

SHORT GUIDE ON PATIENT PARTNERSHIPS IN RARE DISEASE RESEARCH PROJECTS

BASIC
PRE-CLINICAL
TRANSLATIONAL & SOCIAL

Written by the members
of the working group PENREP*
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* **P**atient **E**ngagement in
Biomedical **R**esearch **P**rojects.





Beau - Belgium - Cystic Fibrosis.

1 | INTRODUCTION

- 1.1. BACKGROUND & CONTEXT
- 1.2. GOAL & OBJECTIVES
- 1.3. DEFINITION OF PATIENT PARTNERSHIPS :
ENGAGEMENT, INVOLVEMENT AND PARTICIPATION
- 1.4. BENEFITS OF PATIENT PARTNERSHIPS

2 | GUIDANCE FOR RESEARCHERS, FUNDERS AND PATIENT REPRESENTATIVES

- 2.1. EXAMPLES OF PATIENT PARTNERSHIPS
 - 2.1.1. Throughout the research cycle
 - 2.1.2. Snapshots of Patient Partnerships in research projects
- 2.2. AVOIDING COMMON PITFALLS
- 2.3. INDICATIVE CHECKLIST FOR SELF-EVALUATION
OF APPLICANTS
- 2.4. TESTIMONIALS OF A SUCCESSFUL PATIENT PARTNERSHIP

3 | ACKNOWLEDGEMENTS*

4 | REFERENCES

1

•1 BACKGROUND & CONTEXT

In recent years, several initiatives focused on developing recommendations for processes and frameworks to improve patient engagement / partnerships in developing medicines (e.g. Paradigm¹, PFMD² and others^{3,4,5,6}).

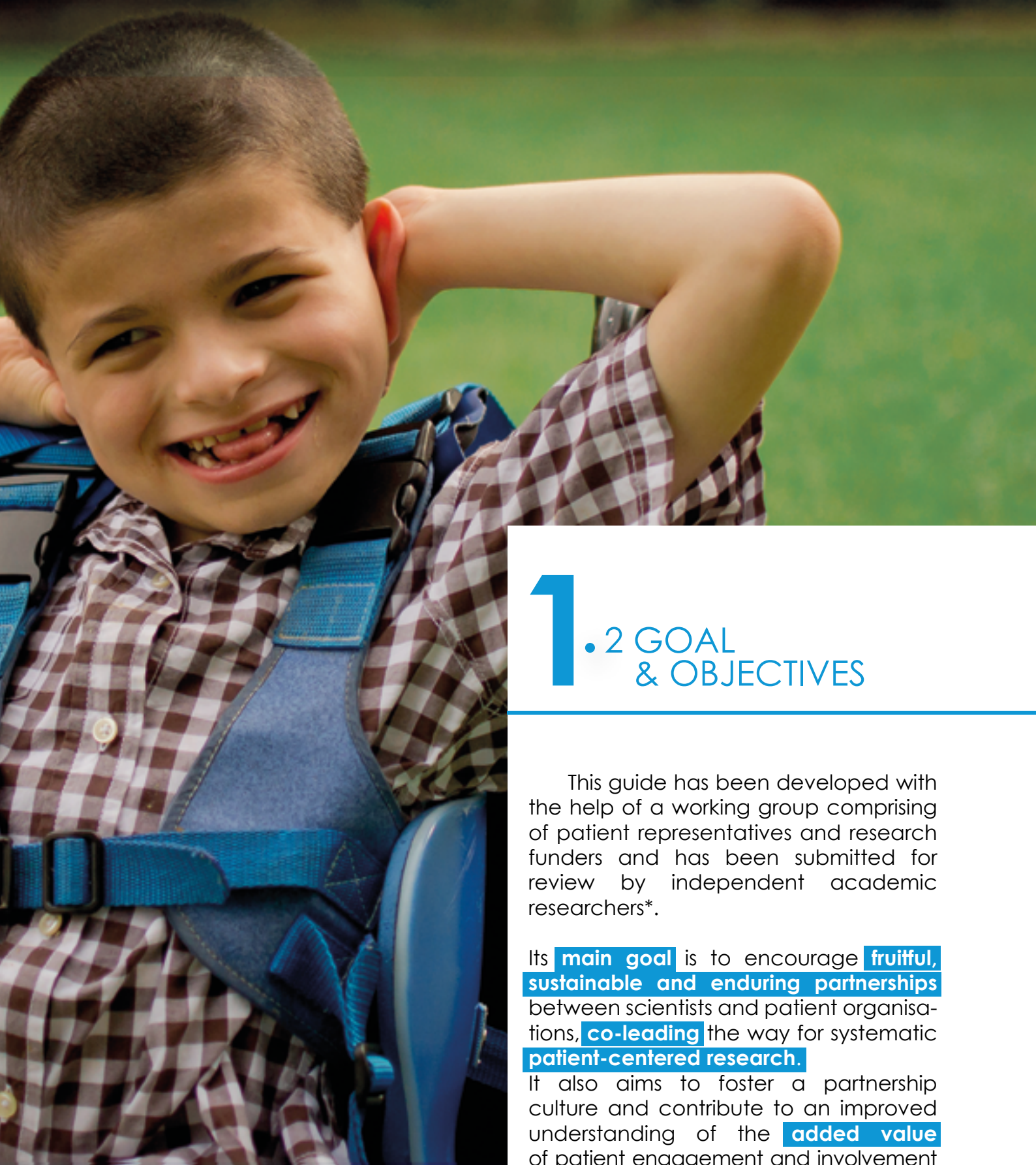
However, these initiatives focus predominantly on engagement within clinical research and the development of medicinal products. There is limited available information that specifically supports engaging patients at an earlier stage in the biomedical research and development process, in projects focused on basic, pre-clinical and translational research as well as those in social sciences and humanities.

Within the Joint Translational Calls (JTC) of the European Joint Programme on Rare Diseases (EJPRD)⁷, patient organisations are eligible to apply as fundable partners of a consortium submitting a research proposal.

Patient involvement is strongly encouraged with the possibility for patient organisations to request a budget in line with their roles and responsibilities within the project.

The experiences of the first call in 2019, which provided patient organisations with this opportunity, evidenced the need for the provision of a guide to support applicants describe the role and added value of patient partnerships in research proposals.





Pierre - France.
Angelman syndrome.

1.2 GOAL & OBJECTIVES

This guide has been developed with the help of a working group comprising of patient representatives and research funders and has been submitted for review by independent academic researchers*.

Its **main goal** is to encourage **fruitful, sustainable and enduring partnerships** between scientists and patient organisations, **co-leading** the way for systematic **patient-centered research**.

It also aims to foster a partnership culture and contribute to an improved understanding of the **added value** of patient engagement and involvement in basic, pre-clinical, translational and social research for the **Rare Disease Community** in Europe and beyond.

With the help of the working group, this guide will be reviewed annually and updated as necessary responding to the learnings from the JTCs of the EJP RD.

1.3 DEFINITION OF PATIENT PARTNERSHIPS

ENGAGEMENT, INVOLVEMENT AND PARTICIPATION

The terms “patients” and “patient representatives” are used interchangeably in this guide and refer to people living with a rare disease and their family members or carers as well as people from organisations that represent the interests of rare disease patients (i.e. from patient organisations or patient groups). These terms do not include people who have a professional role in health and social care services.

Patient Partnerships will often involve the patients and/or patient representatives representing the interests of a group of patients and can contribute the perspectives on behalf of that group (or perhaps make decisions where necessary). Ideally, the patients and patient representatives should be affiliated to a patient organisation (with legal status) or patient group (which can be without legal status) to ensure continuity of representation and direct access to a community of patients for consultation and communication.

In this guide, “Patient Partnerships” is being used as the umbrella term to include the concepts of patient engagement, patient involvement and patient participation. It is worth noting that patient engagement and patient involvement are sometimes used interchangeably and mean different things in different countries and contexts.

Therefore, this guide makes a clear distinction between the three concepts of **PARTICIPATION**, **ENGAGEMENT**, **INVOLVEMENT**, and in order to illustrate the different ways of including patient partnerships in research projects.

To this end, the following definitions will be used within this guide (adapted from INVOLVE^{6,8}) :

Participation :

Where patients take part in a research study as subjects/participants or where patient representatives support recruitment.

Engagement :

Where patients or patient representatives co-create information for dissemination and where information and knowledge about the research project and its results are disseminated by patients.

Involvement :

Where patients or patient representatives are actively involved in research projects, i.e. where research is carried out ‘with’ or ‘by’ patients or patient representatives rather than ‘to’, ‘about’ or ‘for’ them.

These three concepts form a continuum of increasing levels of active & meaningful partnerships :

Patient Participation :

Contribute to the recruitment of patients for the study or as participants themselves.

Patient Engagement :

Review research proposals to ensure feasibility & relevance of study from patient's perspective ; design and / or co-create materials for study participants or for communication about the research study and its results ensuring information accessible to all.

Patient Involvement :

Patient as official partner / Co-Investigator : Identify patient needs, highlight new research directions, design, develop, co-write research proposals, implement research ; contribute to interpretation and findings.

PASSIVE

ACTIVE

PROACTIVE



INTRODUCTION

1.4 BENEFITS FOR PATIENT PARTNERSHIPS

There are multiple benefits of a well-thought out and designed plan for Patient Partnership in research projects.

These benefits illustrate why it is important to involve patients throughout the research process their relevance for different stakeholders involved including researchers, patients, and funders (adapted from “Involving people with arthritis : a researcher’s guide from Arthritis Research UK”⁵).



MOTIVATION & FOCUS

Hearing directly from people living with a specific RD can provide researchers with meaning and context. It can help them make sense of the long hours and days spent in the lab with zebrafish for examples or looking down a microscope in a dark room making the end goal tangible and real. Knowing the difference research makes to the lives of RD patients will provide researchers with that extra level of motivation.



EXPANDED OUTREACH & IMPROVED COMMUNICATION

Patients can assist in the creation of communications, translating information into accessible language to reach a wider community more efficiently. Appropriate communication will help with a better understanding of patient needs which in turn may lead to more research projects that include exemplary patient partnership. Transparency is key to build trust and promote public support for research.



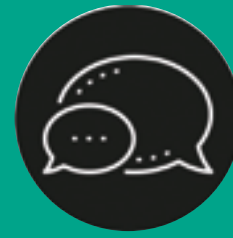
GREATER IMPACT

Carefully considered Patient Partnerships ensure more impactful research as the foundation is grounded in an understanding and prioritisation of patient needs. Patient partners are excellent advocates to generate public interest and impact, raise awareness of the research needs for the benefits of rare disease patients, and facilitate further funding through collaborations with charities.



GREATER RELEVANCE

Involving patients ensures that researchers demonstrate accountability of public money investment as research results translate into concrete benefits and address patients needs ; Successful Patient Partnerships also provide researchers with a competitive advantage and increase chances of securing further funding.



BUILDING REACH

Patients and/or patient representatives can facilitate the creation of research consortia by bringing partners together. Patients can initiate collaborations and support communication between the different partners leading to the development of new expertise in a specific rare disease.



STRONGER FUNDING APPLICATIONS

Applications written by/with patients clearly illustrate patient benefit, study importance and the defined roles and responsibilities of patient partnerships to all evaluation panel members.



NEW IDEAS

Talking to wider groups of patients, particularly in the early stages of research, can identify novel challenges and ideas. These can come from the subtle nuances that only a patient or carer would know from living with the condition.

An example is the observation by a wife of a Parkinson's patient that he smelled similar to other Parkinson's patients she met at a patient group meeting. This observation led to the development of research into a novel way to diagnose this condition using a metabolic biomarker ².

2.1 EXAMPLES OF PATIENT PARTNERSHIPS

2.1.1 THROUGHOUT THE RESEARCH CYCLE

EVALUATING IMPACT

- Collaborate with researchers to evaluate the research process.
- Evaluate the impact to the involvement on the research.
- Patients / public reflect on their role / what they learned

IDENTIFYING & PRIORITISING

- Patients / stakeholders identify relevant research topics through consultation.

IMPLEMENTING

- Increase likelihood of results being implemented due to patient support / lobbying.
- Assessment of value.
- Analysis of benefit / risk.

DISSEMINATING

- Advise on avenues for dissemination.
- Jointly present research findings.
- Contribution to publications.
- Draft lay summaries of results.
- Collaborate in publishing results e.g. via charities / patient groups.

THE RESEARCH CYCLE



DESIGNING & MANAGING

- Be named as co-applicant.
- Ensure protocol / methods are patients appropriate.
- Assist with recruitment & retention strategies.
- Define outcome measures.
- Assist to steer project.
- Develop research tools.
- Review / draft informed consent forms.
- Produce patient / participant friendly research updates (communication).

FUNDING & COMMISSIONING

- Review & draft research funding proposals.
- Ensure the research & methods are ethical.

UNDERTAKING & ANALYSING

- Collaborate in data collection e.g. with phone apps, conducting interviews / surveys.
- Analyse / interpret data & results.

Leave your ego at the door. Think of the bigger, long term picture rather than the immediate benefits. Try to see things from the other one's perspective. Aim for a win-win."

Veronica Popa
MCT8-AHDS Foundation
Romania



From NIHR INVOLVE's Briefing note eight : ways that people can be involved in the research cycle¹⁰,

2.1 EXAMPLES OF PATIENT PARTNERSHIPS

2.1.2 SNAPSHOTS OF PATIENT PARTNERSHIPS IN RESEARCH PROJECT

**MULTICELLULAR ORGANOIDS :
MODELING MECHANISMS
AND THERAPY DEVELOPMENT
OF A SPECIFIC DISEASE
CAUSING MUTATION**

PATIENT INVOLVEMENT

Study design & monitoring :

Key concepts of the research project are identified through close consultation with local / national Patient Organisation to ensure objectives of the research match patients needs.

Patient representative is a member of Project Steering Committee to ensure development of project continues to meet patients' needs.

PATIENT ENGAGEMENT

Co-creation :

Talks, seminars and workshops are co-organised with Patient Organisation and patients involved as speakers. Researchers invited to speak at Patient Organisation meetings.

Videos explaining the research are co-created with patients.

**IMMUNE RESPONSE
AND PATHWAY ANALYSIS
IN A SPECIFIC RARE DISEASE**

PATIENT ENGAGEMENT

Strategic input :

Patient Organisation rep is involved in project steering committee.

Patient Organisation will organise annual strategic workshops for patients to consult the wider community and discuss strategic areas to further engage on specific topics identified through the workshops. These workshops will be connected to consortium meetings to encourage networking between patients and researchers.



**PERSONALISED MEDICINE
FOR A RARE DISEASE :
A MULTI-NATIONAL, LONGITUDINAL
AND TRANSLATIONAL PROJECT**

PATIENT INVOLVEMENT

Study design :

Patients are already involved via survey and ad-hoc workshops to ensure collection of meaningful clinical information reflecting QoL, pain & symptoms relief.

Project monitoring :

Patient Organisation rep included as a member of Steering / governing board. Patient Organisation involved in co-organising kick off meeting.

PATIENT ENGAGEMENT

Co-creation :

Regular newsletter for the patient community to raise awareness of the project and provide regular updates.

Communication :

The partner Patient Organisation is responsible for creating a European Working group of all the disease-specific patient groups in the different countries participating in the consortium.

**RARE INVISIBLE DISEASES
AND SCHOOLING OF CHILDREN :
ENHANCING THE SCHOOL
INCLUSION OF CHILDREN
WITH 3 DIFFERENT RARE DISEASES**

PATIENT INVOLVEMENT

Study design :

The 3 Patient Organisations partners participate in the implementation of experimental scenarios to assess teachers' training.

Project activity :

The 3 Patient Organisations participate in the set up of training and information materials.

PATIENT ENGAGEMENT

Communication :

The 3 Patient Organisations partners design & carry out awareness raising campaigns with their members through various communication activities.

The Patient Organisations partners also support dissemination of the project results to social and economic experts.



2.2 AVOIDING COMMON PITFALLS

Evaluation panel members who specifically review and assess the Patient Partnerships sections and activities within research proposals come across recurrent misconceptions regarding the type and level of information required within proposals. In this section, some of these common pitfalls have been extracted as examples of what is deemed unsatisfactory together with advice explaining how to address the issue and provide an adequate statement.



01

“ Patient engagement / involvement is not applicable or relevant to the proposed project.”

“ We could not find a relevant organisation / a relevant organisation does not exist/the disease is too rare. ”

It is important to explain why it is not applicable. No matter how complicated or technical the research is, there is rarely an example in which patients cannot be involved at all e.g. they can be involved in the dissemination activities at the very minimum.

It is also important to think carefully about whether such dissemination activities are the only activities in which patients can be meaningfully engaged. Sometimes disease-specific patient groups do not exist or might not be organised or equipped to take on Responsibilities.

In these cases, it is acceptable to engage with individual patients living with the disease(s) relevant for the research study even if they are not in a formal patient organisation/group. This would help to understand the relevance and acceptability of the research question.

Alternatively, umbrella patient organisations can be approached. In any case, several resources are available for finding contacts of disease-specific patient organisations^{11,12}.



02

“ Patient organisations will recruit patients as donors for the biobank . ”

Not enough explanation is given as to how this will be achieved. Who ? How ? When ? Was the patient organisation involved in developing the recruitment strategy ?

If involvement / engagement activities are not planned, please provide an explanation as to why it was not possible in this project.



03

“ Patients organisations have been involved in the design of the study ”

It is important to explain how this has been achieved and what has changed / improved in the design of the study as a result of the patient involvement.

04

“ Patient representatives will be invited to attend scientific meetings / conferences ”

Although inviting patients to hear about the results of the research should not be discouraged, it is important to think about :

a) the specific role of patients who have had a meaningful role in the project in presenting the results and information in accessible language (i.e. for non-scientists),

b) supporting patients to attend conferences (e.g. fellowships / bursaries to cover travel expenses) and

c) for patients / Patient Organisation to have a role in the programme of the conference either within the programme committee, as a speaker, session moderator or panel member.

05

“ The applicants are in contact with patients and patient organisations so patients will be engaged / involved throughout the research project ”

“ Patient organisations will be responsible for disseminating the research results to their communities ”

Any specific roles and responsibilities need to be discussed and agreed between the researchers and the patient organisations (or patients) before submitting the proposal and need to be detailed in the proposal.

Generic statements are not useful to evaluators and need to be expanded to include the descriptions of the responsibilities of the different partners.

2.3 INDICATIVE CHECKLIST FOR SELF-EVALUATION OF APPLICANTS



The checklist set out below is purely indicative and not prescriptive.

It is intended to show applicants (researchers and patient organisations) the type of information the evaluation panel will be looking for in a proposal specifically regarding Patient Partnerships. Please note that **not all points will be applicable to all projects.**



- ☐ Have discussions between researchers and patient representatives taken place before identifying the research questions and writing the proposal ?
- ☐ Have you described how the patients/patient representatives were identified and selected ?
- ☐ Has the input of patients/patient representatives been integrated in the development of the proposed research project ? Have you described what changed / improved as a result of this input ?
- ☐ Have clear roles and responsibilities been assigned to the patients / patient representatives in the project ?
- ☐ Have the Patient Partnership activities been clearly explained (who, what and when) ?
- ☐ Have the available resources of respective partners been maximised to the benefit of the research project (e.g. registries, know-how, networks, communication channels) ?
- ☐ Have the approaches through which the patients / patient representatives will be engaged / involved / participate in the project been described (e.g. focus groups, interviews, surveys etc.) ?
- ☐ Has a process been included to ensure two-way communication between the partners throughout the life of the project ?
- ☐ Are patient representatives included in the governance of the research project e.g. as steering committee member, leader or co-leader of a work package ?
- ☐ Are follow up reports (e.g. including feedback from patients / patient representatives) planned within the deliverables of the project to assess the actual Patient Partnership once the project has started ?
- ☐ Are there other specific deliverables relating to the Patient Partnership activities described (e.g. publication of guidelines, analysis of a focus group and/or a survey data, development of a video etc...) ?
- ☐ Has the overall added value of the Patient Partnership for the project been clearly highlighted ?
- ☐ Has a budget been allocated, and costs estimated and justified in line with the proposed specific activities for the Patient Partnership ?
- ☐ Has the monitoring of the Patient Partnership been integrated within the consortium management plan ?
- ☐ Have you planned to include the impact of your Patient Partnership on your study in your publications ?

2.4 TESTIMONIALS OF A SUCCESSFUL PATIENT PARTNERSHIP

In 2019, [Professor Joost Hoenderop](#) was one of the 21 successful applicants to the JTCs of the EJP RD. He proposed a collaborative research project for improving diagnostics and grasping the disease mechanisms of rare hypomagnesemia in patients with CNNM2 mutations with 6 co-applicants including [Antonio Cabrera Cantero](#) from the Spanish Association for information and research on familial hypomagnesemia HIPOFAM. [Joost](#) and [Antonio](#) describe their experiences on this partnership here.



1



How did you approach Patient Partnerships in your research project(s) ?

In our research group we focus on the molecular mechanisms of diseases in which the magnesium homeostasis is disturbed. In this respect we contribute to the identification of novel genes that are mutated in patients with magnesium disturbances.

I had just started a research project with Dr. Alfonso Martinez-Cruz (also investigator in our EJPRD 2019 (consortium) and we had been discussing how the kidneys control magnesium balance.

He informed me about Hipofam (<http://hipofam.org>) in Spain, a patient organisation for information and research on patients with Familial Hypomagnesemia. I met Antonio Cabrera Cantero from Hipofam at the ERKNet meeting organised by Prof. Franz Schaefer in Heidelberg and we discussed hypomagnesemia and the importance of treating patients with these disturbances.

This stimulating setting provided the opportunity to connect, exchange information and build a research network on one of the main causes for hypomagnesemia, mutations in a new gene called CNNM2. We identified patients who were connected to the patient organisation and also from other areas in Europe. This was the start of FIGHT-CNNM2 – a joining of forces with research groups in Spain, Germany, Canada and the Netherlands and the patient organisation Hipofam. **Joost**

Collaboration requires an effort from both sides. The more we understood each other's goals, the better the communication and the collaboration became.”

Veronica Popa
MCT8-AHDS Foundation
Romania

How did you first get involved in this research project ? Were you or your organisation already in contact with the researcher or did they contact you with a proposed plan ? Did you contact them specifically for this research project with your ideas ?

Before this collaboration, we did not know Joost Hoenderop personally, but we did know of his reputation and his work. We even had specific contacts with a collaborator of yours a few years ago. When we met Joost at the ERKNet meeting in Heidelberg, we knew immediately that some kind of collaboration would emerge. He followed up from our meeting and contacted us to invite us to participate in the FIGHT-CNNM2 project.

The mediation of our great collaborator Alfonso Martínez, a researcher at the CICBIOGune in Bilbao and a member of the EJPRD consortium, was also essential. Working together this project has exceeded our expectations. **Antonio**

2

What were the main challenges and how do you think you could overcome these ? What capabilities are required on both sides for a successful partnership ?

The main challenge is developing a strong interaction with patient organisations that have a focus on the same patients.

It was important to the success of our collaborations that Hipofam was aligned with the area we wanted to investigate. An important aspect is to really collaborate, not simply to exchange information, but to learn from each other.

Aim to understand each other's challenges and what would make a difference. Too often you see that patient organisations are involved under certain conditions in research consortia but there are not really the opportunities for them to contribute.

Here, in our EJPRD2019 consortium, they are full partners, for example they have their own budget.

This provides energy and partnership, and opens two-way communication that facilitates realistic deliverables for us as a team. **Joost**

Which were the challenges you encountered when reaching out to researchers ?

The challenges were many.

First of all, Hipofam is a young and small entity, made up of partners who work on a voluntary basis. Most of us are parents of children who have some type of hypomagnesemia and we do not come from the world of science or health. We participate and contribute funding to research projects, but on a smaller or national level. In contrast, FLIGHT-CNNM2 is an international project with European funding, for which we were receiving funds to carry out our part of the work. This in itself changed the nature of our contribution. When we were offered the opportunity to participate in the project, we were clear that it was a unique opportunity, but we had to find a way to fit into the consortium and find our role.

Partnering in this project asked us: to contribute our knowledge about what the patient feels and translate this into the language of the researchers; to combine being parents of sick children with participating in the consortium and finding time to attend meetings, prepare documentation, follow the progress of research, contact patients, etc; to find patients from other countries potentially transforming Hipofam and increasing the impact of the association. Were we ready for this challenge ? **Antonio**

3

Have you had previous experience of Patient Partnerships in your research and if so, is there anything that in retrospect you would have done differently ?

Please describe both positive and negative experiences you might have had.

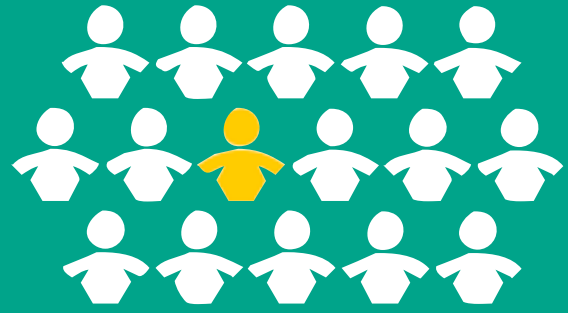
It is important as a biomedical researcher to be in contact with patient organisations. Sometimes you realise that you do not immediately speak each other's language.

The language of advanced research methodology can create barriers and make it difficult to understand what is really important for patients. I learned to share, to exchange, to invest in partnership and explain my background. Only if both parties can understand their challenges will it be possible to have a successful partnership.

Making that first appointment with people from Hipofam to visit them at the ERKNet conference in Heidelberg. This was the start of the EJPRD project FIGHT-CNNM2 and the start of that communication. **Joost**

How did you manage the challenges through the development of the partnership ?

Hipofam members are a great family. We are united because we pursue the same goal - to improve the quality of life of our children.



When we presented the opportunity to participate in FIGHT-CNNM2 in our assembly, we all agreed that it was a great opportunity and at the same time a great challenge. We have encountered the challenges one by one and have discovered together how to overcome them. With the support of Joost and Alfonso little by little we found our fit and importance within the project, both for patients and the researchers.

Hypomagnesemias due to mutations in CNNM2 are extremely rare and we know that the evolution and symptoms can be very different between patients. This vision of how a patient experiences their disease is something that only patients know.

This information does not appear in a genetic study or in a blood test. That is an important contribution that only we can make from Hipofam.

At Hipofam we are also in contact with many patient associations and this may be a way to help researchers find patients in various countries. Furthermore, the budget granted by the EJPRD can serve to professionalize our participation in the consortium and this will help us to combine our family and work life with participation in the project. We have also considered the possibility that other patients may want to be part of Hipofam as a result of our involvement.

We all agree that it would be an incredible thing if we managed to grow our small entity and make it a world leader in this disease. **Antonio**

4

Could you describe in a couple of sentences any unexpected benefits / outcomes that you encountered as a result of the partnership ?

Antonio Cabrera Cantero from Hipofam is a wonderful guy. He is working hard for all patients with hypomanganesemia. It can be difficult in rare disease to find a doctor who knows the disease and who has or has had cases.

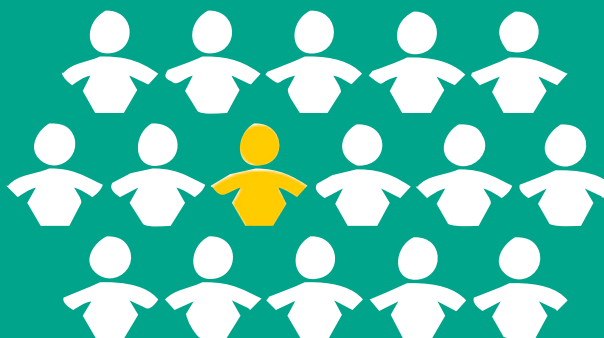
It is even more difficult to find other patients ; there are no patient associations that we know of, neither within nor outside of Spain, specific to Familial Hypomagnesemia in any of its variants. Hipofam aim to change this reality. They want those parents whose children are diagnosed with Familial Hypomagnesemia to have a place where they can find support, information, advice, and fight to find a cure.

I was convinced by their passion and approach and wanted to collaborate with them. It is a great benefit is to witness how our research could have a direct impact on the patients. This is the driving force and stimulates the entire team to be innovative. **Joost**



How much and which input did you have in the research proposal and in developing the specific activities for which you are responsible / involved in ?

We were lucky to have an exceptional person - Joost Hoenderop - who helped us discover how Hipofam could help. While we do not participate in the scientific proposal, we did in the informative part and in the contact with patients. We offered our contacts with patient entities across Europe, to support participation in the research and the possibility of publicizing the project in forums and assemblies of these organizations. **Antonio**



Heta - Finland
Dysmelia

5

Do you have any advice for researchers who are thinking about developing patient partnerships but have never done so before ?

Patient organisations are extremely important. They connect the patients to your research. By listening to their challenges, you will learn how to adapt and fine-tune your research questions to be even more successful and impactful. We also found that exchanges with patient organisations provided inspiration to junior academic researchers. They could see the impact their hard work could have on the quality of a patients' life. My advice would be to invest the time in partnering - talk to each other and try to understand each other's problems. **Joost**

Could you describe in a couple of sentences any unexpected benefit(s) /outcome(s) that you encountered as a result of the partnership ?

We did not expect to contact a patient before the project even started, but we did. Today, thanks to FIGHT-CNNM2, we have a Dutch partner, with whom we regularly exchange messages and who now actively participates in Hipofam. We are planning a face-to-face meeting to further strengthen friendship ties. **Antonio**



6

Do you have any tips or recommendations / advices for researchers and /or Patient Organisation who are thinking of developing patient partnerships in research projects ?

My advice is that if you are thinking about it, just do it ! When we started at Hipofam in 2013, we were only 3 people. We did not know how we would do it but we wanted to help improve the quality of life of these children. We wanted to fund research, but we had no funds. We didn't know anything about all this, but we were excited and wanted to work. In these 7 years we have changed the reality of these diseases in Spain and in Europe. We are doing it. So my advice is, FORWARD. As a great Spanish poet said. Walker, there is no path, the path is made by walking. **Antonio**



3 ACKNOWLEDGEMENTS

This guide has been developed in the framework of the European Joint Programme on Rare Diseases by a dedicated multi-stakeholder working group on :
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(PENREP) chaired by EURORDIS.

This working group includes the following members :

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Any questions concerning this guide and its content ?

Please contact the EJP RD Helpdesk :

<https://www.ejprarediseases.org/index.php/ejp-rd-helpdesk/>

Several photos used in this guide were submitted to the EURORDIS Photo Award ; all photos credit to EURORDIS-Rare Diseases Europe.

Jeanne - France - CDG ALG 11



4

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