TRAINING: USING RARE BAROMETER SURVEY RESULTS FOR ADVOCACY

EURORDIS membership meeting – workshop 3

Jessie Dubief, MPhil Sociology, Social Research Director,
EURORDIS-Rare Diseases Europe

TRAINING: USING RARE BAROMETER SURVEYS FOR ADVOCACY

1. Understanding survey methodology and using survey results

Moderator: Jessie Dubief, Social Research Director, EURORDIS

Friday: 14:00-15:30 ; Saturday: 9:00-10:30

30 minutes break

2. Writing exercises (per table)

With panellists from the community

Friday: 16:00-17:30; Saturday: 11:00-12:30

1. Writing for healthcare professionals and scientists (plan for a scientific poster)
2. Writing for policy makers (4-5 slides for an oral presentation)
3. Developing strategic objectives and indicators for a patient organisation

UNDERSTANDING SURVEY METHODOLOGY AND USING SURVEY RESULTS

1 **WHAT IS A SURVEY?**

To which questions do we want to answer?

2 **WHAT CAN YOU SAY WITH RARE BAROMETER RESULTS?**

How robust is the Rare Barometer information, and can you use it to talk about all the people living with a rare disease?

3 **HOW ARE THE RARE BAROMETER QUESTIONNAIRES DESIGNED?**

How do we go from research questions (*i.e.* questions we ask ourselves), to questions that are asked to the participants?

4 **HOW CAN YOU PRESENT RARE BAROMETER RESULTS?**

Which types of analysis and which types of graphs can you do?

TRAINING: HOW TO USE RARE BAROMETER RESULTS?

	Survey topic	**Scope** (rare diseases, geo)	**Panellist**	**Notions covered**
Table 1	The **diagnosis** odyssey of people living with a rare disease	All rare diseases, **Europe**	**Zoe Alahouzou**, EURORDIS	Mean, median, odd ratios
Table 2	The **diagnosis** odyssey of people living with a rare disease	**Hereditary Hemorrhagic Telangiectasia**, Europe	**Claudia Crocione**, HHT-Europe	Mean, median, boxplot
Table 3	The opinion of people living with rare diseases on **newborn screening**	All rare diseases, **United Kingdom**	**Nick Meade**, Genetic Alliance UK	Percentage, p-value
Table 4	The opinion of people living with rare diseases on **newborn screening**	**Fragile X Syndrome**, Europe	**Kirsten Johnson**, Fragile X International	Percentage, p-value
Table 5	The impact of living with a RD on daily life: **social participation and independent living**	All rare diseases, **Romania**	**Dorica Dan**, Romanian National Alliance for RD	Percentage, p-value
Table 6	The impact of living with a RD on daily life: **social participation and independent living**	**Neurofibromatosis**, Europe	**Claäs Rohl**, NF Patients United	Percentage, p-value

WHAT IS A SURVEY?

To which questions do we want to answer?

WHAT IS A SURVEY?

- A way of **answering questions that you have**, and for which you need to ask people
- **Needs to have clear goals and objectives.**
- **Need to have a clear target population** (or target audience)

OBJECTIVES OF THE SURVEYS

The diagnosis odyssey of people living with a rare disease (2022)

- Measuring the **time necessary to obtain a diagnosis** for people living with a rare disease
- Understand the **different steps** of the diagnosis journey
- Define the **obstacles limiting the access** to diagnosis
- Identify **best practices, tools, support and services** contributing to a faster diagnosis
- Identify the **role** of new technologies

OBJECTIVES OF THE SURVEYS

The opinion of people living with a rare disease on newborn screening (2023)

CREATING A SOUND BASIS TO

- **Advocate** for harmonised criteria and adequate policies for newborn screening.
- **Define** a list of conditions to be tested in the Screen4Care project.

UNDERSTAND

- **The attitudes and perceptions** of people living with rare diseases towards newborn screening for themselves, and for all rare diseases.
- **Which approach of newborn screening** is considered acceptable by people living with rare diseases.
- How the opinion of people living with rare diseases on newborn screening relates to **their characteristics** (age, gender, country, family situation…) and those of **their rare disease**.

OBJECTIVES OF THE SURVEYS

Social participation and independent living (2024)

- Estimate the level of **participation in social activities** such as education, work or leisure.
- Identify **barriers or facilitators** in doing those social activities.
- Understand **preferences and needs** regarding **living arrangements and personal assistance**.
- Collect **experiences with disability assessment**.
- Identify the main difficulties in accessing **social and disability rights**.

WHAT CAN YOU SAY WITH RARE BAROMETER RESULTS?

Do Rare Barometer results give information on the all the people living with a/my rare disease?

KNOW THE TARGET POPULATION

All the people you want to know about, i.e. all the people you should call, interview, or send the questionnaire to, if you could reach them all.

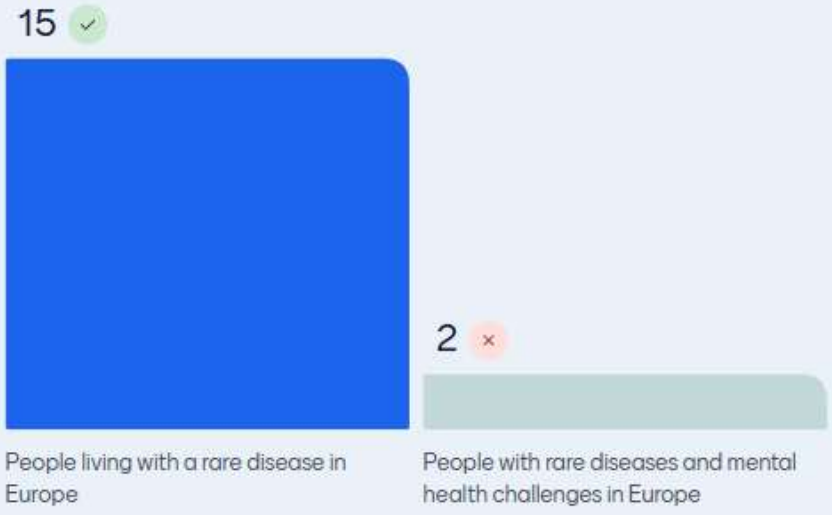
Ex: the 30 million people living with a rare disease in Europe, all patients living with Duchenne Muscular Dystrophy in Europe, all carers of patients living with NF1 in Germany, patients between 12 and 25 y.o. in a specific hospital, patients who have been referred to expert centres for rare kidney diseases, etc.

Join at menti.com | use code 5389 0813

Mentimeter

Target population

The Rare Barometer is launching a survey on the determinants of mental health among people with rare diseases in Europe. Who is the target population?



Menti

My first poll

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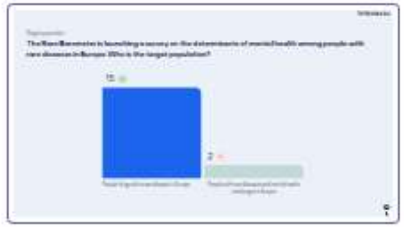

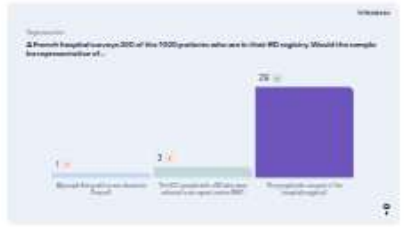

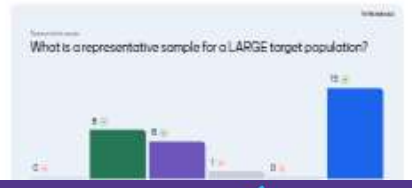

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Target population

A French hospital surveys 300 of the 1000 patients who are in their RD registry. Would the sample be representative of...

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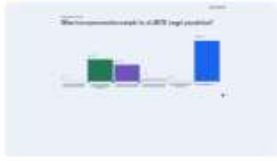

KNOW YOUR SAMPLE

It would be costly (in time and money) to investigate all the target population: instead, we use a **smaller subset of that population**. That group or section of the target population is called a **sample**.

We can then rely on some statistical values that can allow to estimate whether the results that were found in the sample could be **extrapolated** to the target population.

WHAT IS A REPRESENTATIVE SAMPLE?

LARGE target populations

For large target populations (e.g. the 30 million people living with a rare disease in Europe), a representative sample is a sample that…

…EITHER includes a *random* selection of people from the target population, which implies having a database of the target population
e.g. Random selection of voters from a voter list or vehicle owners from a vehicle registration database

…OR mimics the structure of the population, which implies knowing its structure.
e.g. if the target population comprises 52% of women and 15% of people under 25 years old, the sample should be similar to be representative.

…AND includes enough people for the results to be significant
At least 300 people; ideally 1000 people (the gain in robustness is negligible after 1000)

WHAT IS A REPRESENTATIVE SAMPLE?

LARGE target populations

For large target populations (e.g. the 30 million people living with a rare disease in Europe), a representative sample is NOT a sample that…

…corresponds to a certain percentage of the target population

e.g. you do NOT need to have at least 1% of the population, i.e. at least 300 000 people, to represent the 30 million people living with a rare disease in Europe.

…is made up of the largest number of participants you could reach out to

e.g. if your goal is to measure the risk factors of diabetes in your region, surveying all the people in the hospital registries does not give you a sample representative of your target population (adults with or without diabetes in the region).

WHAT IS A REPRESENTATIVE SAMPLE?

SMALL target populations

A good (if not 'representative') sample for small target populations is a sample that…
e.g. the 160 people living with dystonia 28 worldwide (source: Orphanet)

…corresponds to a 'good' percentage of the population
e.g. 100 people out of the 160 people with dystonia 28 worldwide: more than 60% of the target population.

…ideally, it mimics the main characteristics of the population.
e.g. when for a target population (a patient registry of 1000 patients with scleroderma) where 80% are female, the sample is also 80% female.

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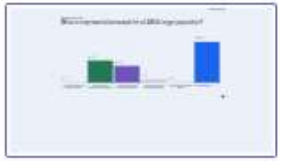
Mentimeter

Representative sample

What is a representative sample for a LARGE target population?

Menti
My first poll

Choose a slide to present



EURORDIS RARE DISEASES EUROPE

Rare Barometer
A EURORDIS INITIATIVE

A REPRESENTATIVE SAMPLE FOR ALL RARE DISEASES?

A representative sample supposes that we already know the characteristics of the population

What would be those characteristics in the context of rare conditions?

e.g. gender balance depending on gender prevalence of rare diseases, age distribution among people living with a rare disease, type of rare disease, diagnosis status, socioeconomic background (probably less relevant given that 70% of rare conditions are genetic)…

What do we know?

- More than 6000 different rare diseases with different prevalences and sociodemographics.
- What epidemiological information do we have on people living with a rare disease?

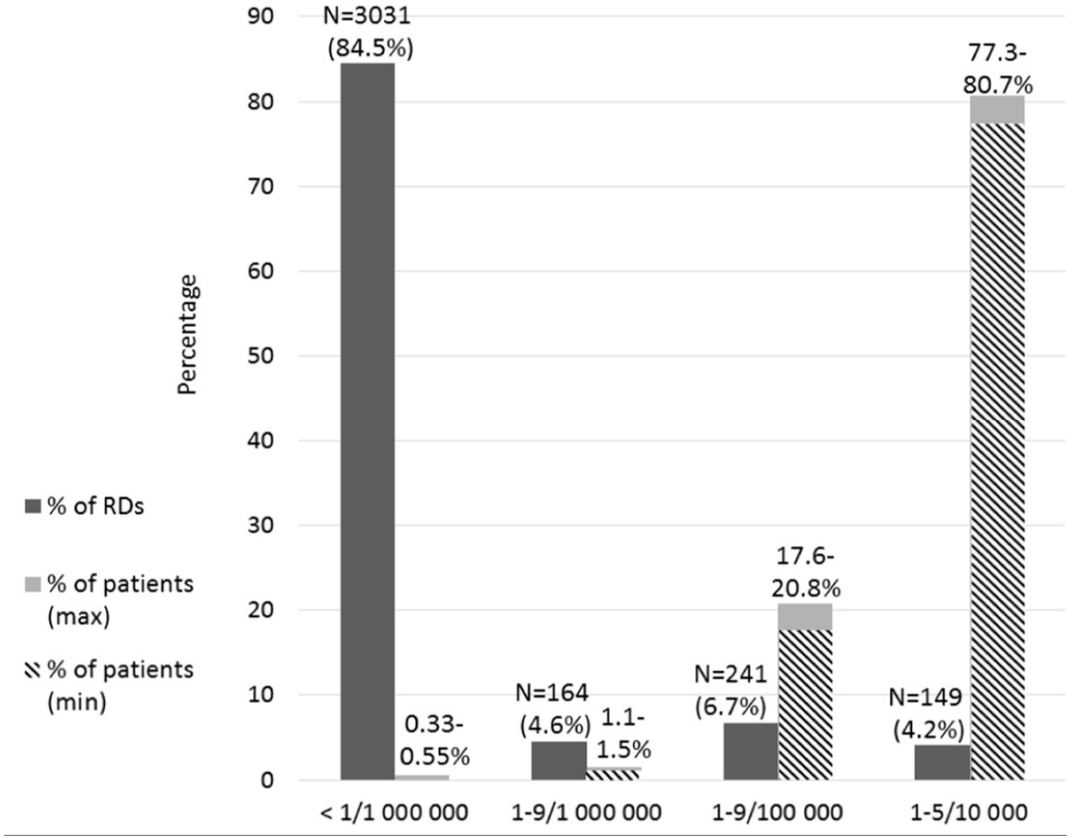
A REPRESENTATIVE SAMPLE FOR ALL RARE DISEASES?

What do we know about all people living with a rare disease in Europe ?

Orphanet study: point prevalence of rare diseases from **3585 conditions** with a known point prevalence (58% of RDs in their data):

- Conditions with a point prevalence between **5/10,000 and 1/100,000** represent **11% of the RDs considered** (390/3585), and **at least 95% of patients**.
- **Ultra-rare conditions** (point prevalence under 1/100,000) represent **89% of the RDs** considered (3195/3585), and **1-2% of the patients.**

That's all we know so far about people living with a rare disease in Europe



A REPRESENTATIVE SAMPLE FOR ALL RARE DISEASES?

Orphanet study:

- Conditions with a point prevalence between **5/10,000 and 1/100,000** represent **11% of the RDs considered** (390), and **at least 95% of patients**.
- **Ultra-rare conditions** (point prevalence under 1/100,000) represent **89% of the RDs** considered (3195), and **1-2% of the patients.**

What is the point prevalence in Rare Barometer samples?

Ex: survey on the impact of living with a rare disease on daily life (pages 35 of the report: tiny.cc/RB_DailyLife:

Table 19. Characteristics of the rare diseases represented.
All participants (n=9591).

Type	Percentage (n)
Diagnosis status	
Diagnosed	95% (9080)
Undiagnosed	5% (510)
Point prevalence of the rare disease (Orphanet)	
Between 5/10,000 and 1/100,000	75% (3796)
<1/100,000	25% (1244)
Not yet diagnosed or unknown point prevalence	47% (4451)

When representativeness is not achievable, we conduct an **exploratory research** by studying a group without making generalisation.

By multiplying exploratory researches, we constitute a **body of evidence**.

INFORMATION ON RARE BAROMETER SAMPLES

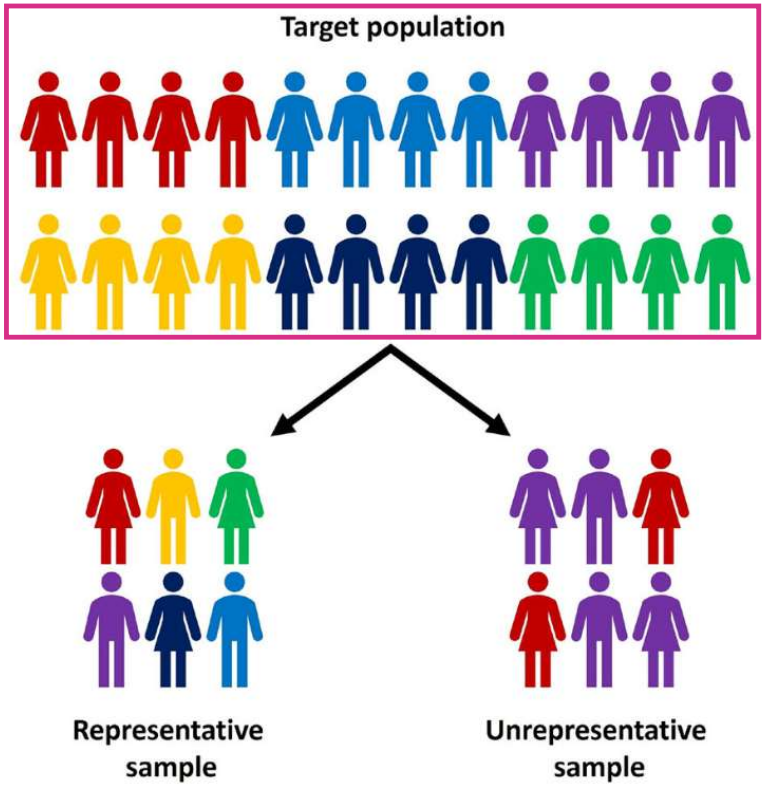
	Survey topic	Where is the sample information?
Table 1 **Table 2**	The **diagnosis** odyssey of people living with a rare disease	**Article:** sections 'Population and recruitment' and 'Characteristics of the respondents. **Factsheet:** top banner **Dashboard:** pages 4-16
Table 3 **Table 4**	The opinion of people living with rare diseases on **newborn screening**	**Report:** page 10-12 **Factsheet:** top banner **Dashboard:** pages 3-15
Table 5 **Table 6**	The impact of living with a RD on daily life: **social participation and independent living**	**Report**: pages 34-36 **Factsheet**: top banner **Dashboard**: pages 4-12

HOW ROBUST ARE OUR RESULTS?

AS ROBUST AS CAN BE!

Are our samples representative?
They cannot be because no one knows the target population (lack of epidemiological studies):
*we talk about **the participants**.*

All people living with a rare disease and their family members, including unsolved cases (undiagnosed)



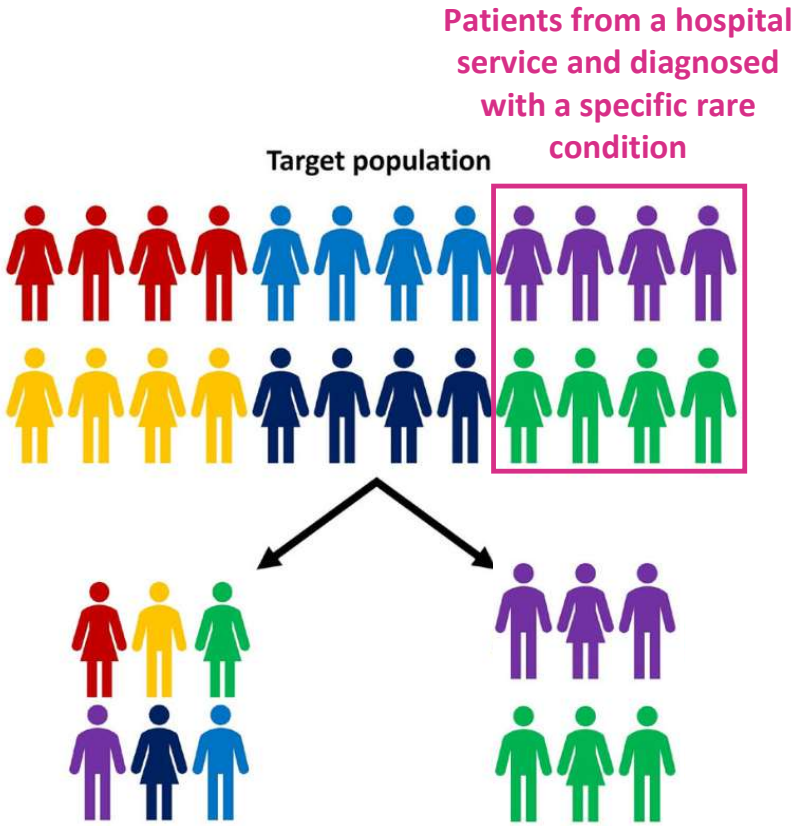
HOW ROBUST ARE OUR RESULTS?

AS ROBUST AS CAN BE!

Are our samples representative?
They cannot be because no one knows the target population (lack of epidemiological studies): we talk about ***the participants****.*

Would our samples be more representative if we reached out to respondents in hospitals?
It would not be the same target population, and we would not reach out to people living with rare diseases who were not referred to Centres of Expertise

Diagnosis survey factsheet



HOW ROBUST ARE OUR RESULTS?

AS ROBUST AS CAN BE!

Are our samples representative?
*They cannot be because no one knows the target population (lack of epidemiological studies): we talk about **participants**.*

Are our sample sizes sufficient?
Yes, at the European level

Population Size	Sample Size Sample Table			
	95% Confidence level		99% Confidence level	
	5% Error	1% Error	5% Error	1% Error
75	63	74	67	75
300	169	291	207	295
800	260	739	363	763
2,500	333	1,984	524	2,173
25,000	378	6,939	646	9,972
100,000	383	8,762	662	14,227
250,000	384	9,248	662	15,555
500,000	384	9,423	663	16,055
2,500,000	384	9,423	663	16,478

One example of calculation of sample size based on population size

HOW ROBUST ARE OUR RESULTS?

AS ROBUST AS CAN BE!

Are our samples representative?
*They cannot be because no one knows the target population (lack of epidemiological studies): we talk about **participants**.*

Are our sample sizes sufficient?
Yes, at the European level

Are our results statistically significant?
We only report on significant results when doing crossings

TABLE 4. Percentage of respondents who agreed or strongly agreed with 'If it is or were possible, I would have liked [the person I care for] to be diagnosed at birth', depending on their country of residence.

'agree' or 'strongly agree' is significantly...	All respondents n=5,569	PLWRD n=2,567	Parents of PLWRD n=2,701
...over-represented	Latvia (93%), Lithuania (92%), Poland (90%), Romania (85%), Spain (84%), Croatia (82%), Czech Republic (80%), Italy (77%)	Poland (87%), Spain (78%), Italy (69%)	Spain (91%), Poland (91%)
...under-represented	Finland (48%), Netherlands (53%), Switzerland (55%), Germany (56%)	Netherlands (46%), Germany (43%), Finland (40%)	France (77%), Germany (76%), Finland (69%), Netherlands (67%), Luxembourg (66%), Switzerland (59%)
Total	73%	63%	82%

Only significant relationships are reported (p-value < 0.05), for countries with at least 20 respondents.

Source: Rare Barometer survey conducted May 24 -July 23, 2023. 'The opinion of people living with a rare disease and their family members on newborn screening'.

INTERPRETING RARE BAROMETER FACTSHEETS

Title and graph: although we seemed to generalise our results in our title for the sake of clarity, we say clearly how we get to these results only based on the participants in the **grey boxes**.

You need to be transparent about how the results were obtained, don't assume that your audience knows.

Factsheets are for policy makers

INTERPRETING RESULTS

Example: In the Daily life survey, sample size = 9591, 8/10 live with a disability.

- If our sample was representative of all people with rare diseases in Europe, we could infer (or extrapolate) that *in Europe, **8/10 people with rare diseases live with disability.***
- As we cannot have a representative sample because we don't know the structure of the target population, we cannot know if the results from our sample are the same as the ones we would obtain if we "surveyed" all people with a rare disease in Europe. Therefore, **it could be misleading to use the same wording.**

➢ Alternative wording (ex. for oral presentations):

- ***In our survey, 8/10 people with rare diseases** live with disability.*
- ***8/10 participants** live with a disability.*

DESIGNING QUESTIONNAIRES

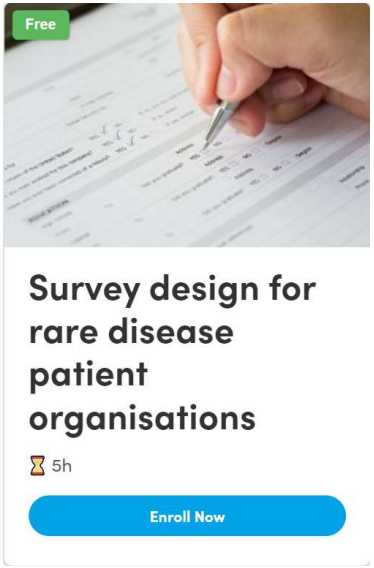
How do we go from questions we ask ourselves (research questions), to questions that are asked to the participants?

RARE BAROMETER RESOURCES TO GO FURTHER

Rare Barometer guide for EURORDIS members

👉 tiny.cc/RBguide_members

Open Academy online course on survey design



👉 openacademy.eurordis.org

Initiated by the ERNs and healthcare team

EURORDIS OPEN ACADEMY

Boston Scientific Foundation Europe

FROM RESEARCH QUESTIONS TO QUESTIONNAIRE QUESTIONS

There is a difference between:

Research question(s) = questions you ask yourselves (and not respondents).
Ex. "What are the most impactful factors on the length of the diagnosis journey?"

Questions that appear in a questionnaire, answered by respondents.
Ex. "As far as you remember, when did you or a healthcare professional first notice the symptoms of the rare disease or think that something was wrong?";
"As far as you remember, when was the name of the rare disease, syndrome or malformation CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test)?"
...

DESIGNING A RARE BAROMETER QUESTIONNAIRE

Background document or literature review

By EURORDIS

Topic Expert Committee

15-20 experts contribute to identifying issues and indicators to include in the questionnaire

Writing the questionnaire

Rare Barometer team

Testing the questionnaire

Topic Expert Committee, EURORDIS staff, council of national alliance (CNA), council of European federations (CEF)

Translating the questionnaire

AI + professional translators
25 langauges

Reviewing the translations

Volunteer reviewers from the RD community

Open Academy course on survey design for rare disease patient organisations:
tiny.cc/survey-design

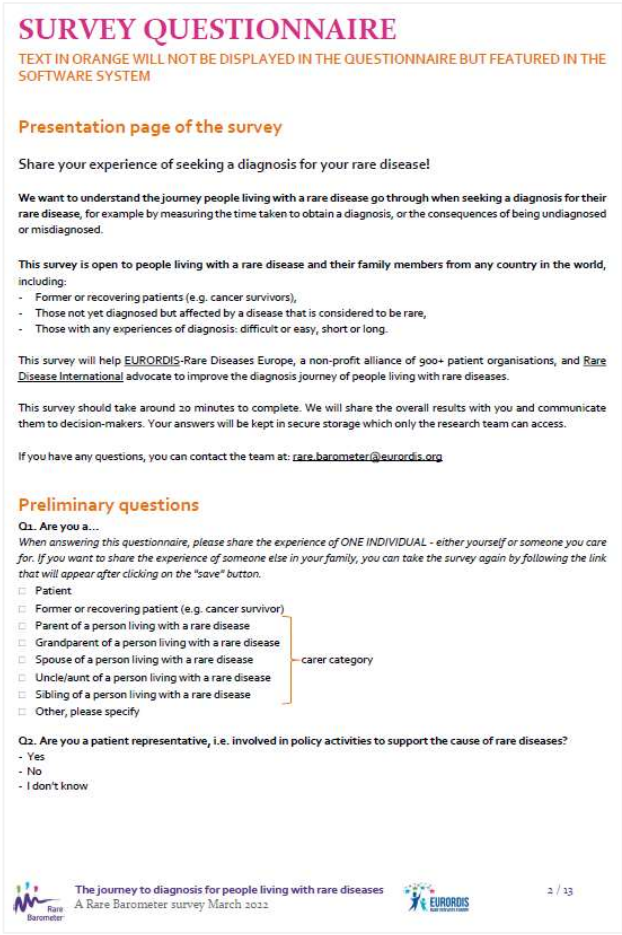
QUESTIONNAIRES ARE AVAILABLE ONLINE

English only

Diagnosis odyssey: tiny.cc/questionnaire_diagRD

Newborn screening: tiny.cc/RB_NBS_questionnaire

Social participation and independent living (daily life): tiny.cc/DailyLife_questionnaire



SURVEY QUESTIONNAIRE

TEXT IN ORANGE WILL NOT BE DISPLAYED IN THE QUESTIONNAIRE BUT FEATURED IN THE SOFTWARE SYSTEM

Presentation page of the survey

Share your experience of seeking a diagnosis for your rare disease!

We want to understand the journey people living with a rare disease go through when seeking a diagnosis for their rare disease, for example by measuring the time taken to obtain a diagnosis, or the consequences of being undiagnosed or misdiagnosed.

This survey is open to people living with a rare disease and their family members from any country in the world, including:

- Former or recovering patients (e.g. cancer survivors),
- Those not yet diagnosed but affected by a disease that is considered to be rare,
- Those with any experiences of diagnosis: difficult or easy, short or long.

This survey will help EURORDIS-Rare Diseases Europe, a non-profit alliance of 900+ patient organisations, and Rare Disease International advocate to improve the diagnosis journey of people living with rare diseases.

This survey should take around 20 minutes to complete. We will share the overall results with you and communicate them to decision-makers. Your answers will be kept in secure storage which only the research team can access.

If you have any questions, you can contact the team at: rare.barometer@eurordis.org

Preliminary questions

Q1. Are you a…

When answering this questionnaire, please share the experience of ONE INDIVIDUAL - either yourself or someone you care for. If you want to share the experience of someone else in your family, you can take the survey again by following the link that will appear after clicking on the "save" button.

- ☐ Patient
- ☐ Former or recovering patient (e.g. cancer survivor)
- ☐ Parent of a person living with a rare disease
- ☐ Grandparent of a person living with a rare disease
- ☐ Spouse of a person living with a rare disease
- ☐ Uncle/aunt of a person living with a rare disease
- ☐ Sibling of a person living with a rare disease
- ☐ Other, please specify

carer category

Q2. Are you a patient representative, i.e. involved in policy activities to support the cause of rare diseases?

- Yes
- No
- I don't know

The journey to diagnosis for people living with rare diseases
A Rare Barometer survey March 2022

2 / 13

FROM QUESTIONS TO DATA

DIAGNOSIS

Q3. Please select your current situation:

a. I know the NAME of the disease, syndrome or malformation, and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (for example, biopsy, blood or urine test) (confirmed diagnosis)

b. I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (initial diagnosis)

c. I only have PARTIAL information on the name of the rare disease, the gene involved, or the type of disease (partial diagnosis)

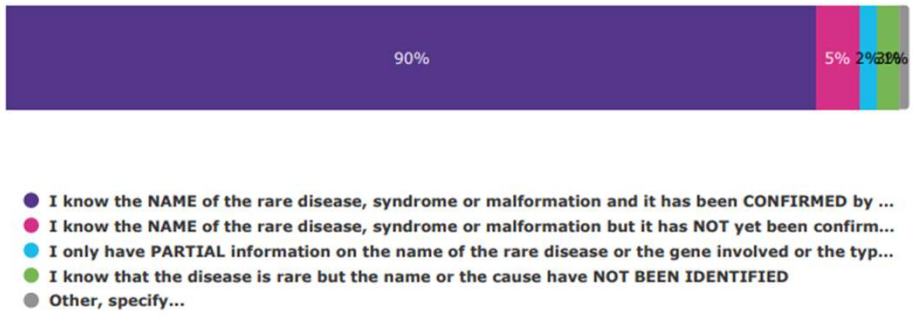
d. I know the disease is rare, but the name or the cause has NOT BEEN IDENTIFIED (unsolved case)

e. Other, please specify

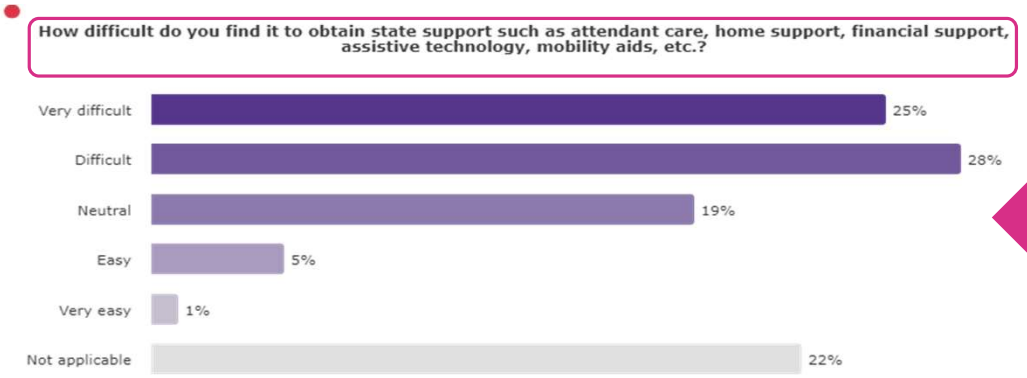
Questions are displayed in the dashboards:

Please select the sentence that best describes your situation:

	N
I know the NAME of the rare disease, syndrome or malformation and it has been CONFIRMED by appropriate genetic, clinical, medical imaging, molecular or biochemical tests (e.g biopsy, blood or urine test)	8,624
I know the NAME of the rare disease, syndrome or malformation but it has NOT yet been confirmed by appropriate genetic, clinical, medical imaging, molecular or biochemical tests	456
I only have PARTIAL information on the name of the rare disease or the gene involved or the type of disease	184
I know that the disease is rare but the name or the cause have NOT BEEN IDENTIFIED	242
Other, specify...	84
TOTAL	**9,590**



FROM THE DASHBOARD TO THE FACTSHEET



All the questions are displayed in our dashboards.
The questions we use in our factsheets are displayed in grey boxes.

53% **found it difficult or very difficult to obtain publicly funded support** such as attendant care support, home support, financial support, assistive technology, mobility aids or other support.

Q *'How difficult do you find it to obtain State support such as attendant care support, home support, financial support, assistive technology, mobility aids, etc.?'- All participants (n=9,591).*

IN THE REPORTS

'Social participation and independent living'

Pages 23-24

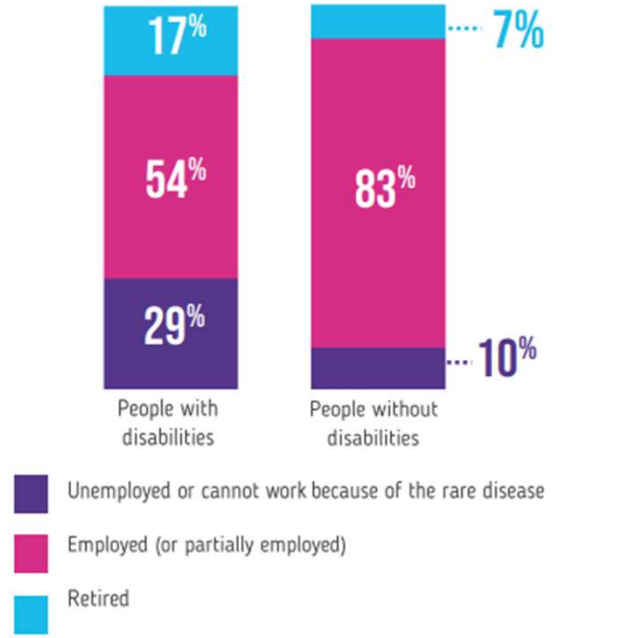
tiny.cc/RB_DailyLife

Table 9. Work-related situation

What is your current situation?	% (n)
Employed	**48%** (2567)
'Employed (or partially employed)'	44% (2329)
'Self-employed'	4% (238)
Unemployed	**23%** (1216)
'Unemployed'	13% (716)
'Cannot work because of the disease'	9% (500)
'Retired'	**13%** (714)
'Student/pupil'	**9%** (500)
Stay-at-home	**3%** (177)
'Not of school age yet.'	**0%** (4)
'Other'	**3%** (154)

Working-aged participants, 'Don't know' excluded (n=5332). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Figure 8. Employment status among people with and without disability.



Working-aged (16-64 y.o.) participants who were employed, unemployed or retired (n=4497). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

DIFFERENCE BETWEEN QUESTIONS AND VARIABLES

A variable is a characteristic of a person, of a place, of a thing, or of a phenomenon that you are trying to measure in some way.
e.g. the age of the participant (i.e. the person answering the questionnaire).

A variable can:

- **correspond to one question in the questionnaire**:
  e.g. "How old are you?" gives directly the variable "Age of the participant'
- **be calculated based on several questions in the questionnaire**.
  e.g. The question "Please, indicate your date of birth" allows for calculating the variable "Age of the participant' when the date of birth is subtracted to another date (e.g. of the survey, of diagnosis…).
- **be calculated based on other data**
  e.g. in Rare Barometer, each respondent gives the name of their rare condition (in a list or in an open text). Then, based on the orphacode of the condition and on Orphanet data, we calculate the point prevalence, mode of inheritance, genetic nature, etc. of the condition of each participant.

INTERPRETING RARE BAROMETER FACTSHEETS

Disability prevalence is calculated based on 3 indicators:

- **Washington Group Short Set (WG-SS)**: sum up of 6 questions
- **Global Activity Limitation Index**: one question
- **Self-identification**: one question

HOW CAN YOU PRESENT RARE BAROMETER RESULTS?

Which types of analysis and which types of graphs can you do?

UNITS OF ANALYSIS: THE SUBJECT OF SENTENCES

The **Unit of analysis** is the main entity that is being studied. It is fundamental to identify it clearly when analysing and communicating the results.

In **Orphanet data**, the unit of analysis is usually **the rare condition**, or group of rare conditions. But it can also be the *treatment*.

In **Rare Barometer**, the selection unit, the reporting unit and the unit of analysis are usually the **participants**, either that they are living with a rare disease or a close family member of someone living with a rare disease.

*But it can also be a **treatment** (e.g. survey on experience of treatment), or a **hospital visit** (e.g. in the H-CARE pilot on patient experience with healthcare).*

The unit of analysis will be the **subject of the sentences**: ex. participants, people living with a rare disease, parents of people living with a rare disease, family members, treatment, hospital visit…

FREQUENCIES AND PERCENTAGES

The **frequency** is the number of times a particular value for a variable has been observed to occur.
Example: In the daily life survey, 5364 participants had undergone a disability assessment.

Percentages are obtained by dividing the frequency by the total and multiplying the result by 100.

Bar graphs and pie charts are a good and simple way to present frequencies and/or percentages

FREQUENCIES AND PERCENTAGES: EXAMPLES

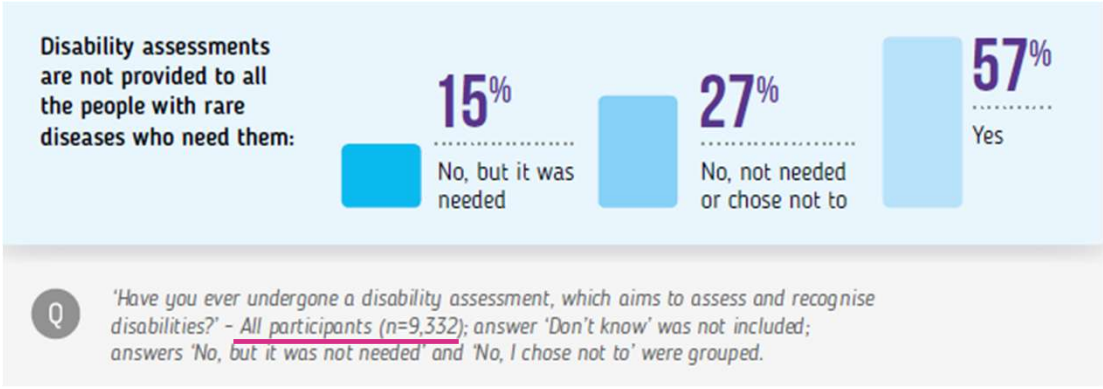
Example: 15% (1435/9332)*100 of participants did not undergo a disability assessment although they needed one.
Always look at the **sample size (n)** corresponding to each question, i.e. the number of people who answered the question, or the number of respondents included in the analysis (subgroups).

In the dashboard, you will find it at the bottom of each table (here in pink).

Have you ever undergone a disability assessment, which aims to assess and recognise disabilities?

	N	%
NO but it is/was needed	1,435	15%
NO but it is/was NOT needed	1,878	20%
No, I chose not to	655	7%
Yes	5,364	57%
TOTAL	**9,332**	**100%**

In the factsheets, it is usually mentioned in the grey box as "n=XXXX".



INTERPRETING CROSS TABULATION RESULTS

	Survey topic	Example of cross-table	What is the p-value? Is it significant?
Table 1	The **diagnosis** odyssey of people living with a rare disease	Dashboard p. 23: - crossing between sex (woman/man) and time to first medical contact; - Crossing between sex and time to diagnosis	- p-value=0.1 (weakly signific.) - p-value<0.01 (very signific.)
Table 2	The **diagnosis** odyssey of people living with a rare disease		
Table 3	The opinion of people living with rare diseases on **newborn screening**	Dashboard p. 18: crossing between status (patient / family member) and willingness to have been diagnosed at birth	p-value<0.01 (very significant)
Table 4	The opinion of people living with rare diseases on **newborn screening**	Dashboard p. 33: crossing between opinion on NBS for all rare diseases depending on access to treatment.	p-value=0.1 (weakly significant.
Table 5	The impact of living with a RD on daily life: **social participation and independent living**	Dashboard p. 17: crossing between disability status (WGSS) and community participation (CASP).	p-value<0.01 (very significant)
Table 6	The impact of living with a RD on daily life: **social participation and independent living**		

CROSSINGS OR CROSS TABULATION: EXAMPLE

Cross: Did you receive or are you receiving TREATMENT(S) OR INTERVENTION(S) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means? / Please tell us how much you agree with the following statements: If it is or were possible, ...I would have liked to be diagnosed AT BIRTH

DID YOU RECEIVE OR ARE YOU RECEIVING TREATMENT(S) OR INTERVENTION(S) TO LESSEN OR CONTROL THE EFFECTS OF THE RARE DISEASE, INCLUDING MEDICATION, SURGERY, DIET OR OTHER MEDICAL MEANS?	PLEASE TELL US HOW MUCH YOU AGREE WITH THE FOLLOWING STATEMENTS: IF IT IS OR WERE POSSIBLE, ...I WOULD HAVE LIKED TO BE DIAGNOSED AT BIRTH							
	STRONGLY AGREE+AGREE		NEITHER AGREE NOR DISAGREE		STRONGLY DISAGREE+DISA...		TOTAL	
	N	%	N	%	N	%	N	%
Yes, even partially (e.g. for one of the symptoms)	1,318	63%	441	21%	333	16%	2,092	100%
No	279	62%	101	22%	71	16%	451	100%

People living with a rare disease were equally willing to be diagnosed at birth regardless of whether they received treatments or not.

63% Treatments or interventions

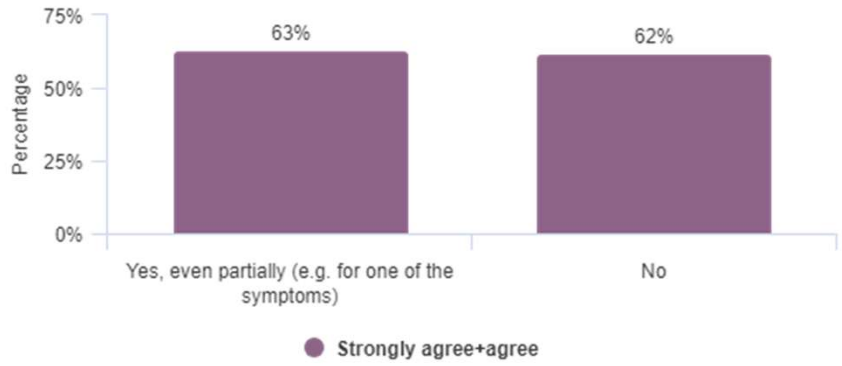
62% No treatment or interventi

"Regardless of available treatments, a diagnosis is often necessary to better understand what is happening to us. It is often indispensable to receive reasonable medical care or help in everyday life.

Person living with a rare disease

Q *Percentage of people living with a rare disease who would have liked to be diagnosed at birth among those who answered either 'Yes, even partially (e.g. for one of the symptoms)' or 'No' to the question 'Did you receive or are you receiving treatment(s) or intervention(s) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means?'.*

Cross: Did you receive or are you receiving TREATMENT(S) OR INTERVENTION(S) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means? / Please tell us how much you agree with the following statements: If it is or were possible, ...I would have liked to be diagnosed AT BIRTH



INTERPRETING CROSS TABULATION RESULTS

The table on the previous slide shows that people living with a rare disease are **equally willing to be diagnosed at birth, regardless of whether they received treatments or not** (63% vs. 62%).

To determine if there is a relationship between the two variables, we need to test the **statistical significance** of the cross-tabulation table:

- If the **variables are independent** (have no relationship), then the results of the statistical test will be "non-significant", the p-value is superior to 0.10 (p>0.10)
- If the variables are related, then the results of the statistical test will be "**statistically significant**" meaning that we can state that there is some relationship between the variables; the p-value is inferior to 0.10 (p<0.10)

***Very significant: p-value <0.01;
**Significant: p-value=0.05;
*Weakly significant: p-value=0.10

CROSSINGS OR CROSS TABULATION

A **crossing is a table** that shows the frequency and/or percentage of respondents who have specific characteristics.

A crossing provides information about the **relationship between the variables**.

Bar graphs can also be used to represent cross tabulation

***Example**: The relationship between having a treatment (or not) and willingness to have been screened at birth among people living with a rare disease.*

INTERPRETING CROSS TABULATION RESULTS

Looking at the p-value below the table (p-value=0.6), we see that there is no relationship between treatment and opinion on screening for rare disease at birth.

The willingness to be diagnosed at birth is not associated with access to a treatment: people living with a rare disease would like to be diagnosed regardless of their access to a treatment. (report page 35)

Did (you / the person you care for) receive or are (you / the person you care for) receiving TREATMENT(S) OR INTERVENTION(S) to lessen or control the effects of the rare disease, including medication, surgery, diet or other medical means?				
Yes, even partially (e.g. for one of the symptoms)	4,503	73% (3,296)	63% (1,315)	82% (1,783)
No	987	72% (706)	62% (277)	80% (398)
Unsure	79	70% (55)	56% (19)	79% (31)
		p=0.46	*p=0.60*	*p=0.57*

MEAN VS. MEDIAN

Descriptive statistics for numerical variables

The **mean (M)** is the average of the value of a variable.

The **standard deviation (SD)** is a measure of how dispersed the data is in relation to the mean.

The **median (Mdn)** is the value that cut a data set in 2 part so that 50% of data points (people) have a value smaller or equal to the median and 50% of data points (people) have a value higher or equal to the median.

The **interquartile range (IQR)** is the measure of dispersion around the median; it's the distance between the 1st quartile (25% of the sample with the lowest value) and the 3rd quartile (25% of the sample with the highest values).

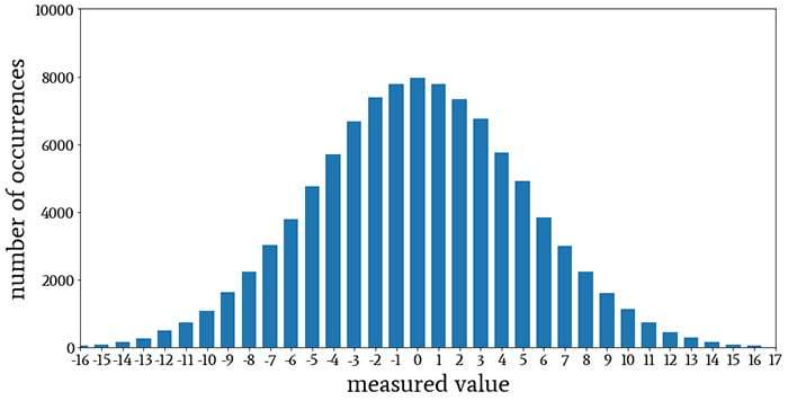
The mean is the most common statistics. For that reason, when the 2 values are closed, you can use the mean.

MEAN VS. MEDIAN

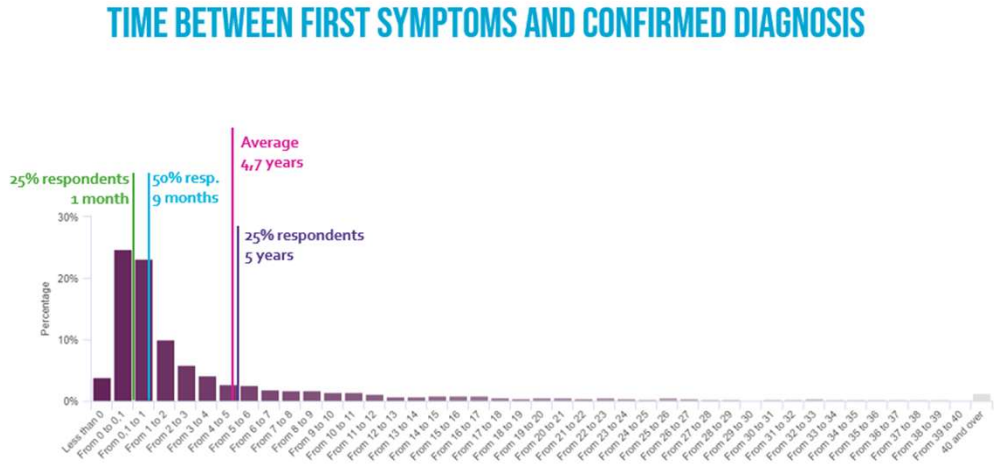
Descriptive statistics for numerical variables

If the mean and the median are very different, it indicates a non-normal distribution which can be caused by outliers or extreme values.

Example: diagnosis survey (explain how extreme time to diagnosis tend to stretch the mean, therefore much higher than median)



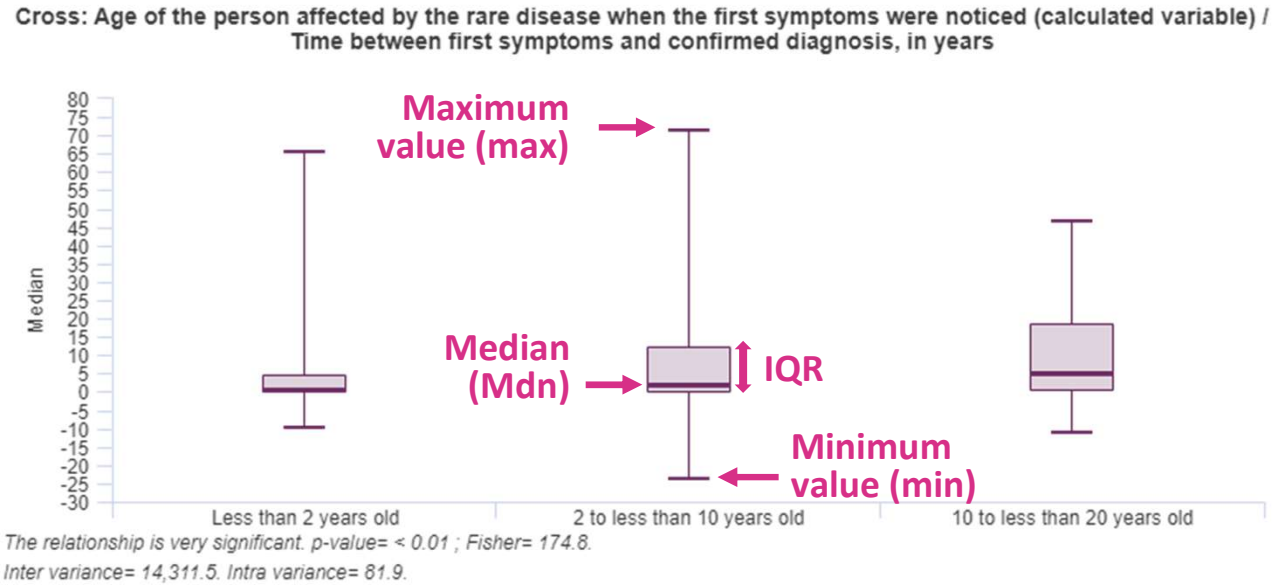
Normal distribution



GRAPHIC REPRESENTATIONS

Graph for numerical variables

Boxplots are a good way to represent mean, median and IQR.

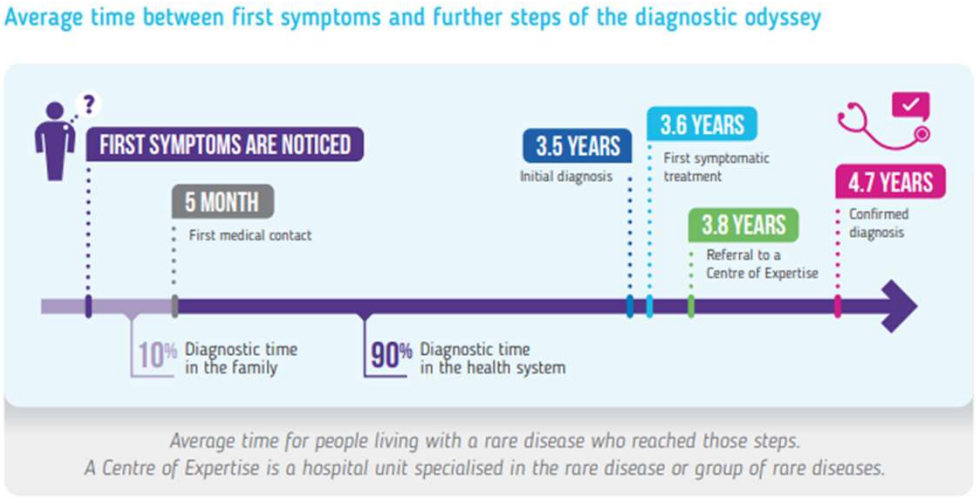


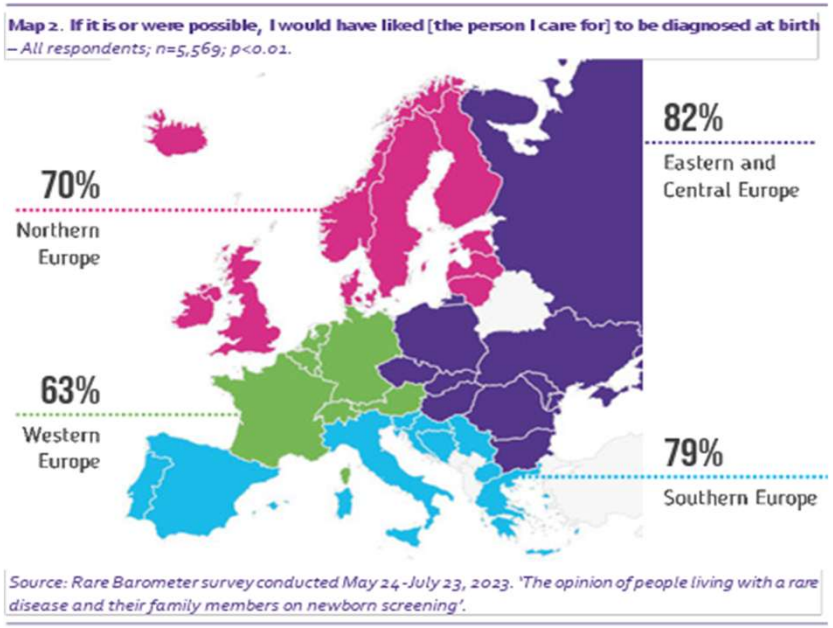
These boxplots represent the median time (**horizontal line**) to diagnosis for children and adolescents. Median can be replaced by mean. You can also present both.

VISUAL REPRESENTATIONS

In sum, depending on the type of data and what you want to represent, you can use visual presentation for your presentation.

- For example: bar graphs or pie chart for percentages, box plots to capture mean, median or interquartile range for numerical variables…
- You can also design your own visual representations:





MULTIVARIATE ANALYSIS

Multivariate analyses are used to identify potential association between **many variables**.

It allows to assess the association between **a dependent variable** (e.g. time to diagnosis) and **multiple independent variables** such as age at first symptoms, geographic areas, gender, type of rare diseases, etc.

Examples:

- Diagnosis survey (article): table 2 (ordinal logistical regression = odd ratios) shows that **age at symptom onset** is a predictor of diagnostic delays; people who were adolescents at symptom onset were 4.7 times more likely to have a delayed diagnosis (> 1 year) than people who were 50 years old or more at symptom onset.
- Daily Life survey (report page 34): table 16 (linear regression) shows that **the average community participation score of participants with disabilities was 12.182** less than that of participants without disabilities.

CONCLUSION

ANALYSING RESULTS

ONLY SAY WHAT THE SURVEY DESIGN ALLOWS YOU TO SAY

When you write and communicate on the results, be mindful of your **unit of analysis** (people living with a rare condition / family members / participants / hospital visits…) and of the **structure of your sample** (and what it allows you to say):

- Only communicate on results when the frequency is high enough
- When we look at the relation between 2 or more variables, non-significant results (based on p-value) can be as relevant as significant ones. **Whether you communicate on them or not depends on your hypothesis** (e.g. opinion on newborn screening and access to treatments)

ADAPT YOUR COMMUNICATION TO YOUR TARGET AUDIENCE:

- Scientists and healthcare professionals
- Policy makers
- Members / People living with a rare disease

SUM UP OF RARE DISEASE SPECIFICITIES

Samples are usually not representative: only talk about respondents for descriptive statistics (do not refer to the whole population) and use significant data for crossings and multivariate analysis.

During results analysis and communication, **the subject of your sentences should correspond to your survey unit**: participants / patients or people living with a rare disease / carers or family members… If your titles extrapolate on the results, always remind the number of participants and how you got those results (methods section, grey boxes).

Respondents are usually contacted through patient organisations, and online: be mindful of the **social characteristics of respondents** (probably more educated, usually more respondents in Western Europe, more engaged…).

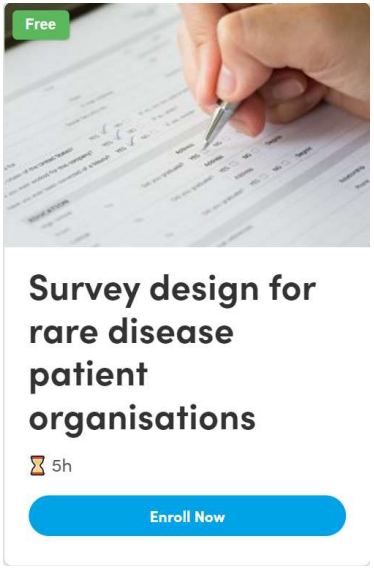
/!\ SURVEY FATIGUE > we encourage you to disseminate rare barometer questionnaires (common efforts), but if you do not wish to or you cannot, you can also use some of our questions and compare your results with ours (open academy, guide…).

RARE BAROMETER RESOURCES TO GO FURTHER

Rare Barometer guide for EURORDIS members

👉 tiny.cc/RBguide_members

Open Academy online course on survey design



👉 openacademy.eurordis.org

Initiated by the ERNs and healthcare team

EURORDIS OPEN ACADEMY

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