

How to get started in research

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EURORDIS Open Academy Meet up

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GETTING TO THE PRACTICE OF PATIENT PARTNERSHIPS IN RARE DISEASE PROJECTS

Brief overview



- How to manage/coordinate a research project as a small patient organization for a disease with little focus



- Initiating and leading research instead of being just invited to join a consortium



- Your responsibilities as an advocate/patient representative



- Co-developing and co-writing a research proposal: from theory to practice



- Learnings and tips



2019-2021



POLL

DRUG DISCOVERY

The most important thing to make a research project concretely happen is.....



POLL

DRUG DISCOVERY

For my rare disease, efforts are currently needed in:

- **Basic Research:** starting from zero and learn of a biological target (e.g., an enzyme, protein, gene, etc.) thought to be dysfunctional
- **Pre-clinical Research:** investigate a new target
- **Early Clinical Research:** entering Phase I
- **Late clinical research**
- **I don't know**



CHALLENGES AND POSSIBILITIES

Theory



**Research
participant**

MY CHALLENGES AND POSSIBILITIES

Theory

Practice



**Research
participant**

**Taking action as
partner
and decision
maker**



MAIN CHALLENGES

STAGNATION OF RESEARCH



- Projects fail or are dropped at some stage of the R&D process
- Disease models do not fully recapitulate human diseases
- Academic-industrial collaborations not including patient involvement in early research stages
- Funding: we do not have a fundraising culture and “financial hubs” like in North America
- Initial research, often occurs in academia: ***How to start and generate interest?***

STAGNATION OF RESEARCH



FACTS

- Too many products fail in Phase 2, indicating weaknesses in predictivity of preclinical studies
- Population has not been defined properly
- No early patient involvement

Where/how to start?

**Identify the main actors at each research stage
(it is not always Industry)**



Development of novel findings into meaningful medicinal products

Is not an easy task and requires many disciplines



Your role:

Finding and bringing together different disciplines

Involving the right expert at the right time can save time and money



**But, How do you select your collaborators?
Where do you find the expertise you require
at a specific point in time?**

- Matchmaking tool
- Learning/reading about other rare diseases/research studies: you learn about disciplines and expertise that may work for your consortium as well

01

**KNOW WHERE EFFORTS
ARE NEEDED**

DRUG DISCOVERY



Initial Target (protein, genes..) **identification**

Validation: generates increased confidence in the relationship between target and disease

Compound Screening

Lead identification and optimisation: discovering a small molecule or large molecule lead compound with the desired activity and progressing it towards a therapy

Needed to justify drug discovery effort:

=

Turn a small or large molecule into a preclinical candidate

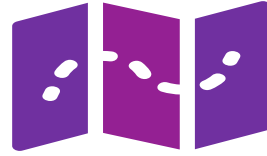


Selection of a candidate for clinical development



Many compounds may get candidate status which should not have been.

Consequent failure in pre-clinical or clinical studies with compounds progressing through the discovery pipeline with obvious cost and time investment



INITIAL RESEARCH (OFTEN OCCURRING IN ACADEMIA)

Data is generated to develop a hypothesis that a protein or pathway will result in a therapeutic effect in a disease

IDEA FOR A TARGET

Can come from a variety of sources: academic, clinical research and from commercial sector

Input is required from scientists from a variety of disciplines and backgrounds AND YOU ARE AMONG THESE!

Patient-led

ADVANCING RESEARCH



**Initiated and
coordinated by
patients**



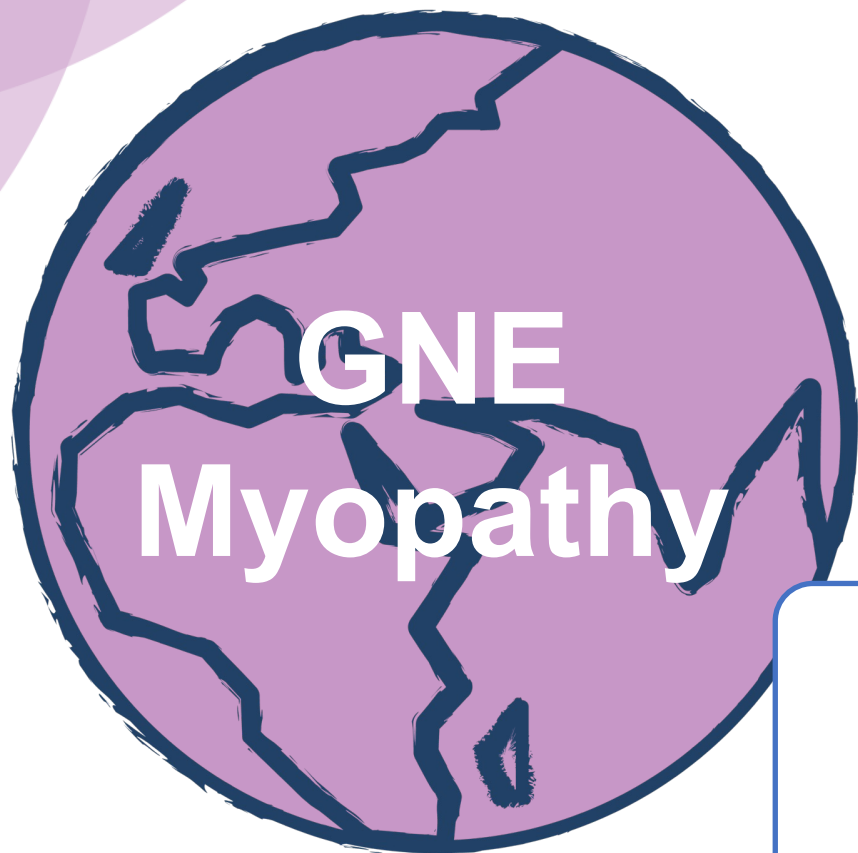
MY PATIENT ORGANISATION

- Run by 2 patients: Myself and the president
- Annual income: ~ 5,000 EUR
- Language barrier
- Patients: Lots of requests and no desire to work in advocacy

YOUNG ADULT ONSET GENETIC MUSCLE DISEASE

Prevalence ~1-6/1,000,000

- Mutations in the GNE gene ▶ Initiates and regulates sialic acid biosynthesis pathway
- Initially identified in Japanese and Middle Eastern populations, now reported world-wide
 - 1,100 cases reported in literature



Community needs ▶ Committed and educated advocates

Only 2 patient organisations (Italy and India) + 2 Research Foundation (US)

Little research, little funding, in need of increased global efforts and transnational collaboration



DRUG DISCOVERY

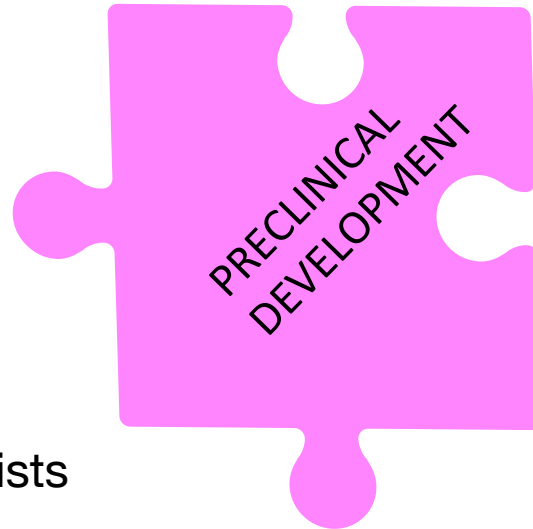
Chemistry optimization to further improve the properties of the compound

Patients

Chemists

Pharmacologists

Drug metabolism specialists



Regulatory
experts

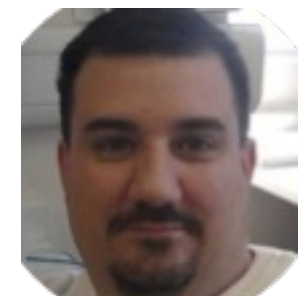
Formulation
experts

Clinical experts

How I started contacting People



Michela Onali



Fabrizio Pertusati



Pierluigi Caboni



CHEO RESEARCH INSTITUTE
INSTITUT DE RECHERCHE

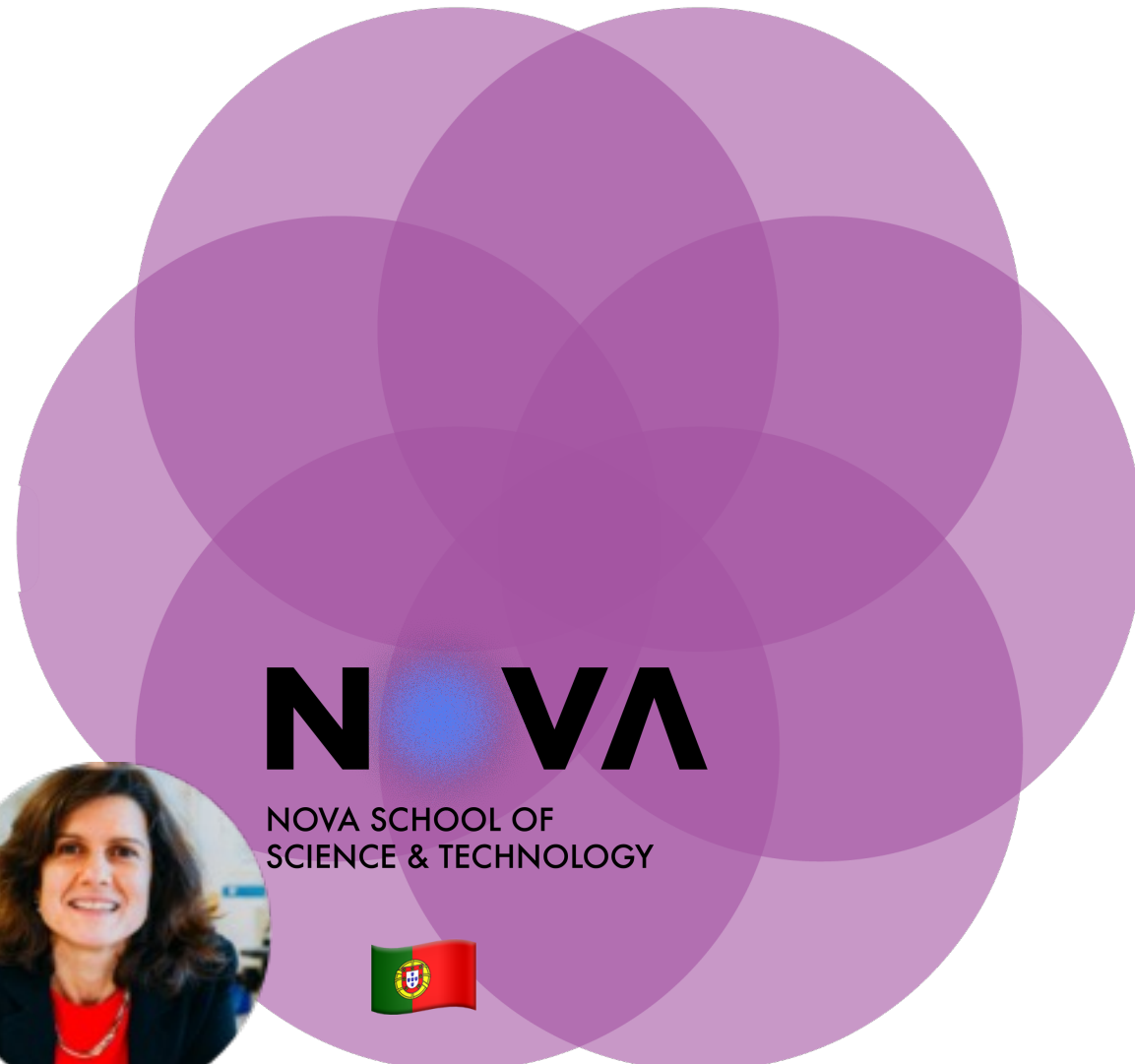


Hanns Lochmüller





Rüdiger Horstkorte



NVA

NOVA SCHOOL OF
SCIENCE & TECHNOLOGY



Paula Videira



ProD **GNE**



02

IDENTIFY YOUR OBJECTIVES

Objectives in ProDGNE



1

DEVELOP

An improved substrate replacement approach

2

IMPROVE DRUG ABSORPTION AND THERAPEUTIC EFFECT

Address gastrointestinal AEs encountered in clinical trials

3

IDENTIFY

Biomarkers through a multi-omic approach

4

DETERMINE

Safest and most promising compound. Modeling GNEM in zebrafish using morpholino and CRISPR technology

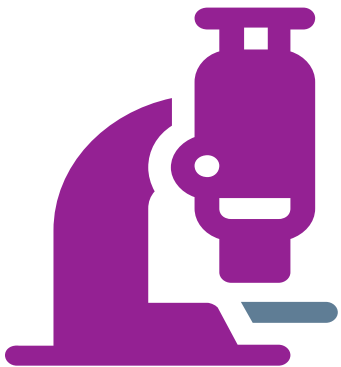
03

**IDENTIFY A POSSIBLE CALL
FOR FUNDING
+
REASON ON THE
CRITERIA/REQUIREMENTS OF
THE CALL**

JOINT TRANSNATIONAL CALL 2020

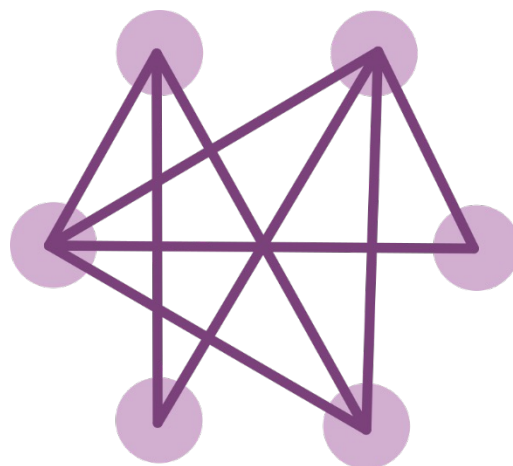
Pre-clinical Research to Develop Effective Therapies for Rare Diseases

- Open to research teams from countries with national/regional funders involved - min of 4 teams from 4 countries
- Funding transnational **collaborative research** to enhance the cooperation between RD scientists **across Europe and beyond**
- Each country/region funds their own national/regional component of the transnational research project
- Funding from the **European Commission** distributed through the national/regional funding agencies
- INSERM funding PAOs: 50,000EUR per consortium



PROPOSAL SUBMISSION

**Basis to build mutual trust,
team spirit, long-lasting
relationships**





Joint Transnational Call 2020

Pre-clinical Research to Develop Effective Therapies for Rare Diseases

EJP RD Joint Transnational Call 2020

Pre-clinical Research to Develop Effective Therapies for Rare Diseases PRE-PROPOSAL AND FULL-PROPOSAL

1. *Background, state of the art in the research field and preliminary results obtained by the consortium members*
2. *Description of the working program including objectives, the rationale and the methodology, highlighting the novelty, main hypothesis(es), originality and feasibility of the project*
3. *Description of the unmet medical and patient need that is addressed by the proposed work and the potential health impact that the results of your proposed work will have;*
4. *Added value of the transnational collaboration*
5. *Description of patient organizations within the proposal, including their role and contribution*
6. *Risk management: identification of possible bottlenecks*
7. *Feasibility of the project given requested resources*
8. *Realistic timelines*



CONSORTIUM MAKE UP

Your role as applicant

- Before contacting your first partner, be concrete, short and include the call for proposals + deadline
- Read carefully the pre-announcement, eligibility and call criteria to gain more time to contact possible partners and initiate discussions
- Partner finding tool can help if available
- Check eligible countries/beneficiaries
- Involve Early Career Researchers and partners from countries usually underrepresented (Czech Republic, Slovakia, Hungary, Lithuania, Poland, and Turkey)
- Discuss the role of PAO in the project

THINGS THAT “COULD” HAVE STOPPED ME

1
My idea will never be taken seriously

2
I am not a scientist, this is something too big for me

3
It is not worth the effort. Success rate of these calls is less than 10%

4
There is a Minimal number of experts in participating countries

5
The deadline is too close

6
I do not have the time

7
This is not realistic

8
THIS IS AN ULTRA-RARE DISEASE it is just too complicated



**How to coordinate and manage a
Research team and position yourself as
a primary partner?**

ATTRACT RESEARCHERS TOWARD YOUR IDEA



Especially the ones new to your disease

Make the idea interesting and stimulating (Researchers spend already a lot of their time writing grant proposals)

Be present. Do not underestimate the impact of your motivation and the value of your ideas

Show your ideas have the biggest foundation: you live the disease every day, you have (re) read all possible scientific publications about your disease

You are an advocate so you follow research efforts world-wide

You may have a direct experience as a research participant

You go beyond statistics: You talk to patients and collect their individual experience and needs

- YOU KNOW ABOUT R&D -

SHORT GUIDE ON PATIENT PARTNERSHIPS IN RARE DISEASE RESEARCH PROJECTS

BASIC PRE-CLINICAL
TRANSLATIONAL & SOCIAL

Written by the members
of the working group PENREP*
Guide first published in July 2020 on
www.ejprarediseases.org

* **P**atient **E**ngagement in
Biomedical **R**esearch **P**rojects.





WHAT IS EJP RD?

OUR ACTIONS AND SERVICES

NEWS AND EVENTS

HELPDESK



FUNDING OPPORTUNITIES

Open Funding Opportunities

Past Funding Opportunities

Funded projects

PATIENTS IN RESEARCH

VIRTUAL PLATFORM OF DATA AND RESOURCES

What is the virtual platform?

What do we do?

Resources for RD Research

Services and Events

TRAINING & EDUCATION

General Information on Training

RD Research Training

Patients Training

ERN Workshops

Online Academic Education Course

RESEARCH TRANSLATION SUPPORT OFFICE

Innovation Management Toolbox

Mentoring for Translational Research

Follow-on funding support

CLINICAL STUDIES SUPPORT OFFICE

Support office

Rare Diseases Clinical Trials Toolbox

ETHICS & REGULATORY ADVICE

Introduction to AREB

Key Ethics & Regulatory Documents

ERN Registries Generic Informed Consent Forms

1200 people

EJP RD in numbers

87 beneficiaries





Online Courses / Healthcare & Medicine



From Lab to Clinic: Translational Research for Rare Diseases

Gain an overview of the issues, challenges, and opportunities in translating research into treatments for rare disease patients.


★★★★★ 4.8 (13 reviews) 1,784 enrolled on this course


Educators are currently **active** on this course

From Lab to Clinic: Translational Research for Rare Diseases

★★★★★ 4.8 (13 reviews)

1,784 enrolled on this course

 5 weeks

 4 hours per week

 Digital certificate when eligible

 Introductory level

Join course





DISCUSSION

**SAVE THE DATE! Final ProDGNE
public meeting: in Sardinia on
19-20 April 2024**

X @ProDGNE