EURORDIS
RARE DISEASES EUROPE

Rare Barometer
A EURORDIS INITIATIVE

IMPROVE OUR EXPERIENCE OF HEALTH CARE!

Key findings from a survey on patients' and carers' experience of medical care for their rare diseases

January 2021

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

NO CURE
for the vast majority of diseases and few treatments available

Coordination and continuity of care is of particular importance in the field of rare diseases. Care pathways are often complex and involve a large number of healthcare professionals that need to share information and collaborate, among themselves and with the patient, along the entire pathway to deliver safe and effective quality care.

By measuring patients' and carers' experience of health care, the H-CARE Survey helps us understand how to improve medical care to address the needs of people living with a rare disease and how **multidisciplinary care teams specialised in the treatment of rare diseases** can help to reach this goal.

WITHIN THIS REPORT YOU WILL FIND:

1. Key results of the H-CARE Survey on the experience of medical care for rare disease patients and their family members
2. Three recommendations to improve the healthcare experience for all people living with a rare disease
3. Methodology of the H-CARE Survey

1 THE HEALTHCARE EXPERIENCE OF PEOPLE LIVING WITH A RARE DISEASE

Patients and carers were asked to rate how often, on a scale from 1 (never) to 5 (always), healthcare professionals considered aspects of their healthcare experience. **Questions included aspects related to follow-up after a consultation, information on treatment, care coordination or how to manage their health in their daily life.** By combining these answers into one average score, the H-CARE Survey allows to propose and compare a measure which relates to patients' and carers' healthcare experience.

Rare disease patients give their healthcare experience a medium-low score

Rare disease patients on average rate their healthcare experience...

on a scale from 1 to 5

> *"Often in the context of rare and complex diseases, professionals are powerless, for lack of information or lack of knowledge about an unusual disease. Despite their good will, they discover the disease at the same time as the patient discovers or develops its symptoms."*
> Rare disease patient

> *"My experience in different hospitals throughout my life is that only medical aspects are attended to. The psychological and emotional part is not taken into account nor is there a joint multidisciplinary treatment."*
> Rare disease patient

Rare disease patients seem to have a worse experience of health care than patients with chronic diseases: **70%** of studies that used the same questionnaire reported a higher average score for chronic disease patients' experience of health care[1].

Q *The score for rare disease patients' healthcare experience was obtained by combining answers to 11 questions detailed in page 7.*

Carers score the healthcare experience higher on average than patients

Carers (family members...) of rare disease patients on average rate their healthcare experience...

on a scale from 1 to 5

> *"It is a complex disease that requires the intervention of a multidisciplinary team from the health, social and school fields. There is usually no communication within the team and there is no coordination, which usually falls on the main carer."*
> Rare disease carer

> *"Fragmentary treatment of the complex rare syndrome, also focusing only on drugs and not holistic treatment and management. Ignoring the needs and difficulties of carers."*
> Rare disease carer

Q *The score for rare disease carers' healthcare experience was obtained by combining answers to 12 questions detailed in page 7.*

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[1] Based on the results of 52 peer-reviewed studies using the PACIC or the PACIC-S questionnaires. These questionnaires were developed by the MacColl Centre for Health Care Innovation to measure the healthcare experience of chronic disease patients.

2 HOW CAN HEALTHCARE EXPERIENCE BE IMPROVED?

Patients and their carers pointed out that their healthcare team often did not support them **beyond the clinical aspects of their treatment or beyond the time dedicated to the medical consultation**, for example by ensuring follow-up after visits (including after announcing a diagnosis), providing emotional support or telling them how to reach patient groups to help them cope with their disease.

To ensure a better healthcare experience the top 3 areas that need to be improved are:

1

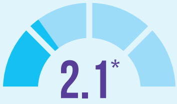
Contacting patients or carers after a visit to see how things are going

> *No multidisciplinary follow-up is proposed as in more well-known or classic serious diseases."*
> **Rare disease patient**

> *Since the diagnosis, about 4 years ago, the facility has not contacted me for any follow-up."*
> **Rare disease patient**

2

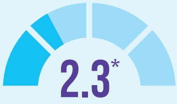
Encouraging patients or carers to go to a specific group or class to help them cope with the rare disease



> *I have no psychological or social help; I do all my steps alone thanks to the support of an association. It is a shame that we are not advised to approach certain associations."*
> **Rare disease patient**

3

Helping patients and carers deal with emotions related to the patient's health status



> *Despite the good technical competence of the facility, I feel very alone and not well supported psychologically."*
> **Rare disease patient**

> *I lack support regarding the psychological or emotional level of the disease, the side effects of medications and how this affects my private life."*
> **Rare disease patient**

Q *Over the past 6 months, when I/the person I care for received medical care for my/his/her rare disease, I was:*

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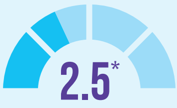
* Average score on a scale from 1 (never) to 5 (always).

In order to support patients and carers, healthcare professionals could also **improve coordination with other doctors** and provide recommendations on **how to manage the rare disease in daily life,** for instance by setting specific goals or by making a list of what could be done to manage the rare disease in everyday life.

Everyday self-management of the disease and care coordination could also be improved by helping patients and carers to:

1

Know how their visits with other specialists help the treatment

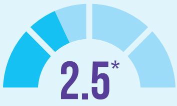


> *My doctor is excellent and offers an adapted treatment but I have no overall support."*
> **Rare disease patient**

> *Coordination between the family and the different services that see the patient, as symptoms appear, with an interlocutor from the hospital who can coordinate all actions and care plan would be good."*
> **Rare disease carer**

2

Plan ahead so they could take care of the patient's rare disease even in hard times

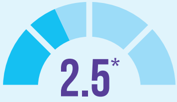


> *I would love to understand what can be done about what symptoms, what I just need to live with and what I can do myself to makes things as best as they can be."*
> **Rare disease patient**

> *I often have have to try to find solutions by myself in order to live with my disease."*
> **Rare disease patient**

3

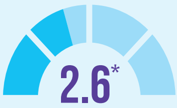
Make a treatment plan[2] that they could do in their daily life



> *Doctors do not have much information themselves so giving a treatment plan is more of a guessing game. I do not blame them but at the same time it is scary when even your own doctor does not quite know what to do when there is no support around."*
> **Rare disease patient**

4

Set specific goals to improve the patient's eating or exercise



> *Consultations focus exclusively on seeing if the disease has progressed without offering options to achieve improvements based on an adequate physical exercise plan nor in relation to the patient's self-esteem"*
> **Rare disease carer**

Q *Over the past 6 months, when I/the person I care for received medical care for my/his/her rare disease, I was:*

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* Average score on a scale from 1 (never) to 5 (always).

[2] A treatment plan is a list, made with the care team, of what needs to be done to take care the patient's health.

3

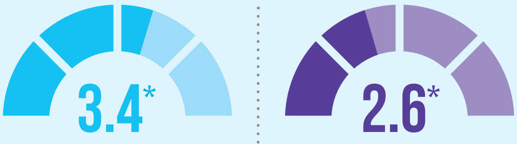
THE CRUCIAL ROLE OF CENTRES OF EXPERTISE FOR RARE DISEASES

In Europe, **European Reference Networks (or ERNs[3])** are virtual networks that bring together hospital units from across the European Union to ensure that people living with rare or complex diseases can benefit from the best treatment and advice available for their condition. Typically, hospital units that are members of a European Reference Network are Centres of Expertise that bring together **multidisciplinary care teams specialised in the treatment of a given rare or complex disease.**

The H-CARE Pilot Survey allowed to specifically measure the healthcare experience of **1,319 patients and carers living with rare diseases affecting kidneys, lungs, the urogenital area and genetic tumour risk syndromes**, whether they were treated in Centres of Expertise that belong to a European Reference Network or not. Further research is needed to measure differences in healthcare experience depending on the type of hospital unit for other rare disease areas.

Patients and carers living with a rare disease affecting kidneys, lungs, the urogenital area and genetic tumour risk syndromes have a better healthcare experience when they are treated by hospital units that are part of a European Reference Network

On a scale from 1 to 5, rare disease patients' and carers' average experience of health care receives a rating of...



when patients are treated in hospital units that are...

✓ **PART** of a European Reference Network (centres of expertise for rare diseases)

✕ **NOT PART** of a European Reference Network

Chronic disease patients also report a better experience of healthcare when their care team is multi-disciplinary or trained in chronic care and when they are part of a disease-management programme[4].

> *"Before getting taken on by [Centre of Expertise] as a rare disease patient, I had over twenty years of poor care. But [Centre of Expertise] now looks after me incredibly well."*
> Rare disease patient

> *"Since I was followed by the [Centre of Expertise], my quality of life has improved considerably, as has my mental approach to my pathology and life. I have been given treatments that allow me to lead a full life, as if I did not suffer from any disease. Without the support of the specialists and their staff, this would not have been possible."*
> Rare disease patient

Q *The score for rare disease patients' and carers' average care experience was obtained by combining answers to 11 questions (for patients) or 12 questions (for carers) detailed in page 7.*

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[3] ec.europa.eu/health/ern_en

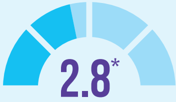
[4] Based on the results of 52 peer-reviewed studies using the PACIC or the PACIC-S questionnaires. These questionnaires were developed by the MacColl Centre for Health Care Innovation to measure the healthcare experience of chronic disease patients.

Improving experience of healthcare in Centres of Expertise

Patients and carers whose rare disease is treated in hospital units that belong to a European Reference Network give a higher score across all the aspects of healthcare experience that were measured in the H-CARE Pilot Survey. It is worth noting that the areas for improvement for Centres of Expertise that belong to an ERN and for non-expert centres are the same.

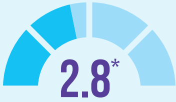
The main aspects of healthcare experience that should be improved in the Centres of Expertise for rare diseases are:

1



Contacting patients or carers after a visit to see how things are going

2



Encouraging patients or carers to go to a specific group or class to help them cope with the rare disease

3

Helping patients and carers deal with emotions related to the patient's health status

4

Telling patients or carers how their visits with other specialists helped the treatment

Q *Over the past 6 months, when I/the person I care for received medical care for my/his/her rare disease, I was:*

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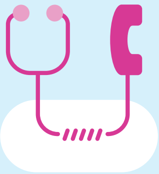
* Average score on a scale from 1 (never) to 5 (always).

4 RECOMMENDATIONS TO IMPROVE THE HEALTHCARE EXPERIENCE FOR PEOPLE LIVING WITH A RARE DISEASE

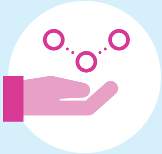
RECOMMENDATION 1



GO BEYOND DIAGNOSIS AND MEDICAL TREATMENT

Increase focus on follow-up after consultations, on psychological support and on aspects of healthcare that allow patients to manage their health in their daily life.

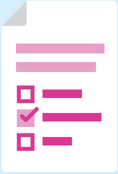
RECOMMENDATION 2



ENSURE ACCESS TO MULTIDISCIPLINARY AND NETWORKED HEALTHCARE

Allow every person living with a rare disease to have **access to quality highly specialised care** by simplifying the national referral pathways to Centres of Expertise; by facilitating timely access to face to face and virtual cross-border healthcare and virtual expert advice when expertise is not available at local level; and by facilitating the sharing of knowledge assets produced by Centres of Expertise with other healthcare providers.

RECOMMENDATION 3



DEVELOP AND VALIDATE A QUESTIONNAIRE TO MEASURE THE HEALTHCARE EXPERIENCE OF RARE DISEASE PATIENTS AND CAREGIVERS

Such a questionnaire designed specifically for people living with a rare disease would contribute to **set quality standards for rare diseases health care**. It would need to include dimensions that are specific to rare diseases and to be flexible enough to adapt to the diversity of situations and profiles of people living with a rare disease.

5 SURVEY METHODOLOGY

The H-CARE Pilot Survey[5], conducted from 2019 to 2020, was designed with patient representatives and healthcare providers from European Reference Networks ErkNet, eUROGEN, Genturis and LUNG. The goal was to measure the healthcare experience of people living with a rare disease using a **validated questionnaire**, which is an instrument used by scientists to measure subjective concepts, such as anxiety or care experience, through a fixed series of questions that are all related to the concept being measured and that are combined to give a numerical score.

In the absence of a validated questionnaire to measure experience of healthcare for rare and complex diseases, the PACIC-S questionnaire was used. This is the short form of the Patient Assessment of Care for Chronic Conditions (PACIC) questionnaire, which was developed in 2004 by the MacColl Centre for Health Care Innovation to measure the care experience of patients living with chronic conditions. Since its creation, this questionnaire has been validated into 11 languages and has been used in more than 130 peer-reviewed studies worldwide, setting international standards of care quality for chronic conditions.

The wording of the PACIC-S questionnaire was adapted to patients and carers living with a rare or a complex disease: questions of these adapted PACIC-S questionnaires are listed in the table below, as well as the average score that rare disease patients and carers gave to each question.

Over the past 6 months, when I/the person I care for received medical care for my/his/her rare or complex disease:			Average score on a scale from 1 (never) to 5 (always)
Questions of the adapted PACIC questionnaire	1	*[I was]* Given choices about treatments to think about	2.8
	2	*[I was]* Satisfied that my / the patient's care was well organized	3.5
	3	*[I was]* Helped to set specific goals to improve my / the patient's eating or exercise	2.6
	4	*[I was]* Given a copy of my / the patient's treatment plan[6]	2.8
	5	*[I was]* Encouraged to go to a specific group or class to help me cope with my / the patient's rare or complex disease	2.1
	6	*[I was]* Asked questions, either directly or on a survey, about my / the patient's health habits	2.8
	7	*[I was]* Helped to make a treatment plan[6] that I / the patient could do in my / his / her daily life	2.5
	8	*[I was]* Helped to plan ahead so I could take care of my / the patient's rare or complex disease even in hard time	2.5
	9	*[I was]* Asked how my / the patient's rare or complex disease affects my / the patient's life	2.8
	9C[7]	*[I was]* Asked how the patient's rare or complex disease affects my *[the carer's]* life	2.2
	10	*[I was]* Contacted after a visit to see how things were going	2.0
	11	*[I was]* Told how my visits with other specialists, like a geneticist or cardiologist, helped my / the patient's treatment	2.5
Additional questions on psychological support	12	*[Healthcare professionals]* Helped me / the patient deal with emotions related to my / his / her health status	2.3
	12C[7]	*[Healthcare professionals]* Helped me *[the carer]* deal with emotions related to the patient's health status	2.2

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[5] http://www.eurordis.org/guidelines_hcaresurvey

[6] A treatment plan is a list, made with your care team, of what needs to be done to take care of your health.

[3] Questions 9C and 12C were only asked to carers.

Patients and carers who responded to the H-CARE Survey were approached online but also onsite, in 36 participating hospital units, where healthcare professionals distributed posters and leaflets to encourage them to participate in the survey.

605 respondents

European patients and carers treated in Centres of Expertise for rare kidney diseases, rare urogenital diseases, rare lung diseases or genetic tumour risk syndromes.
They were contacted through EURORDIS networks and through 36 hospital units that are part of the European Reference Networks ErkNet, eUROGEN, Genturis or LUNG:

European Reference Network | Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERKNet The European Rare Kidney Disease Reference Network

European Reference Network | ERN-LUNG European Reference Network for rare respiratory diseases

European Reference Network | eUROGEN *Urogenital Diseases*

EURORDIS RARE DISEASES EUROPE

Rare Barometer A EURORDIS INITIATIVE

3300 respondents

Patients and carers living with all types of rare diseases in the world.
They were contacted through EURORDIS networks:

EURORDIS RARE DISEASES EUROPE

Rare Barometer A EURORDIS INITIATIVE

THE FIRST EVER LARGE SCALE QUANTITATIVE SURVEY MEASURING THE HEALTHCARE EXPERIENCE OF PEOPLE LIVING WITH A RARE DISEASE

3905 respondents

23 languages

December 2019 to march **2020**

TARGET POPULATION:
patients living with a rare disease or family members (parents and close relatives) over 12 years old

65 countries and
900+ diseases represented

THANK YOU to all people living with a rare disease who participated in the survey, to the healthcare providers and patient representatives of the European Reference Networks ErkNet, eUROGEN, LUNG and Genturis who participated in the design and the dissemination of the survey, and to Rare Barometer partners!

Rare Barometer A EURORDIS INITIATIVE

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