**European Parliament**
2024-2029

*Subcommittee on Public Health*

16.12.2024

# WORKING DOCUMENT

on consultation on rare disease

Subcommittee on Public Health

# INTRODUCTION

Thank you for launching the European Parliament survey on **Rare diseases**. The survey is open from X to X.

A disease is called a rare disease if it affects no more than five persons in 10 000. While each rare disease affects a very limited number of people, the number of different rare diseases is estimated to be between 6 000 and 8 000. Thus, the total number of people living with a rare disease in the EU is estimated to be between 27 and 36 million.

The low number of patients per rare disease creates multiple problems, in particular regarding their diagnosis, care and treatment. One of the issues is low market incentives for the development of treatments for rare diseases, limited expertise for research and for the diagnosis and treatment of the rare diseases as well as high costs of the existing medicines.

The purpose of this public consultation is to provide a basis for the forthcoming work of the Committee of Public Health in bringing a better understanding and detailed knowledge of the challenges of persons affected by rare diseases and views of persons working with or involved in rare diseases.

**This survey is voluntary and anonymous**.

We do not ask for names, registration numbers or similar data. If you provide such information in the free text fields, it will removed before your contribution is included in the detailed results of the survey. If you believe that any question would allow a person to be identified, or if you simply do not feel comfortable answering it, please, skip it without replying to it.

Click here to read our **Data Protection Notice** about how we will use the information you provide when completing this survey.

We greatly appreciate your participation in the survey.

## *[Consultation on Rare Diseases - healthcare professionals and researchers]*[1]

*Initial sorting question*

1. Are you are replying as:[2]

a. an individual
b. a patient/support group
**c. a healthcare professional, researcher, healthcare provider, research institute or university**
d. an international organisation, government or other public body
e. a stakeholder or interest group

*About you and your organisation*

1. What is your current professional role?

- ○ Doctor
- ○ Nurse
- ○ Physiotherapist
- ○ Midwife
- ○ Pharmacist
- ○ Dietician
- ○ Dentist
- ○ Psychologist
- ○ Other healthcare professional
- ○ Researcher in the field of health
- ○ Representative of a healthcare provider
- ○ Representative of a research institute

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[1] Not to be translated.
[2] Repeats, so only translate once

- ○ Representative of a university
- ○ Other

*If the answer for Q1 is Representative of a healthcare provider, Representative of a research institute or Representative of a university display the following questions then proceed for all with*

*2.* If you represent a healthcare provider/research institute/university,

*If answer is healthcare provider/research institute/university:*

3. Where is your organisation based?
(List of countries)

4. Has your organisation received funding from the EU in the last five years?

- Yes
- No
- Prefer not to answer

5. Is your organisation registered in the EU Transparency Register? (https://transparency-register.europa.eu/)

- Yes, under the following registration number: ________________
- No

Q2. Are rare diseases your primary area of research/clinical expertise?

a. Yes, it is my/our primary area of research/clinical expertise
b. It is one of my/our areas of research/clinical expertise
c. It is not one of my/our areas, but I have worked with rare disease patients or cases
d. No, it is not my/our area of research/clinical expertise / I/we do not have any research or clinical experience with rare diseases

*If Q2 is answered with options **a,b,c,** the questions below (Q3-Q12) are displayed*

Q3. What is your field of expertise with rare diseases?

- ○ Allergy and immunology
- ○ Anesthesiology
- ○ Dermatology
- ○ Diagnostic radiology
- ○ Emergency medicine
- ○ Family medicine/General Practitioner
- ○ Internal medicine
    - General internal medicine
    - Endocrinology
    - Gastroenterology
    - Cardiology
    - Cardiovascular disease
    - Pulmonology
    - Rheumatology
    - Haematology
    - Infectious disease
    - Critical care medicine
    - Nephrology
    - Geriatrics
    - Oncology
- ○ Medical genetics
- ○ Neurology
- ○ Nuclear medicine
- ○ Obstetrics and gynecology
- ○ Ophthalmology
- ○ Pathology
- ○ Pediatrics
- ○ Physical/sports medicine and rehabilitation
- ○ Preventive medicine
- ○ Psychiatry
- ○ Radiation oncology
- ○ Surgery
- ○ Urology
- ○ Other

Q4. What is the patient population of your research/clinical expertise with rare diseases?

- ○ Children
- ○ Adults (over 18)
- ○ Both

Q5. How many years have you been involved in rare disease research/patient care?

- ○ Less than 5 years
- ○ 5-10 years
- ○ More than 10 years

Q6. In which country do you conduct your primary research/work?
(Scroll down list of countries)

Q7. How often do you collaborate with other research centres/healthcare professionals/institutions <u>in your country?</u>

- ○ Never
- ○ At least once a year
- ○ At least once a month
- ○ At least once a week
- ○ Prefer not to answer

Q8. How often do you collaborate with other research centres/healthcare professionals/institutions <u>outside your country</u>?

- ○ Never
- ○ At least once a year
- ○ At least once a month
- ○ At least once a week
- ○ Prefer not to answer

Q9. How do you keep up to date with the latest developments in the field? (Multiple choice)

- ○ Medical conferences, webinars, and seminars
- ○ Medical journals, newsletter or medical databases
- ○ Educational courses / medical trainings
- ○ Online discussion forum
- ○ Social media
- ○ Other

Q10. In your opinion, is there sufficient research on rare disease?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q11. Do you agree with the opinion that in general the diagnosis of rare diseases are efficient and does not take too long.

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q12. What are the biggest challenges you face in researching/managing rare diseases? (Max 3 choices)

- ○ Lack of funding and investments
- ○ Lack of financial incentives
- ○ Lack of collaboration and networking
- ○ Lack of policy support
- ○ Geographical barriers
- ○ Lack of data
- ○ Limited patient population
- ○ Lack of disease awareness
- ○ Lack of adequate treatments
- ○ Lack of specialised healthcare centres/teams

- Consent and Ethical Considerations
- Other__________

Q13. What types of resources or support services do you find most helpful for rare disease patients? (Max 3)

- Rare disease registries and databases
- Specialised centres and networks
- Genetic testing
- Telemedicine and digital health tools
- Multidisciplinary care teams
- Mental health and social support services
- Patient support groups
- Single contact point for all medical and administrative needs
- Information on ongoing and new clinical trials for patients and doctors
- Access to clinical trials
- Other

Q14. What types of resources or support services do you think need improvements for rare disease patients? (Max 3)

- Rare disease registries and databases
- Specialised centres and networks
- Genetic testing
- Telemedicine and digital health tools
- Multidisciplinary care teams
- Mental health and social support services
- Patient support groups
- Single contact point for all medical and administrative needs
- Information on ongoing and new clinical trials for patients and doctors
- Access to clinical trials
- Other

Q15. If you are engaged in rare disease research, what are your primary sources of funding? (Choose as many as applies)

- Government agencies
- European Union research programs
- International organisations
- Non-profit organisations and foundations
- Philanthropy
- Academic institutions
- Pharmaceutical and biotech companies
- Private sources
- Other

Q16. What funding opportunities for cross-border collaboration do you consider most important for advancing research on rare diseases?

- Public research funding in your country
- Public research funding from another country
- Horizon Europe projects
- Other EU grants
- International organisations
- Academic networks
- Industry cooperation
- Other

Q17. In your opinion, should cross-border data sharing be improved within the rare disease research/healthcare community?

- Yes, within the EU
- Yes, internationally
- No
- Prefer not to answer

*If the answer for Q17 was yes*

Q18. How can cross-border data sharing be improved?

| Free Text |
|---|

Q19. Are you aware of EU actions in the area of rare diseases?

(Choose as many as applies):

- ○ I am not aware of any EU actions in the area of rare diseases
- ○ European Experts group on rare diseases
- ○ Screen4care project
- ○ European Reference Networks
- ○ EUROCAT network
- ○ Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)
- ○ Funding for R&D
- ○ EUROBIO Bank
- ○ EU RD platform
- ○ European Rare Diseases Research Alliance (ERDERA)
- ○ Other

Q20. In your opinion, how could the EU improve its support for rare disease research?

(Choose as many as applies)

- ○ Financing of EU networks for rare disease
- ○ Coordination of legislation in member states
- ○ Enhance cooperation between member states
- ○ Increased international cooperation
- ○ Awareness raising
- ○ Expansion of EU wide database with anonymised patients data
- ○ More funding for research
- ○ No improvement is needed
- ○ Other

Q21. In your opinion, how could the EU improve its support for treatment development?

| |
|---|
| Free Text |

Q22. What further matters would you like to raise in relation to the question of rare diseases?

| |
|---|
| Free text |

## *[Consultation on Rare Diseases - Individuals]*

*Initial sorting question*

1. Are you are replying as:
   - **f. an individual**
   - g. a patient/support group
   - h. a healthcare provider, researcher, research institute or university
   - i. an international organisation, government or other public body
   - j. a stakeholder or interest group

**For everyone**
1. In which country do you live? *Choose from list*

2. Date of birth

3. What is your gender?
   - ○ Woman
   - ○ Man
   - ○ Non-binary
   - ○ Prefer not to answer

4. What is your highest level of education?
   - ○ Less than secondary school
   - ○ Secondary school graduate (includes equivalency)
   - ○ Bachelor's degree
   - ○ Master or Ph.D.
   - ○ Graduate or professional degree from vocational school
   - ○ Prefer not to answer

5. What is your current occupation?

- ○ Student
- ○ Full time employed
- ○ Part time employed
- ○ Self employed
- ○ Unemployed or temporarily not working
- ○ Retired
- ○ Other: ________________________

6. Where do you live?

- ○ Large city or suburb of a large city
- ○ Smaller city/town or on a rural area
- ○ Countryside

Q1. Which category describes you best?

A. A person living or having lived with a rare disease

B. A person at risk of a rare disease

C. A parent of a person with a rare disease

D. A relative of or close to a person living with a rare disease

E. Other

*If the answer is A, B, C, D, the questions below are displayed.*

Q2. Do you (or your close person) have a confirmed diagnosis for the disease?

- ○ Yes
- ○ No
- ○ Prefer not to answer

Q3. Does the condition have a specific name?

- ○ Yes
- ○ No
- ○ I don’t know
- ○ Prefer not to answer

Q3. To which category does belong the rare disease that you (or your close person) have? (Choose as many as applies)

- ○ Genetic tumour risk syndromes
- ○ Hematological disorders
- ○ Pediatric cancers (including hematological cancers)
- ○ Solid tumors/cancers
- ○ Neurological conditions
- ○ Neuromuscular disorders
- ○ Craniofacial anomalies and Ear, Nose, and Throat (ENT) conditions
- ○ Skin disorders
- ○ Eye conditions
- ○ Heart conditions
- ○ Kidney disorders
- ○ Liver (Hepatological) conditions
- ○ Respiratory disorders
- ○ Urogenital disorders
- ○ Endocrine system
- ○ Connective tissue and musculoskeletal disorders
- ○ Malformation syndromes, intellectual disabilities, and neurodevelopmental disorders
- ○ Immunodeficiency, autoinflammatory and autoimmune diseases
- ○ Hereditary metabolic disorders
- ○ Inherited and congenital (digestive and gastrointestinal) anomalies
- ○ Multisystemic vascular diseases

Q5. How many years have you (or your close person) been living with a rare disease?

- ○ Less than 1 year
- ○ 1 - 5 years
- ○ More than 5 years
- ○ Prefer not to answer

Q6. Do you think that you (or your close person) have sufficient information about the disease?
Very sufficient - Sufficient - Fair - Insufficient - Very insufficient - Prefer not to answer

Q7. How long did it take to receive the first diagnosis after the symptoms first started?

- ○ Less than 1 year
- ○ 1 - 2 years
- ○ 2 - 5 years
- ○ More than 5 years
- ○ Prefer not to answer

Q8. Have you (or your close person) received all the necessary support for the diagnosis in your country?
agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

Q9. In your opinion, would it shorten the time necessary for the diagnosis if you (or your close person) had access to the healthcare facilities (doctors, laboratories) outside your country?
agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

Q10. Is there currently a treatment available for your (or your close person's) disease?

- ○ Yes
- ○ No
- ○ Prefer not to answer

*If the answer is yes, the question Q11, Q12 and Q13 are displayed*

Q11. Does the treatment

- ○ Cure the disease definitively
- ○ Cure the disease temporarily
- ○ Treat only the symptoms

○ Prefer not to answer

Q12. Do you (or your close person) have access to the treatment?

○ Yes, the treatment is in a healthcare facility within my country
○ Yes, the treatment is in a healthcare facility in another EU country
○ Yes, the treatment is in a healthcare facility outside the EU
○ No, there is no access to the treatment
○ Prefer not to answer

Q13. Is the cost of this treatment covered by your (or your close person's) national healthcare insurance?

○ Yes
○ Yes, but only partly
○ No
○ Prefer not to answer

Q14. Do you (or your close person) have easy access to the health services you need from your place of residence?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q15. Do you (or your close person) receive any kind of support from public institutions? (Choose as many as applies)

a. Personal assistance
b. Financial support
c. Mobility aid
d. Psychological support
e. Housing adaptation
f. other
g. I do not receive any kind of support

h. Prefer not to answer

*If any except g:*

Q16. How difficult was it to obtain the support you (or your close person) needed?

- very difficult - quite difficult - neither difficult, nor easy - quite easy - very easy - prefer not to answer

Q17. Do you (or your close person) receive any kind of support from any Non-Governmental Organisations? (Choose as many as applies)

i. Personal assistance

j. Financial support

k. Mobility aid

l. Psychological support

m. Housing adaptation

n. Other

o. I do not receive any kind of support

p. Prefer not to answer

*If any except g or h:*

Q18. How difficult did you (or your close person) find it to obtain the support you needed?

very difficult - quite difficult - neither difficult, nor easy - quite easy - very easy - prefer not to answer

Q19. Which barriers are you (or your close person) facing in obtaining support?
(Choose as many as applies)

- Lack of information about available support services
- Limited availability of support services
- High cost of support services
- Long waiting times

- Geographical barriers
- Difficult processes for accessing support services
- Other__________________
- I do not face any barriers
- I do not need any kind of support
- Prefer not to answer

Q20. Have you ever experienced discrimination related to rare disease or disability?

- Prefer not to answer
- Never
- Yes, but rarely
- Yes, often
- Always

*If the answer is yes, but rarely; yes, often or always, then:*

Q21. In what field?
(Choose as many as applies)

- At work
- Education
- Healthcare
- Public services
- Financial sector (e.g. banks, insurance)
- Public spaces
- Private spaces (e.g. family, friends)
- Other: _________________
- Prefer not to answer

Q22. Does the rare disease affect your (or your close person's) daily life?

- Prefer not to answer
- No, it doesn't
- Yes, inability (or difficulty) to work
- Yes, difficulty to find the work adequate to the professional experience or educational

background

- Yes, inability (or difficulty) to take care of own household (e.g. shopping, cleaning, cooking...)
  - Yes, difficulty in accessing social and cultural events
  - Yes, inability (or difficulty) to care of family members (e.g. children, spouse, other relatives)
  - Yes, other: _____________

Q23. Do you feel that your (or your close person's) participation in social life is limited by rare disease?

- Prefer not to answer
- I (or my close person) fully participate without any limitations
- I (or my close person) participate, but with some limitations
- I (or my close person) rarely participate
- I (or my close person) do not participate
- Other_______

Q24. In your opinion, are the healthcare facilities in your country adequately prepared for treating your (or your relative's) disease?

- Yes
- Partially
- No
- Prefer not to answer

Q25. In your personal experience, what impact have the changes and health restrictions during the COVID-19 pandemic had on your (or your relative's) access to care?

- The access got a lot worse

- The access got worse
- The access stayed about the same
- The access got better
- The access got a lot better
- Prefer not to answer

Q26. In your personal experience, in the last 2-3 years, the access to your (or your relative’s) care has gotten:

- A lot worse
- Worse
- Stayed about the same
- Better
- A lot better
- Prefer not to answer

Q27. In your opinion, what are your (or your relative’s) biggest difficulties in care-seeking? (Several answers possible)

- No difficulty
- Lack of general practitioners/family doctors
- Lack of specialists
- No adequate referral
- Lack of adequate equipment within healthcare setting
- Difficulty in obtaining an accurate diagnosis
- Limited treatment options

- ○ High cost of care/medicines
- ○ Lack of support
- ○ No care or limited care within your country
- ○ No care or limited care in own region
- ○ Other: ___________________________
- ○ Prefer not to answer

Q28. In your opinion, what is the best solution to address these challenges?

- ○ Collaboration and coordination between healthcare professionals
- ○ Increased funding for healthcare
- ○ More specialised centres
- ○ Increased access to general practitioners/family doctors
- ○ Increased access to specialists
- ○ Increased funding for research on rare diseases
- ○ Faster approval of new treatments/therapies
- ○ Introduction (in your country) of existing treatment that until now has been unavailable
- ○ Increase access to existing treatments abroad that are not available in your country
- ○ Greater patient engagement
- ○ Creation or expansion of support networks
- ○ Other: ___________________________

- Prefer not to answer

Q29. Are you aware of EU actions in the area of rare diseases?
(Choose as many as applies):

- I am not aware of any EU actions in the area of rare diseases
- European Experts group on rare diseases
- Screen4care project
- European Reference Networks
- EUROCAT network
- Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)
- Funding for R&D
- EUROBIO Bank
- EU RD platform
- European Rare Diseases Research Alliance (ERDERA)
- Other

Q31. Do you think that the EU should be doing more for rare disease patients?

- Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q32. Would you agree to share your medical records (anonymously) if this would help the treatment of other persons living with rare diseases?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q33. Is there anything else you would like to share regarding your experience with care for rare diseases?

| Free text |
|---|

***If in Q1 selected answer E (other)***

Q6. Do you agree that it is important to invest in rare disease research?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q7. In your opinion, do people with rare diseases have access to the right kind of support?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

*If disagree or partially disagree*:

Q8. What is the main lack in support for persons with rare diseases?
(Choose as many as apply or none if nothing is lacking).

- o Lack of general practitioners/family doctors
- o Lack of specialists
- o No adequate referral
- o Lack of adequate equipment within healthcare setting
- o Difficulty in obtaining an accurate diagnosis
- o Limited treatment options
- o High cost of care/medicines
- o Lack of support
- o No care or limited care within your country
- o No care or limited care in own region
- o Other: ____________________________
- o Prefer not to answer

Q9. In your opinion, what would be the most effective way to raise public awareness about rare diseases?

○ Online promotion campaigns

○ Off-line promotion campaigns

○ TV or radio awareness

○ Conference speeches

○ Studies and publications

○ Press articles

○ Organising events

○ Trainings

○ Advertisements

○ Presentations in schools

○ Gadgets distribution

○ Other _________

Q10. Are you aware of EU actions in the area of rare diseases?
(Choose as many as applies):

○ I am not aware of any EU actions in the area of rare diseases

○ European Experts group on rare diseases

○ Screen4care project

○ European Reference Networks

○ EUROCAT network

○ Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)

- ○ Funding for R&D
- ○ EUROBIO Bank
- ○ EU RD platform
- ○ European Rare Diseases Research Alliance (ERDERA)
- ○ Other

Q12. Do you think that the EU should be doing more for rare disease patients?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q13. Would you agree to share your medical records (anonymously) if this would help the treatment of persons living with rare diseases?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q14. Is there anything else you would like to share regarding your experience with care for rare diseases?

| |
|---|
| Free text |

## *[Consultation on Rare Diseases - Patient and support groups]*

*Initial sorting question*

1. Are you are replying as:

k. an individual
**l. a patient/support group**
m. a health care provider, researcher, research institute or university
n. an international organisation, government or other public body
o. a stakeholder or interest group

*About your organisation*

1. What is the name of your organisation?

2. How many employees/volunteers does your organisation have?

- 1-9
- 10-49
- 50-249
- 250-499
- 500 or more

3. Where is your organisation?
(List of countries)

4. Has your organisation received funding from the EU in the last five years?

- Yes
- No

5. Is your organisation registered in the EU Transparency Register? (https://transparency-register.europa.eu/)

- Yes, under the following registration number: ________________
- No

6. Is your organisation mainly focusing on:

- ○ patients of all needs
- ○ patients with rare diseases
- ○ other specific group of patients

7. Is your organisation active in:

- One of the EU Member State
- In multiple (or all) EU Member States
- Not active in any EU Member State

*Needs of rare diseases patients*

Q1. What are the major issues of rare disease patients according to your organisation?
(Choose as many as applies)

- Difficulty in accessing care
- Hinders for accessing cross-border healthcare
- Difficulty in diagnosis
- Limited access to treatments
- High cost of care
- High cost of treatments
- High costs of accessing care (travel costs, etc)
- Access to support in the daily life
- Access to psychological support
- Other ________

Q2. Do you take actions to raise awareness about rare diseases and if so through which means?
(Choose as many as applies)

- We do not take such actions
- Online promotion campaigns
- Off-line promotion campaigns
- TV or radio awareness
- Conference speeches
- Studies and publications
- Press articles
- Organising events

- Trainings
- Advertisements
- Presentations in schools
- Gadgets distribution
- Other _________

Q3. What are the major issues that your organisation faces?
(Choose as many as applies)

- Reaching patients
- Raising awareness on the importance of rare diseases
- Raising funds
- Mobilisation volunteers
- Finding staff with adequate knowledge about rare diseases
- Other ______________

Q4. Do you collaborate with other stakeholders?
(Choose as many as applies)

- We do not collaborate with other stakeholders
- Public entities in your country
- Public entities outside your country
- International bodies (including the EU)
- Patient and support groups
- NGOs/Charity organisations
- Healthcare institutions (hospitals, clinics...)
- Health professionals organisations
- Industry
- Media

Q5. What types of resources or support services do you find most helpful for rare disease patients?

(Choose as many as applies)

- Rare disease registries and databases
- Specialised centres and networks
- Genetic testing
- Telemedicine and digital health tools
- Multidisciplinary care teams
- Mental health and social support services
- Patient support groups
- Single contact point for all medical and administrative needs
- Information on ongoing and new clinical trials for patients and doctors
- Access to clinical trials
- Other ____________

Q6. In your opinion, what types of resources or support services for rare disease patients need improvements?
(Choose as many as applies)

- Rare disease registries and databases
- Specialised centres and networks
- Genetic testing
- Telemedicine and digital health tools
- Multidisciplinary care teams
- Mental health and social support services
- Patient support groups
- Single contact point for all medical and administrative needs
- Information on ongoing and new clinical trials for patients and doctors
- Access to clinical trials
- Other

Q7. In your opinion, in which fields can EU improve its actions?
(Choose as many as applies)

- Level of EU funding for research
- Time necessary for approval of new treatments/therapies
- Access to cross border healthcare
- Collaboration and coordination between healthcare professionals
- Collaboration between healthcare institutes/centres
- Patient engagement
- Specialised centres for rare disease
- Support networks
- Harmonisation of Member States’ legislation
- Coordination of multinational research
- Other

Q8. In your opinion, in which fields can EU intervene more efficiently than the Member States alone?

(More than one option can be chosen)

- Support for research networks
- Support for development of database for research
- Support for development of database (information repository) for patients
- Coordination of national legislation
- Regulating the market
- Increased international cooperation
- Funding for research/medicines/treatments
- Awareness raising
- Other

Q9. Are you aware of EU actions in the area of rare diseases?

(Choose as many as applies):

- I am not aware of any EU actions in the area of rare diseases
- European Experts group on rare diseases

- ○ Screen4care project
- ○ European Reference Networks
- ○ EUROCAT network
- ○ Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)
- ○ Funding for R&D
- ○ EUROBIO Bank
- ○ EU RD platform
- ○ European Rare Diseases Research Alliance (ERDERA)
- ○ Other
- ○

Q10. Is there anything else you would like to share regarding your views on care for rare diseases?

| |
|---|
| Free text |

***[Consultation on Rare Diseases - Stakeholder and interest groups]***

*Initial sorting question*

1. Are you are replying as:

p. an individual
q. a patient/support group
r. a health care provider, researcher, research institute or university
s. an international organisation, government or other public body
**t. a stakeholder or interest group**

*About your organisation*

1. What is the name of your organisation?

Q1. Please indicate the type of organisation or company:

- ○ Non-governmental organisation
- ○ Think tank
- ○ Media
- ○ Consumer organisation
- ○ Industry association
- ○ Company/SME/Start-up
- ○ Interest group
- ○ Trade union
- ○ Other_______

4. How many employees/volunteers does your company/organisation have?

- 1-9
- 10-49
- 50-249
- 250-499
- 500 or more

5. Where is your organisation/company based?
(List of countries)

6. Has your organisation/company received funding from the EU in the last five years?

Yes - No

7. Is your organisation registered in the EU Transparency Register (https://transparency-register.europa.eu/)?

- Yes, under registration number:_____________________
- No

### *Policy of the European Union*

Q1. What are the major issues of rare disease patients according to your organisation?
(Choose as many as applies)

- Difficulty in accessing care
- Hinders for accessing cross-border healthcare
- Difficulty in diagnosis
- Limited access to treatments
- High cost of care
- High cost of treatments
- High costs of accessing care (travel costs, etc)
- Access to support in the daily life
- Access to psychological support
- Other ________

Q2. Do you take actions to raise awareness about rare diseases and if so through which means?
(Choose as many as applies)

- We do not take such actions
- Online promotion campaigns
- Off-line promotion campaigns
- TV or radio awareness
- Conference speeches
- Studies and publications
- Press articles
- Organising events
- Trainings
- Advertisements
- Presentations in schools
- Gadgets distribution

- Other _________

Q3. Do you collaborate with other stakeholders?
(Choose as many as applies)

- We do not collaborate with other stakeholders
- Public entities in your country
- Public entities outside your country
- International bodies (including the EU)
- Patient and support groups
- NGOs/Charity organisations
- Healthcare institutions (hospitals, clinics...)
- Health professionals organisations
- Industry
- Media

Q4. What types of resources or support services do you find most helpful for rare disease patients?
(Choose as many as applies)

- Rare disease registries and databases
- Specialised centres and networks
- Genetic testing
- Telemedicine and digital health tools
- Multidisciplinary care teams
- Mental health and social support services
- Patient support groups
- Single contact point for all medical and administrative needs
- Information on ongoing and new clinical trials for patients and doctors
- Access to clinical trials
- Other ____________

Q5. In your opinion, what types of resources or support services for rare disease patients need improvements?
(Choose as many as applies)

- ○ Rare disease registries and databases
- ○ Specialised centres and networks
- ○ Genetic testing
- ○ Telemedicine and digital health tools
- ○ Multidisciplinary care teams
- ○ Mental health and social support services
- ○ Patient support groups
- ○ Single contact point for all medical and administrative needs
- ○ Information on ongoing and new clinical trials for patients and doctors
- ○ Access to clinical trials
- ○ Other

Q6. In your opinion, in which fields can EU improve its actions?
(Choose as many as applies)

- ○ Level of EU funding for research
- ○ Time necessary for approval of new treatments/therapies
- ○ Access to cross border healthcare
- ○ Collaboration and coordination between healthcare professionals
- ○ Collaboration between healthcare institutes/centres
- ○ Patient engagement
- ○ Specialised centres for rare disease
- ○ Support networks
- ○ Harmonisation of Member States' legislation
- ○ Coordination of multinational research
- ○ Other

Q8. In your opinion, does the EU and its industry possess the necessary knowledge and technologies to satisfy patients’ needs?

Yes - Mostly - Partly - No - Prefer not to answer

*If the answer is Mostly - Partly - No, the following question is displayed*

Q9. What skills or infrastructure are missing in the EU?

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| Free text |

Q10. In your opinion, how important is the role of the EU in supporting rare disease research and treatment development?

Very important - Quite important - Not important - Counterproductive - Prefer not to answer

Q11. In your opinion, does the EU adequately support research on medicines and treatment for rare diseases?

agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

*If the answer is agree partly - neither agree, nor disagree - disagree partly - disagree fully, the following question is displayed*

Q12. What should be changed?

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| Free text |

Q13. In your opinion, does the EU adequately support introduction of new medicines and treatment for rare diseases to the market?

agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

*If the answer is agree partly - neither agree, nor disagree - disagree partly - disagree fully, the following question is displayed*

Q14. What should be changed?

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| Free text |

Q15. In your opinion, does the industry in the EU have the necessary production potential for rare disease medicines?

agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

Q16. In your opinion, does the EU adequately support production potential of the EU industry for rare diseases medicines?

agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

*If the answer is agree partly - neither agree, nor disagree - disagree partly - disagree fully, the following question is displayed*

Q17. What should be changed?

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| Free text |

Q18. In your opinion, does the EU have a sufficiently large market for economically sustainable commercialisation of new medicines for rare diseases?

agree fully - agree partly - neither agree, nor disagree - disagree partly - disagree fully - prefer not to answer

Q19 . In your opinion, what could be done to make medicines more affordable and accessible to patients?

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| Free text |

*If the answer to Q1 is Company/SME/Start-up display Q*

*Research and development of medicines and treatments for rare diseases.*

QC1. Is your company directly involved in researching or developing of medicines or treatments for rare diseases?

Yes - No

*If the answer is Yes*

QC2. Does your company cooperate in the research on rare diseases with partners from other EU Member States?

Yes - No

QC3. What do you consider the main challenges in the research and development of medicines or treatments for rare disease?

- ○ Limited funding and investments
- ○ Regulatory restrictions
- ○ Limited access to patients data
- ○ Lack of experienced staff
- ○ Lack of incentives for the commercialisation of new medicines or treatments
- ○ Difficulty in accessing the market of other Member States
- ○ Competition from outside the EU
- ○ Other__________

QC4. Is your company producing or distributing medicines for rare diseases?

Yes - No

*If the answer is Yes*

QC5. Is your company producing or distributing medicines based on proprietary technologies?

Yes - No

*If the answer for QC5 is No*

QC6. What is the origin of your technology (multiple choice possible):

- ○ EU
- ○ US
- ○ China
- ○ India
- ○ Other countries

QC7. In your opinion, do market prices cover the necessary costs of production and distribution (excluding research costs) of the medicines for rare diseases?

- ○ Yes
- ○ Mostly
- ○ Partly
- ○ No
- ○ Prefer not to answer

*To all respondents*

Q20. Are you aware of EU actions in the area of rare diseases?
(Choose as many as applies):

- ○ I am not aware of any EU actions in the area of rare diseases
- ○ European Experts group on rare diseases
- ○ Screen4care project
- ○ European Reference Networks
- ○ EUROCAT network
- ○ Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)
- ○ Funding for R&D
- ○ EUROBIO Bank
- ○ EU RD platform
- ○ European Rare Diseases Research Alliance (ERDERA)
- ○ Other

Q21. What further matters would you like to raise in relation to the question of rare diseases?

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| Free text |

***[Rare Diseases Consultation - Public and international organisations]***

*Initial sorting question*

1. Are you replying as:

u. an individual
v. a patient/support group
w. a health care provider, researcher, research institute or university
**x. an international organisation, government or other public body**
y. a stakeholder or interest group

## d. INTERNATIONAL ORGANISATION, GOVERNMENT OR OTHER PUBLIC BODY

*About your organisation*

Q1. Please indicate which type of public authority or international organisation you represent:

○ Government
○ Regional or local authority
○ International organisation
○ Other public authority or body

Q2. The name of your public authority/ international organisation:

________________________

Q3. In which country are you based?

<u>Scroll down list of countries</u>

*Policy of the European Union*

Q3. Are the current EU policies and legislation in the field of rare disease effective?

> Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q4. What are the most important gaps in the current healthcare system for rare disease patients that require urgent policy intervention? (Max 3 choices)

- ○ Limited competences of healthcare professionals
- ○ Lack of specialists
- ○ Limited budget of your healthcare system in your country
- ○ Difficult access to adequate healthcare services in your country
- ○ Difficult access to adequate cross-border healthcare services
- ○ Low level of public awareness about rare diseases
- ○ Lack of collaboration between healthcare centres in your country
- ○ Lack of international collaboration between healthcare centres from different countries
- ○ Unavailability of medicines and treatment
- ○ Insufficient access to medicines and treatment
- ○ High cost of care
- ○ Data sharing issues
- ○ Stigmatisation of rare diseases
- ○ Other

Q5. Are you satisfied with the support of your local government to rare disease research?

> Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q6. Are you satisfied with the support to rare disease research from the EU?

> Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q7. Are you collaborating with partner organisations, advocacy groups, international organisations or global partners in the rare disease field?

- ○ Yes, always
- ○ Yes, often
- ○ Yes, sometimes
- ○ Yes, but rarely
- ○ No
- ○ Prefer not to answer

Q8. In your opinion, are there fields where the EU can improve its actions?
(Choose as many as applies)

- ○ Level of EU funding for research
- ○ Time necessary for approval of new treatments/therapies
- ○ Access to cross border healthcare
- ○ Collaboration and coordination between healthcare professionals
- ○ Collaboration between healthcare institutes/centres
- ○ Patient engagement
- ○ Specialised centres for rare disease
- ○ Support networks
- ○ Harmonisation of Member States’ legislation
- ○ Coordination of multinational research
- ○ Other

Q10. In your opinion, how can the EU improve the framework for <u>rare disease treatment</u>?

- ○ Harmonise Member States’ legislation
- ○ Facilitate the access to cross-border rare disease diagnosis and treatment
- ○ Finance more research networks
- ○ Build an incentive and flexible-based framework to boost investment in research for rare disease treatments
- ○ Directly finance rare disease treatment

- ○ Introduce of a price cap for rare disease treatment and medicines
- ○ Enhance cooperation with non-EU countries
- ○ Facilitate data sharing between EU countries
- ○ Facilitate data sharing between EU and non-EU countries
- ○ Improve the cooperation between EU research centres and patients
- ○ Other:_________

Q11. How can the EU improve the cooperation between research centres conducting clinical trials and patients all over the EU?
(Choose as many as applies)

- ○ Enhance the coherence and coordination for the existing clinical trial databases
- ○ Facilitate cross-border access to clinical trials
- ○ Improve the existing incentive framework for clinical trials
- ○ Reduce the administrative burden
- ○ Support the implementation of more patient-centred clinical trial
- ○ Support research and investment into clinical trials to develop a more efficient diagnostic system
- ○ Harmonise national requirements for clinical trials'
- ○ Support research partnerships between different stakeholders (academia, research centres, industry and health care institutions)
- ○ Other: __________________
- ○ No need for improvement

Q14. Would joint purchasing by the Member States of rare diseases medicines/treatments be a good idea?

Fully agree - Partially agree - Neither agree, nor disagree - Partially disagree - Fully disagree - Prefer not to answer

Q15. Are you aware of EU actions in the area of rare diseases?
(Choose as many as applies):

- I am not aware of any EU actions in the area of rare diseases
- European Experts group on rare diseases
- Screen4care project
- European Reference Networks
- EUROCAT network
- Mechanism of Coordinated Access to Orphan Medicinal Products (MoCA-OMP)
- Funding for R&D
- EUROBIO Bank
- EU RD platform
- European Rare Diseases Research Alliance (ERDERA)

Q16. Do you have any further remarks?

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| Free text |