY ON THE FUTURE OF RARE DISEASES

A large scale quantitative survey conducted by Rare Barometer

3 DECEMBER 2020 → 17 JANUARY 2021

3770 respondents in Europe · 23 languages · 70 countries · 978 diseases represented

TARGET POPULATION: patients living with a rare disease or family members (parents and close relatives)



1 PEOPLE LIVING WITH A RARE DISEASE THINK THEIR QUALITY OF LIFE COULD BE IMPROVED BY INTEGRATED HEALTH CARE AND SOCIAL CARE

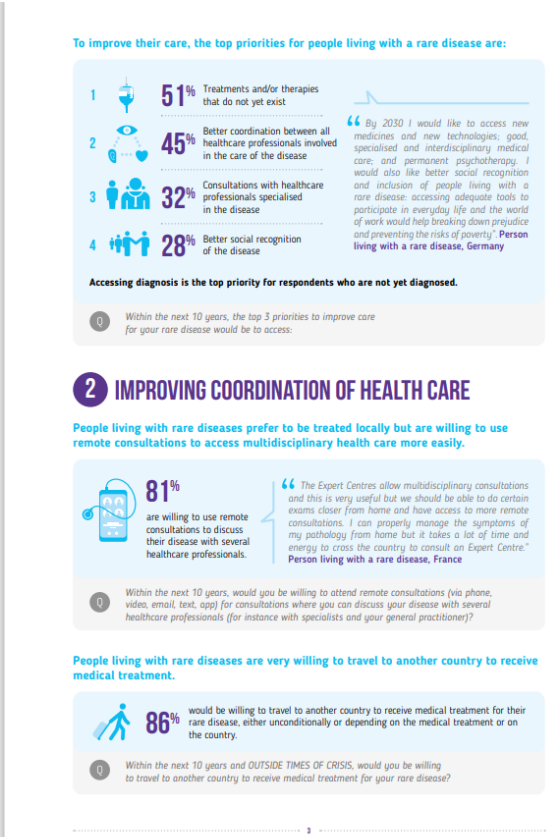
Most people living with a rare disease do not expect to be cured within the next 10 years, but consider it possible to improve their quality of life by accessing integrated health care and social care.

79% of people living with a rare disease do not expect to be cured from their rare disease within the next 10 years, but they hope to:

- **58%** be supported to manage the psychological or emotional aspects of the rare disease
- **53%** have their rare disease stabilised
- **49%** manage the symptoms of the rare disease even if they are still progressing
- **44%** access adapted and accessible employment as well as flexible work arrangements
- **39%** not be discriminated against due to their rare disease or due to their disabilities, in the various aspects of their daily life

“Obviously I would like the cure to be found but if that is not possible (which in 10 years I doubt it), then at least to be treated as chronic disease patients, which we already are, so that we can have access to the same benefits as them”. **Person living with a rare disease, Spain**

Q *Do you think it is possible and realistic that within 10 years, you or the person you care for could...*



To improve their care, the top priorities for people living with a rare disease are:

1. **51%** Treatments and/or therapies that do not yet exist
2. **45%** Better coordination between all healthcare professionals involved in the care of the disease
3. **32%** Consultations with healthcare professionals specialised in the disease
4. **28%** Better social recognition of the disease

“By 2030 I would like to access new medicines and new technologies; good, specialised and interdisciplinary medical care; and permanent psychotherapy. I would also like better social recognition and inclusion of people living with a rare disease: accessing adequate tools to participate in everyday life and the world of work would help breaking down prejudice and preventing the risks of poverty”. **Person living with a rare disease, Germany**

Accessing diagnosis is the top priority for respondents who are not yet diagnosed.

Q *Within the next 10 years, the top 3 priorities to improve care for your rare disease would be to access:*

2 IMPROVING COORDINATION OF HEALTH CARE

People living with rare diseases prefer to be treated locally but are willing to use remote consultations to access multidisciplinary health care more easily.

81% are willing to use remote consultations to discuss their disease with several healthcare professionals.

“The Expert Centres allow multidisciplinary consultations and this is very useful but we should be able to do certain exams closer from home and have access to more remote consultations. I can properly manage the symptoms of my pathology from home but it takes a lot of time and energy to cross the country to consult an Expert Centre.” **Person living with a rare disease, France**

Q *Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app) for consultations where you can discuss your disease with several healthcare professionals (for instance with specialists and your general practitioner)?*

People living with rare diseases are very willing to travel to another country to receive medical treatment.

86% would be willing to travel to another country to receive medical treatment for their rare disease, either unconditionally or depending on the medical treatment or on the country.

Q *Within the next 10 years and OUTSIDE TIMES OF CRISIS, would you be willing to travel to another country to receive medical treatment for your rare disease?*

3 PEOPLE LIVING WITH A RARE DISEASE STRONGLY SUPPORT NEWBORN SCREENING FOR RARE CONDITIONS

95% support newborn screening for rare conditions

“I have bronchiectasis and was told when it was diagnosed that I probably had it for many years. Earlier diagnosis and treatment would have resulted in less damage to my lungs and lower use of medications. With early diagnosis it would be possible for future people with rare diseases to be treated appropriately and quickly.” **Person living with a rare disease, United Kingdom**

Q *In your opinion, in order to diagnose rare diseases at an early stage, should tests for rare diseases be performed at child's birth (e.g. blood tests, genetic screening)?*

4 PATIENT ORGANISATIONS ARE WILLING TO BE ACTIVELY INVOLVED IN RESEARCH FOR RARE DISEASES

For patient representatives, patient organisations should contribute in research for rare diseases by:

- **93%** being official partners or co-investigators
- **78%** raising funds for research on their disease
- **72%** leading their own research projects

“We need a lot of research through clinics, doctoral theses, studies - also internationally please! We also need the work of patient organisations to be recognised and more involved in the research and supply processes.” **Person living with a rare disease, Germany**

Q *As a patient representative, do you think that PATIENT ORGANISATIONS should contribute in research for rare diseases by...*

For more information on the questions of the survey or on its results, see the full report in English or contact rare.barometer@eurordis.org

To know more about the Rare 2030 Foresight Study or to participate in the Rare 2030 campaign to implement its recommendations, please visit action.eurordis.org/rare2030action

THANK YOU to all people living with a rare disease who participated in the survey, and to Rare Barometer and Rare 2030 partners!

Rare Barometer A EURORDIS INITIATIVE

EURORDIS-Rare Diseases Europe regularly surveys the rare disease community via its Rare Barometer programme to identify patients' perspectives and needs on a number of issues in order to be their voice within European and international initiatives and policy developments. Rare Barometer brings together close to 15,000 patients, carers and family members to make the voice of the rare disease community stronger. For more information please visit eurordis.org/voices.

EURORDIS RARE DISEASES EUROPE

Rare Barometer A EURORDIS INITIATIVE

UPCOMING SURVEYS

Social participation & independent living
Mental health

SOCIAL PARTICIPATION AND INDEPENDENT LIVING

OBJECTIVES:

- Understanding the unmet needs of PLWRD regarding disability assessment, access to disability rights and independent living.
- Understanding the impact of rare diseases on PLWRD who are not disabled.
- Shaping our advocacy and initiatives on social policy.

TIMELINE:

[**February**: Topic Expert Committee]
[**March**: Writing the questionnaire]
April: English questionnaire sent to National Alliances for them to test it
May: review the translations in your native language (email us: rare.barometer@eurordis.org)
May-June 2024: Questionnaire distribution (fieldwork)
2nd Semester 2024: communication of survey results

MENTAL HEALTH AND WELL-BEING

MENTAL HEALTH AND WELL BEING

- Understanding the specific and the unmet needs of PLWRD regarding mental health and wellbeing.
- Shaping the EURORDIS Mental Health and Well Being Initiative.

2nd semester 2024: questionnaire distribution (fieldwork)

1st semester 2025: communication of survey results

THANK YOU!

to the Rare Barometer participants, partners and corporate donors in 2023!

rare.barometer@eurordis.org

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