RARE 2030

D6.1 Report on the results of the survey

January 2021

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Rare disease patients' opinion on the future of rare diseases

A Rare Barometer survey for the Rare 2030 Foresight Study

January 2021

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Introduction

The Rare 2030 Survey on the future of rare diseases was conducted by EURORDIS-Rare Diseases Europe via the Rare Barometer program in the scope of the Rare 2030 Foresight Study co-funded by the European Union Pilot Projects and Preparatory Actions Programme (2014- 2020).

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 700 rare disease patient organisations from more than 60 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting patients, families and patient groups, as well as bringing together stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

The Rare Barometer Program is a EURORDIS-Rare Diseases Europe initiative created to systematically collect patients' opinions on transversal topics and introduce them into the policy and decision-making process. The objective is to transform patients' and families' opinions and experiences into facts and figures that can be shared with a wider public and policymakers.

Rare 2030 is a foresight study that gathers the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that will lead us to improved policy and a better future for people living with a rare disease in Europe. The final outcomes of this project include Rare 2030 Recommendations "The Future of Rare Diseases Starts Today- Recommendations from the Rare 2030 Foresight Study"

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- Patients, carers and patient representatives who took the time to complete the survey and share their needs and opinions for the benefit of the rare disease community;
- All National Alliances of patient organisations for rare diseases, patient organisations and patient representatives for their voluntary contribution in recruiting participants for this survey;
- All EURORDIS staff contributing to the design of the questionnaire;
- Proof-readers of the original version of the questionnaire and its translations.

Objectives of the survey

The overall goal of this survey was to gather the opinion of rare disease patients and carers on policies that may impact their lives and take these perspectives into account in the Rare 2030 recommendations.

Executive summary

3 998 patients, carers and patient representatives expressed their needs and opinions on priorities for rare diseases within the next 10 years, regarding medical and social care; early screening to diagnose rare diseases; access to remote healthcare and cross-border healthcare; the best criteria for research incentives and the participation of patient organisations in research.

Accessing treatments and therapies is the top priority for people living with rare diseases:

- They put great hopes in **advanced therapies**, and especially in gene therapy.
- **Early diagnosis** is often crucial for rare disease patients and respondents who **strongly support screening for rare conditions at a child's birth** (95%), both for treatable conditions (79%) and for actionable conditions such as hereditary conditions (70%) or conditions for which appropriate disease management can improve the health and quality of life of the patient (67%).
- **Respondents do not expect to be cured from their rare disease within the next 10 years** (70%) or that they would no longer be limited by their disease to handle routine needs (42%). However, they believe **there is hope for them to stabilise their disease** (53%); to **manage their symptoms** with new medicines and several types of therapies; to receive appropriate **emotional support** (58%); to access **adapted employment** (44%); and **not to be discriminated against** due to their rare disease (39%).

Accessing high quality and multidisciplinary healthcare is also very important for people living with a rare disease who often become specialists of their rare disease out of necessity. They usually **prefer to be treated near their home** but given the scarcity and geographical dispersion of health care experts who can treat them, they are very willing to use **remote health care**, first of all to save time but also to access high quality care, when remote health care is appropriate for the care needed and when they have already met their care team. They are also more willing to **travel to another country in the European Union to receive care or treatments** than the general population.

When asked about research priorities, their answered showed that **they would like research to benefit every rare disease**, including diseases with:

- no curative or symptomatic treatments (81%);
- scarce or inexistent knowledge of the scientific mechanisms of the disease (81%);
- very high associated costs for the patient or for society (71%);
- very low prevalence/incidence (68%).

Patient representatives are **very willing to be proactively involved in research projects as an official partner or co-investigator** (93%). They also support patient organisations to **lead their own research projects** (70%) and **raise funds for research** on their disease (78%).

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1. Methodology

1. 1 Questionnaire design and translation

As part of the Rare 2030 Foresight Study, the questionnaire has been designed based on:

- Key issues for patients defined within the Rare 2030 consultations;
- Results of previous Rare Barometer surveys that could be used to support the Rare 2030 recommendations[1];
- Existing studies that would allow comparison with the general population;
- Relevance for the Rare 2030 recommendations.

The main goal of the survey is to support the Rare 2030 recommendations with evidence-based figures. Previous EURORDIS surveys already covered several aspects of the Rare 2030 recommendations: questions for this survey were chosen in order to focus on aspects that were not yet covered. Hence, this report often makes references to EURORDIS previous surveys.

The questionnaire was written in English and translated in the 22 following languages by professional translators specialized in health-related issues: Bulgarian, Croatian, Czech, Danish, Dutch, Finnish, French, German, Greek, Hungarian, Italian, Latvian, Lithuanian, Norwegian, Polish, Portuguese, Romanian, Russian, Slovak, Slovenian, Spanish, Swedish. Translations were then reviewed by native speakers to verify their consistency with the original English version.

1. 2 Timing and organisation

The survey was open from 3 December 2020 to 17 January 2021. During this period, the COVID-19 pandemic was still ongoing in Europe and several countries applied measures such as lockdowns or restrictions in national or international travels. This situation has been taken into account in the wording of the questions related to remote healthcare and cross-border healthcare.

1. 3 Survey sample

3 998 responses were received:

- 54% respondents were contacted through the Rare Barometer Voices database. Rare Barometer Voices is a tool to carry out EURORDIS quantitative surveys on issues affecting people living with a rare disease. It is made up of a community sample of over 15,000 people living with a rare disease who commit to regularly participate in surveys and studies.
- 46% respondents were contacted through social media, patient organisations and several online networks.

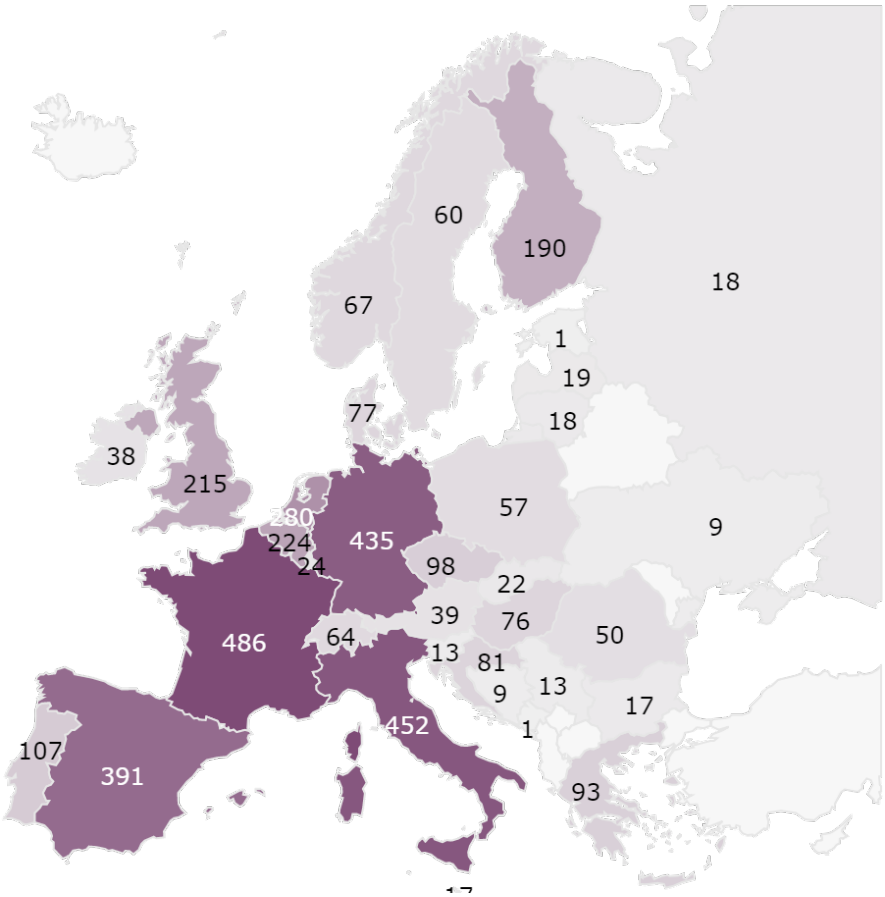
The Rare 2030 survey was only disseminated online. Hence, respondents are all already equipped and used to using the internet. This should be taken into account when interpreting results on remote healthcare and cross-border healthcare.

The Rare Barometer Voices survey software enables high-quality, secure data collection and analysis.

[1] https://www.eurordis.org/voices

Country

The geographical distribution of the sample is as follows:



Geographical distribution of respondents in large part corresponded to the number of country inhabitants: countries with the most respondents were also those with the largest populations (Germany, France, Italy, Spain, Netherlands, Belgium, the United Kingdom), with the exception of Poland and Romania where there were few respondents in comparison to their population.

Geographical distribution also depended on existing networks of patient organisations that disseminated the survey to their members, and other cultural particularities.

Given the relatively low number of respondents in some countries, only significant results (p-value <0.05) were taken into account when analysing differences in answers between countries.

Status

More than 2 in 3 respondents were patients: the proportion of patients and of caregivers is similar to other surveys carried out within the rare disease community. Among the 252 patient representatives who responded to this survey, 43% were also patients and 38% were also caregivers.

TABLE 1. Composition of the Rare 2030 Survey sample

Type	Percentage
Respondent status (several answers possible)	
Patient	69 %
Caregiver of a person living with a rare disease (parent, spouse, family member…)	33 %
Patient representative	7 %
Gender	
Male	22 %
Female	78 %
Age	
Under 18 years old	1 %
18-24 years old	3 %
25-34 years old	10 %
35-49 years old	39 %
50-64 years old	34 %
65 years old and above	13 %
Diagnosed	
Yes	97%
No	3%

Gender

The female proportion of respondents (78/22) was high compared to the general population (52/48) but similar to other surveys carried out among the rare disease community[2].

Age

Most respondents were between 35 and 64 years old. Despite the large paediatric onset of rare diseases, only a few patients under 25 years old responded to the survey, which is consistent with the fact that young people are more reluctant to be surveyed.

Diagnosis

Only 3% of respondents did not yet receive a diagnosis for their rare disease.

Diseases and therapeutic areas

The rare disease population is very diverse: there are over 6,000 distinct rare diseases and a range of disease groups. The sample of this survey represents this diversity and is composed of more than 820 diseases.

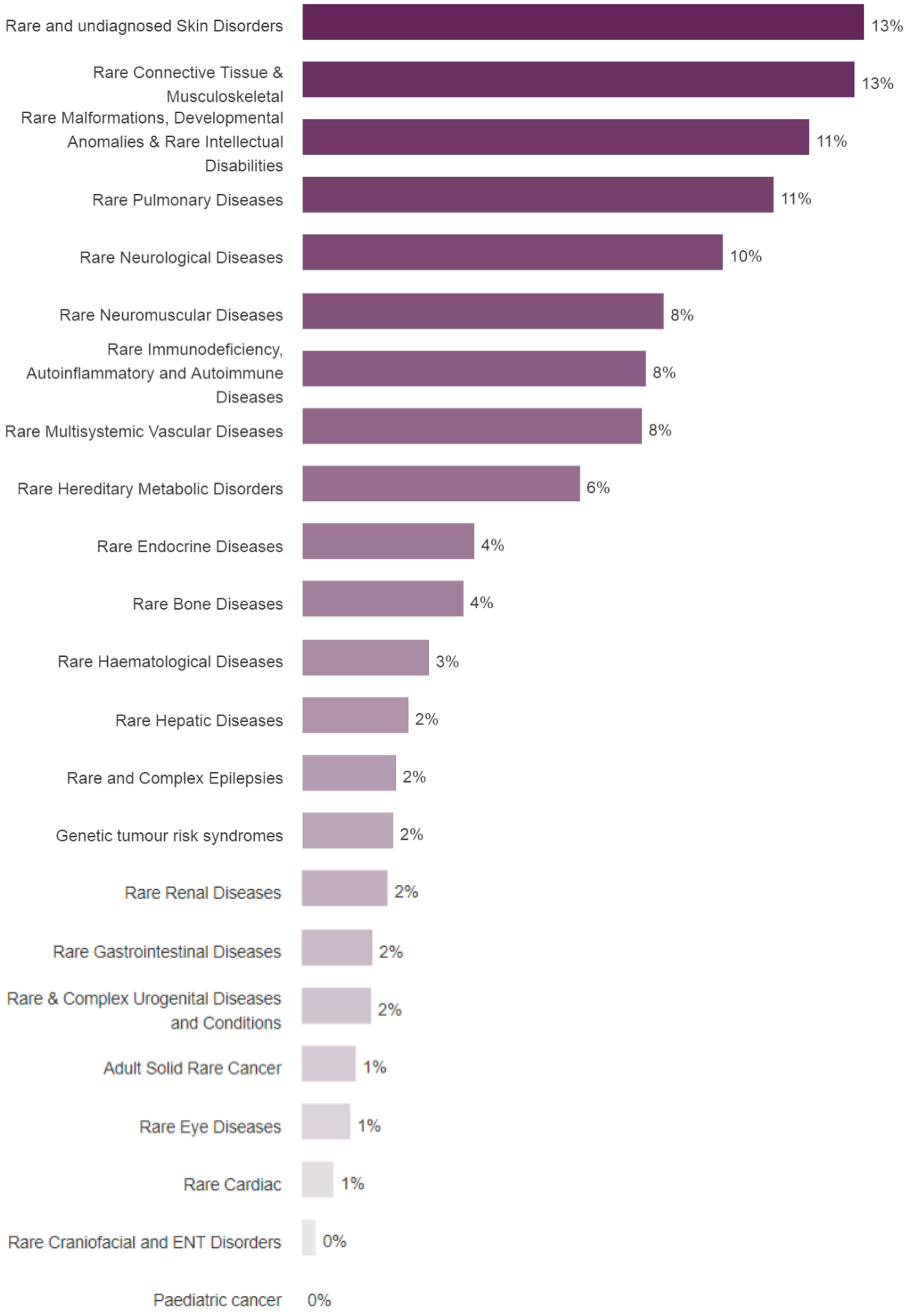
Rare Barometer distinguishes 23 therapeutic areas corresponding to groupings of diseases that could be treated within European Reference Networks[3].

Every rare disease that respondents declared has been classified in up to two therapeutic areas based on:

- The list of rare diseases treated in each European Reference Network, checked with ERNs and patient advocacy groups in 2017.
- The Orphanet classification of each rare disease.

[2] Rare Barometer, *Juggling care and daily life. The balancing act of the rare disease community*, May 2017, p. 7.
[3] https://ec.europa.eu/health/ern_en

Graph 1. Percentage of respondents per therapeutic area



2. Treatment, care coordination and access to healthcare specialists pointed out as top three priorities

Results presented below show that **therapeutic research stays**[4] **the priority for people living with rare diseases** for the upcoming 10 years. Respondents also call for a **better coordination of their health and social care**[5] for instance by being accompanied by one doctor, one person or one expert centre to design **a treatment plan**, together with all professionals involved in their care. This claim is aligned with the proposition of the INNOVCare project to integrate **case managers**, *i.e.* professionals supporting rare disease patients and carers to access health and social care services and support[6]. In the absence of such case managers, care coordination is a burden that widely falls on the patient or on the main carer.

> Within the next 10 years, I would like the knowledge used by health professionals to guide management and treatment to be regularly updated. I would also need a better coordination and integration of carer for my rare disease, as well as case management. **Rare disease patient, Austria**
>
> One doctor who treats everything would be nice. Now treatment is fragmented between internist and general practitioner. **Rare disease patient, Netherlands**

Accessing treatments and therapies is the top priority for people living with rare diseases as they believe it is the main factor to improve their care (see graph 11). Open comments show that respondents have great hopes that **advanced therapies** can help cure their disease. Likewise, only a small percentage of the open comments refer to the possibilities offered by drug development (see annex 2).

> We want genetic research to know the exact cause of the genetic mutation in order to be able to control and correct it to find a normal discomfort. **Rare disease carer, France**
>
> Disease control, innovative therapies, availability of innovative drugs, genetic therapies, social support, financial support. **Rare disease carer, Serbia**
>
> That they finally find a genetically correct treatment or at least a more effective treatment than the existing one. **Rare disease carer, Spain**
>
> Treatment, symptom treatment, recognition of rights, flexible working hours and free aids. **Rare disease patient, Italy**
>
> Findings about possible causes (for other parents) and treatment options against the causative genetic change or at least to alleviate the symptoms. **Rare disease carer and patient representative, Germany**

[4] Rare Barometer, *Rare disease patients' participation in research*, February 2018.

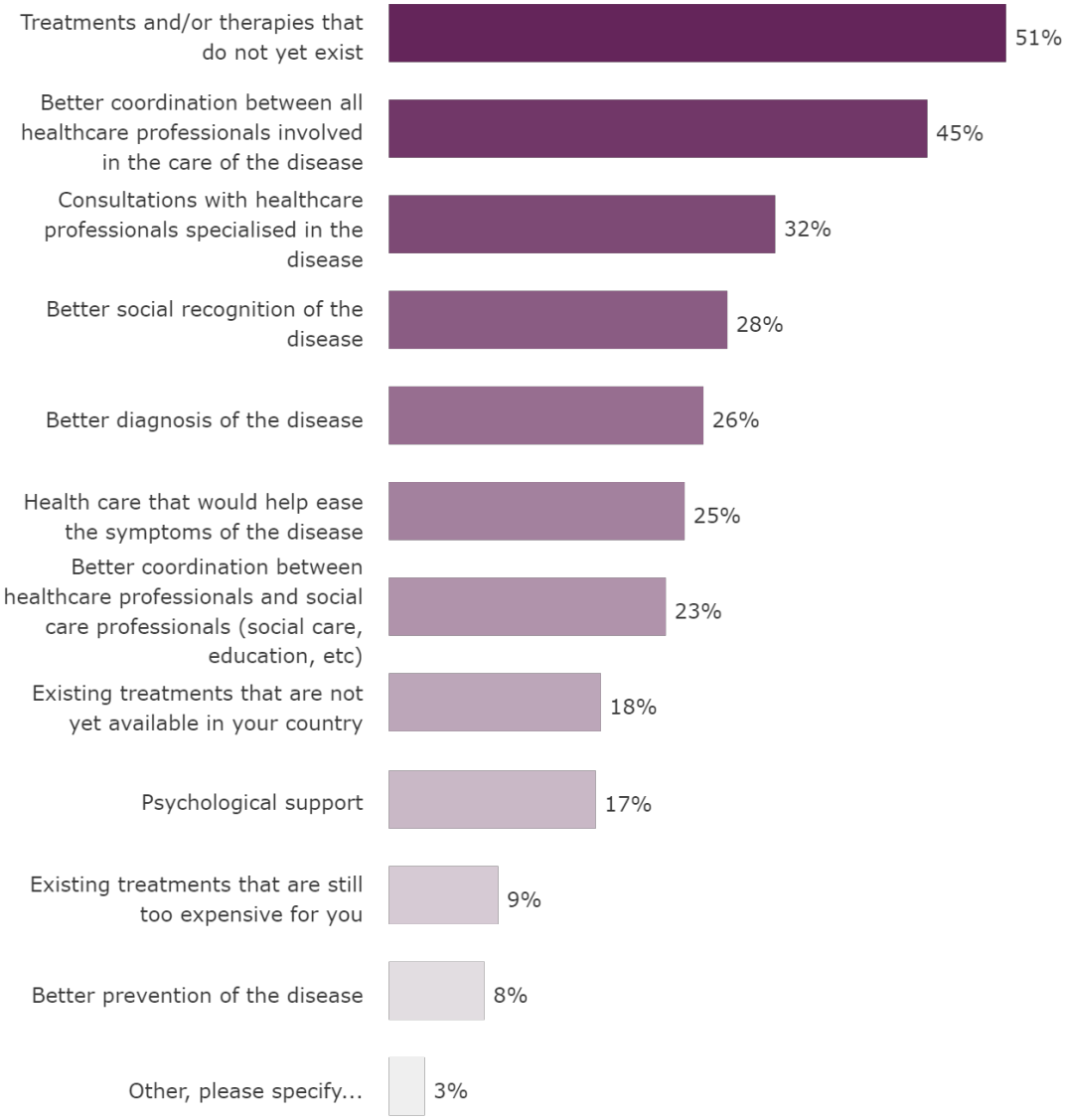
[5] Rare Barometer, *Improve our experience of heath care! Key findings from a survey on patients' and carers' experience of medical care for their rare diseases*, January 2021.

[6] INNOVCare, *Case managers for rare diseases: Roles and training outlines. An outcome of the INNOVCare Workshop on Advancing Holistic & Innovative Care for Rare Diseases and Complex Conditions in Cluj-Napoca, Romania, June 2017*.

The **actual clinical experience** (coordination between healthcare professionals, access to specialists…), along with the **social recognition of the disease** and its **psychological impact** are also highly valued by patients and carers.

> My rare disease is downplayed, the signs and symptoms of the disease are overlooked. By 2030, I would like to raise awareness for the disease, train doctors and specialists as well as medical students, send doctors to internships in countries where my disease is treated, support research into the disease and subsequent treatment, collect data from the patients themselves. The creation of a specialized centre for rare diseases would be most beneficial. **Rare disease patient, Czech Republic**
>
> In my case a program in which you get more information about medication and side effects, exercise, restrictions, nutrition and course of the disease, and advice about work. **Rare disease patient, Netherlands**
>
> The psychosocial care and psychoeducation of people with rare diseases in Europe must be improved and must be a standard of care. **Rare disease carer, Austria**

Graph 11. Within the next 10 years, the top 3 priorities to improve care for your rare disease would be to access to:



Several answers possible; 3 answers maximum.
The order in which modalities appeared was random and different for each respondent.

Access to diagnosis only appears as the 6th priority for respondents, 97% of which are already diagnosed. However, diagnosis was considered the second priority for research on rare disease in a 2018 Rare Barometer survey[7]. Answers to the open question also point out how crucial early and efficient diagnosis is.

> Diagnosis should be established as quickly as possible so that the person can be treated. I suffer from a rare heart disease that could be fatal if left untreated. I know that it is difficult to diagnose in children but at the slightest suspicion they should be followed up. I am 15 years old and it took doctors 13 years to find out. **Rare disease patient, Spain**
>
> That the syndrome becomes better known by health personnel and that genetic testing is taken of the child as early as possible so that mental and physical help and facilitation can start as early as possible in the child's life. A child with a diagnosis can develop many additional psychiatric diagnoses if they do not receive help quickly enough. **Rare disease carer, Norway**
>
> L1 cam syndrome is very rare, but numbers still do not stack up. We need more research and awareness to support early diagnosis in children. This will prevent misdiagnosis and enable families to plan for future children. **Rare disease carer, United Kingdom**

2. 1 Use of screening to diagnose rare diseases

It is estimated that 72% of rare diseases have a genetic origin and almost 70% of rare diseases have an exclusively paediatric onset[8]. For many rare diseases which can be chronic and progressive, signs may be observed at birth or in early childhood. Respondents strongly support screening for rare conditions at a child's birth (95%) and implementing newborn screening programmes across Europe. They also favour the use of new technologies to diagnose rare conditions during pregnancy (87%) and around the time of conception (80%). These findings were consistent for respondents from every country, and especially for more than 90% of respondents living in Romania, Greece, Portugal, Spain, Italy and France.

Screening infants for certain disorders can significantly improve their quality of life by allowing families to better address current needs, anticipate future needs and seek the most appropriate care, treatments and solutions. **Respondents strongly supported newborn screening for treatable conditions (79%) and for actionable conditions** such as hereditary conditions (70%) hence allowing parents to have reproductive options during subsequent pregnancies; or conditions for which appropriate disease management can improve the health and quality of life of the patient (67%).

40% to 47% respondents also supported newborn screening for degenerative and untreatable conditions, which "enables disease prevention through the diagnosis of severe and rare disorders as early as possible [...]. It also provides opportunities for patients and families to plan their future"[9].

> I hope diagnosis will happen at an earlier stage in life for everyone so that as many preventative actions may be taken by the individuals affected by the disease and the people surrounding them. **Rare disease patient, Finland**
>
> Unfortunately, I transmitted one of my diseases to my three daughters because I did not know what I had. I would like research to allow my future grandchildren not to have one of my diseases. **Rare disease patient and carer, France**
>
> 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

[7] Rare Barometer, *Rare disease patients' participation in research*, February 2018.
[8] Nguengang Wakap, S., Lambert, D.M., Olry, A. *et al.* Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* **28,** 165–173 (2020).
[9] EURORDIS, *Key Principles for Newborn Screening. A EURORDIS Position paper*, January 2021.

medications. With early diagnosis it would be possible for future people with rare diseases to be treated appropriately and quickly. **Rare disease patient, UK**

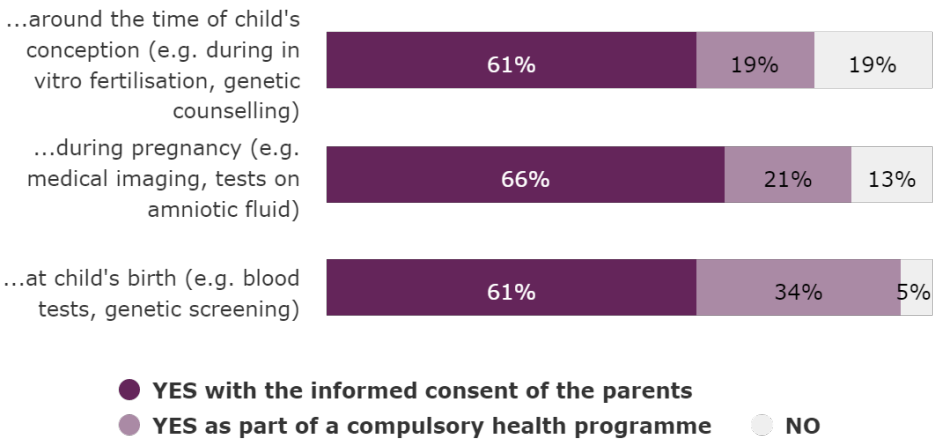
I would like better prevention in the context of pre-implantation and prenatal control by 2030. **Rare disease patient representative, Greece**

I would like people and healthcare professionals to be more relaxed regarding my disease: if one is not seriously affected, one can live with it for a long time, wholly or fairly, without restriction. That is why I am not for diagnosis for foetuses, because I am glad I did not have an abortion! A life counselling would be desirable: how can I behave, what can my surroundings do, what must they do and when - so that I can still live good, because there are many small things that can be done. You stand there alone in a wide hallway. **Rare disease patient, Germany**

Graph 20. New technologies are being developed at a rapid pace and could be used to diagnose some rare diseases in children at a very early stage, improving their treatment options and quality of life. The next few questions will be about your opinion on the use of those technologies in the future.

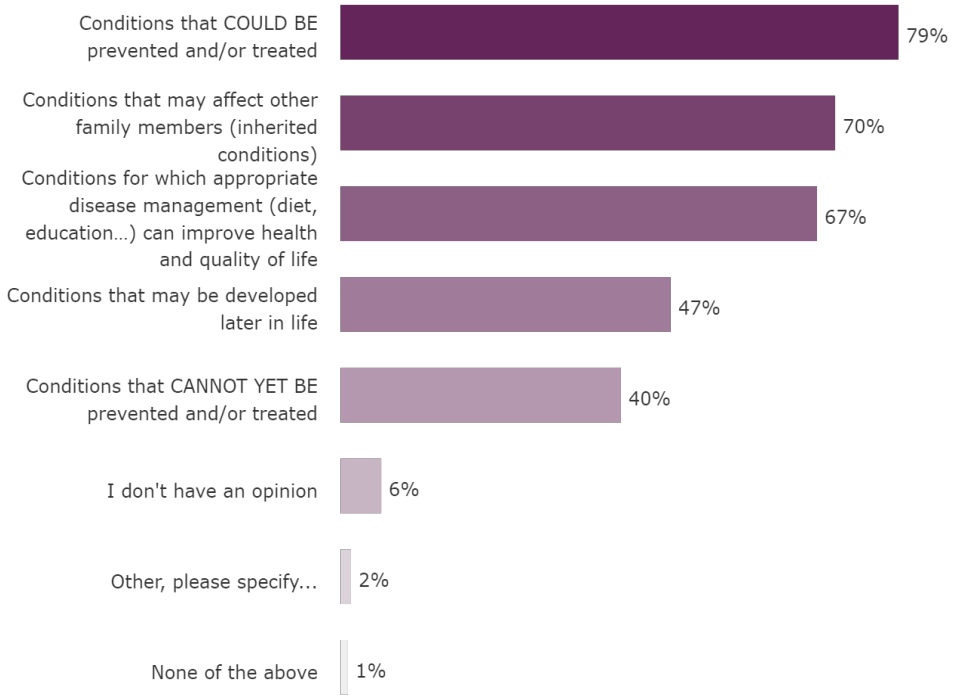
Your answers will help EURORDIS-Rare Diseases Europe voice the opinion of the rare disease community on this delicate matter.

In your opinion, in order to diagnose rare diseases at an early stage, should tests for rare diseases be performed...



Graph 21. Many countries have developed compulsory health programmes to test newborns and diagnose rare diseases at an early stage. However, there are still discussions about what types of diseases should be tested as part of those programmes.

In your opinion, for which conditions should newborns be tested as part of a health programme?



Several answers possible.

2. 2 Most respondents do not expect a cure by 2030 but consider it is possible to improve their quality of life

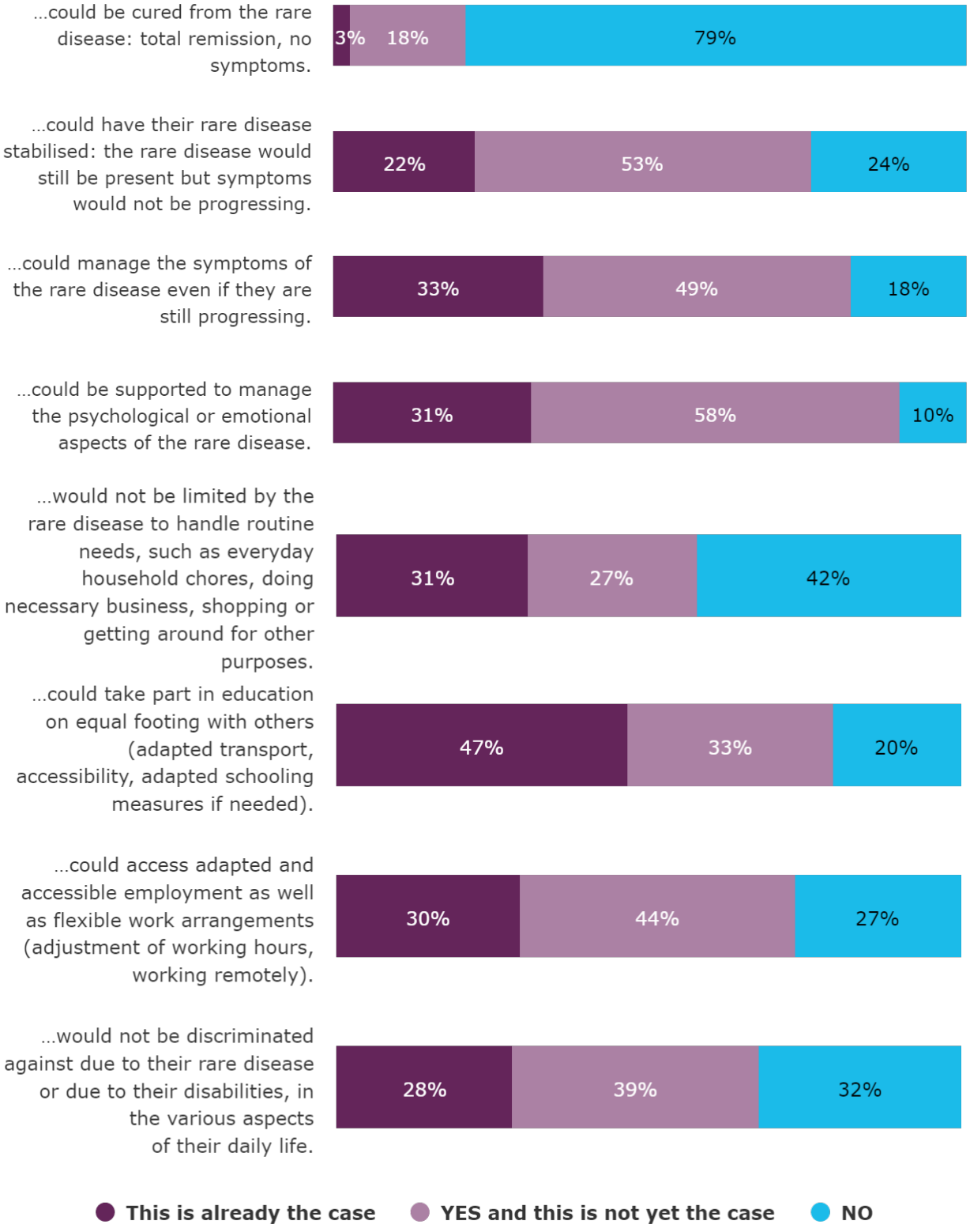
People living with rare diseases do not expect to be cured from their rare disease within the next 10 years (70%) or that they would no longer be limited by their disease to handle routine needs (42%).

However, they rely on research because they believe there is hope for them to **stabilise their disease** (53%); to **better manage their symptoms** with new medicines and several types of therapies (see graph 13); to receive appropriate **emotional support** (58%); to access **adapted employment** (44%); and to **not be discriminated against** because of their rare disease (39%).

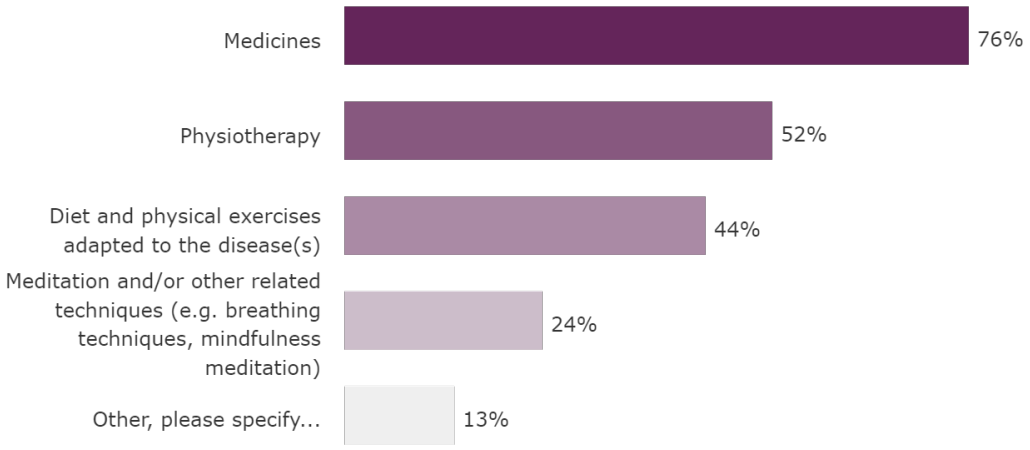
> Obviously I would like the cure to be found but if that is not possible (which in 10 years I doubt it), then at least they treat us as chronic disease patients, which we already are, so that we can have access to the same benefits as chronic disease patients. **Rare disease patient and patient representative, Spain**
>
> I am an adult, in my illness there is no cure, I just hope that in the coming years the research will make progress, for this reason all rare diseases will find a cure at least to stop the disease. **Rare disease patient, Italy**
>
> Affordable therapy to cure or to greatly improve quality of life. Unambiguous definition, tests, management of the disease with objective measures, social acceptance of the disease, no discrimination in the workplace due to part-time work or other limitations unrelated to the quality of the work, coordination between medical workers (doctors, specialists, nursing, …) so that they

all say the same thing. Make additional training mandatory for doctors / specialists who treat patients with such diseases or that they refer fairly if they do not know themselves. Do not get fallacies for the disease if they are not scientifically founded and have not been tested (e.g. you are overweight, it is psychological, ...). **Rare disease patient, Belgium**

Graph 12. Do you think it is possible and realistic that within 10 years, you or the person you care for...



Graph 13. Within the next 10 years, in order to help ease the symptoms of your rare disease, you would need access to (N= 924*)



**Only respondents who cannot yet manage the symptoms of their rare disease but consider it possible and realistic within 10 years.*

In the open question (see annex 2), respondents ask for a **better knowledge** of rare diseases for patients, caregivers, healthcare professionals (general practitioners as well as specialists), social professionals and the whole society: they propose to improve education for all.

> I would like rare diseases to be better known and to be more easily recognized so that patients are not only considered lazy or depressed and addressed to a psychiatrist. Overall, I want early recognition, early genetic testing (preferably from birth) and for the doctors to believe the patients, no matter how weird their symptoms sound and even if they are depressed young women. **Rare disease patient, Finland**
>
> Greater knowledge of the disease on the part of health professionals, an association that supports and informs patients, greater support for using specialists from other countries, which can be done through online consultations. **Rare disease patient, Portugal**
>
> Avoid trial and error in the treatment of epilepsy related to the disease by documenting treatment data (existing mutation, medication, effect) in databases to which the treating physicians and, if applicable, patients have access. **Carer and patient representative, Germany**
>
> More education! **Patient, Germany**

Some respondents pointed out that **teleworking** allows them to enhance their quality of life by working for a sufficient number of hours while better managing their physical symptoms and being more included socially:

> I would like the organization culture to change for the better. With COVID-19, I have been able to work longer hours and maintain a full time job because I am working from home all the time and thus can manage my physical symptoms imminently as they occur. Previously I would be struggling far more and working much reduced hours when they included travel and working seated for hours. I would love to work full time and primarily from home and I hope this becomes more socially acceptable. **Rare disease patient, UK**

Several respondents also raised the issue of **transition from paediatric to adult units**:

> I would like reference centres for adults to be created for my son's disease, they do not exist. It is a great difficulty to find specialists for adults and a great loss of energy. Being able to bring everything together in one place would be a blessing. **Rare disease carer, France**

There is an urgent need for adequate and well-founded transition programs for rare diseases that affect patients from childhood. Synergies should urgently be used better and patient organizations should receive basic funding and be involved in decisions on health policy at national and international level. **Rare disease carer, Austria**

3. Improving access to health care

People living with rare diseases often have little information available about their diseases and very few treatment options. They usually prefer to be treated near their home[10] but given the scarcity and geographical dispersion of health care experts who can treat them, they are very willing to use remote health care (see graphs 2, 3 and 6), first of all to save time (see table 2) but also to access high quality care (see graph 3), when remote health care is appropriate for the care needed (see graph 4) and when they have already met their care team (see graph 6).

A face to face consultation is always the best option especially if it is the first meeting of a patient with a specialist. There is the human contact which helps us to better explain and understand each other so as to establish the right diagnosis and protocol. There is also the delicate matter of how to break the news to a patient that s/he has a rare disease which in most cases, causes a lot of distress to the patients. But some follow-up consultations can then be held remotely. **Rare disease patient, Malta**

People living with rare diseases are also more willing to travel to another country to receive care or treatments than the general population (see table 3).

I don't think it will be a big problem. In fact, I was searching for specialists/experts from other countries to understand my condition. Ideally they are around and you can meet them from time to time. But I prefer to talk with knowledgeable experts via the phone compared to meeting a doctor/specialist who doesn't understand my condition. It will make diagnosis and treatments much easier! **Rare disease patient, Netherlands**

However, those who are not willing to travel or to use remote consultations should not be overlooked. They are usually older (65 years old or above). One of the main obstacles to remote consultations are concerns about the safety of the care (27%), be it missing important symptoms that only physical examination can spot or concerns regarding confidentiality of online consultations. Other limitations include language difficulties if the healthcare professional does not live in the same country (50%), technical obstacles like difficulties accessing the internet or using the phone (21%), difficulties in being reimbursed for remote consultation (17%) but also the lack of human contact (open text question).

The lack of human contact makes me feel abandoned. **Rare disease patient, France**

It is hard to find a place at home with peace to be able to participate. **Rare disease carer, Denmark**

The best response to my needs has come in close contact at the reception with a specialist familiar with my disease. Virtual interpretations are difficult and misconclusions have already complicated my treatment. **Rare disease patient, Finland**

Being hearing impaired, I have difficulty understanding in videoconferencing or on the phone. **Rare disease patient and patient representative, France**

[10] EURORDIS, *The voice of 12 000 patients: experiences and expectations of rare disease patients on diagnosis and care in Europe. A report based on the EurordisCare3 surveys*, Paris, 2009.

The Rare 2030 survey was disseminated online, introducing positive bias with respondents already equipped and used to using online devices and services. Willingness to use remote consultations may have been overestimated within the general rare disease population and digital divide should be taken into account when interpreting the results of this survey.

3. 1 Saving time

Most rare diseases are complex, requiring a multidisciplinary approach to their care: in the EurordisCare3 survey[11] conducted between 2006 and 2008, respondents needed an average of 9.4 different medical services, including four kinds of consultations, three types of medical exams and 2.4 types of additional care services. The daily life of people living with a rare disease often revolves around locating service providers, securing appointments and obtaining the quality care required. Care-related tasks have a serious impact on everyday life as they represent more than 2 hours a day for 42% of people living with rare diseases[12].

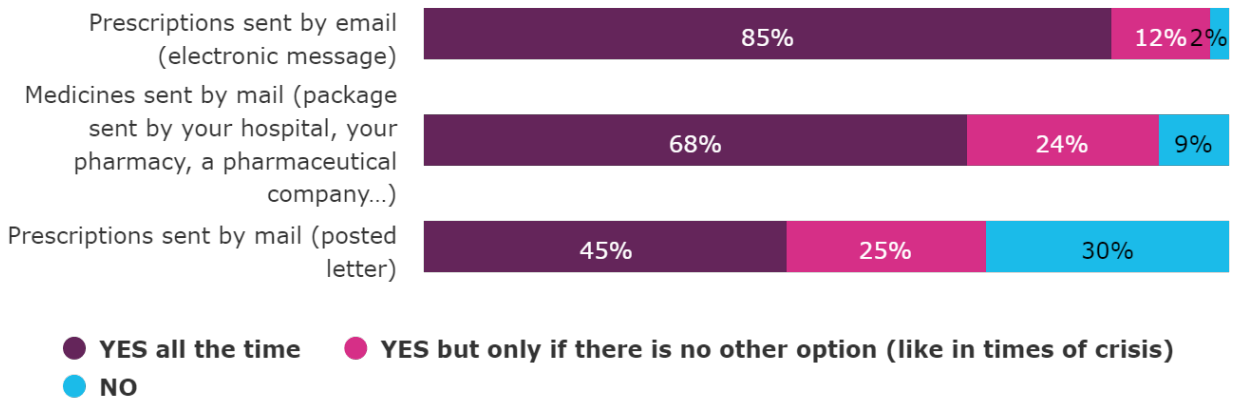
As a consequence:

- **Saving time is the main reason** for people living with a rare disease to use remote consultations and remote prescriptions.
- **84 %** respondents are willing to use electronic prescriptions all the time.
- **65%** respondents are also willing to receive medicines by mail all the time.

TABLE 2. Remote healthcare services, such as virtual consultations and electronic prescriptions, have been developing in daily life and are playing a crucial role during the COVID-19 pandemic.
The next few questions will be about how you are willing to use these remote healthcare services within the next 10 years.
Within the next 10 years, what would be the two main reasons for you to use…

	Saving time	Accessing higher quality care	Having no other option	Safety	Saving money
…remote consultations	**53%**	39%	28%	21%	12%
…remote prescriptions (sent by email or by mail)	**75%**	17%	15%	17%	16%

Graph 2. Within the next 10 years, would you be willing to use the following services:



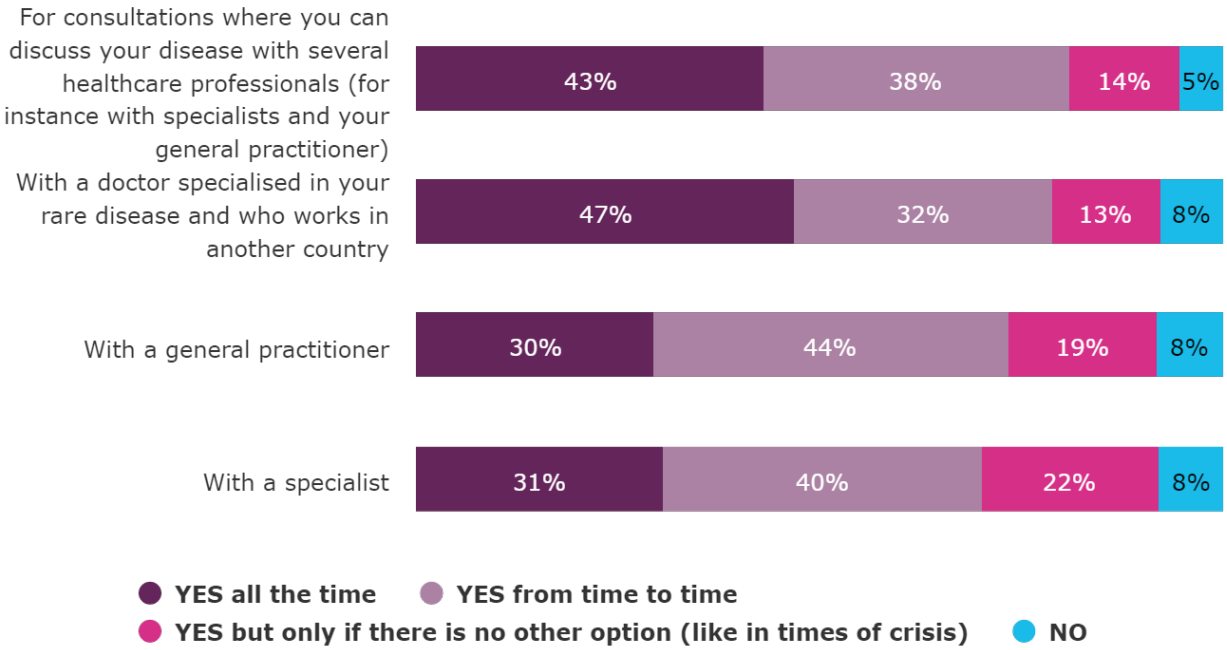
[11] EURORDIS, *The voice of 12 000 patients, op. cit.*
[12] Rare Barometer, *Juggling care and daily life. op. cit.*

3. 2 Accessing high quality and multidisciplinary care

Remote consultations should not be the rule: only a minority of people living with rare diseases would be willing to use them all the time. Face to face consultations and local treatments should be favoured, through local centres of expertise or by helping local doctors to treat rare disease patients through European Reference Networks.

Accessing higher quality care is the second main reason reported by people living with rare diseases for using remote consultations, either for **multidisciplinary consultations** (it is easier to gather several specialists during a remote consultation than to gather them in one room) or for **consultations with a specialist working in another country**. In the latter case, language difficulties were reported as a major potential obstacle to remote consultations (by 50% of respondents) and adequate measures should be taken to address this barrier, such as inviting an interpreter or using automated translations.

Graph 3. Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app):



3. 3 Remote consultations are not appropriate for all types of health care

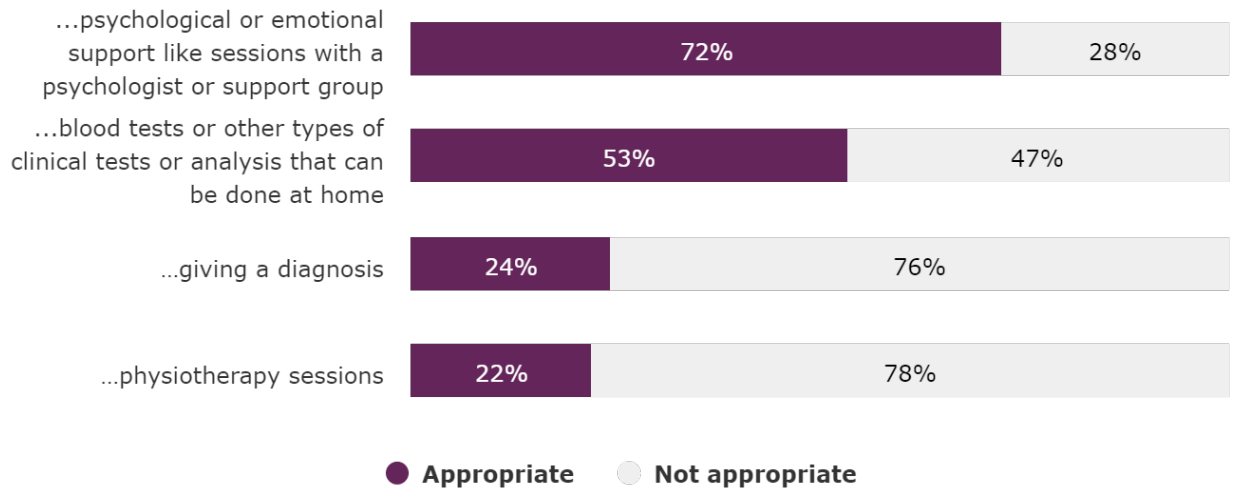
Most respondents consider that remote consultations are appropriate for psychological support and for clinical tests that can be done at home. However, 3 in 4 respondents consider that remote consultations are not appropriate to give a diagnosis or for physiotherapy sessions. Inappropriate care is one of the main obstacles for people living with rare diseases to use remote consultations (for 49% of respondents).

> When you have a lump under your skin and the doctor is on the phone, how should the doctor know what it is? **Rare disease patient, Luxembourg**
>
> Remote diagnosis without proper investigation can lead to misdiagnosis. **Rare disease carer, Germany**

It is difficult to know, for example, if you do an exercise correctly in physiotherapy before you have done it. But remote consultations can be excellent for follow-up or as a complement! **Rare disease patient, patient representative and carer, Sweden**

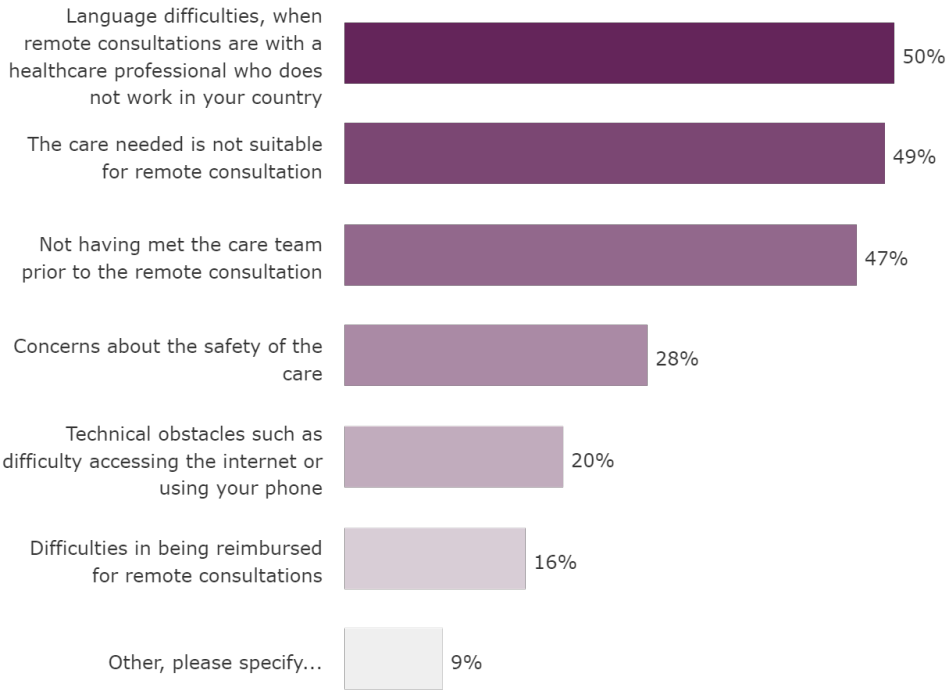
In person, the doctor is able to notice a lot that you might not notice during a video call. Psychological aspects can change the outcome of the consultation and are practically ignored during the virtual consultation. **Rare disease patient, Latvia**

Graph 4. Do you think that remote virtual consultations are appropriate for…



Some modalities have been grouped:
- *Appropriate = very appropriate + fairly appropriate*
- *Not appropriate = not very appropriate + not at all appropriate*
- *Answers to the modality "no opinion" are excluded from the analysis*

Graph 5. What would be the 3 main obstacles for you to use remote consultations within the next 10 years?



Three answers maximum.

3. 4 People living with rare diseases are more willing to use remote consultations if they have already met their care team

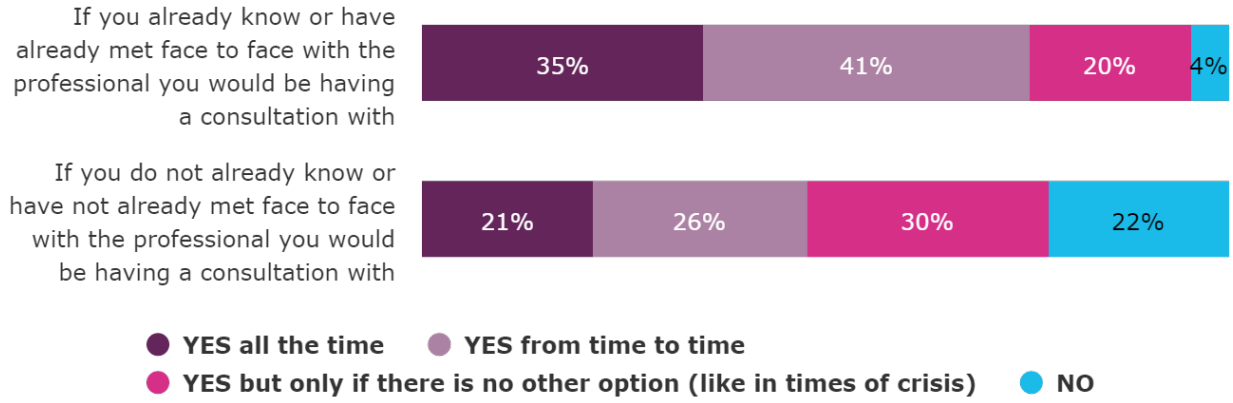
People living with rare diseases are more willing to use remote consultations if they have already met face to face with the professional they would be having a consultation with. **Not having met the care team is also one of the main obstacles to using remote consultations** within the next 10 years (for 47% of respondents).

In general, respondents are also more willing to use remote consultations:

- **from time to time** (as compared to all the time), hence keeping some face to face consultations.
- **when they have no other option** (like in times of crisis), even when they have not already met face to face with the healthcare professionals.

22% respondents would not have a remote consultation, even in times of crisis, if they have never met the professional.

Graph 6. Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app, etc.):



Answers to modality "I don't know" are excluded from the analysis.

3. 5 Willingness to access cross-border healthcare

Responses to the Rare 2030 survey were gathered from December 2020 to January 2021, during the COVID-19 pandemic and while many countries had imposed restrictions regarding national and/or international travels.

Questions on cross-border healthcare were taken from the special Eurobarometer 425 published in 2014[13], where questions were asked by phone and respondents were offered the possibility to answer "yes" or "no". Whenever respondents spontaneously answered "it depends on the medical treatment or on the country", this answer was also registered. The methodology of the Rare 2030 survey differs from the one of Eurobarometer as questions were asked online and there was no possibility to add a spontaneous "it depends" answer. This probably explains part of the differences between the percentage of respondents who answered "it depends" in the Rare 2030 survey and in the Special Eurobarometer 425.

Directive 2011/24/EU on patients' rights in cross-border healthcare establishes the legal framework to receive healthcare in another country of the European Union. This directive is complementary to the EC regulation Nos. 883/04 and 987/09 which entitles insured individuals to receive healthcare elsewhere within the European Union or the European Economic Area, as well as in Switzerland, and be covered for the care received by their home country's healthcare system according to that system's specific guidelines. These legislations allow European citizens to have access to cross-border healthcare, which can be critical when patients do not find healthcare professionals specialised in their rare disease in their country.

86% of respondents would be willing to travel to another country to receive medical treatment, either unconditionally or depending on the medical treatment or on the country; they were 49% in the general EU population in 2014. Only 9% of people living with rare diseases would not be willing to travel to another country to receive medical treatment; they were 46 % in the general EU population in 2014.

TABLE 3. Within the next 10 years [and OUTSIDE TIMES OF CRISIS], would you be willing to travel to another country in the European Union to receive medical treatment [for your rare disease]?[ɣ] n=3 365

	Percentage of People Living With Rare Diseases (PLWRD)	Percentage in the general EU population	Countries where the modality was significantly over-represented among people living with a rare disease[†]
Yes	44%	33%	Ireland (79%), Romania (69%), Croatia (69%), Sweden (66%), Greece (64%), Austria (64%), Poland (61%), Portugal (59%), Czech Republic (56%), Italy (50%).
It depends on the medical treatment or on the country	42%	16%	Germany (54%), Netherlands (53%), France (47%)
No	9%	46%	Finland (14%), Netherlands (12%), France (12%), Germany (12%)
I don't know	5%	5%	France (9%), Spain (8%)
Number of respondents	3 365	27 868	3 365

[ɣ]Text between brackets was only included into the Rare 2030 survey and not into the Eurobarometer survey.
[†]Percentages between brackets refer to the number of people living with a rare disease who live in the country and who chose the corresponding modality. Only significant relationships are taken into account (p-value < 0.05).

[13] We thank the Eurobarometer for sending us the translations of their questions and for allowing us to reproduce them in the Rare 2030 survey. Special Eurobarometer 425, *Patients' rights in cross-border healthcare in the European Union*, May 2015.

People living with rare diseases are mainly willing to be treated in another EU country to **access treatments that are not available in their country** (84%), to access **better quality treatment** (59%) and **treatment from a renowned specialist** (45%). Treatment costs are less cited as a reason to look for cross-border healthcare than in the general EU population.

TABLE 4. For which of the following reasons would you be willing to go to another country in the European Union to receive medical treatment [for your rare disease(s)]?[ɣ] n=3 075

	Percentage of People Living With Rare Diseases (PLWRD)	Percentage in the general EU population	Countries where the modality was significantly over-represented among people living with a rare disease[†]
To receive treatment that is not available in your country	84%	71%	Netherlands (91%)
To receive better quality treatment	59%	53%	Croatia (68%), Italy (62%)
To receive treatment from a renowned specialist	45%	38%	Sweden (79%), Austria (79%), Germany (76%), France (51%)
To receive treatment more quickly	31%	34%	Hungary (46%), Netherlands (39%), Italy (35%)
To receive cheaper treatment	11%	23%	Finland (23%), Belgium (19%)
To receive treatment from a provider that is closer to home	9%	6%	Germany (15%), France (13%)
I don't know	3%	2%	France (6%)
Other, please specify…	2%	2%	Romania (8%), Czech Republic (7%)
Number of respondents	3 075	13 503	3 075

Several answers possible

[ɣ] Text between brackets was only included into the Rare 2030 survey and not into the Eurobarometer survey.

[††] Percentages between brackets refer to the number of people living with a rare disease who live in the country and who chose the corresponding modality. Only significant relationships are taken into account (p-value < 0.05).

Base: Respondents who would be willing to travel to another country in the EU to receive medical treatment for their rare disease, unconditionally or depending on the country or on the medical treatment ("yes" or "it depends on the country or on the medical treatment").

Although most people living with rare diseases are willing to travel abroad to receive medical treatment, many continue to discard this option, reporting that **it is more convenient to be treated near home** (50%) or that they are **already satisfied with the treatments they receive where they live** (49%). They also see the lack of information on their rights, reimbursements, patients' safety standards in the country of treatment as valid reasons not to use cross-border healthcare (43% to 48%).

TABLE 5. For which of the following reasons would you not be willing to go to another country in the European Union to receive medical treatment [for your rare disease(s)]?[¥] n=1 878

	Percentage of people living with rare diseases (PLWRD)	Percentage in the general EU population	Countries where the modality was significantly over-represented among people living with a rare disease[†]
It is more convenient to be treated near your home	50%	49%	Spain (53%)
You are satisfied with the medical treatments you receive in your country	49%	55%	Netherlands (65%)
You do not have enough information about the availability and quality of medical treatments abroad	48%	21%	/
You have no information on patient safety and quality of care abroad	44%	20%	/
You are not aware of your rights in case things go wrong	43%	23%	Denmark (77%)
You would have issues understanding the language	43%	27%	Czech Republic (60%)
You cannot afford to receive medical treatment abroad	39%	20%	Poland (78%), Croatia (64%), Finland (58%), Portugal (43%)
You are not sure that you would be reimbursed	34%	16%	Germany (53%)
You prefer not to travel[‡]	15%	/	Belgium (22%), Italy (16%)
Your doctor advised you not to[‡]	7%	/	Romania (19%), Netherlands (14%)
I don't know	3%	4%	Greece (9%)
Other, please specify…	5%	4%	Portugal (11%)
Number of respondents	1 394	12 964	1 394

Several answers possible

[¥]Text between brackets was only included into the Rare 2030 survey and not into the Eurobarometer survey.

[†]Percentages between brackets refer to the number of people living with a rare disease who live in the country and who chose the corresponding modality. Only significant relationships are taken into account (p-value < 0.05).

[‡]These modalities did not appear in the Special Eurobarometer 425 survey and were added especially in the Rare 2030 survey.

Base: Respondents who would not at all or not always be willing to travel to another country in the EU to receive medical treatment for their rare disease ("no" or "it depends on the country or on the medical treatment").

Awareness of the right to access cross-border healthcare in the European Union (EU)

Respondents living in the European Union were given four statements about their rights to medical treatment in another EU Member State and were asked whether they were true or false.

Three of those statements were true: "You have the right to receive planned medical treatment in another country in the European Union and to be reimbursed for that treatment by national health authority or healthcare insurer", "You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union" and "Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network".

One item was false, "You cannot get a prescription from your doctor to use in another country in the European Union".

The findings show that **people living with rare diseases only have partial knowledge of their rights regarding cross-border healthcare and are less aware of these rights compared to the general EU population**. However, 86% of respondents answered correctly to at least one statement.

Contrary to the general EU population, only a minority of people living with rare diseases in the EU know that they have the right to be reimbursed for medical treatment in another EU country.

Graph 7. Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?

You have the right to receive planned medical treatment in another country in the European Union and to be reimbursed for that treatment by national health authority or healthcare insurer.

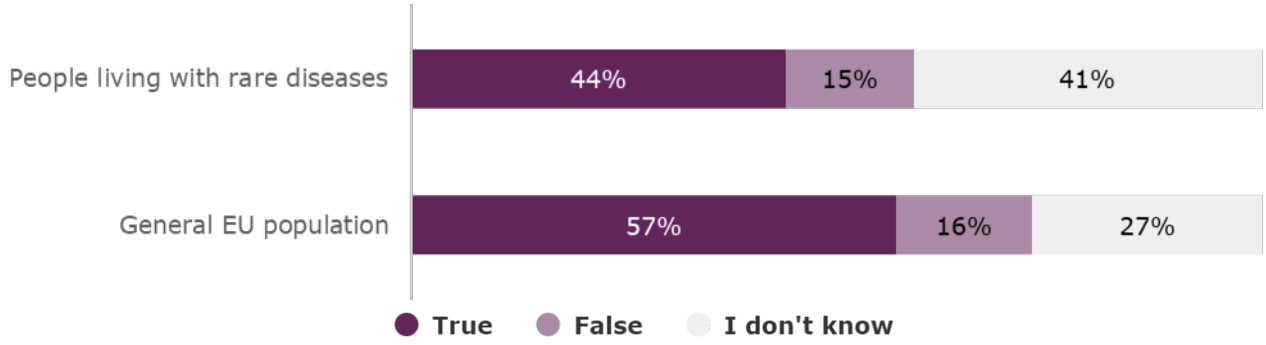


TABLE 6. You have the right to receive planned medical treatment in another country in the European Union and to be reimbursed for that treatment by national health authority or healthcare insurer. n=3 365

	Percentage of respondents	Countries significantly over-represented†	Countries significantly under-represented†
True (correct answer)	44%	Sweden (59%), Hungary (55%), Italy (51%), Finland (51%)	Czech Republic (31%), Belgium (33%), Netherlands (34%), Germany (38%)
False	15%	Croatia (34%), Czech Republic (28%), Belgium (27%), Poland (27%)	Finland (7%), Spain (11%), Italy (12%)
I don't know	41%	Spain (50%), Netherlands (48%)	Austria (19%), Italy (37%)

†Only significant relationships are taken into account (p-value < 0.05).

Only 1 in 4 respondents know that they can get a prescription form a doctor to use in another EU country.

Graph 8. Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?

You cannot get a prescription from your doctor to use in another country in the European Union.

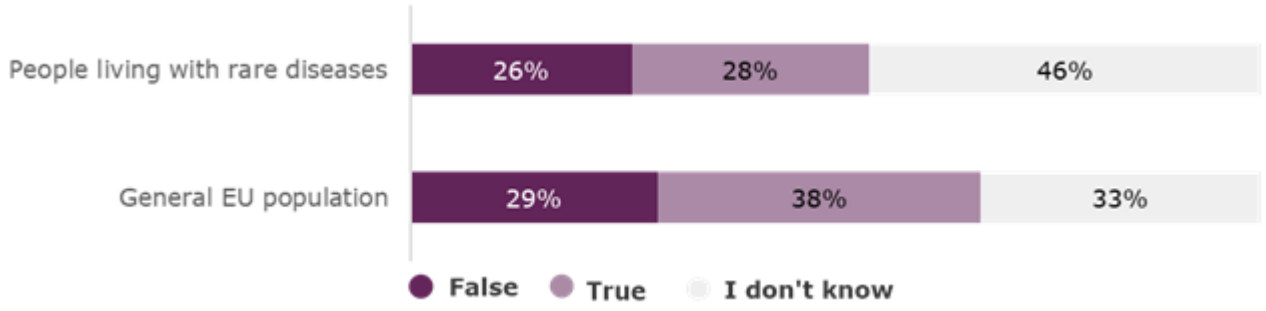


TABLE 7. You cannot get a prescription from your doctor to use in another country in the European Union. n=3 365

	Percentage of respondents	Countries significantly over-represented†	Countries significantly under-represented†
False (correct answer)	26%	Croatia (42%), Italy (38%), Belgium (24%), Spain (32%), Germany (32%)	Finland (10%), Netherlands (15%), France (18%)
True	28%	Finland (46%), Netherlands (42%), France (32%)	Germany (15%), Spain (16%), Italy (18%)
I don't know	46%	Germany (52%), Spain (52%)	Belgium (35%)

†Only significant relationships are taken into account (p-value < 0.05).

3 in 4 respondents know that they have the right to receive a copy of their medical record when they seek healthcare in another EU country.

Graph 9. Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?

You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union.

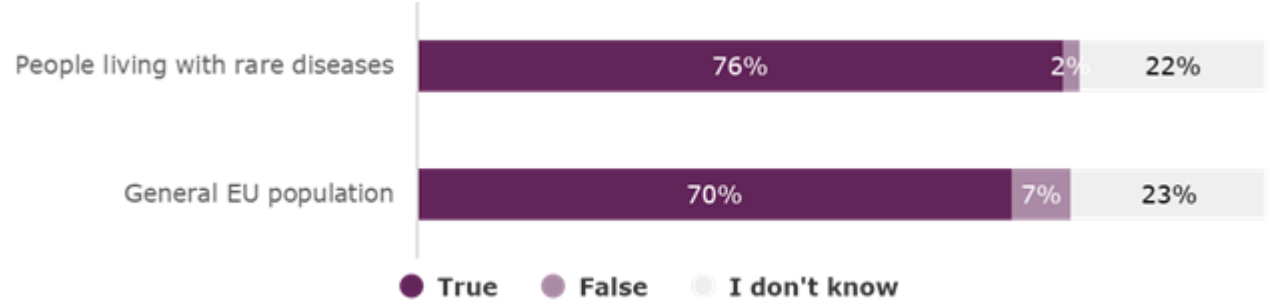


TABLE 8. You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union. n=3 365

	Percentage of respondents	Countries significantly over-represented†	Countries significantly under-represented†
True (correct answer)	76%	Hungary (89%), Czech Republic (85%), Netherlands (84%), Finland (82%)	Croatia (62%), France (64%)
False	2%	Croatia (9%)	Spain (0%)
I don't know	22%	France (34%)	Hungary (8%), Czech Republic (11%), Netherlands (16%)

†Only significant relationships are taken into account (p-value < 0.05).

43% respondents do not yet know that their doctor can ask for help in treating them locally by contacting a European Reference Network (ERN).

Graph 10. Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?

Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network.

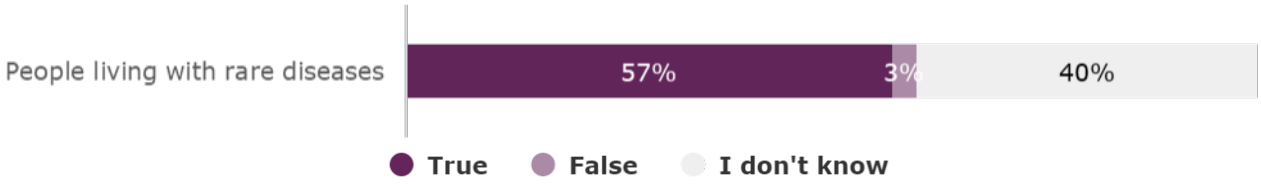


TABLE 9. Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network. n=3 365

	Percentage of respondents	Countries significantly over-represented†	Countries significantly under-represented†
True (correct answer)	57%	Austria (81%), Germany (65%), Netherlands (64%), Belgium (63%)	Poland (29%), Croatia (38%)
False	3%	Croatia (16%), Poland (11%)	France (1%)
I don't know	40%	Poland (61%), 55% (Romania), Finland (47%), France (45%)	Austria (17%), Germany (32%), Netherlands (34%)

†Only significant relationships are taken into account (p-value < 0.05).

3. 6 Remote consultations and cross-border healthcare are means to access higher quality care and better care coordination

In 2017, 2 in 3 people living with rare diseases reported that they visited different health, social and local services in a short period of time, and that they felt that these services badly communicated between each other[14]. As a consequence, when openly describing the changes they want to see for their rare disease by 2030, respondents extensively asked for better access to healthcare and especially for better care coordination[15]. **Remote consultations and access to cross-border healthcare then only appear as a means to receive better quality and more multidisciplinary care**.

> Provide access to professionals, quality healthcare, medicines, psychological and social assistance in the home country and in other countries, if it is in the better interest of the patient. **Rare disease carer, Bosnia**

[14] Rare Barometer, *Juggling care and daily life.* op. cit.

[15] See annex 2 describing the text analysis of the open question "Please describe here, as precisely as possible, what changes you want to see for your rare disease within the next 10 years".

4. Research for all rare diseases

Respondents were asked to express their opinion on how financial resources available for research across all rare diseases should be distributed based on four criteria: available body of knowledge on the scientific mechanisms of the diseases; available curative or symptomatic treatments; severity of the diseases (burden of the diseases for patients, their family and society); number of people living with the diseases (prevalence/incidence).

In order to avoid forcing them to choose between diseases and/or criteria, respondents were asked to say whether they agreed or disagreed with several statements related to each of those four criteria. Answers are quite complicated to analyse because most respondents agreed with all the statements proposed, even when those statements were contradicting each other. This could be interpreted as rare disease patients' and carers' willingness **not to choose between rare diseases** or, in other words, as their willingness to **maintain equity between all rare diseases in terms of research and development**.

Yet, some rare disease areas are historically disregarded during the development and the launch of orphan drugs on the market[16]. Prioritising research in those areas may help achieve greater focus for these disease areas and restore the equilibrium. Especially, **respondents agree that it is important to have research studies conducted on rare diseases for which no curative or symptomatic treatments exist yet (81%), for which knowledge of the scientific mechanisms is scarce or inexistent (81%), for rare diseases with very high associated costs for the patient or for society (71%) or with very low prevalence/incidence (68%).**

> I would like public-private partnership programs in Europe to have a very clear focus on rare diseases, as these as a whole represent a health economic challenge for all of Europe, and the vast majority of rare diseases urgently require intensive research. **Rare disease carer, Austria**
>
> My disease is extremely rare as there are only 9 people living with this disease today, me included. I would like to see the creation of a worldwide network of patients (patient database) as a prerequisite to launch clinical trials and to improve research and diagnosis of my disease. **Rare disease patient, Slovakia**
>
> In the case of hereditary diseases, I would like that by 2030, as soon as the patient expresses the desire, s/he is diagnosed as quickly as possible in terms of genetic research to be able to have immediate access to follow-up and support throughout his degenerative journey. **Rare disease patient, France**
>
> Research for the other 20% - non-genetic, adult onset rare diseases. **Rare disease patient, Austria**
>
> More being done for research. There should be more money available! **Rare disease patient, Germany**

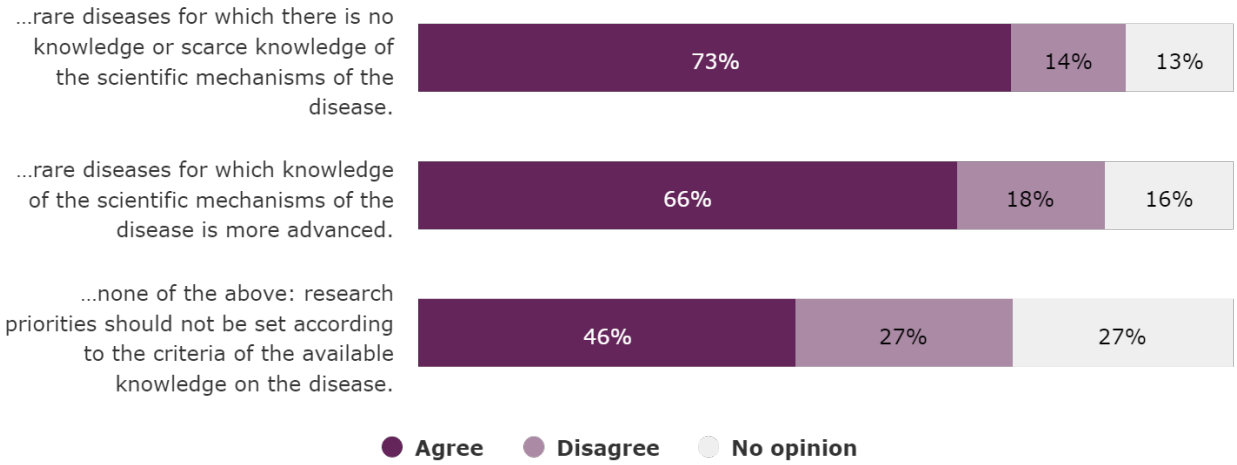
[16] Miraldo M., Sassi F., Shaïkh M., Simmons B., Vrintten C., *The development and market launch of orphan drugs from 1980-2019: q quantitative analysis. RARE2030 Foresight study, Work Package 5*, December 2020.

Knowing that financial resources for research on rare diseases are limited, research funding agencies and organisations might set priorities based on four criteria: *available knowledge on the disease, available treatments, severity of the disease and number of people living with the disease*.

The next few questions will be about priorities for RESEARCH ACROSS ALL RARE DISEASES regarding each of those criteria.

Graph 14. AVAILABLE BODY OF KNOWLEDGE (n=1 956)

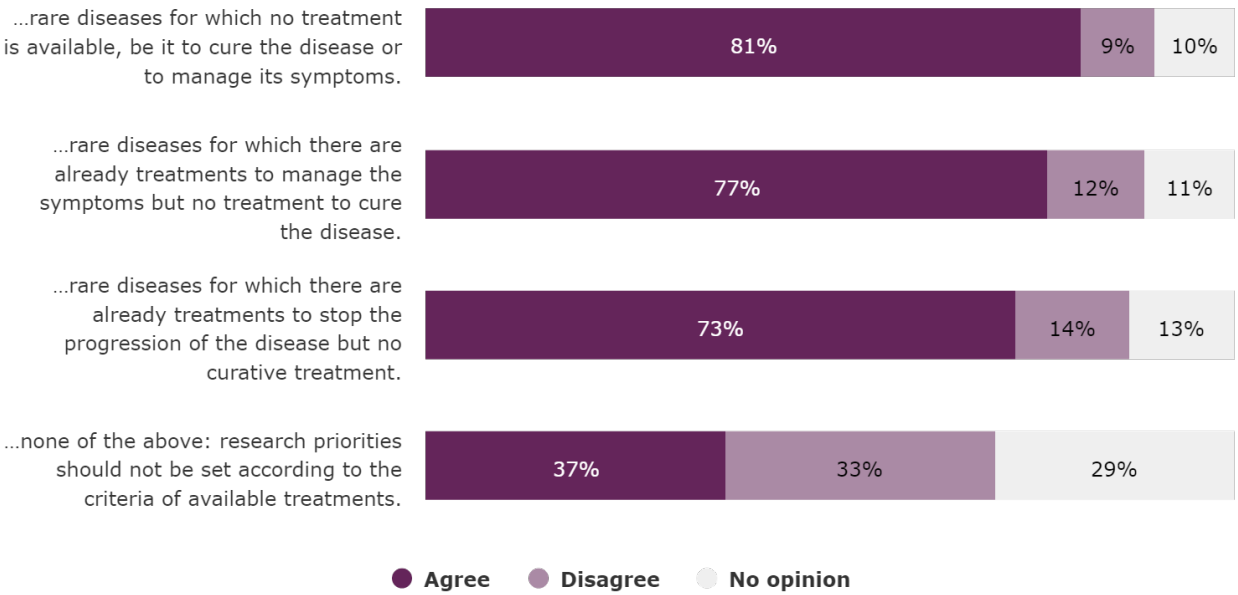
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



Some modalities have been grouped:
- Agree = strongly agree + agree
- Disagree = strongly disagree + disagree

Graph 15. AVAILABLE TREATMENTS (n=2 029)

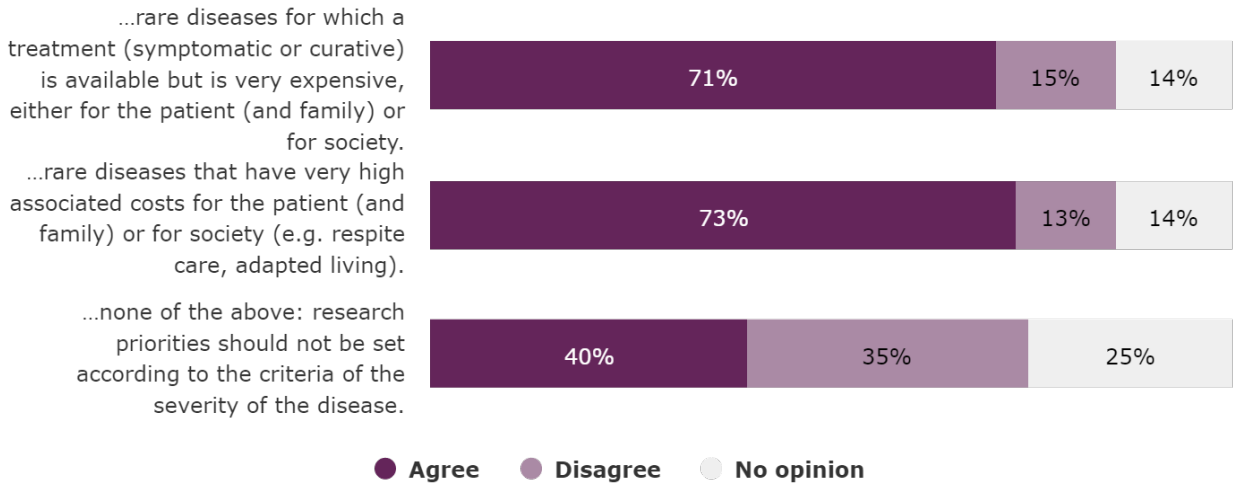
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



Some modalities have been grouped:
- Agree = strongly agree + agree
- Disagree = strongly disagree + disagree

Graph 16. SEVERITY OF THE DISEASE (BURDEN OF THE DISEASE – n=1 973)

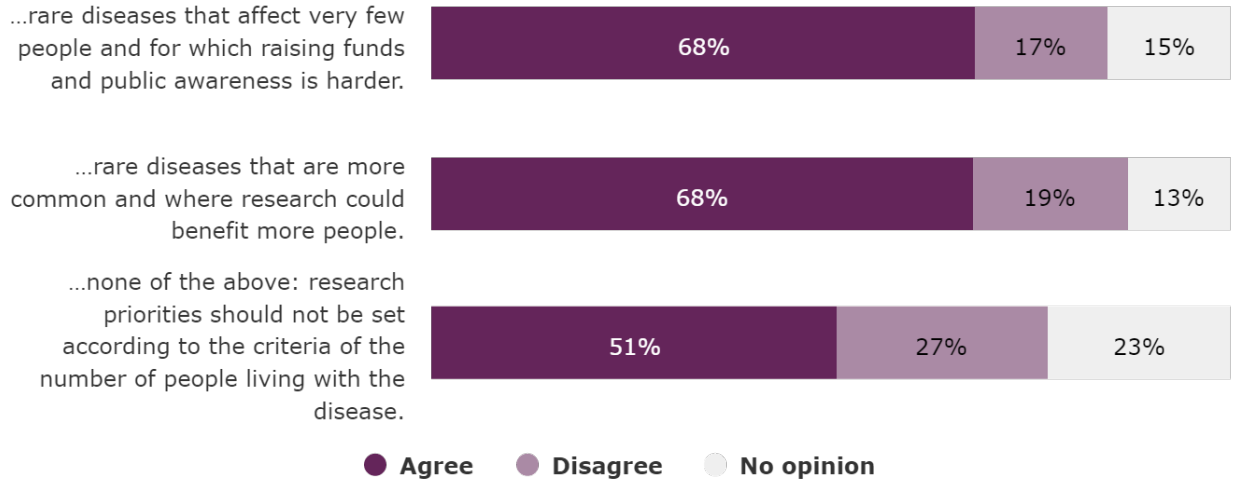
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



Some modalities have been grouped:
- Agree = strongly agree + agree
- Disagree = strongly disagree + disagree

Graph 17. NUMBER OF PEOPLE LIVING WITH THE DISEASE (PREVALENCE – n=1 977)

Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



Some modalities have been grouped:
- Agree = strongly agree + agree
- Disagree = strongly disagree + disagree

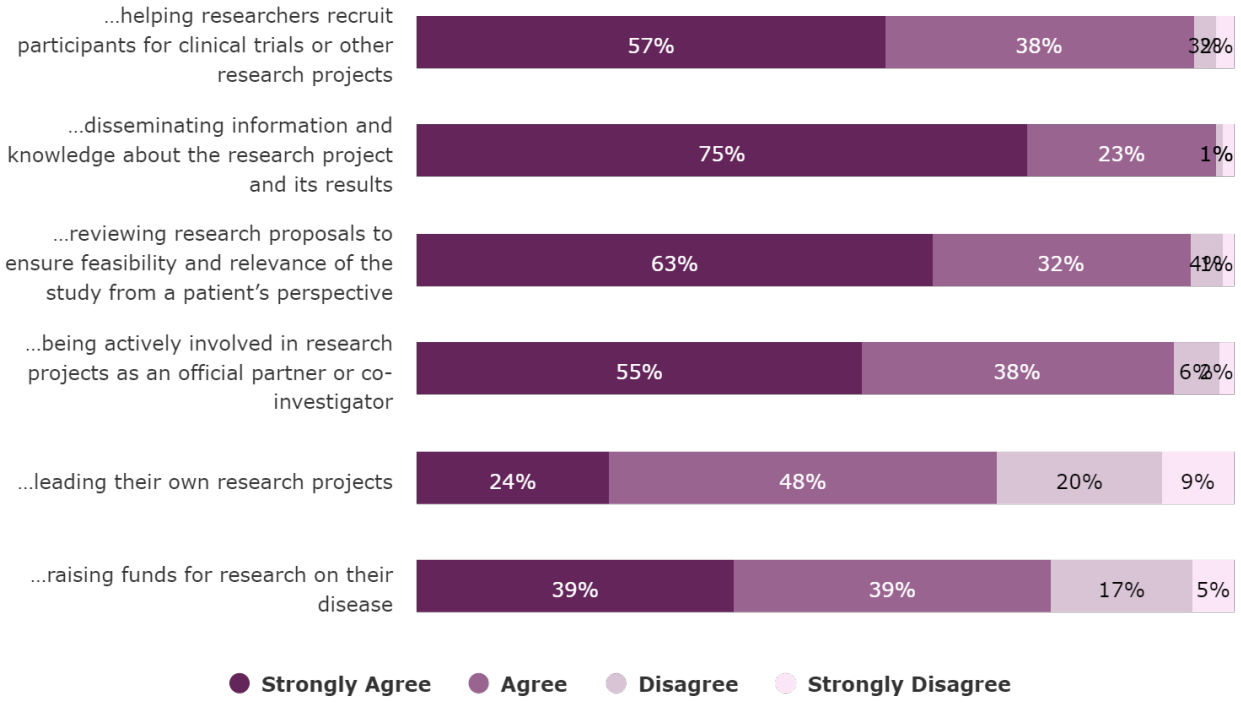
Patient organisations are willing to be involved in research for rare diseases

Only 18% people living with rare diseases have participated in research to develop treatments and therapies. This lack of patient participation in research is mainly due, according to patients and carers, to the lack of public and private funding and to the small patient population[17]. To increase research opportunities, **a majority of patient representatives are very willing to be proactively involved in the research process** (see graph 18), as defined by the EJP-RD short guide on patient partnership in rare disease research projects (see Table 10). They are also in support of patient organisations to **lead their own research projects** (70%) and **raise funds for research** on their disease (78%).

In order to be fully involved in research, a majority of patient organisations need financial support, human resources and knowledge or capabilities that they could acquire with training and capacity building (see graph 19).

> Patients and patient organizations should be involved when researching the disease (research subject, methodology, implementation ...). **Rare disease patient, Germany**
>
> That professionals and parents/patients dialogue more and that families are more involved in new developments and that information is better disseminated. **Rare disease carer, Luxembourg**
>
> We need a lot of research through clinics, doctoral theses, studies - also internationally please! Recognition of the work in the self-help associations and groups and the increased involvement of these in the supply processes. **Rare disease patient representative, Germany**

Graph 18. As a patient representative, do you think that PATIENT ORGANISATIONS should contribute in research for rare diseases by... (n=252*)



**Only patient representatives.*
Answers to the modality "no opinion" are excluded from the analysis.

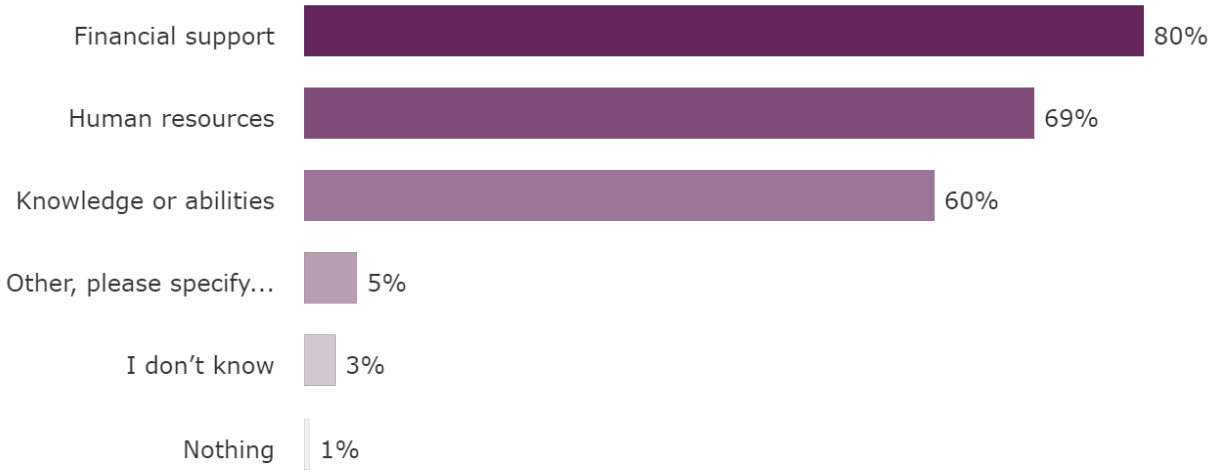
[17] Rare Barometer, *Rare disease patients' participation in research*, February 2018.

Table 10. Definition of patient partnerships according to the EJP-RD Short Guide and link with the questions of the Rare 2030 survey[18]

Concept of patient partnership	Definition of the concept	Type of patient implication	Corresponding questions in the Rare 2030 survey
Patient participation	Patients contribute to the recruitment of patients for the study or as participants themselves.	Passive	Helping researchers recruit participants for clinical trials or other research projects.
Patient engagement	Patients review research proposals to ensure feasibility and relevance of study from the patient's perspective. Patients design and/or co-create materials for study participants or for communication about the research study and its results ensuring information accessible to all.	Active	Disseminating information and knowledge about the research project and its results. Reviewing research proposals to ensure feasibility and relevance of the study from a patient's perspective.
Patient involvement	Patient as official partner / co-investigator: identify patient needs, highlight new research directions, design, develop, co-write research proposals, implement research; contribute to interpretation and findings.	Proactive	Being actively involved in research projects as an official partner or co-investigator.

Patient participation, patient engagement and patient involvement form a continuum of increasing levels of active and meaningful partnership.

Graph 19. In order to be fully involved in research for the rare disease(s) you represent, what type of support would your organisation need most? Several answers possible (n=252*)



**Only patient representatives.*

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Annex 1: questionnaire of the Rare 2030 Survey

TEXT IN ORANGE WON'T BE DISPLAYED IN THE QUESTIONNAIRE BUT FEATURED IN THE SOFTWARE SYSTEM

TEXT IN BLUE COMES FROM OTHER SURVEYS AND COULD BE USED FOR COMPARISON (IN THAT CASE IT SHOULD NOT BE CHANGED)

TEXT IN PURPLE IS RARE BAROMETER FRAMEWORK AND SHOULD NOT BE CHANGED FOR COMPARISON PURPOSES

As a reminder, we agreed to:

- Whenever possible, propose statements for respondents to give their opinion on a scale similar to "strongly disagree / partially disagree / partially agree / strongly agree" or others (preferably with an even number of modalities).
- Take into account differences between times of crisis and others.
- Tackle the place of patient organisations in the future of rare diseases.

In this proposition, **39 questions in total** *(= max number if all sociodemographic questions, patient representative and EU citizen)*:

- 4 preliminary questions for further filters (status, country & patient journey)
- 5 questions on remote health care
- 3 questions on cross-border health care / + 1 question only for EU citizens (=90-95% respondents)
- 3 questions on research / + 6 questions only for patient representatives (=5-10% respondents)
- 2 questions on prenatal & new born screening
- 1 open question
- 8-14 sociodemographic questions - only to respondents who are not registered in RBV – *these questions can't be removed*

Patients (EU): 27-33 questions
Patient representatives (EU): 33-39 questions
Non-EU citizens: minus 1 question
RBV participants: minus 8-14 questions

PRESENTATION PAGE OF THE SURVEY

What should the future of rare diseases look like?

Give us your opinion on the possible ways to shape the future of rare diseases regarding your access to health care, medical and social research as well as early diagnosis for children.

This survey will take around 15 minutes to complete and will help EURORDIS-Rare Diseases Europe, a non-profit and non-governmental alliance of 900+ patient organisations, to improve the lives of people with rare diseases.

We will share the overall results of the survey with you and communicate them (without communicating your individual responses) to decision-makers. Your contact details and any written records of your responses during the study will be kept in secure storage which only the research team can access.

This survey is part of the Rare 2030 Foresight Study. For more information please visit rare2030.eu

If you have any questions while taking part in this survey, you can contact rare.barometer@eurordis.org

PRELIMINARY QUESTIONS

Q1. How would you like to answer this questionnaire? In your capacity as a… *(several answers possible)*

- ☐ patient
- ☐ patient representative
- ☐ Parent of a child living with a rare disease
- ☐ Grandparent of a person living with a rare disease
- ☐ Spouse of a person living with a rare disease
- ☐ Uncle/aunt of a person living with a rare disease
- ☐ Sibling of a child or adult living with a rare disease
- ☐ Other, please specify

carer category

Q2. In which country do you live? [select one country in the list of world countries]

Q3. Are you or te person you care for: *Patient journey*	Yes	No	Not applicable
Diagnosed	☐	☐	☐
Currently receiving care/treatment to cure or to significantly stabilise the progression of the rare disease	☐	☐	☐
Currently receiving care/treatment to manage the symptoms of the rare disease	☐	☐	☐
Receiving psychological or emotional support to help deal with the rare disease	☐	☐	☐
In contact with a group of patients or a patient organisation	☐	☐	☐

ONLY TO THOSE WHO ANSWERED "NO" TO "RECEIVING TREATMENT OR CARE TO CURE OR STABILISE THE DISEASE", OR TO "MANAGE SYMPTOMS" AT Q3

Q4. You or the person you care for are not receiving treatment for the rare disease because: ***Patient journey 2***

- ☐ The treatment was stopped
- ☐ No treatment exists for the disease
- ☐ Existing treatments are not accessible in your country
- ☐ Existing treatments are too expensive
- ☐ Other, specify

REMOTE HEALTH CARE

Remote healthcare services, such as virtual consultations and electronic prescriptions, have been developing in daily life and are playing a crucial role during the COVID-19 pandemics.

The next few questions will be about how you are willing to use these remote healthcare services within the next 10 years.

Q5. Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app, etc.):	YES all the time	YES from time to time	YES but only if there is no other option (like in times of crisis)	NO	I don't know
With a general practitioner	☐	☐	☐	☐	☐
With a specialist	☐	☐	☐	☐	☐
With a doctor specialised in your rare disease who works in another country.	☐	☐	☐	☐	☐
For consultations where you can discuss your disease with several healthcare professionals (for instance with specialists and your general practitioner)	☐	☐	☐	☐	☐
If you already know or have already met face to face with the professional you would be having a consultation with	☐	☐	☐	☐	☐
If you do not already know or have not already met face to face with the professional you would be having a consultation with	☐	☐	☐	☐	☐

Q6. Do you think that remote consultations are appropriate for…	Not at all appropriate	Not very appropriate	Fairly appropriate	Very appropriate	No opinion
…giving a diagnosis	☐	☐	☐	☐	☐
…physiotherapy sessions	☐	☐	☐	☐	☐
…blood tests or other types of clinical tests or analysis that can be done at home	☐	☐	☐	☐	☐
…psychological or emotional support like sessions with a psychologist or support group	☐	☐	☐	☐	☐

Q7. What would be the 3 main obstacles for you to use remote consultations within the next 10 years?

- ☐ Technical obstacles such as difficulty accessing the internet or using your phone
- ☐ Not having met the care team prior to the remote consultation
- ☐ The care needed is not suitable for remote consultations
- ☐ Concerns about the safety of the care
- ☐ Difficulties in being reimbursed for remote consultations
- ☐ Language difficulties, when remote consultations are with a healthcare professional who does not work in your country
- ☐ Other, please specify…

Q8. Within the next 10 years, would you be willing to use the following services:	YES all the time	YES but only if there is no other option (like in times of crisis)	NO	I don't know
Prescriptions sent by email (electronic message)	☐	☐	☐	☐
Prescriptions sent by mail (posted letter)	☐	☐	☐	☐
Medicines sent by mail (package sent by your hospital, your pharmacy, a pharmaceutical company…)	☐	☐	☐	☐

Q9. Within the next 10 years, what would be the 2 main reasons for you to use…	Saving money	Saving time	Safety	Accessing higher quality care	Having no other option
…remote consultations	☐	☐	☐	☐	☐
…remote prescriptions (sent by email or by mail)	☐	☐	☐	☐	☐

CROSS-BORDER HEALTH CARE

Accessing expertise, treatment or health care in another country can sometimes be critical for rare disease patients.

The next few questions will be about your willingness to travel to another country to access health care for your rare disease.

Text in blue is coming from the 2014 Special Eurobarometer 425 on *Patients' Rights in Cross-Border Healthcare in the European Union* – percentages between brackets correspond to answers of the general EU population (Eurobarometer results)

Q10. Different wording for EU and for non-EU

WORDING FOR EU: Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease?

WORDING FOR non-EU: Within the next 10 years and OUTSIDE TIMES OF CRISES, would you be willing to travel to another country to receive medical treatment for your rare disease?

- ☐ Yes (33%)
- ☐ No (46%)
- ☐ It depends on the medical treatment or on the country (16%)
- ☐ I don't know (5%)

> **IF 'YES" OR "IT DEPENDS ON TREATMENT OR ON THE COUNTRY" TO Q10**
>
> **Q11. Different wording for EU and non-EU**
>
> **WORDING FOR EU: For which of the following reasons would you be willing to travel to another country in the European Union to receive medical treatment for your rare disease?** *Several answers possible*
>
> **WORDING FOR non-EU: For which of the following reasons would you be willing to travel to another country to receive medical treatment for your rare disease?** *Several answers possible*
>
> ☐ To receive treatment that is not available in your country (71%)
> ☐ To receive better quality treatment (53%)
> ☐ To receive treatment from a renowned specialist (38%)
> ☐ To receive treatment more quickly (34%)
> ☐ To receive cheaper treatment (23%)
> ☐ To receive treatment from a provider that is closer to home (6%)
> ☐ I don't know (2%)
> ☐ Other, please specify (2%)

IF "NO" OR "DEPENDS ON TREATMENT OR ON THE COUNTRY" TO Q10

Q12. Different wording for EU and for non-EU

WORDING FOR EU: For which of the following reasons would you not be willing to go to another country in the European Union to receive medical treatment for your rare disease? *Several answers possible*

WORDING FOR non-EU: For which of the following reasons would you not be willing to go to another country to receive medical treatment for your rare disease? *Several answers possible*

☐ You are satisfied with the medical treatments you receive in your country (55%)
☐ It is more convenient to be treated near your home (49%)
☐ You would have issues understanding the language (27%)
☐ You are not aware of your rights in case things go wrong (23%)
☐ You do not have enough information about the availability and quality of medical treatments abroad (21%)
☐ You cannot afford to receive medical treatment abroad (20%)
☐ You have no information on patient safety and quality of care abroad (20%)
☐ You are not sure that you would be reimbursed (16%)
☐ I prefer not to travel
☐ My doctor advised me not to
☐ I don't know (4%)
☐ Other, please specify (4%)

ONLY TO RESPONDENTS LIVING IN EU27

Q13. Here are some statements related to health care received in another country of the European Union. For each of the following, could you please say whether you think it is true or false?	True	False	I don't know
You have the right to receive planned medical treatment in another country in the European Union and to be reimbursed for that treatment by national health authority or healthcare insurer. *True*	☐	☐	☐
You cannot get a prescription from your doctor to use in another country in the European Union. *False*	☐	☐	☐
You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union. *True*	☐	☐	☐
Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network. *True*	☐	☐	☐

MEDICAL AND SOCIAL RESEARCH

Medical and social research can greatly improve the quality of life of people living with a rare disease.

The next few questions will be about what areas should research focus on to help you live with your rare disease in the future.

Q14. Do you think it is possible and realistic that within 10 years, you or the person you care for…	This is already the case today	YES and this is not yet the case	NO	I don't know
…could be cured from the rare disease: total remission, no symptoms.	☐	☐	☐	☐
…could have their rare disease stabilised: the rare disease would still be present but symptoms would not be progressing.	☐	☐	☐	☐
…could manage the symptoms of the rare disease even if they are still progressing.	☐	☐	☐	☐
…could be supported to manage the psychological or emotional aspects of the rare disease.	☐	☐	☐	☐

...would not be limited by the rare disease to handle routine needs, such as everyday household chores, doing necessary business, shopping or getting around for other purposes. Based on CDC's HRQOL questionnaire[19]	☐	☐	☐	☐
...could take part in education on equal footing with others (adapted transport, accessibility, adapted schooling measures if needed).	☐	☐	☐	☐
...could access adapted and accessible employment as well as flexible work arrangements (adjustment of working hours, working remotely).	☐	☐	☐	☐
...would not be discriminated against due to their rare disease or due to their disabilities, in the various aspects of their daily life.	☐	☐	☐	☐

Q15. **Different wording for patients and for patient representatives; randomised order for answers**

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

3 answers maximum

WORDING FOR PATIENT REPRESENTATIVES: **Within the next 10 years, the 3 top priorities to improve care for PATIENTS LIVING WITH THE RARE DISEASE(S) YOU REPRESENT would be access to:**

3 answers maximum

- ☐ Better diagnosis of the disease
- ☐ Better prevention of the disease
- ☐ Better social recognition of the disease
- ☐ Consultations with healthcare professionals specialised in the disease
- ☐ Better coordination between all healthcare professionals involved in the care of the disease
- ☐ Health care that would help ease the symptoms of the disease
- ☐ Better coordination between healthcare professionals and social care professionals (social care, education, etc.)
- ☐ Treatments and/or therapies that do not yet exist
- ☐ Existing treatments that are not yet available in your country
- ☐ Existing treatments that are still too expensive for you
- ☐ Psychological support
- ☐ Other, please specify

[19] https://www.cdc.gov/hrqol/hrqol14_measure.htm

ONLY TO THOSE WHO ANSWERED "SYMPTOMATIC TREATMENTS" AT Q15

Q16. Different wording for patients and for patient representatives

WORDING FOR PATIENTS: Within the next 10 years, in order to help ease the symptoms of your rare disease, you would need access to:

WORDING FOR PATIENT REPRESENTATIVES: Within the next 10 years, in order to help ease the symptoms of the disease(s), PATIENTS LIVING WITH THE RARE DISEASE(S) YOU REPRESENT would need

access to:

- ☐ Medicines
- ☐ Physiotherapy
- ☐ Diet and physical exercises adapted to the disease(s)
- ☐ Meditation and/or other related techniques (e.g. breathing techniques, mindfulness meditation).
- ☐ Other, please specify

ONLY TO PATIENT REPRENSENTATIVES

Wording of modalities of Q12 and Q13 are based on EJP's "Short guide on patient partnerships in RD research projects".

Q17. As a patient representative, do you think that patient organisations should contribute in research for rare diseases by…	Strongly disagree	Fairly disagree	Fairly agree	Strongly agree	No opinion
…helping researchers recruit participants for clinical trials or other research projects [participation]	☐	☐	☐	☐	☐
…disseminating information and knowledge about the research project and its results [engagement]	☐	☐	☐	☐	☐
…reviewing research proposals to ensure feasibility and relevance of the study from a patient's perspective [engagement]	☐	☐	☐	☐	☐
…being actively involved in research projects as an official partner or co-investigator [involvement]	☐	☐	☐	☐	☐
…leading their own research projects	☐	☐	☐	☐	☐
…raising funds for research on their disease	☐	☐	☐	☐	☐

Q18. In order to be fully involved in research for the rare disease(s) you represent, what type of support would your organisation need most? *(several answers possible)*

- ☐ Financial support
- ☐ Human resources
- ☐ Knowledge or abilities
- ☐ Nothing
- ☐ I don't know
- ☐ Other, please specify…

Knowing that financial resources for research on rare diseases are limited, research funding agencies and organisations might set priorities based on four criteria: *available knowledge on the disease*, *available treatments*, *severity of the disease* and *number of people living with the disease*.

The next few questions will be about priorities for RESEARCH ACROSS ALL RARE DISEASES regarding each of those criteria.

Q19. AVAILABLE KNOWLEDGE ON THE DISEASE **Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…**	Strongly disagree	Fairly disagree	Fairly agree	Strongly agree	No opinion
…rare diseases for which there is no or scarce knowledge of the scientific mechanisms of the disease.	☐	☐	☐	☐	☐
…rare diseases for which knowledge of the scientific mechanisms of the disease is more advanced.	☐	☐	☐	☐	☐
…none of the above: research priorities should not be set according to the criteria of the knowledge on the disease.	☐	☐	☐	☐	☐

Q20. AVAILABLE TREATMENTS **Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…**	Strongly disagree	Fairly disagree	Fairly agree	Strongly agree	No opinion
…rare diseases for which no treatment is available, be it to cure the disease or to manage its symptoms.	☐	☐	☐	☐	☐
…rare diseases for which there are already treatments to manage the symptoms of the disease but no curative treatment.	☐	☐	☐	☐	☐
…rare diseases for which there are already treatments to stop the progression of the disease but no curative treatment.	☐	☐	☐	☐	☐
…none of the above: research priorities should not be set according to the criteria of available treatments.	☐	☐	☐	☐	☐

Q21. SEVERITY OF THE DISEASE **Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…**	Strongly disagree	Fairly disagree	Fairly agree	Strongly agree	No opinion
…rare diseases for which a treatment (symptomatic or curative) is available but is very expensive, either for the patient (and family) or for society.	☐	☐	☐	☐	☐
…rare diseases that have very high associated costs for the patient (and family) or for society (e.g. respite care, adapted living)	☐	☐	☐	☐	☐
…none of the above: research priorities should not be set according to the criteria of the severity of the disease.	☐	☐	☐	☐	☐

Q22. NUMBER OF PEOPLE LIVING WITH THE DISEASE Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…	Strongly disagree	Fairly disagree	Fairly agree	Strongly agree	No opinion
…rare diseases that affect very few people and for which raising funds and public awareness is harder.	☐	☐	☐	☐	☐
…rare diseases that are more common and where research could benefit more people.	☐	☐	☐	☐	☐
…none of the above: research priorities should not be set according to the criteria of the number of people living with the disease.	☐	☐	☐	☐	☐

EARLY DIAGNOSIS FOR CHILDREN

New technologies are being developed at a rapid pace and could be used to diagnose some rare diseases in children at a very early stage, improving their treatment options and quality of life.

The next few questions will be about your opinion on the use of those technologies in the future. Your answers will help EURORDIS-Rare Diseases Europe voice the opinion of the rare disease community on this delicate matter.

Q23. In your opinion, in order to diagnose rare diseases at an early stage, should tests for rare diseases be performed…	YES with the informed consent of the parents	YES as part of a compulsory health programme	NO	I don't have an opinion
…around the time of child's conception (e.g. during in vitro fertilisation, genetic counselling)	☐	☐	☐	☐
…during pregnancy (e.g. medical imaging, tests on amniotic fluid…)	☐	☐	☐	☐
…at child's birth (e.g. blood tests, genetic screening…)	☐	☐	☐	☐

Many countries have developed compulsory health programmes to test newborns and diagnose rare diseases at an early stage. However, there are still discussions about what types of diseases should be tested as part of those programmes.

Q24. In your opinion, for which conditions should newborns be tested at birth as part of a compulsory health programme? *(several answers possible)*

- ☐ Conditions that COULD BE prevented and/or treated
- ☐ Conditions that CANNOT YET BE prevented and/or treated
- ☐ Conditions for which appropriate disease management (diet, education…) can improve health and quality of life
- ☐ Conditions that may affect other family members (inherited conditions)
- ☐ Conditions that may be developed later in life
- ☐ None of the above
- ☐ I don't have an opinion
- ☐ Other, please specify

HAVE YOUR SAY

Q25. Please describe here, as precisely as possible, what changes you want to see for your rare disease within the next 10 years: *Open question*

SOCIODEMOGRAPHIC INFORMATION

This information will allow us to analyse the data in more depth

IF RESPONDENTS ARE NOT REGISTERED IN THE RARE BAROMETER VOICES DATABASE

Q26. How old are you?
- Under 15
- 15-17
- 18-24
- 25-34
- 35-49
- 50-64
- 65 or older

If you are younger than 18 years old, your parent's consent is needed for your registration. Please ask your parents to respond to the following questions.

TO ALL RESPONDENTS WHO ARE NOT REGISTERED YET

Q31. Are you:
- Female
- Male

Q32. Please select in the following list the centre or hospital in which you or the person you look after is treated *(Several answers possible).* **If you cannot find the relevant hospital, please skip this question.** [select a hospital in the list of hospitals attached to an ERN, by country]

IF RESPONDENTS ARE DIAGNOSED

Q33. How many rare diseases are you living with?
- 1
- 2
- 3
- 4
- 5 or more

Q34. Which rare disease are you living with?

Please start typing the disease below, select it, and then click outside of the list to proceed to the next page. If you cannot find the rare disease in this list below, please type it yourself in the "Cannot find the disease" box.

MESSAGE AFTER SAVING THE QUESTIONNAIRE

Thank you for participating in this survey!

To make the voice of rare disease patients stronger, it would be great to have you as an ambassador so to help us to spread the word about Rare Barometer Voices!

Share the dedicated webpage with people living with a rare disease and patient organisations around you: eurordis.org/voices

In accordance with the Data Protection laws, you can access, modify, or suppress your information at any time. If you want to exercise this right and obtain information about your data, please contact rare.barometer@eurordis.org

Regarding access to cross-border health care for European Union citizens, please note that:

- To know more about your right to receive planned medical treatment in another European Union country and to be reimbursed for that treatment, please go to your national contact point[20].
- You can get a prescription from your doctor to use in another country in the European Union.
- You have the right to receive a copy of your medical record from your doctor when you seek to receive healthcare in another country in the European Union.
- Your doctor may be able to get help in treating you locally by contacting a network of doctors specialised in specific rare disease(s) called a European Reference Network. For more information, please visit ec.europa.eu/health/ern_en

[20] https://europa.eu/youreurope/citizens/health/planned-healthcare/get-more-info/index_en.htm

Annex 2: synthesis of the textual analysis of the open question

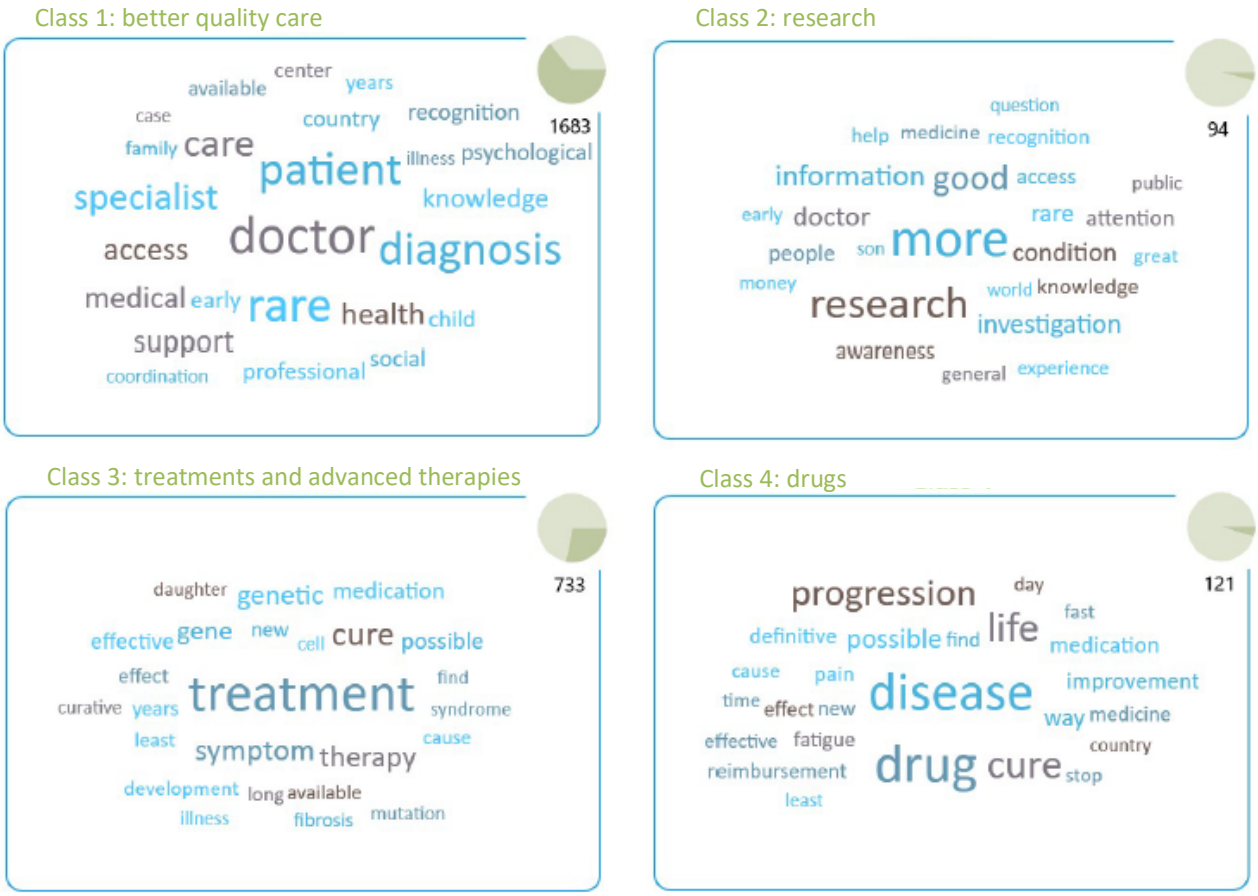
At the end of the questionnaire, respondents were asked to openly answer the following question: **please describe here, as precisely as possible, what changes you want to see for your rare disease within the next 10 years**.

Corpus of answers (55 608 words) was automatically translated into English and a synthetic textual analysis was conducted with the Sphinx software.

Graph 22. Word cloud of all responses to the open question (55 608 words)

The corpus was cut into segments of a median length of 10 words. Those segments were then grouped into four classes, depending on the similarities in the words that compose these segments.

Graph 23. Word cloud for each of the four classes of text segments



Better quality care

Respondents most frequently requested a better quality care for their rare diseases (class 1, 64% of the segments), which includes:

- First of all, a better and earlier diagnosis accompanied by psychological support, more follow-up and a treatment plan.
- Better care coordination, which could be taken care of by a person (doctor, social worker, case manager) or an expert centre, or ensured through a treatment plan and more follow-up.
- More psychological support all along the patient journey.
- Better and more frequently shared knowledge of rare diseases for patients, caregivers, healthcare professionals (general practitioners as well as specialists), social professionals and society as a whole.
- Improved education regarding their disease.

Respondents also evoked research and organisational aspects of accessing better quality care (class 2, 4% of segments):

- More investment in research for rare diseases;
- Better knowledge of rare diseases from doctors (general practitioners or specialists).
- Improving recognition of rare diseases by public authorities by adapting organisational structures to rare disease healthcare (specialised centre) and improving the overall quality of life of rare disease patients and carers (leaner bureaucracy, better working conditions).

Finding and treating the causes of rare diseases

Respondents hope that research can help find the causes and the scientific mechanisms of rare diseases (class 2, 28% of segments) in order to cure them or at least alleviate their symptoms and improve patients' quality of life. Respondents put great hopes in advanced researches and therapies such as early screening (during conception, at child's birth), pre-implantation genetic diagnosis or gene therapies.

Although less segments directly refer to medicines (class 4, 5% of the segments), respondents also rely on drugs to cure their disease, stabilise it or at least significantly improve their quality of life. To do so, they would like to access drugs that do not yet exist or that are not yet available in their country.

List of the most specific text segments for each class

List of the 20 most specific segments of class 1

64% of segments - Better quality care: diagnosis, care coordination, psychological support, better knowledge of the disease, improved education on the disease.

Provide access to professionals, quality health care, medicines, psychological and social assistance in the home country and in other countries, if it is in the better interest of the patient.

Greater specific knowledge of them; especially by health, social services and education professionals in addition to the global knowledge of society in general.

For the rare illness of Rett Syndrome, I would like my child to have more knowledge of specialists, dissemination of knowledge about Rett Syndrome, recognition of the achievements - physical and psychological - of family members.

Greater knowledge of the disease on the part of health professionals, an association that supports and informs patients, greater support for using specialists from other countries, which can be done through online consultations.

Increased timeliness of diagnosis, improved treatment options, better knowledge of all medical professionals, cure, education opportunities for patients, caregivers and medical professionals.

In addition, more attention to the psychological aspect and more support for people who suffer from several 'rare' disorders at the same time, whose specialists work in different hospitals.

Assessments and plans made nationwide in a centralized multidisciplinary care team with expertise in rare diseases, in close collaboration with local social and health care professionals.

Greater disclosure of the pathology both in the medical and social fields, so as not to have to hide under a social aspect, and for a doctor to have more easily a first help as soon as they know they have symptoms / problems.

More coordination between specialists, more involvement of specialists, access to professionals specialized in my pathology and referral centres, access to treatments for adults that are administered in the United States.

Definitely a seamless collaboration of doctors from different fields or that there would be one doctor who would bring together the studies and opinions of the doctors treating over time and contribute to the advancement of the patient's rare disease research.

Easy access to a rare disease healthcare system and low costs for people traveling from other countries, with no healthcare insurance.

Recognition of patients with financial assistance for care and psychological follow-up, help in schools that accept children with rare diseases.

Have a referral centre for my child's rare illness that provides multiple medical, psychological and equipment and support.

An early diagnosis to avoid years of diagnostic wandering like that was my case: care management: to improve; psychological help systematically offered after the diagnosis

Access to treatments and technologies already in use in the United States; greater clarity and ease of access to social benefits available to persons with disabilities; better acceptance of disability in schools and work

Recognition of society, social inclusion, recognition and government assistance, such as the provision of all treatment free of charge by SUS, financial aid for complementary treatment.

Where possible, an early diagnosis and treatment plan (if available) and care follow-up by checking the child and offering the right care and treatment options in a timely manner

Early diagnosis, specialist doctors, coordination of services and insurance agencies, psychological support, quality of life improvement

Rapid diagnostics, connection of specialists for individual diagnoses and their cooperation, information in one place regarding the possibilities of therapies and necessary services, organized support for family and caregivers.

Appropriate diagnosis: general practitioners / dermatologists referring patients to expert centres for appropriate diagnosis and treatment (currently this does not happen in some cases, with terrible consequences to the patients)

List of the 20 most specific segments of class 2

4% of segments - Better quality care: more research and a better organisational structure.

Better recognition by public authorities and better consideration in access to rights.

That more could be investigated with public money, more help to these people

Better treatment and organization of structures and departments, leaner bureaucracy and better working conditions

Faster research although research takes too long because there is no money

Investigation of new treatments; access to clinical trials; longitudinal investigation of disease effects

Greater presence of specialized centres in the territory and greater dissemination of information and awareness.

More attention must be paid to this, knowledge level in primary care is very poor.

More research into and therefore clarity about the immune system, so that it can respond better to my condition.

That there is more information on rare diseases, as well as better access to their treatments

That more doctors be trained about this disease and that doctors already in practice have an obligation to provide information about it.

Greater understanding of rare disease physician teams

More awareness in general medicine.

More knowledge on how to make life easier to lev3 with the condition.

Educating society and improving inclusion opportunities for people with disabilities

If my life expectancy is less than a year, I won't answer that kind of question.

Recognition of the patient's experience in the therapeutic strategy (in the broad sense)

More world research ...

Greater knowledge and information

Recognition and knowledge among all people

More attention to the consequences of the disease

List of the 20 most specific segments of class 3

28% of segments – Finding and treating the causes of the diseases, including through genetic research and therapies.

Would love to see the genetic aspect of treating and curing (genetic editing, gene therapy etc.) become a mainstream form of treatment ... and affordable.

We want genetic research to know the exact cause of the genetic mutation in order to be able to control and correct it to find a normal discomfort.

I am an adult, in my illness there is no cure, I just hope that in the coming years the research will make progress, for this reason all rare diseases will find a cure at least to stop the disease.

An effective treatment, not only to eliminate symptoms in severe cases, but also so that these symptoms do not appear and you can lead a normal life.

Findings about possible causes - for other parents, and treatment options against the causative genetic change or at least to alleviate the symptoms.

A genetic treatment that cures a hereditary disease or a treatment that prevents damage to the organs caused by this disease

Support research to find therapies that alleviate symptoms and it would be desirable to have complete symptom relief, for example through gene therapy

I also hope that a cure will be found for a cure and will succeed in reversing the damage of the autoimmune disease when one finds a treatment to stop the autoimmune disease.

That it is known and easier to diagnose, that it be investigated, its causes are known and how to alleviate its effects, and if a cure could be found, or at least better treatments that minimize its effects.

A possible regression of symptoms to improve the quality of life, this I would like to be possible by reducing the daily medication or a therapy that has a lasting effect.

A method capable of modifying the diseased gene: for example, the Crisp case 9 method, and a pharmacological treatment that blocks the symptoms

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 they treat us as a chronic disease, which it already is and we have access to the benefits that diseases enjoy Chronicles.

To find a cure that will CURE Myasthenia, not just treat its symptoms and by the method of trying to succeed or fail to bring the symptoms under control.

The development of treatments is primarily by medical methods and / or by the easiest possible physical methods.

Treatment plan so I know in advance when what will happen: participation in research, diagnostic and treatment research; new, more effective treatment

Disease control, innovative therapies, availability of innovative drugs, genetic therapies, social support, financial support.

To find either targeted treatment for the whole syndrome or for some individual symptoms, or at least to stop the progression of the disease.

The next step would then be genetic engineering therapy that corrects the genetic defect, ideally also in the germ cells.

That they finally find a genetically correct treatment or at least a more effective treatment than the existing one

Treatment options and cures are so important for disease that are rare but cause severe symptoms and impact quality of life.

List of the 20 most specific segments of class 4

5% of segments – Drugs to cure or stabilise the diseases, and to improve quality of life.

Faster time to market new drugs and faster reimbursement

I just wish the researchers could find a way to stabilize my son's rare disease.

Planning for the future life of our disabled adult son when we are no longer there

Finding a drug that definitely cures me and makes it unnecessary to take hormone replacement drugs every day.

If a complete cure is not possible, then at least a significant improvement in quality of life.

Have access to expensive drugs, and find more drugs to have a better quality of life

I wish it were possible to limit the damage resulting from the progression of the disease.

It would be nice if the exact cause was known and a drug to stop the continuation of the condition could be found

Finding the definitive cure for not having epileptic seizures that do not allow you to live a normal life

I wish there was a definitive cure that would stop the progression of the disease forever.

I would like a drug that helps with cystic kidneys and does not have severe side effects.

Find a drug with fewer chronic side effects.

I would like a drug (with as few side effects as possible) that can treat and improve my rare disease.

Complete healing or a drug that works longer and does not have to be taken 3 times a day.

May new drugs approved in other countries arrive in Spain to cure it!

I would like my disease (alopecia universalis) to find a workable treatment that has no side effects

Hope for a cure or prevention by being faster.

Less fatigue. Best drugs

Advances in medications that stop the disease (CF)

That a medication is found to prevent progression.

www.eurordis.org/voices

Thank you to all people living with a rare disease who participated in the survey and to Rare Barometer partners!