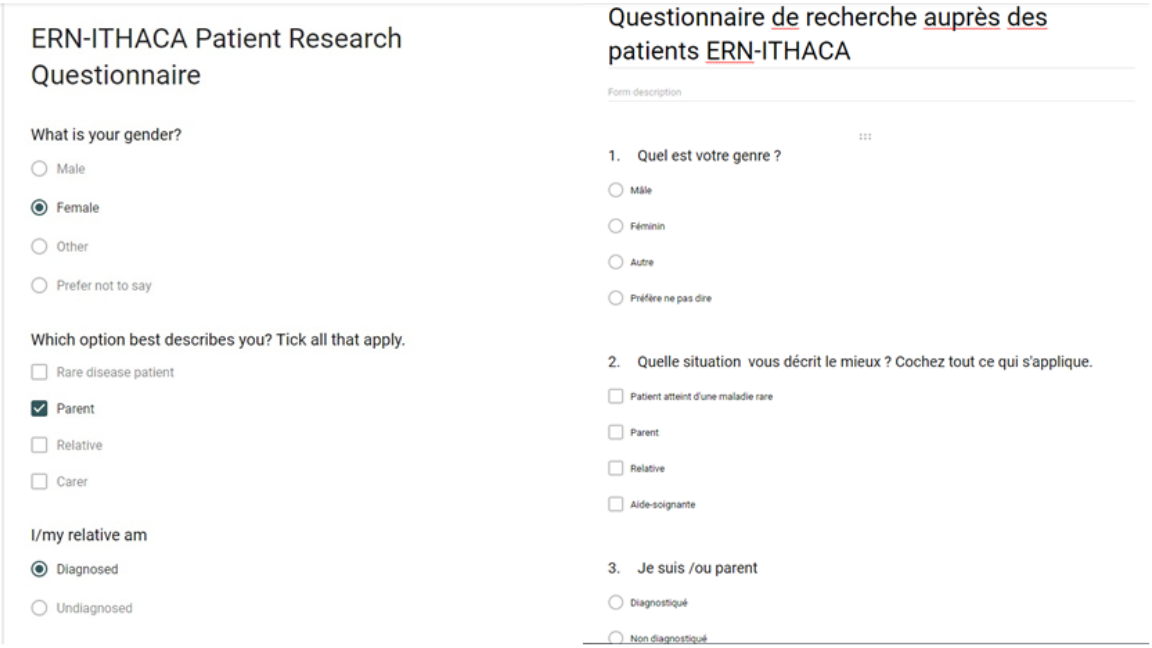
ERN ITHACA Year 2 Deliverable

Date: February 18th 2019

Report on Patient Research Survey

European Reference Network
for rare or low prevalence complex diseases

Network Intellectual Disability and Congenital Malformations (ERN ITHACA)



ERN-ITHACA Patient Research Questionnaire

What is your gender?

- Male
- Female
- Other
- Prefer not to say

Which option best describes you? Tick all that apply.

- Rare disease patient
- Parent
- Relative
- Carer

I/my relative am

- Diagnosed
- Undiagnosed

Questionnaire de recherche auprès des patients ERN-ITHACA

Form description

1. Quel est votre genre ?

- Mâle
- Féminin
- Autre
- Préfère ne pas dire

2. Quelle situation vous décrit le mieux ? Cochez tout ce qui s'applique.

- Patient atteint d'une maladie rare
- Parent
- Relative
- Aide-soignante

3. Je suis /ou parent

- Diagnostiqué
- Non diagnostiqué

ERN-ITHACA Patient Research Survey

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1.1 Executive Summary

In 2018, the European Reference Network on Congenital Malformations and Rare Intellectual Disabilities (ERN-ITHACA) designed a survey to establish the research priorities of the network's large base of patients and carers. As a patient-centred network, the

coordination thought it crucial that patients were given a voice in deciding the type of research the network participated in. The survey, launched in December 2018, was made available in four languages (English, Italian, French and Dutch) and was completed by a total of 189 participants from fourteen countries. This document provides a summary of the results, along with supporting material on rationale, methodology, target audience and how the results will be utilised in the future. The Manchester team plan to write a reflective review of the survey and its findings for peer-reviewed publication in the near future.

1.2 Rationale

Alongside teaching and training, telehealth, patient registries and care guidelines, research was one of the five key areas identified when ERN-ITHACA was originally proposed in 2016. While the restrictions on industry collaboration combined with the limited amount of dedicated funding mean that it is not practical for ERNs to commission or independently undertake large-scale research, there are still many ways that ERNs can fulfil active research agendas. Examples of these are given in the box below:

How might ERNs facilitate research?

- Networking researchers with shared interests electronically and physically
- Calls for rare disease cohorts through ERN communications
- Include research on ERN agendas at annual meeting
- Facilitating trainee research attachments, courses and tools
- Contributing to the drawing up of the European Joint Plan (EJP)
- Funding of small research workshops
- Participation in EU research projects e.g. H2020
- Use of CPMS for research purposes
- Setting up patient registries as research resources
- Admin help with preparation of ERN research publications

The first year of ITHACA's research activities was focused on cataloguing the existing active research projects which our 37 HCP members were involved with, the main purpose being to establish a network research profile which could be used to identify areas of overlap and potential collaboration. The results of this scoping exercise were disseminated throughout the network and uploaded as a Year 1 deliverable.

After establishing this 'state of play', the ITHACA five-year plan set out to use the remaining four years of the network's initial funding period to actively engage in research. The collaborative ethic between patient and health care provider with the emphasis on patient involvement which underpins European Reference Networks meant that it was essential to find a way to accurately record the views of patients on how ITHACA should proceed with their research agenda and, considering the wide geographical footprint of our network, a web-based survey seemed like the most cost effective and logical way to begin this task.

Furthermore, there has been a modern tendency for rare disease networks to undertake patient research via survey, as evidence by largescale patient experience surveys conducted through the patient groups EURORDIS (2017)[1] and Rare Disease UK (2010)[2], as well as the UK-based clinician-led services the National Institute for Health Research (annually since 2015/16)[3] and General Dental Council (2017)[4]. Other European Reference Networks have also undertaken patient surveys; ERN-Reconnet (2019)[5] surveyed their patient stakeholders on ethical, legal and privacy issues while ERN-RND (2018)[6] conducted a similar survey to that detailed here, only with both patient and clinical respondents.

1.3 Methodology

The survey was initially designed by the Manchester team, primarily ERN-ITHACA coordinator Prof. Jill Clayton-Smith, Research workpackage lead Dr Siddharth Banka and project manager Michael Smith and was developed using Google Forms, a free-to-use software with no restrictions on either number of questions or respondents. The survey was written in English and then translated into French, Italian and Dutch by network HCP members.

The purpose of the survey was to find out the research priorities of patient stakeholders and as such the questions focused on two broad areas:

- demographic information (age, gender, country of residence, prior involvement in research)
- opinions on research (how can ERNs contribute to research, which areas do they feel are most important to focus on, what should the roles of patients be)

With the exception of the final two questions (free-text fields asking respondents to list any research-specific skills they and for general comments), the questions all followed the same format: multiple-choice, either single answer or all that apply, with the option to select 'Other' and answer in free-text in the case that none of the provided answers were applicable. This semi-closed format had two advantages. Firstly, completing the survey was a relatively fast process, with internal trial runs seeing both English and non-English first-language speakers taking between 5-7 minutes to complete the survey. Secondly, having mostly fixed answer options made analysis much easier, as the data could be grouped and presented visually – as it is in this paper.

The authors were aware that, while they had drafted the survey with patients in mind, as people from the clinical side, they were not themselves the target audience of recipients.

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[1] https://www.eurordis.org/news/3000-rare-disease-patients-carers-voice-difficulties-balancing-care-life

[2] https://www.raredisease.org.uk/our-work/experiences-of-rare-diseases-an-insight-from-patients-and-families-2010/

[3] https://www.nihr.ac.uk/patients-and-public/documents/Patient%20Research%20Experience%20Survey%202017-18.pdf

[4] https://www.gdc-uk.org/about/what-we-do/research/patient-and-public-survey

[5] http://reconnet.ern-net.eu/2019/01/23/patients-survey-ethical/

[6] http://www.ern-rnd.eu/poster-ern-rnd-survey-research-priorities/

With this in mind, the survey was sent to Miriam Ingram (Comms Manager) and Lauren Roberts (Director) of SWAN-UK, a patient group with which the network has worked with successfully in the past. The SWAN staff members provided invaluable feedback, in particular pointing out that there was some medical terminology used which non healthcare professionals may struggle to understand and commenting that several questions could be interpreted ambiguously. By implementing their suggested revisions, we were able to disseminate a survey which was able to be understood and answered accurately by its target audience; out of the 189 respondents, only one queried the meaning of a question.

The surveys can be found here:

https://goo.gl/forms/vbkNw4yjMI8lcJOK2 (Dutch)

https://goo.gl/forms/A0gtNqw1VQIh57J22 (Italian)

https://goo.gl/forms/DYmiJGfARLlCQUsH2 (English)

https://goo.gl/forms/9izKcha0da6o71PW2 (French)

1.4 Target Audience and Dissemination

The survey was aimed at patients, carers and family members (these are not always mutually exclusive categories) who have been impacted by the disease areas which fall under the remit of ERN-ITHACA.

We utilised various means to disseminate the survey:

- all clinical, ePAG and independent patient representatives on the ERN-ITHACA mailing lists were sent a direct email encouraging them to disseminate the survey amongst their networks as widely as they thought appropriate
- the Italian language survey was publicised by the patient groups Associazione Nazionale Macrodattilia and Uniamo, while the French language survey was disseminated by the national networks AnDDI-Rares and DéfiScience. There was a direct positive correlation between the timing of the patient groups' announcements and the uptake of the surveys in their languages.
- The survey was also shared via ERN-ITHACA's monthly newsletter and Twitter account

With the help of SWAN-UK., the survey authors also drafted an introductory text which was sent with the English language version of the survey:

> Will you help us find out about the research priorities of patients supported by the **European Reference Network for Rare Congenital Malformations and Intellectual Disability (ERN-ITHACA)?**
>
> We have developed a questionnaire to explore some of the areas we felt might be important, as well as seeking some general comments on research. The questionnaire can be accessed at:
> https://goo.gl/forms/q3FQpon59lCalcru2

As well as completing the questionnaire personally, we would appreciate if you **could disseminate it throughout your networks.**

Research is particularly important to our ERN because:
* We have a larger proportion of undiagnosed patients
* We have a high proportion of ultra-rare disorders
* We have a larger number of different disorders
* We include a significant proportion of non-genetic disorders
* We have many disorders for which there is no diagnostic test
* We do not have a large number of disorders for which there are currently treatments or treatment trials in progress
* Many of the disorders covered by our ERN have only recently been described

As emphasised above, ERNs do not have the resources to fund research specifically, but there are several ways that the networks can support it. ERN-ITHACA has so far sought to support research in the following ways:
* Establishing a research working group to bring together those with a greater interest in research
* Participation in the cross-ERN Working Group on Research
* Contribution to writing of the European Joint Programme on Rare Disease<http://ec.europa.eu/research/participants/portal/desktop/en/opportunities/h2020/topics/sc1-bhc-04-2018.html>
* Participation as an ERN in the H2020 SOLVE-RD<http://solve-rd.eu/> project
* Circulation of calls for rare disease patients to participate in funded studies being undertaken by partner HCPs with evidence that these calls are helping to build patient cohorts
* Creating opportunities for trainee research attachments
* Aiming to foster new research collaborations and to write joint grant applications for future funding

1.5 Evaluation and Future Plans

A summary of the results is presented below and we plan to conduct a full analysis with the aim of peer-reviewed publication later in 2019. This report will be circulated to the ERN-ITHACA membership and uploaded on the ERN public website and European Collaborative Portal.

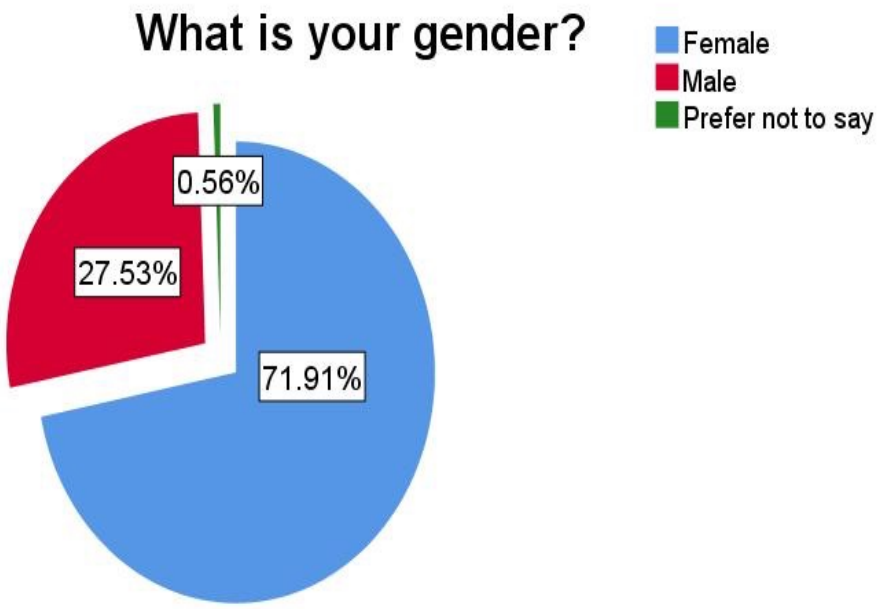
1.6 Preliminary observations from the study

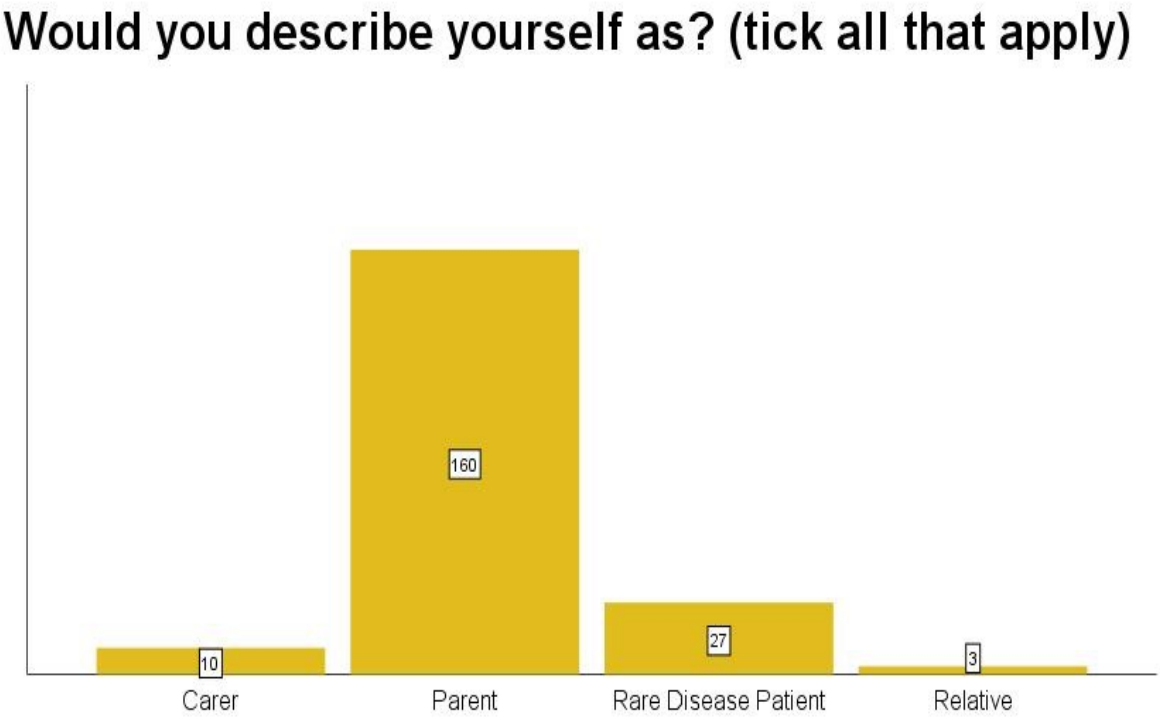
- Mothers of patients are more likely to be the responders to patient surveys of this type
- Translating surveys into different languages is a key way to engage patients from different countries
- A large percentage of the patients surveyed had some experience of being involved in research
- Up until now a lot of studies involving patients have been studies to find a diagnosis. This may change as we move into an era where genomic technologies have been able to diagnose more patients

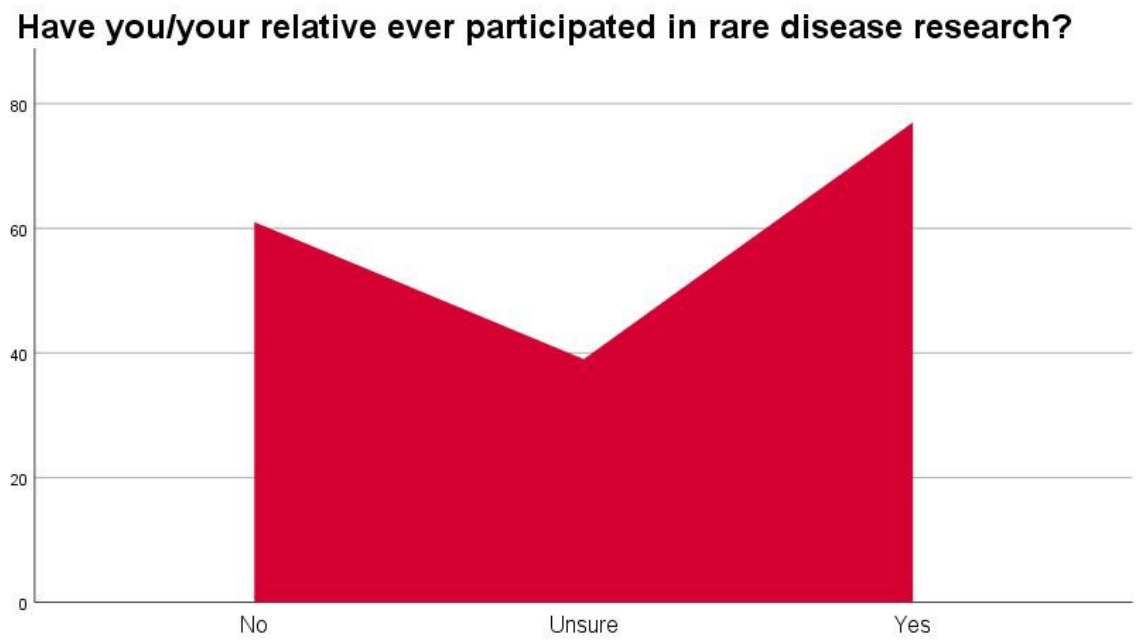
- There was clear support for ERNs working on natural history studies and quality of life studies in the future, both research areas where funding is difficult . For natural history studies a good register infrastructure will be needed.
- There is still a perception that ERNs have the wherewithall to actually fund research studies and it is not clear to some that they were not set up to be primary research networks
- The most important function of ERNS in research was that of networking researchers to work on joint research projects and thinking of ways to achieve this with the financial and IT resources available to the ERNS will be important going forward.

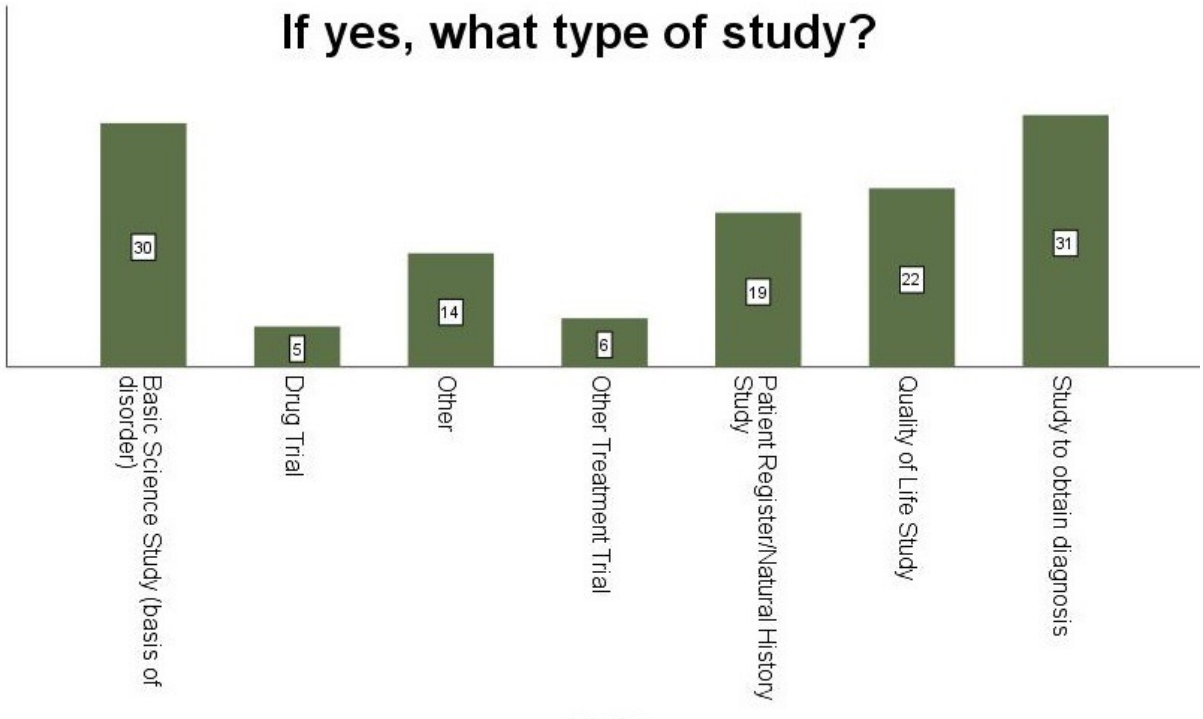
The results of the survey will be discussed in more detail and more broadly on a webcall with the Research WP members, the ERN-ITHACA Coordination team and Patient Representatives. The call is due to take place on Friday 1st March and its annotated minutes will form the second research deliverable for Year 2 ‘Report from Patient Research Group.’

Appendix I: Summary of Survey

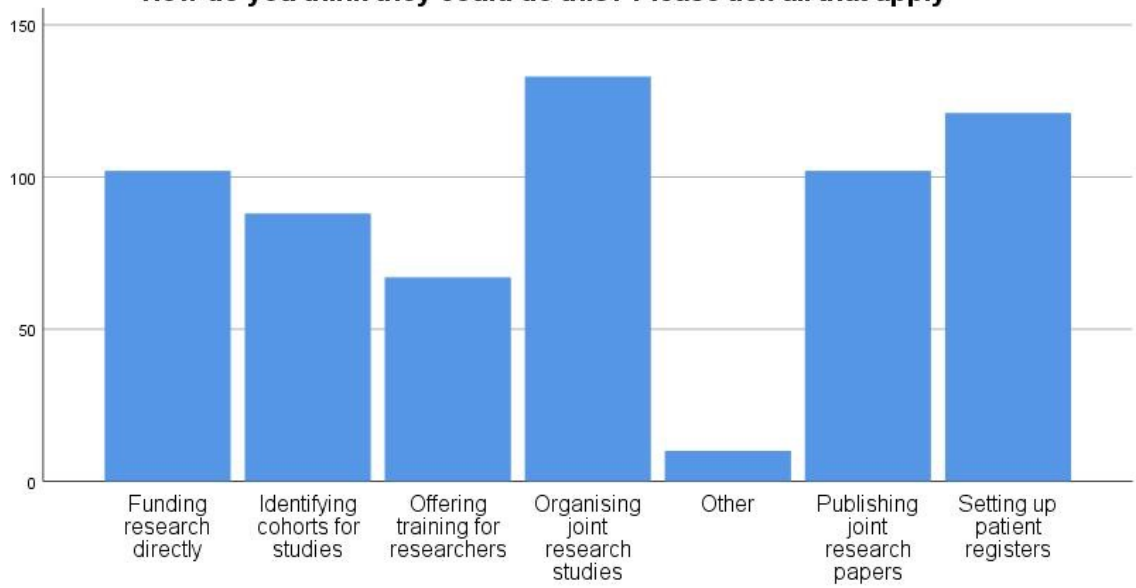




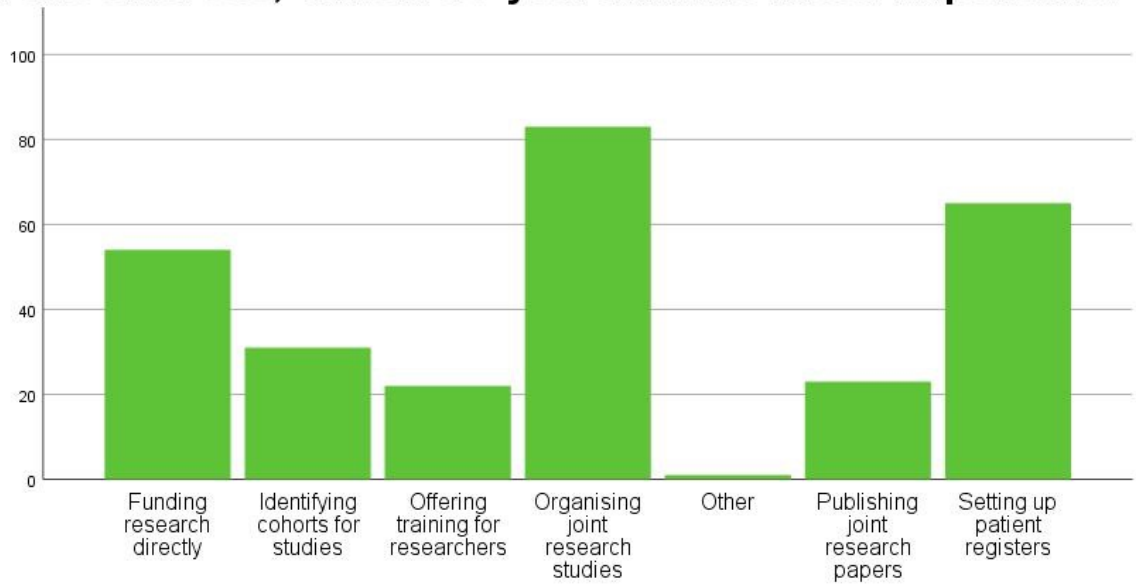

Have you/your relative ever participated in rare disease research?
80
60
40
20
0
No
Unsure
Yes

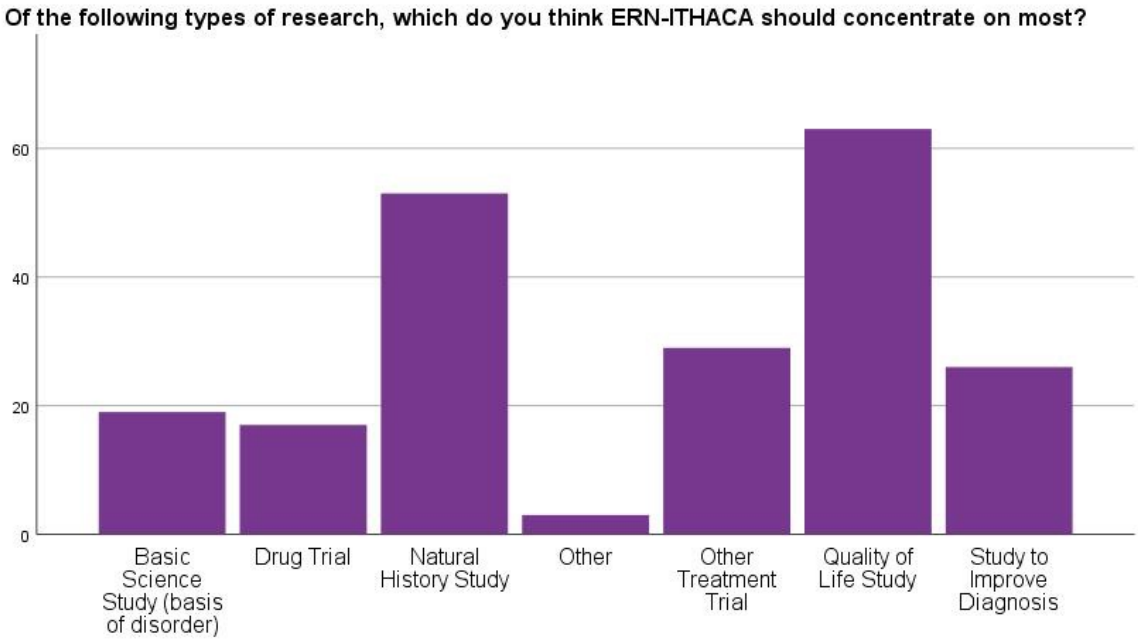

If yes, what type of study?
30
5
14
6
19
22
31
Basic Science Study (basis of disorder)
Drug Trial
Other
Other Treatment Trial
Patient Register/Natural History Study
Quality of Life Study
Study to obtain diagnosis

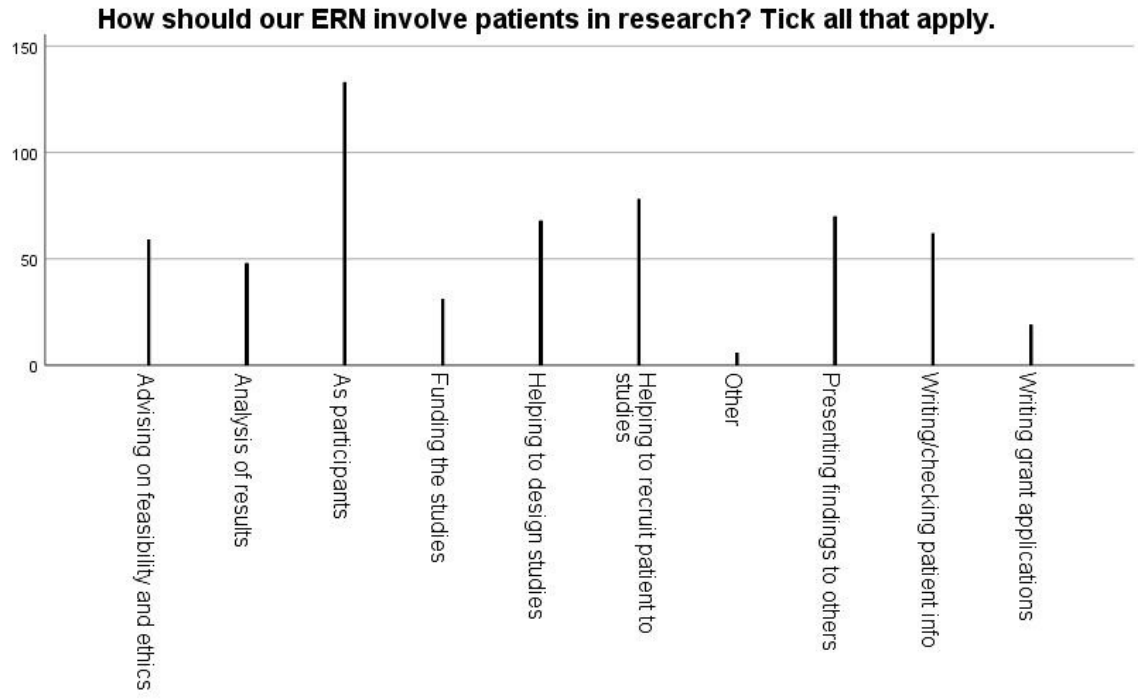
European Reference Networks for rare diseases aim to facilitate rare disease research. How do you think they could do this? Please tick all that apply



Of the choices, which do you think is most important?




Of the following types of research, which do you think ERN-ITHACA should concentrate on most?
60
40
20
0
Basic Science Study (basis of disorder)
Drug Trial
Natural History Study
Other
Other Treatment Trial
Quality of Life Study
Study to Improve Diagnosis


How should our ERN involve patients in research? Tick all that apply.
150
100
50
0
Advising on feasibility and ethics
Analysis of results
As participants
Funding the studies
Helping to design studies
Helping to recruit patient to studies
Other
Presenting findings to others
Writing/checking patient info
Writing grant applications