Rare disease patients' opinion on the future of rare diseases

A Rare Barometer survey for the Rare 2030 Foresight Study

June 2021

Rare Barometer

Rare 2030
Foresight in Rare Disease Policy

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The Rare 2030 Survey on the future of rare disease policy was conducted by EURORDIS-Rare Diseases Europe via the Rare Barometer programme. This survey is part of the Rare 2030 Foresight Study, which is 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 900 rare disease patient organisations from more than 70 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 Programme 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 gathered the input of a large group of patients, practitioners and key opinion leaders to propose policy recommendations that result in improved policy and ultimately 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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- 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 objective 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

While rare diseases were nearly invisible in the European health and care systems in previous decades, they are now recognised as a public health and human rights priority. With this recognition have come treatments, new knowledge and hope. Yet for the 30 million people in Europe living with a rare disease, challenges remain and the need for an updated policy framework that allows a true concerted approach to rare diseases across Europe has emerged.

The Rare 2030 Foresight Study was supported by the European Parliament, Commission and key opinion leaders across Europe to prepare a better future for patients and families living with a rare disease. For two years, it called upon a panel of more than 250 experts: doctors, companies, researchers, young citizens, advocates, patients, payers and policy makers. These experts reflected on remaining challenges, identified predictable trends, constructed plausible future scenarios for rare disease policy and finally drafted recommendations to reach the preferred scenario characterised by patient needs-led innovation and solidarity.

To ensure that the direct patient perspective was included in shaping final recommendations, the Rare 2030 Foresight Study took into account the experience, needs and opinion of thousands of people living with a rare disease who have contributed their opinion during the last decade through the EurordisCare and the Rare Barometer survey programmes. The Rare 2030 survey gathered the needs and priorities of **3 998 patients, carers and patient representatives** on the following topics: medical and social care; early screening to diagnose rare diseases; access to remote health care and cross-border health care; criteria to incentivize research funding; and the participation of patient organisations in research.

Results of the Rare 2030 survey on the future of rare diseases

1. Accessing treatments and better care coordination are the top priorities for people living with rare diseases

Access to treatments and therapies that do not yet exist (50%), better **coordination of health care** (44%) and **access to healthcare specialists** (33%) are pointed out as top three priorities for rare disease patients by 2030. Early diagnosis is often crucial for rare disease patients and respondents **strongly support screening for rare conditions at birth** (95%), both for **treatable conditions** (79%) and for **actionable conditions** for which appropriate disease management can improve the health and quality of life of the patient (67%). Respondents also put great hopes in advanced therapies, and especially in gene therapy.

2. Respondents do not expect to be cured from their rare disease within the next 10 years but hope to improve their quality of life by accessing integrated health care and social care

79% respondents do not expect to be cured from their rare disease within the next 10 years and only 42% of them think that they would no longer be limited by their disease to handle routine needs by then. But they believe **there is hope for them to stabilise their disease** (53%); to **manage their symptoms** (49%) with new medicines and several types of therapies; to receive appropriate **emotional support** (58%); to access **adapted employment** (44%); and **not to be discriminated because of** their rare disease (39%).

3. Accessing high quality and multidisciplinary health care is crucial for people living with a rare disease

Besides considering better coordination of care as their second priority (44%) and integrated health and social a pillar to achieve for quality of life improvements in the next 10 years, respondents usually **prefer to be treated near to their home**. But given the scarcity and geographical dispersion of healthcare experts who can treat them, they are very willing to use **remote health care, when it is appropriate for the care needed** and **when they have already met their care team**. The daily life of persons living with a rare disease often revolves around care related tasks and remote health care allows them to save time. It also allows them to access high quality care, such as a remote consultation with a specialist working in another country or a collective consultation that reunites their local physician and one or several specialists. Because accessing high quality and multidisciplinary care is a priority, persons living with a rare disease are also more willing to **travel to another country in the European Union to receive care or treatments** than the general population, even if they prefer to be treated locally.

4. A call for research that benefits every rare disease and that involves patient organisations

When asked about research priorities, respondents' answers 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 pathogenic mechanisms of the disease (81%);
- very high associated costs for the patient or for society (71%);
- very low prevalence (68%).

Patient representatives are **very willing to be proactively involved in research projects as official partners or co-investigators** (94%). They also support patient organisations to **raise funds for research** on their disease (78%) and to **lead their own research projects** (71%).

What should the future of rare diseases look like? Recommendations of the Rare 2030 Foresight Study

Thanks to the work of the panel of experts and to the thousands of patients and families who responded to EURORDIS surveys, the voice of the rare disease community is loud and clear: we want a future where everyone can live to their fullest potential, and where policies are built upon the principles of social responsibility, equity and collaboration.

After two years of consultation and dialogue, the rare disease community has identified the need for a new European policy framework for rare diseases that recognises new technological opportunities, modern values and the current healthcare policy context. This framework must:

1. A European policy framework guiding the implementation of consistent national plans and strategies, monitored and assessed by a multistakeholder body on a regular basis.
2. Earlier, faster and more accurate diagnosis of rare diseases through better and more consistent use of harmonised standards and programmes across Europe, new technologies and innovative approaches driven by patient-needs.
3. A highly specialised healthcare ecosystem, with political, financial and technical support at European and national levels, which leaves no person living with a rare disease in uncertainty regarding their diagnosis, care or treatment.
4. Guarantee the integration of people living with a rare disease in societies and economies by implementing European and national actions that recognise their social rights.
5. A culture encouraging meaningful participation, engagement and leadership of people living with a rare disease in both the public and private sectors.
6. Rare disease research maintained as a priority - across basic, clinical, translational and social research.
7. Data use and re-use optimised to improve the health and well-being of people living with a rare disease.
8. Improve the availability, accessibility and affordability of rare disease treatments, by attracting investments, fostering innovation and collaboration across countries, to address inequalities.

Existing specific European policies on orphan drugs and on European Reference Networks do not make a European policy for rare diseases. They are essential pieces, but they only refer to means and instruments: new products, networking, activities. They are not enough to achieve the kind of impact and improvement that we expect to see in 10 years' time in diagnosis, survival and quality of care for persons living with a rare disease. To achieve these goals, the rare disease community needs a holistic approach that encompasses research, medical and social care, and that considers new technological opportunities. The European strategy for rare diseases has to integrate those three elements at national and European levels. Without such an integrated approach, policies on rare diseases will stay fragmented and even with good money and good will, they will not be enough to reach the goal to leave no one behind.

To know more about the Rare 2030 recommendations, please visit: rare2030.eu/recommendations

Table of contents

1. Methodology

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 Rare 2030 survey is to develop evidence-based recommendations. 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 refers to EURORDIS previous surveys.

1. 1 Translation of the questionnaire

The questionnaire was written in English and translated in the 22 following languages by professional translators specialised 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 and Swedish. Native speakers then reviewed translations 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 health care and cross-border health care.

1. 3 Survey sample

3,998 responses were received worldwide and **3,770** in Europe:

- 54% respondents were contacted through the Rare Barometer database. The Rare Barometer database 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 around 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 health care and cross-border health care.

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

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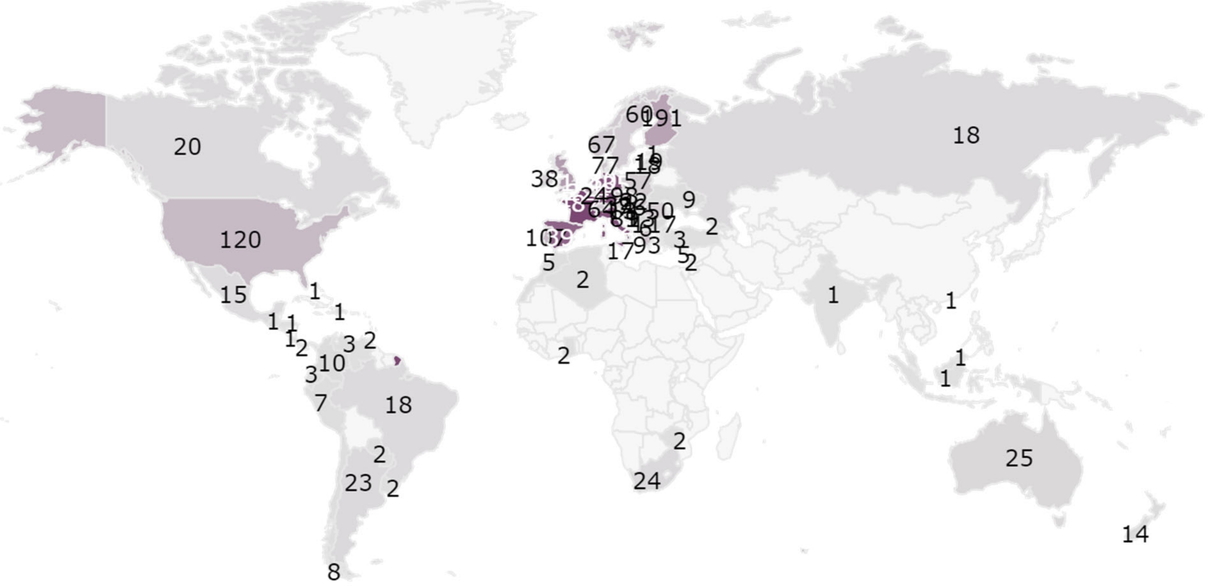
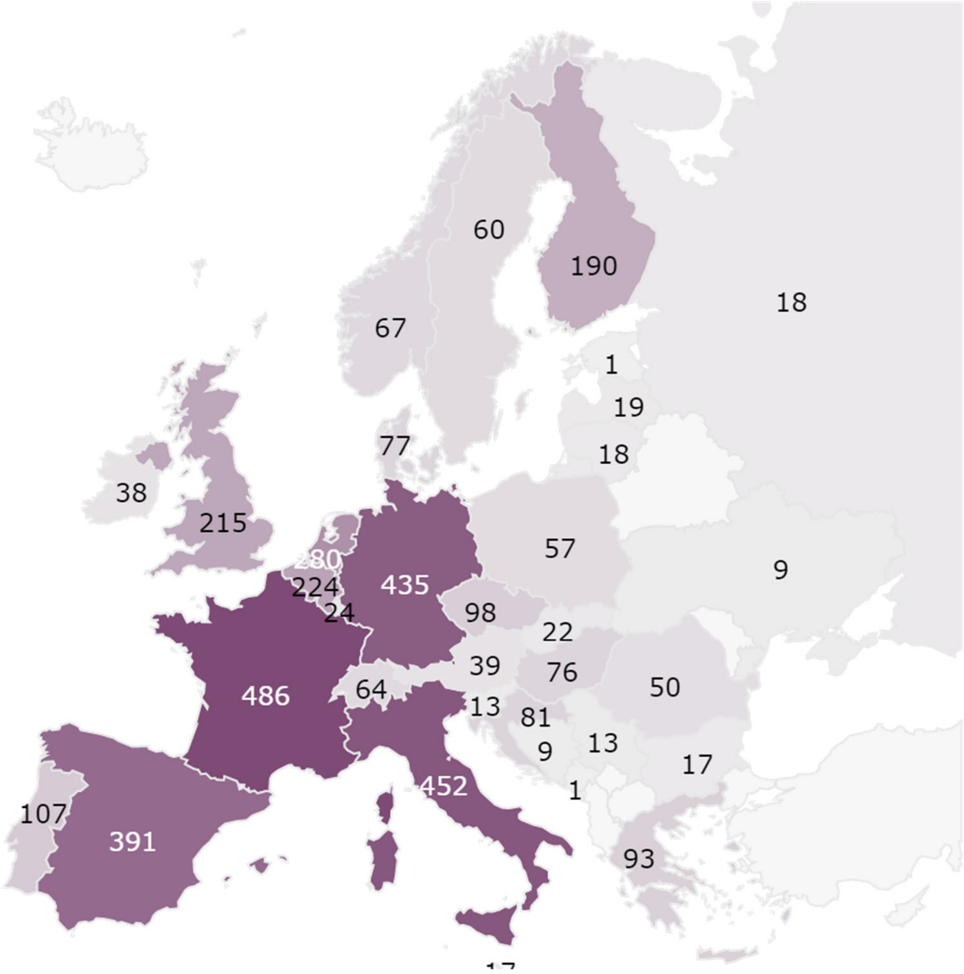
[1] https://www.eurordis.org/voices

Geographical distribution of the sample

Map 1. Number of respondents per country in the world



Map 2. Number of respondents per country in Europe



Geographical distribution of respondents in Europe corresponds in large part 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 depends 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 countries with more than 20 respondents and with 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 carers 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 carers.

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

73% 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.

TABLE 1. Composition of the Rare 2030 Survey sample (n=3998)

Type	Percentage
Respondent status *(several answers possible)*	
Patient	69 %
Carer 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	11 %
35-49 years old	39 %
50-64 years old	34 %
65 years old and above	12 %
Diagnosed	
Yes	97%
No	3%

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[2] Rare Barometer, *Juggling care and daily life. The balancing act of the rare disease community*, May 2017, p. 7.

Point prevalence

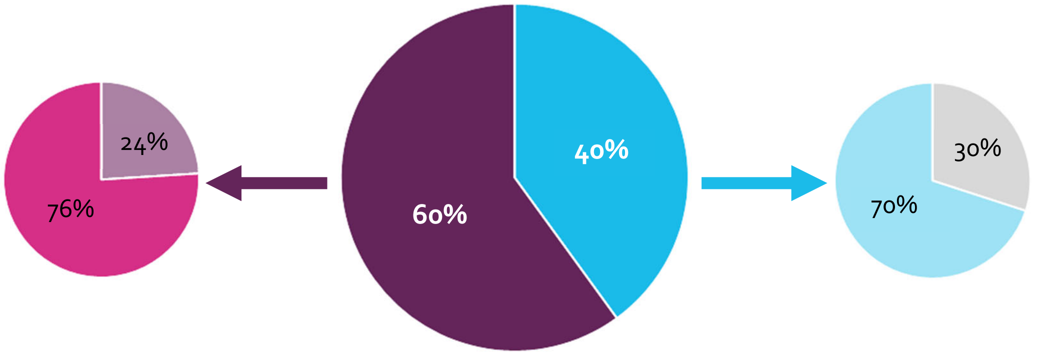
Point prevalence is the proportion of a particular population found to be affected by a given disease at a specific time. It is defined by comparing the number of people found to have a disease within the population of a country, of a larger geographical area or worldwide. It is expressed as the number of cases per 10,000 or 100,000 people.

For each respondent of the Rare 2030 survey who declared their rare disease and their country, the corresponding point prevalence was assigned using Orphanet epidemiological data, which is the most complete and up to date database on prevalence and incidence of diseases that fall into the European definition of rare conditions (less than 2 cases for 5,000 people)[3]. Specifically, for respondents of the Rare 2030 survey, point prevalence was assigned:

- in priority for the country, when data is available;
- per large geographical area (Africa, Eastern Mediterranean Asia, Europe, Latin America, North America, Oceania, South East Asia, Western Asia) if point prevalence is not known for the country;
- worldwide if point prevalence is not known for the country or for the large geographical area.

Point prevalence is known for 60% respondents of the Rare 2030 survey, of which **24% are living with a very rare disease** (less than 1 case for 100,000 people) and **76% are living with a more common rare disease** (from 2 cases for 5,000 people to 1 case for 100,000 people). Point prevalence is still unknown for 40% of respondents, of which 30% did not declare their rare disease and 70% are living with a rare disease for which epidemiological data is not yet available.

Graph 1. Point prevalence for respondents of the Rare 2030 survey (n=3,998)



● **Respondents with known point prevalence:**

● Respondents living with more common rare diseases (from 2 cases for 5,000 people to 1 case for 100,000 people)

● Respondents living with very rare diseases (less than 1 case for 100,000 people)

● **Respondents with unknown point prevalence:**

● Respondents living with a rare disease for which point prevalence is not yet known

● Respondents who did not specify 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 **978 diseases**. Rare Barometer distinguishes 23 therapeutic areas corresponding to groupings of diseases that could be treated within European Reference Networks[4].

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.

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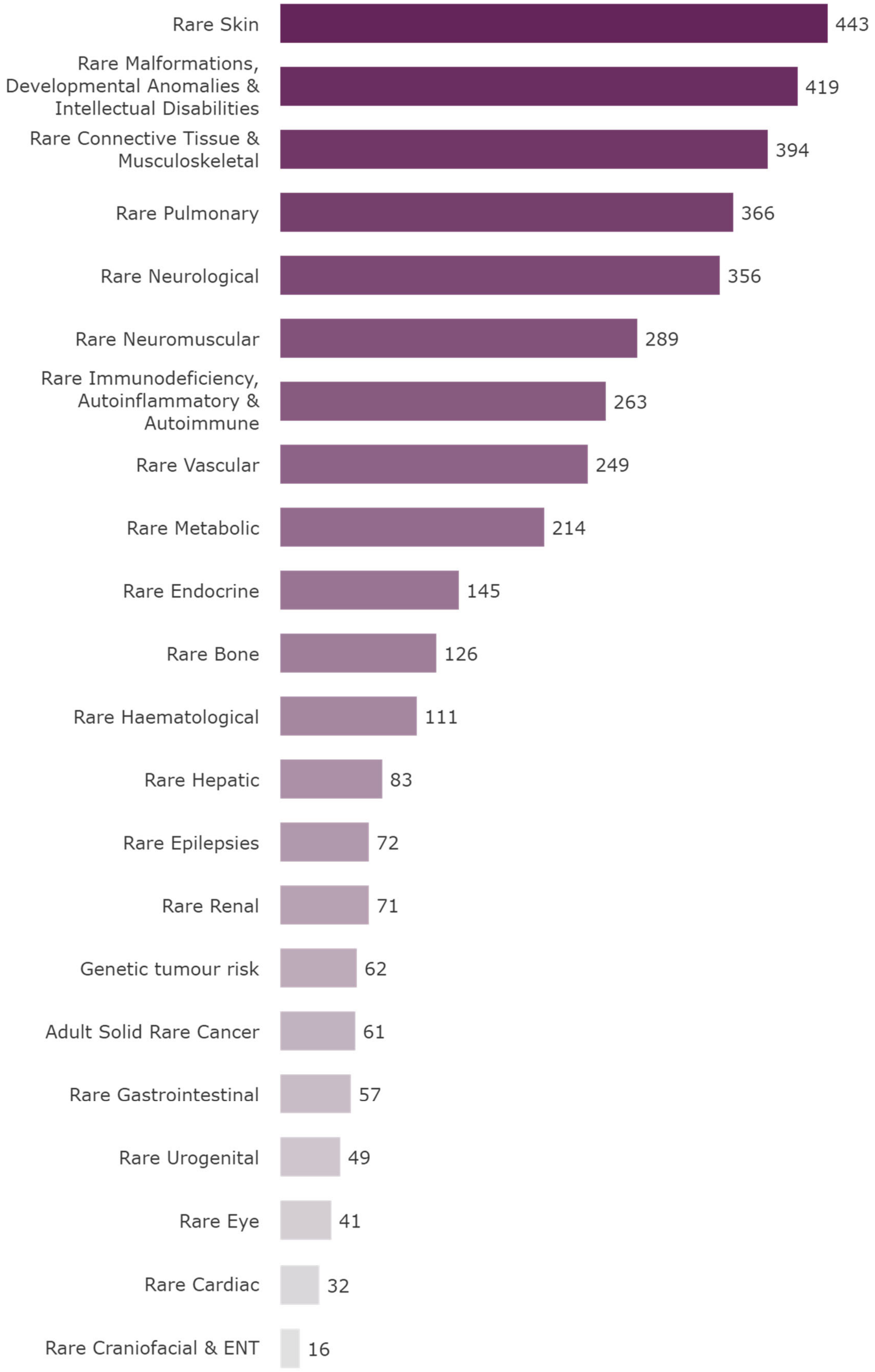
[3] http://www.orphadata.org/cgi-bin/epidemio.html; https://www.nature.com/articles/s41431-019-0508-0
[4] https://ec.europa.eu/health/ern_en

Graph 2. Number of respondents per therapeutic area (n=3,522)



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

Results presented below show that **therapeutic research remains**[5] **the priority for people living with rare diseases** for the upcoming 10 years. **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 3). Open comments show that respondents have great hopes that **advanced therapies** can help cure their disease. A small percentage of the open comments also refer to the possibilities offered by drug development.

> 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**

Respondents also call for a **better coordination of their health and social care**[6] 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[7]. In the absence of such case managers, care coordination is a burden that widely falls on the patient or on the main carer: **coordination of health care is the second priority for carers, while better coordination between healthcare professionals and social care professionals is their third priority** (Table 2).

> 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 care for my rare disease, as well as case management. **Rare disease patient, Austria**

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

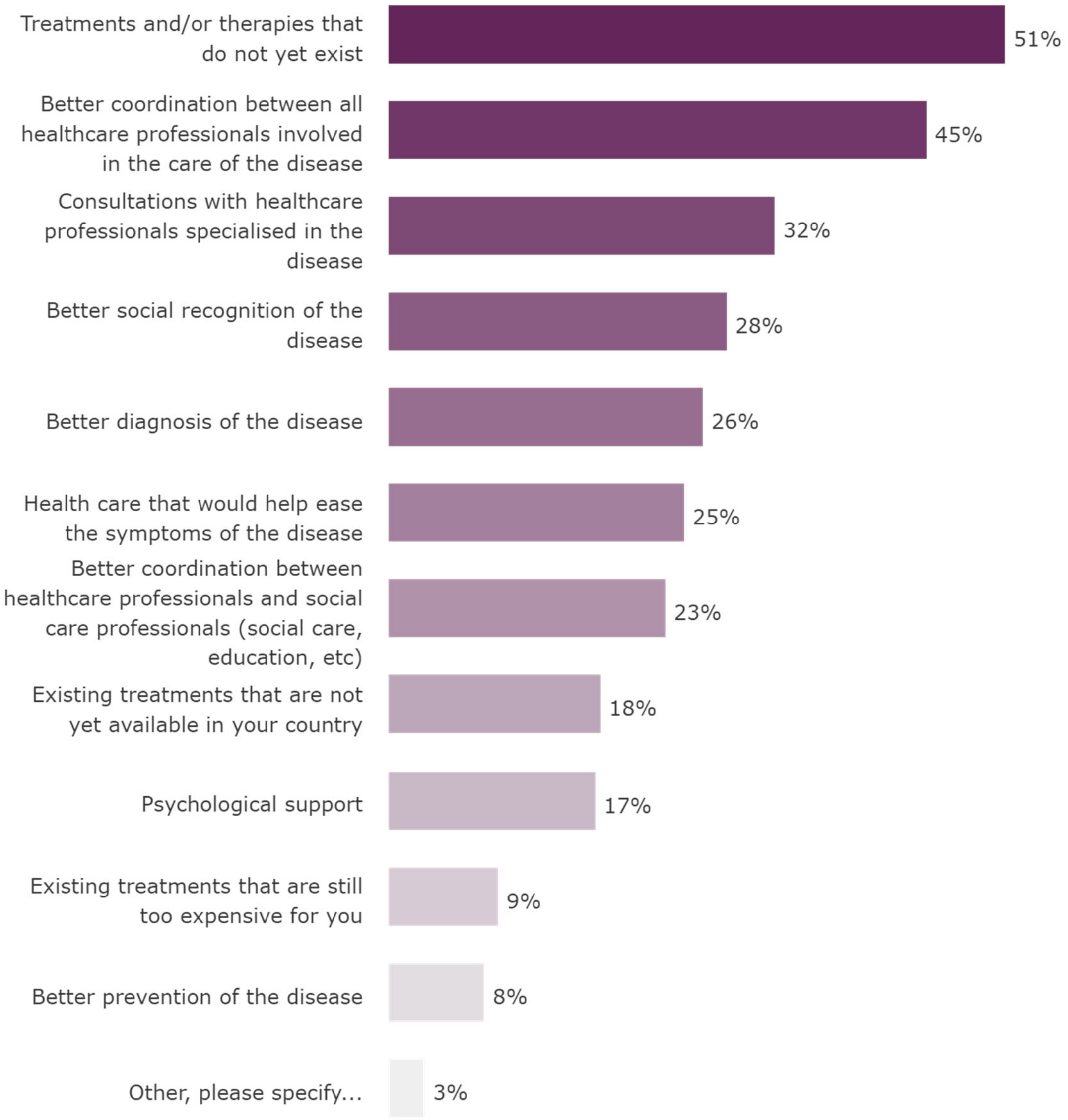
[6] Rare Barometer, *Improve our experience of heath care! Key findings from a survey on patients' and carers' experience of medical care for their rare diseases*, January 2021.

[7] 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. Better social recognition of the disease is especially valued by people living with very rare diseases (Table 2).

> 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 3. Within the next 10 years, the top 3 priorities to improve care for your rare disease would be to access to: (n=3,998)



Several answers possible; 3 answers maximum.

The order in which modalities appeared was random and different for each respondent.

In a 2018 survey on rare disease patients' participation in research[8], respondents engaged in advocacy activities rated their policy priorities within the rare disease research fields. The results showed that the top 3 priorities for patient representatives were research on therapeutics (with a grade of 8.5 out of 10), research on diagnosis (with a grade of 8.0 out of 10) and research on mechanisms and causes of the disease (with a grade of 7.8 out of 10).

If accessing better diagnosis does not appear in the top 3 priorities of all respondents to improve the care for rare diseases by 2030, we can see that (Table 2):

- Patient representatives give more importance to diagnosis than respondents who are not engaged in advocacy activities: better diagnosis is their third priority to improve care for rare diseases.
- 97% respondents of the Rare 2030 survey are already diagnosed and may not see a need to improve diagnosis of their disease. Indeed, accessing better diagnosis is the first priority for respondents who are not yet diagnosed.

TABLE 2. Top 3 priorities to improve care for rare diseases by 2030 depending on respondents' characteristics

	All respondents (n=3,998)	People living with very rare diseases (less than 1 case for 100,000 people - n=582)	Carers (parent, spouse, family members) of patients (n=1,346)	Patient representatives (n=242)	Respondents who are not yet diagnosed (n=109)
First priority	**Treatments and/or therapies that do not yet exist (51%)**	Treatments and/or therapies that do not yet exist (54%)	Treatments and/or therapies that do not yet exist (53%)	Better coordination between all healthcare professionals involved in the care of the disease (56%)	Better diagnosis of the disease (66%)
Second priority	**Better coordination between all healthcare professionals involved in the care of the disease (45%)**	Better coordination between all healthcare professionals involved in the care of the disease (41%)	Better coordination between all healthcare professionals involved in the care of the disease (42%)	Treatments and/or therapies that do not yet exist (50%)	Treatments and/or therapies that do not yet exist (44%)
Third priority	**Consultations with healthcare professionals specialised in the disease (32%)**	Better social recognition of the disease (32%)	Better coordination between healthcare professionals and social care professionals (32%)	Better diagnosis of the disease (39%)	Better coordination between all healthcare professionals involved in the care of the disease (39%)

Several answers possible; 3 answers maximum.
The order in which modalities appeared was random and different for each respondent.
Percentages between brackets refer to the percentage of respondents who chose the corresponding modality.

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

Answers to the open question also point out how crucial early and efficient diagnosis is for people living with rare diseases.

> 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[9]. 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 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%).

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** for which appropriate disease management can improve the health and quality of life of the patient (67%). They also support screening for hereditary conditions (70%) hence allowing parents to have reproductive options during subsequent pregnancies. **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" [10].

> 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 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**

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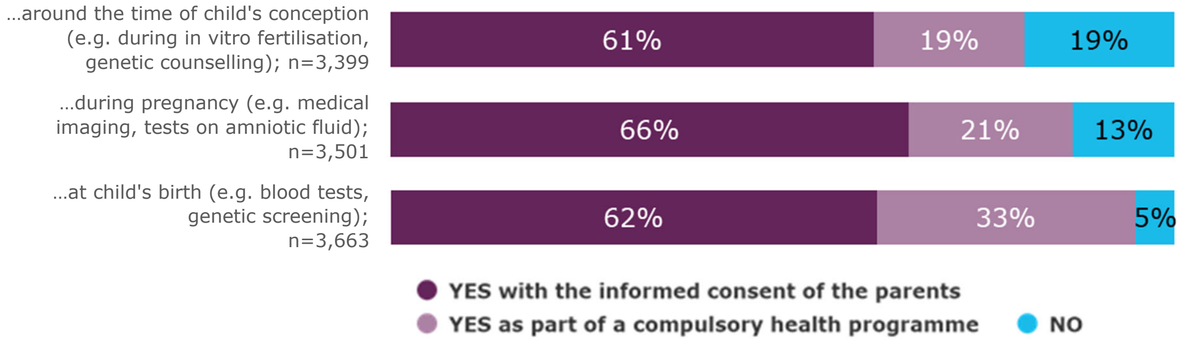
[9] Nguengang Wakap, S., Lambert, D.M., Olry, A. *et al.* Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. *Eur J Hum Genet* 28, 165–173 (2020).

[10] EURORDIS, *Key Principles for Newborn Screening. A EURORDIS Position paper*, January 2021.

Graph 4. 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.

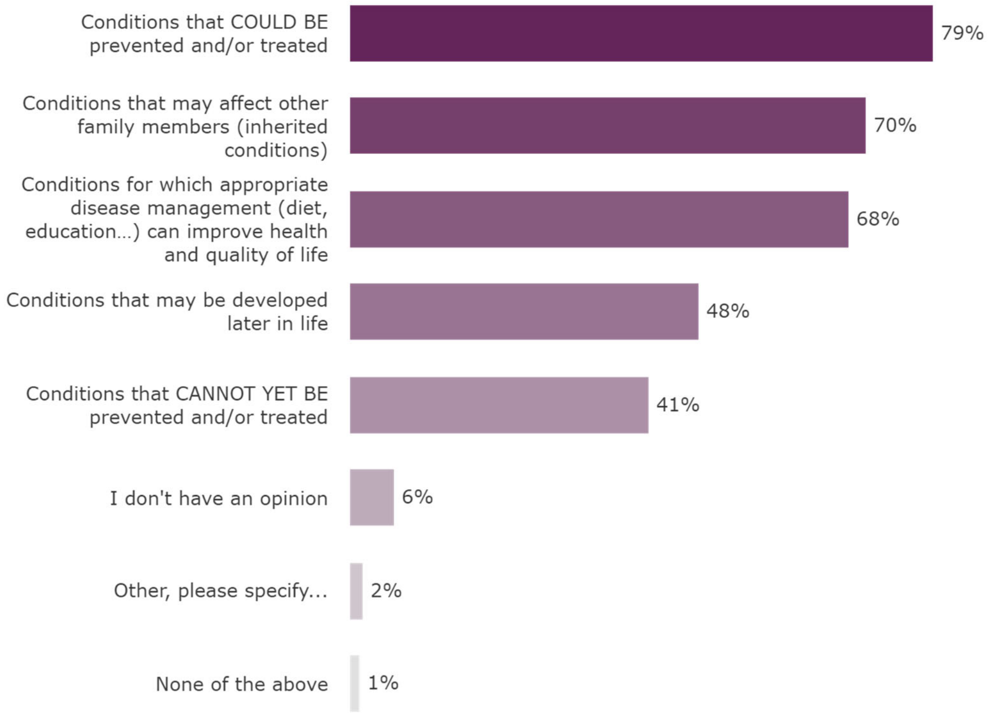
Your answers will help EURORDIS-Rare Diseases Europe voice the opinion of the rare disease community on this delicate matter.

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



Graph 5. 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? n=3,981



Several answers possible.

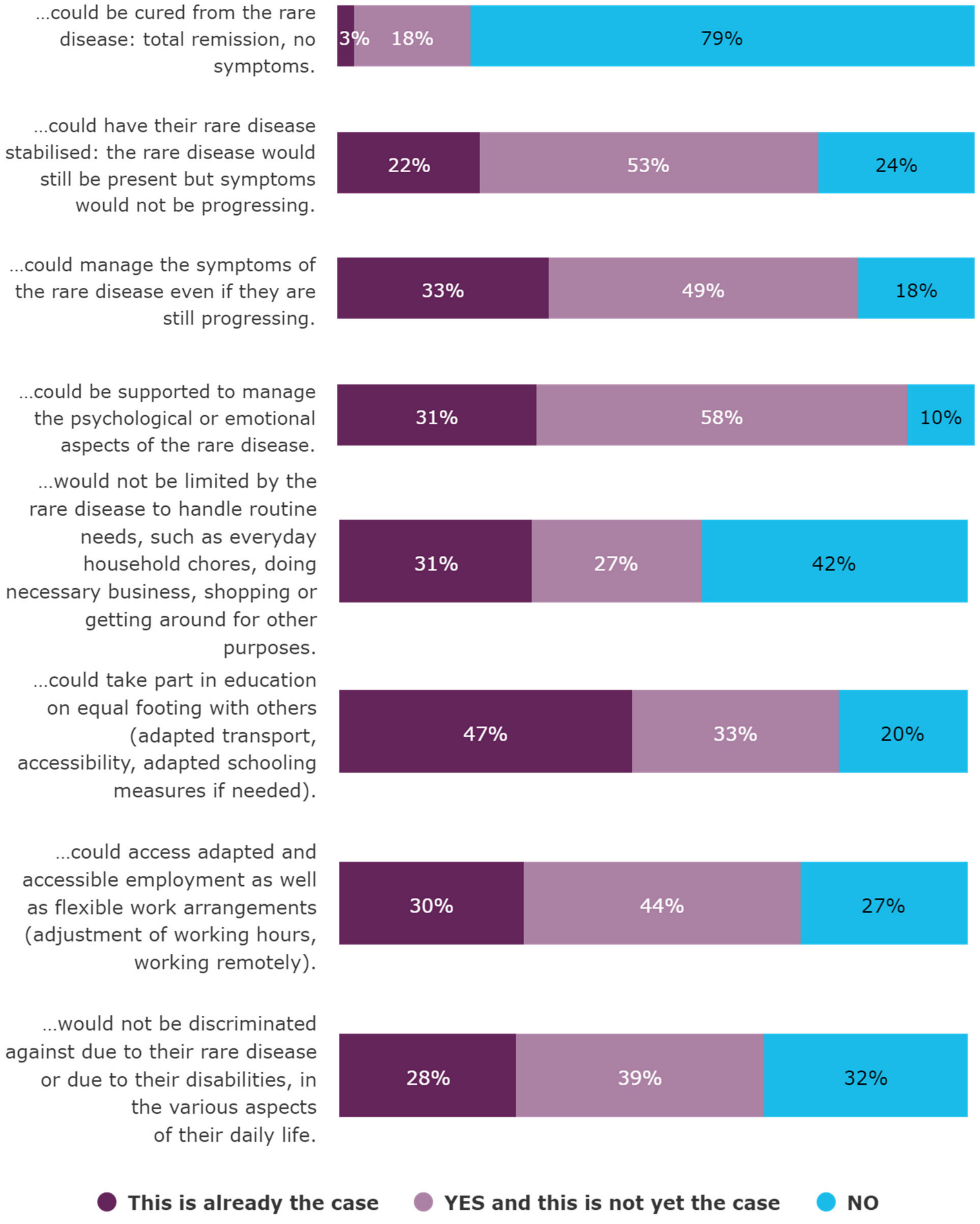
2. 2 Most respondents do not expect a cure by 2030 but consider it possible to improve their quality of life

People living with rare diseases do not expect to be cured from their rare disease within the next 10 years (79%) and 42% of them think that they will continue to be limited by their disease to handle routine needs.

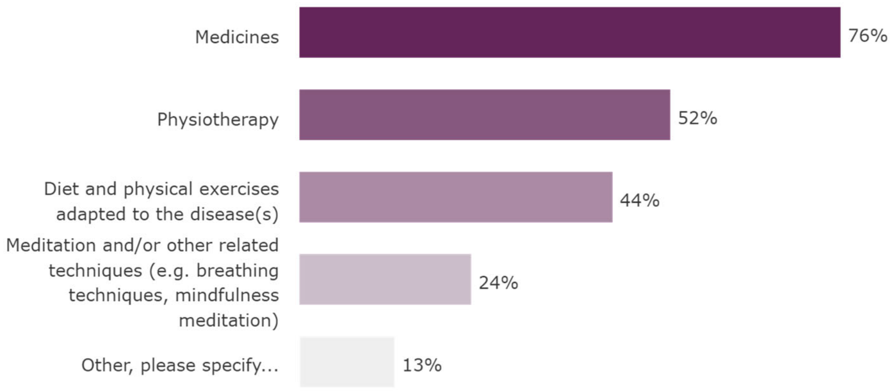
However, they rely on research because they believe there is hope for them to **stabilise their disease** (53%); to **better manage their symptoms** (49%) with new medicines and several types of therapies (see graph 7); to receive appropriate **emotional support** (58%); to access **adapted employment** (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**
>
> By 2030, I would like:
> - 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.
> - To make additional training mandatory for doctors / specialists who treat patients with such diseases or that they refer fairly if they do not know themselves.
> - Not to 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 6. Do you think it is possible and realistic that within 10 years, you or the person you care for… n=3,998



Graph 7. Within the next 10 years, in order to help ease the symptoms of your rare disease, you would need access to (n=924)



Base: respondents who cannot yet manage the symptoms of their rare disease but consider it possible and realistic to do so within 10 years.

Compared to people living with "more common" rare diseases, people who live with very rare diseases currently feel more limited in their access to the human and social rights listed below, and have less hope to access those rights by 2030:

- Taking part in education on equal footing with others;
- Accessing adapted and accessible employment as well as flexible work arrangements;
- Not being discriminated against due to their rare disease or to their disabilities;
- Not being limited to handle routine needs.

TABLE 3. Respondents' opinion on possible improvements in quality of life depending on the point prevalence of their rare disease n=2,112

Do you think it is possible and realistic that within 10 years, you or the person you care for...	Respondents living with...	This is already the case‡	YES and this is not yet the case	NO‡	I don't know
...could take part in education on equal footing with others (adapted transport, accessibility, adapted schooling measures if needed).	...more common rare diseases†	42% ***	26%	15% ***	17%
	...very rare diseases†	33% ***	27%	23% ***	16%
...could access adapted and accessible employment as well as flexible work arrangements (adjustment of working hours, working remotely).	...more common rare diseases†	27%***	36%	19% ***	17%
	...very rare diseases†	20% ***	34%	27% ***	19%
...would not be discriminated against due to their rare disease or due to their disabilities, in the various aspects of their daily life.	...more common rare diseases†	26% ***	33%	24%***	17%
	...very rare diseases†	18% ***	33%	31% ***	19%
...would not be limited to handle routine needs, such as everyday household chores, doing necessary business, shopping or getting around for other purposes.	...more common rare diseases†	27% ***	24%	34%**	15%
	...very rare diseases†	20% ***	23%	40%**	16%

†Very rare diseases = point prevalence of less than 1 case for 100,000 people; More common rare disease = point prevalence from 2 cases for 5,000 people to 1 case for 100,000 people.

*‡Asterisks indicate when percentages are significantly different between respondents with very rare diseases and with more common diseases: ** = significant (p-value<0.05), *** = highly significant (p-value<0.01).*

In the open question, respondents asked for **better knowledge** on rare diseases for patients, carers, healthcare professionals (general practitioners as well as specialists), social professionals and the whole society: they propose to improve education and awareness 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**
>
> I would like 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**
>
> I would like to 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 flexible work arrangements, such as **working from home** allow 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**

As part of the measures that could improve quality of life in the next 10 years, respondents also pointed the need to **improve the 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**[11] but given the scarcity and geographical dispersion of the healthcare experts who can treat them, they are **very willing to use remote health care** (see graphs 8, 10 and 13). This allows them to **save time** (see table 3) and to **access high quality care** (see graph 10), **when remote health care is appropriate for the care needed** (see graph 11) and **when they have already met their care team** (see graph 13).

> 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**

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.

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[11] 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.

3. 1 Saving time

Most rare diseases are complex, requiring a multidisciplinary approach to their care: in the EurordisCare3 survey[12] 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[13].

Therefore:

- **Saving time is the main reason** for people living with a rare disease to use remote consultations and remote prescriptions.
- **85 %** respondents are willing to use electronic prescriptions all the time.
- **68 %** respondents are also willing to receive medicines by mail all the time.

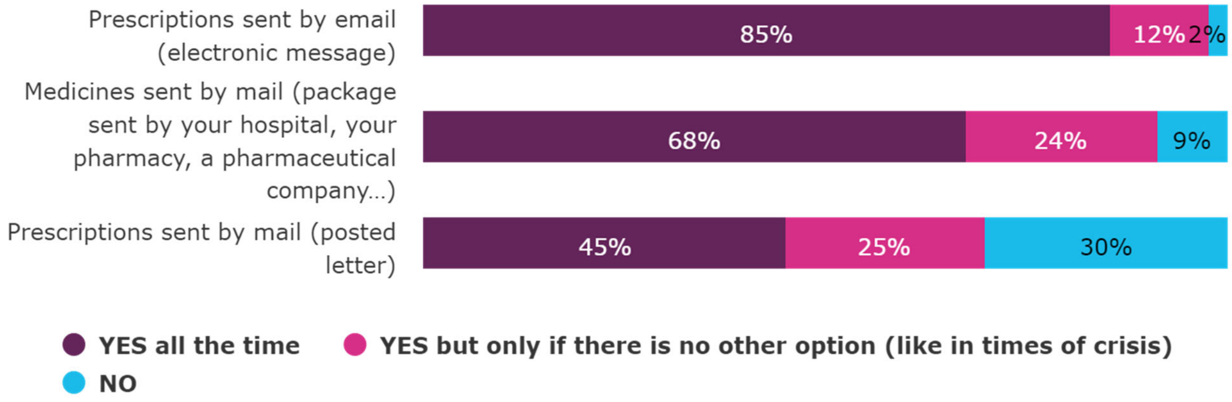
TABLE 4. Remote healthcare services, such as virtual consultations and electronic prescriptions, have been developing in daily life and are playing a crucial role during the COVID-19 pandemic.

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... n=3,998

	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 8. Within the next 10 years, would you be willing to use the following services: n=3,998



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[12] EURORDIS, *The voice of 12 000 patients, op. cit.*

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

Respondents with very rare diseases (less than 1 case for 100,000 people) are significantly more willing to use remote consultations for safety issues than those with more common rare diseases (at least 1 case for 100,000 people).

TABLE 5. Within the next 10 years, what would be the two main reasons for you to use remote consultations? n=3,273

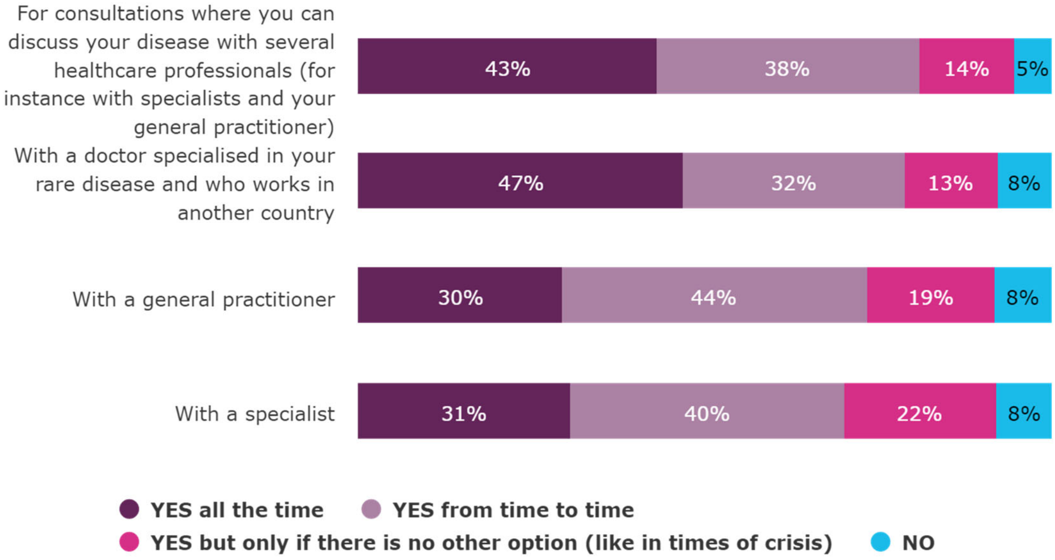
	Saving time	Accessing higher quality care	Having no other option	Safety	Saving money
Respondents living with more common rare diseases (from 2 cases for 5,000 people to 1 case for 100,000 people)	56% ***	39%	26%	21% ***	12%
Respondents living with very rare diseases (less than 1 case for 100,000 people)	49% ***	37%	17%	27% ***	14%

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 9. Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app) n=3,998

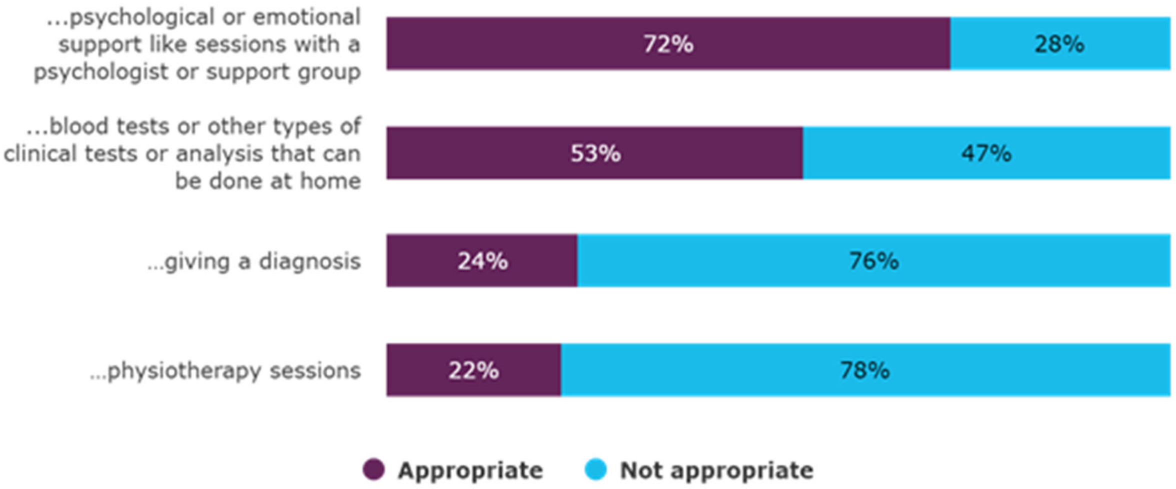


3. 3 Remote consultations are not appropriate for all types of health care

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).

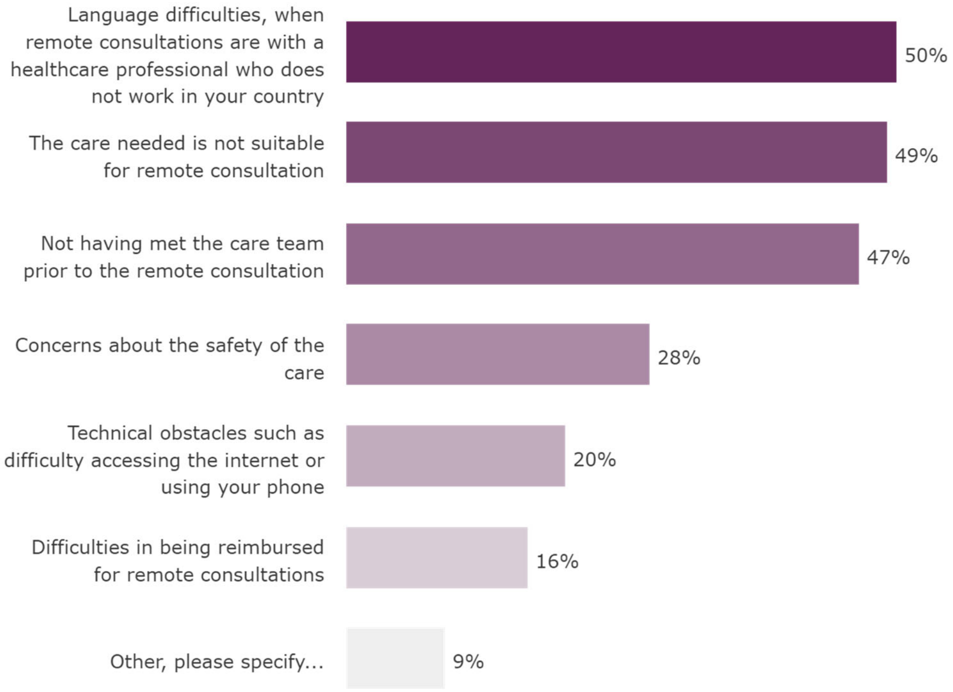
> "When you have a lump under your skin and the doctor is on the phone, how should the doctor know what it is? **Rare disease patient, Luxembourg**
>
> Remote diagnosis without proper investigation can lead to misdiagnosis. **Rare disease carer, Germany**
>
> It is difficult to know, for example, if 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 10. Do you think that remote virtual consultations are appropriate for... n=3,998



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 11. What would be the 3 main obstacles for you to use remote consultations within the next 10 years? N=3,998



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

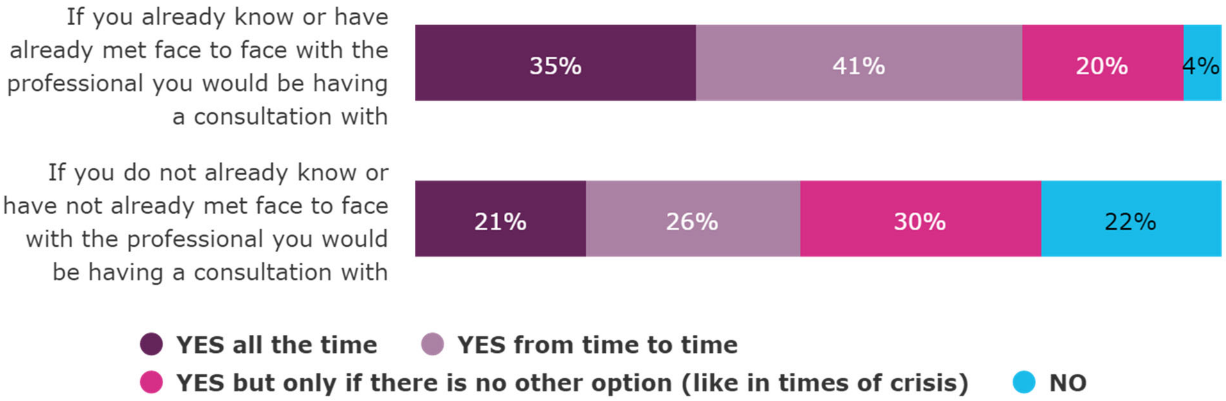
People living with rare diseases are more willing to use remote consultations if they have already met face to face with the professional they would be having a consultation with. **Not having met the care team is also one of the main obstacles to using remote consultations** within the next 10 years (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 12. Within the next 10 years, would you be willing to attend remote consultations (via phone, video, email, text, app, etc.) n=3,998



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

3. 5 Willingness to access cross-border health care

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 (EU). This directive is complementary to the EC regulations Nos. 883/04 and 987/09 which entitle 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 EU citizens to have access to cross-border health care, which can be critical when patients do not find healthcare professionals specialised in their rare disease in their country.

This chapter will focus on the willingness to access cross-border health care by people living with a rare disease, for EU people living in EU and non-EU countries. The responses provided by people living in the EU will be compared to those given by the general EU population within the special Eurobarometer 425.

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 health care were taken from the special Eurobarometer 425 published in 2014[14], 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.

Willingness to access cross-border health care

86% of respondents living in the EU 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.

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[14] 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.

TABLE 6. 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]?¥ 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 PLWRD†
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	

¥Text between brackets was only included into the Rare 2030 survey and did not appear in the questions of the Eurobarometer survey.
‡Percentage in the EU population is taken from Special Eurobarometer 425.
†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).

Overall responses of people living in EU and non-EU countries are aligned as **84% of respondents not living in the EU would be willing to travel to another country to receive medical treatment**, either unconditionally or depending on the medical treatment or on the country. 12% of them would not be willing to travel to another country to receive medical treatment, which is significantly higher than for respondents who are living in EU countries.

TABLE 7. Within the next 10 years and OUTSIDE TIMES OF CRISIS, would you be willing to travel to another country to receive medical treatment for your rare disease? n=731

	Percentage of respondents‡	Countries where the modality was significantly over-represented†	Countries where the modality was significantly under-represented†
Yes	**41%**	Argentina (65%), Norway (49%), Switzerland (47%)	United States of America (23%), United Kingdom (25%)
It depends on the medical treatment or on the country	**43%**	United States of America (60%)	South Africa (25%), Argentina (26%)
No	**12% ****	United Kingdom (22%)	Norway (6%)
I don't know	**4%**	/	/

*‡Asterisks indicate when percentages are significantly different from the ones obtained within EU citizens: ** = significant (p-value<0.05), *** = highly significant (p-value<0.01).*
†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), in countries where at least 20 people responded to the Rare 2030 survey.

Reasons to be willing to access cross-border health care

People living with rare diseases in the EU 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 8. 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)]?[¥] 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 PLWRD[†]
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	

Several answers possible

[¥] Text between brackets was only included into the Rare 2030 survey and did not appear in the questions of the Eurobarometer survey.

[‡] Percentage in the EU population is taken from Special Eurobarometer 425.

[†] 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 ("yes" or "it depends on the country or on the medical treatment").

Respondents who are living in non-EU countries are mainly willing to be treated in another country to **access treatments that are not available in their country** (83%), **treatment from a renowned specialist** (64%) and to **access better quality treatment** (62%).

TABLE 9. For which of the following reasons would you be willing to go to another country to receive medical treatment for your rare disease(s)? n=646

	Percentage of respondents‡	Countries where the modality was significantly over-represented†	Countries where the modality was significantly under-represented†
To receive treatment that is not available in your country	**83% *****	/	United States of America (75%)
To receive better quality treatment	**62%**	/	/
To receive treatment from a renowned specialist	**64% *****	/	/
To receive treatment more quickly	**33%**	Canada (61%)	United States of America (31%)
To receive cheaper treatment	**13% *****	United States of America (17%)	/
To receive treatment from a provider that is closer to home	**5%**	/	/
I don't know	**3%**	/	/
Other, please specify…	**3%**	/	/

Several answers possible
*‡Asterisks indicate when percentages are significantly different from the ones obtained within EU citizens: ** = significant (p-value<0.05), *** = highly significant (p-value<0.01).*
† 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), in countries where at least 20 people responded to the Rare 2030 survey.
Base: Respondents who do not live in the European Union and who would be willing to travel to another country to receive medical treatment for their rare disease ("yes" or "it depends on the country or on the medical treatment").

Reasons for not wanting to access cross-border health care

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 10. 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 PLWRD[†]
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	

Several answers possible

[¥] Text between brackets was only included into the Rare 2030 survey and did not appear in the questions of the Eurobarometer survey.

[‡] Percentage in the EU population is taken from Special Eurobarometer 425.

[†] 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").

Among respondents who are not living in the EU and who would not be willing to travel abroad to receive medical treatment, 55% report that **it is more convenient to be treated near home**, 55% that **they cannot afford to receive medical treatment abroad** and 52% that they are **satisfied with the treatments they receive in their country**. They also see the **lack of information** on their rights, reimbursement policies and patients' safety standards in the country of treatment as valid reasons not to use cross-border healthcare (49% to 51%).

TABLE 11. For which of the following reasons would you not be willing to go to another country to receive medical treatment [for your rare disease(s)]?¥ n=429

	Percentage of respondents[‡]	Countries where the modality was significantly over-represented[†]	Countries where the modality was significantly under-represented[†]
It is more convenient to be treated near your home	**55%**	/	/
You cannot afford to receive medical treatment abroad	**55%** ***	South Africa (83%)	Switzerland (21%)
You are satisfied with the medical treatments you receive in your country	**52%**	/	United States of America (40%)
You are not aware of your rights in case things go wrong	**51%**	/	/
You do not have enough information about the availability and quality of medical treatments abroad	**50%**	/	/
You have no information on patient safety and quality of care abroad	**49%**	/	/
You would have issues understanding the language	**37%** ***	/	/
You are not sure that you would be reimbursed	**30%** **	/	United Kingdom (23%)
You prefer not to travel[‡]	**25%** ***	/	/
Your doctor advised you not to[‡]	**10%** **	/	/
I don't know	**1%** **	/	/
Other, please specify...	**6%**	/	/

Several answers possible

*[‡] Asterisks indicate when percentages are significantly different from the ones obtained within EU citizens: ** = significant (p-value<0.05), *** = highly significant (p-value<0.01).*

[†] 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), in countries where at least 20 people responded to 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 health care in the European Union (EU)

Respondents living in the EU 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 than 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.

TABLE 12. 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. 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 PLWRD[†]	Countries where the modality was significantly under-represented among PLWRD[†]
True (correct answer)	**44%**	57%	Sweden (59%), Hungary (55%), Italy (51%), Finland (51%)	Czech Republic (31%), Belgium (33%), Netherlands (34%), Germany (38%)
False	**15%**	16%	Croatia (34%), Czech Republic (28%), Belgium (27%), Poland (27%)	Finland (7%), Spain (11%), Italy (12%)
I don't know	**41%**	27%	Spain (50%), Netherlands (48%)	Austria (19%), Italy (37%)

[‡] Percentage in the EU population is taken from Special Eurobarometer 425.
[†] Only significant relationships are taken into account (p-value < 0.05).

Only **26%** respondents know that they can get a prescription from a doctor to use in another EU country.

TABLE 13. 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. 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 PLWRD†	Countries where the modality was significantly under-represented among PLWRD†
False (correct answer)	**26%**	29%	Croatia (42%), Italy (38%), Belgium (24%), Spain (32%), Germany (32%)	Finland (10%), Netherlands (15%), France (18%)
True	**28%**	38%	Finland (46%), Netherlands (42%), France (32%)	Germany (15%), Spain (16%), Italy (18%)
I don't know	**46%**	33%	Germany (52%), Spain (52%)	Belgium (35%)

‡Percentage in the EU population is taken from Special Eurobarometer 425.
†Only significant relationships are taken into account (p-value < 0.05).

76% respondents know that they have the right to receive a copy of their medical record when they seek healthcare in another EU country.

TABLE 14. 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. 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 PLWRD†	Countries where the modality was significantly under-represented among PLWRD†
True (correct answer)	**76%**	70%	Hungary (89%), Czech Republic (85%), Netherlands (84%), Finland (82%)	Croatia (62%), France (64%)
False	**2%**	7%	Croatia (9%)	Spain (0%)
I don't know	**22%**	23%	France (34%)	Hungary (8%), Czech Republic (11%), Netherlands (16%)

‡Percentage in the EU population is taken from Special Eurobarometer 425.
†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).

TABLE 15. 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. n=3,365

	Percentage of people living with rare diseases (PLWRD)	Countries where the modality was significantly over-represented among PLWRD†	Countries where the modality was significantly under-represented among PLWRD†
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 health care are one of the 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[15]. Therefore, 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. **Remote consultations and access to cross-border health care then only appear as a means to receive better quality and more multidisciplinary care**.

> 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. **Rare disease carer, Bosnia**

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

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 pathogenic 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).

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 complicated to analyse because most respondents agreed with all the statements proposed, even when those statements contradicted 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 when it comes to clinical research and drug development[16]. Prioritising research in those areas may help achieve greater focus for these disease areas and restore the equilibrium. Respondents agree that it is important to have research studies conducted on rare 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 (68%).**

> 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**
>
> 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**
>
> 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**

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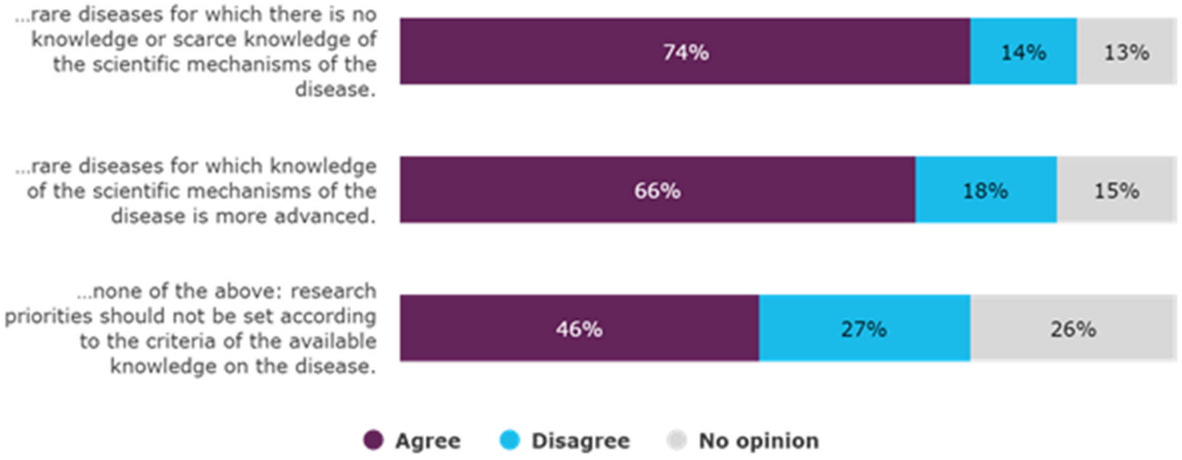
[16] Miraldo M., Sassi F., Shaïkh M., Simmons B., Vrintten C., *The development and market launch of orphan drugs from 1980-2019: q quantitative analysis. RARE2030 Foresight study, Work Package 5*, December 2020.

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 13. AVAILABLE BODY OF KNOWLEDGE (n=1,956)

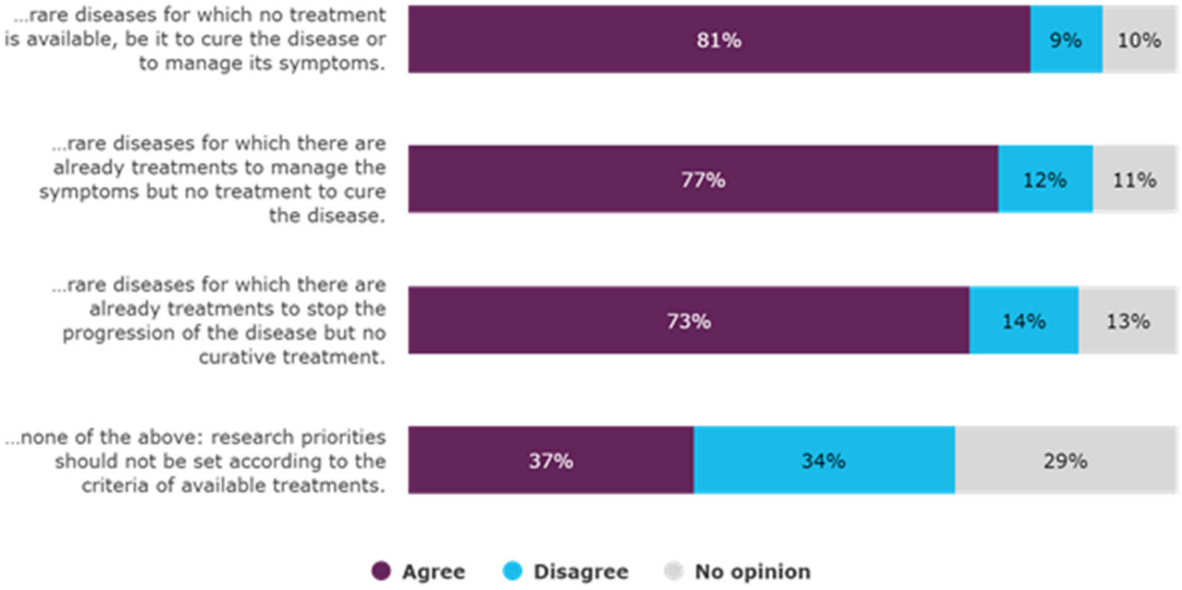
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



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

Graph 14. AVAILABLE TREATMENTS (n=2,029)

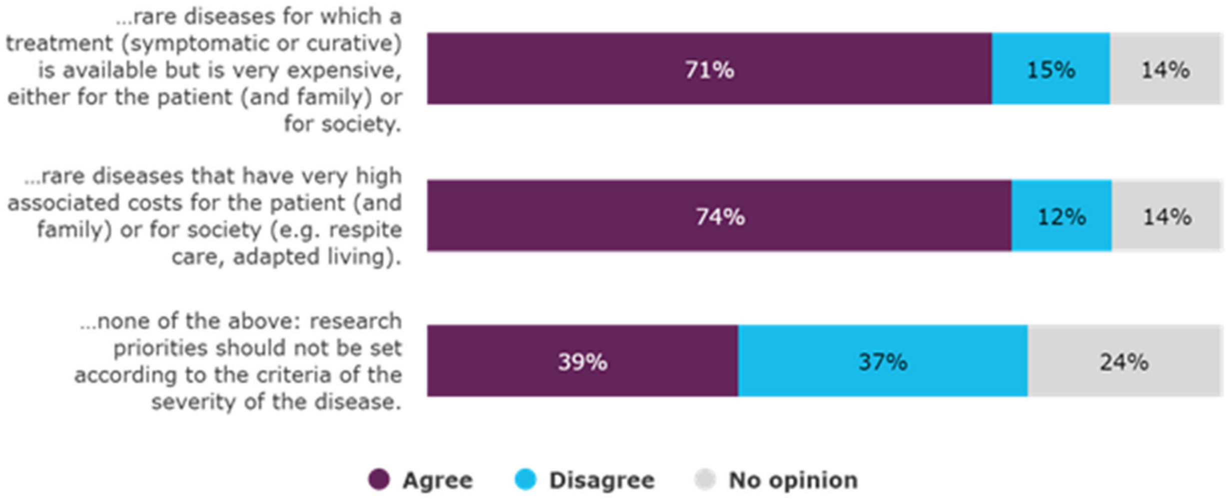
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



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

Graph 15. SEVERITY OF THE DISEASE (BURDEN OF THE DISEASE – n=1,973)

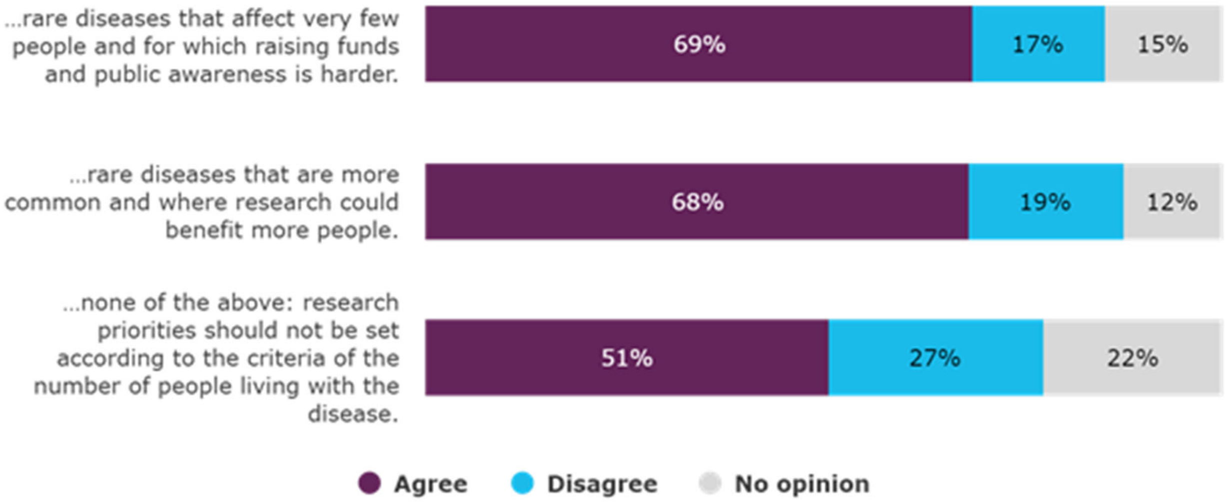
Within the next 10 years, do you think that available resources for RESEARCH ACROSS ALL RARE DISEASES should focus on…



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

Graph 16. 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.

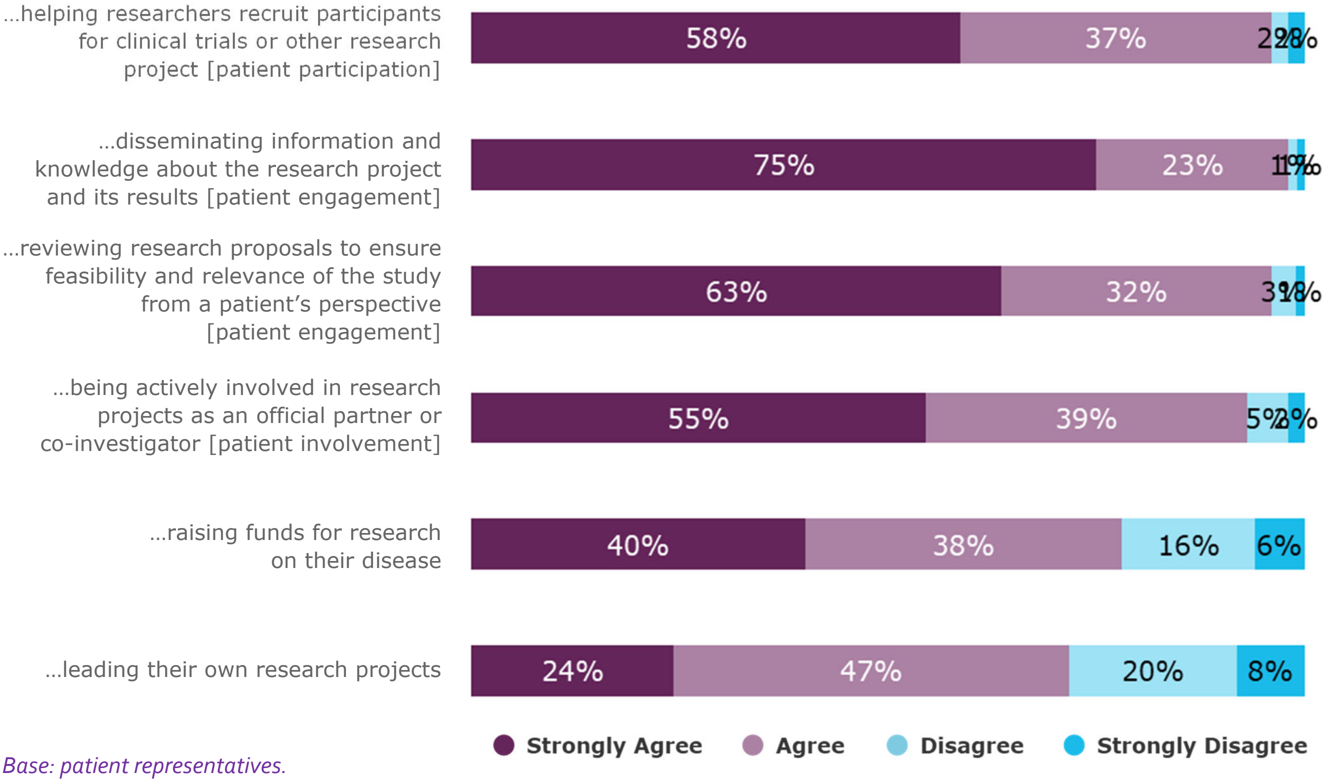
Patient organisations are willing to be involved in research for rare diseases

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 14). They are also in support of patient organisations to **raise funds for research** on their disease (78%) and to **lead their own research projects** (70%).

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**
>
> I would like professionals and parents/patients to dialogue more, families to be more involved in new developments and information to be 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 17. As a patient representative, do you think that PATIENT ORGANISATIONS should contribute in research for rare diseases by… (n=252)



Base: patient representatives.
Answers to the modality "no opinion" are excluded from the analysis.

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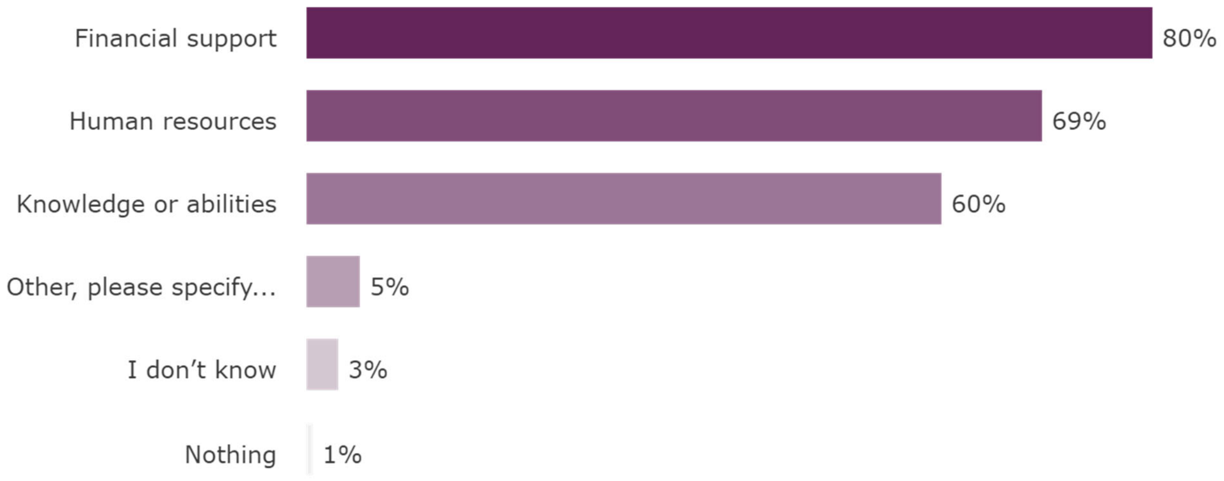
[17] Rare Barometer, *Rare disease patients' participation in research*, February 2018.

Table 16. Definition of patient partnerships according to the EJP-RD Short Guide and corresponding questions of the Rare 2030 survey[18]

Concept of patient partnership	Description of patient partnership	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 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.
	Patients review research proposals to ensure feasibility and relevance of study from the patient's perspective.		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 18. 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)



Base: patient representatives.

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