European
Commission

Patient Advisory Board Workshop
Dec 8th – 2022 Budapest

Chair Dorica Dan, CoChair Gabor Pogany,

Coordination Anne Hugon

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Intellectual Disability
and Congenital
Malformations (ERN ITHACA)

Agenda – am Patient Council, Internal Board

- 9h00   Welcome in Budapest, Introduction , Anne Hugon, Dorica Dan

  Added value of ERN to engage Patient organisation at National level ", Gabor Pogany (represented)
- 9h20 A case study within ITHACA from national to EU scale: history of our journey in Rasopathies, Ioel Detton
- 9h30 Round Table presentation & my plans with Ithaca All participants

10h30 Coffee break

- 11h00 5 years of involvement and evaluation, achievements and challenges, Dorica Dan
- 11h15 Activity report & Work Group, What can I do, Anne Hugon
- 11h30 EURORDIS in supporting Patient Partnership : A spotlight on ERN ITHACA, Rita Francisco

  European Patient Advocacy Groups (ePAG) in numbers / A spotlight on ITHACA ePAG / Team support patient partnership/ New patient involvement framework in ERN governance / New application process + Resources for newcomers / Empowerment and training (guides) (new EURODIS website) / ePAG horizontal Working Group: A spotlight on the Patient Partnership Working Group
- 12h00 Feed back on the Team building session "UBUNTU “ & and Discussion, Dorica Dan
- 12h30 Election
  - Patient Board 2022 - 2023
  - Chairs of Patient Board 2022 – 2023

13h00 Lunch time

Agenda – pm Focus on Work Group with clinicians, sharing expertise and expectations

- 14h00 Introduction
- 14h15 Focus WG Guidelines
  - When and how should be Patients be involved in guidelines, asking your opinion

  Agnies van Eeghen, Charlotte Gaasterland, Mirthe Klein Haneveld
- 15h45 Focus WG Teaching & Education
  - What resources can I rely on, what can we develop as educational support needed, asking your opinion

  Laurence Faivre, Giuzeppe Zampino, Anne Hugon

16h15 Coffee Break

- 16h45 Focus WG NDD Neuro Developmental Disorders
  - Over view on Neuro-developmental Disorders work Group, Tjitske Kleefstra, Anne Hugon
  - World Café “Clinical genetics & Psychiatry (Psychiatric phenotyping) from a multidisciplinary approach, including the perspective of the expert by lived experience”

  Tjitske Kleefstra, Claudine Laurent-Levinson, Jolanda Van Golde
- 18h00 Final discussion, Wrap up and conclusion Dorica Dan, Gabor Pogany

Represented …
Gabor Pogany, UFERDIS, Rare Diseases Hungary

Added value of ERN to engage Patient organisation at National level”

THE ADDED VALUE OF ERNs TO ENGAGE PATIENT ORGANIZATION AT NATIONAL LEVEL

„Start by doing what's necessary then do what's possible and suddenly you are doing the impossible” *Saint Francis of Assisi*

Pogány Gábor Ph.D.
President of HUFERDIS.
member of HAPO, ePAG and Orphanet International Advisory Board

www.rirosz.hu www.orpha.net www.bemosz.hu

ERN ITHACA Patient Council Workshop, 2022. 12. 08.

Welcome to Budapest

Here is a short video about the city: [Budapest.mp4 parancsikonja.lnk]

What is HUFERDIS?

- Rare Diseases Hungary - The Hungarian Federation of People with Rare and Congenital Diseases - (HUFERDIS) was created on 05.05.2006 after several years of preparatory work.
- It is a network of 60 Hungarian patient organisations and several individuals active in the field of rare diseases.
- The federation is dedicated to helping people with rare diseases and assisting the organisations that serve them.
- Our community has a rich knowledge base and experience on listening and capturing the needs and perspectives of people living with a rare disease in different contexts.

There is only one way to reach our goals!

More effective, professional operation with strategic approach!
The best way to achieve it by the help of national and international cooperation!

- **Utilize common experience,**
- **Best practices,**
- **Financial support,**
- **Possibility of common political pressure!**

Therefore, we participated in the advocacy work to get ERNs form the beginning.

No country is rich enough to deal effectively with all the thousands of rare diseases, to build enough knowledge and capacity for each of them. At least continent level cooperation is required which results in huge added value!

HUFERDIS Advocacy : bridging diversity to support rare diseases patients

- Empowerment of RD patients and families
- Share cases and life stories to better support them
- Promote and disseminate activities through collaboration and networking
- Providing a voice of consensus to stakeholders
- Promotion of multidisciplinary advocacy at EU/national level

Coordination & Advocacy

- Support & inform on rare diseases and ERNs
- Advocate – only patients can truly represent their needs
- Enhance research – unique contribution

European level

National level

RD problems

The perspective of a RD National Alliance is different:

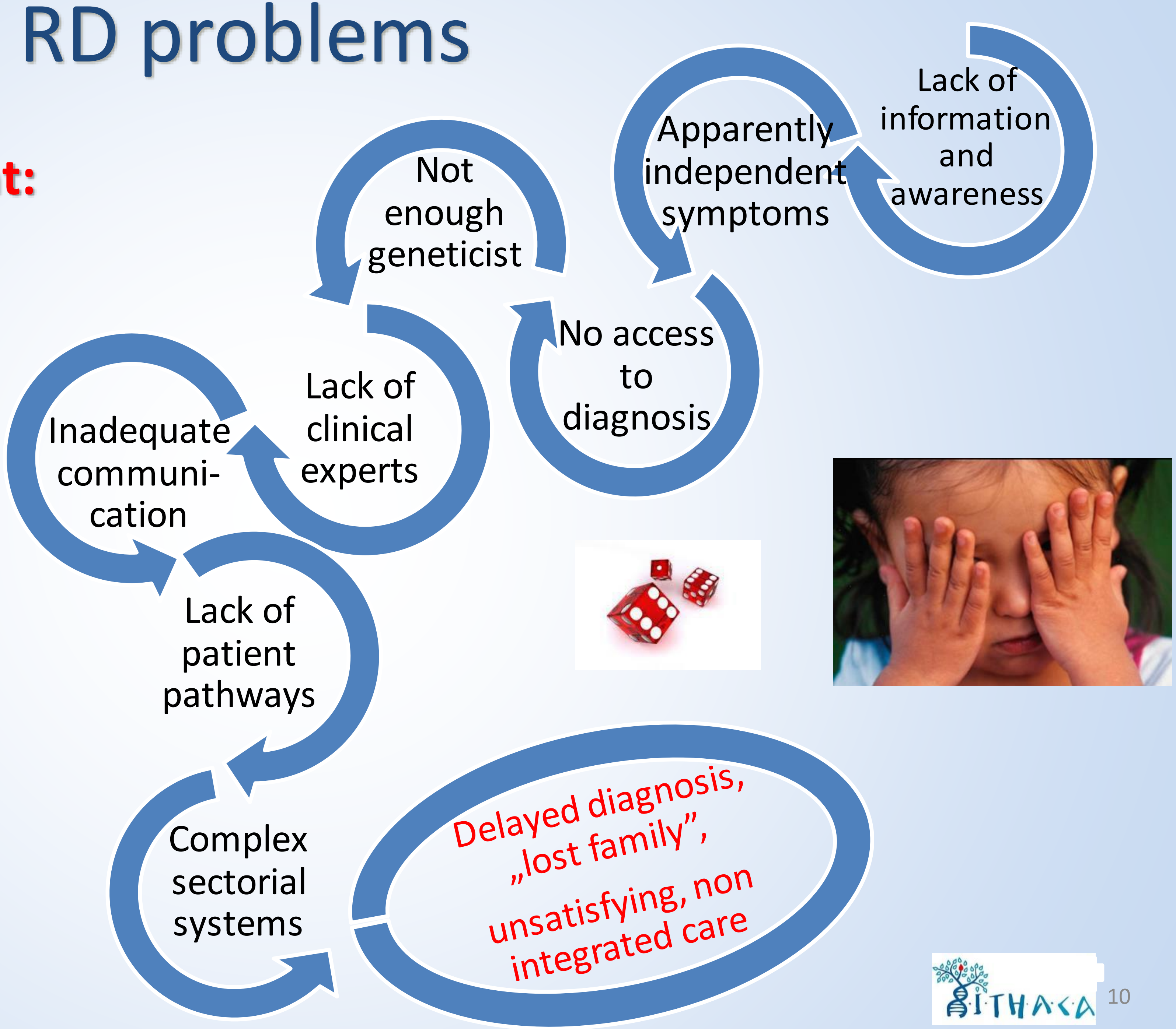
We represent all rare disease patients and their families

European Reference Networks

National Rare Disease Plan

EUROPLAN
European Project for Rare Diseases National Plans Development

We leave no one behind!



Lack of information and awareness

Apparently independent symptoms

No access to diagnosis

Not enough geneticist

Lack of clinical experts

Inadequate communication

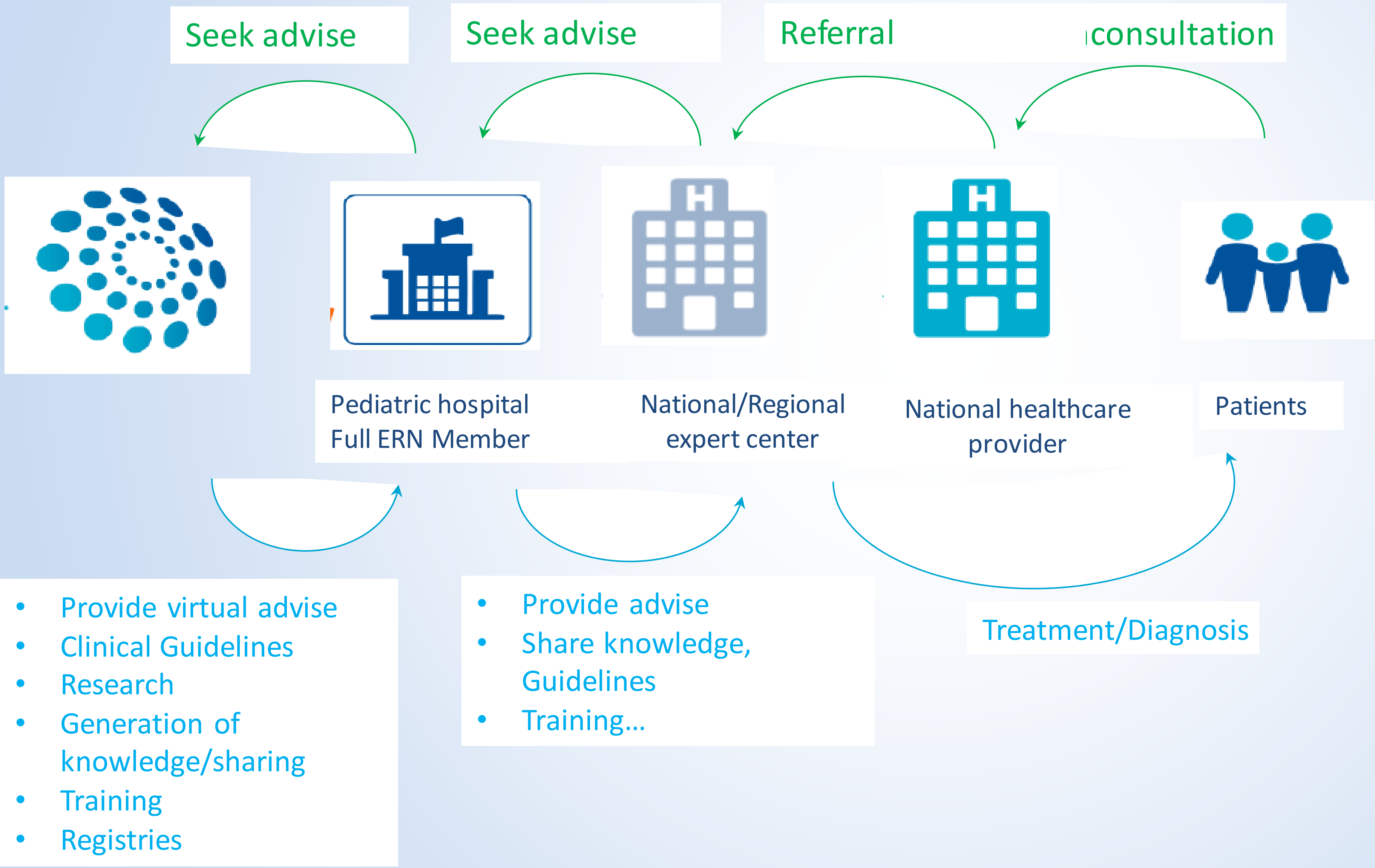
Lack of patient pathways

Complex sectorial systems

Delayed diagnosis, „lost family”, unsatisfying, non integrated care

ITHACA

In order to function effectively, the ERNs must be integrated into the National Health Systems!



Seek advise

Seek advise

Referral

consultation

Pediatric hospital
Full ERN Member

National/Regional
expert center

National healthcare
provider

Patients

- Provide virtual advise
- Clinical Guidelines
- Research
- Generation of knowledge/sharing
- Training
- Registries

- Provide advise
- Share knowledge, Guidelines
- Training…

Treatment/Diagnosis

A revolutionary new culture of helping!

It has historical significance, as the EU has thus begun to harmonize the very different healthcare systems of the member states, even though it has not financed such a thing until now, as healthcare is a competence of the member states.

In 2021, a second round of expansion – AMEQUIS program on the assessment of applicants, continuous monitoring of operations, and evaluation every 5 years.

What can we do as a patient representative generally

- Articulate the perspective of patients
- Provide insight on issues, problems, and/or questions appropriate to the viewpoint of patients and family members
- Ensure the exchange of information between patients and other stakeholders
- Protect patient's rights and help them learn how to obtain information and services
- Provide tools and information to help patients become empowered

It is not simple

You should have:

- Personal experience with and/or be knowledgeable about the specific illness or condition
- The ability to disseminate information about your experience to the patient community
- Experience as a patient advocate
- The ability to understand and actively participate in discussions with other stakeholders
- Formal affiliation with a patient advocacy organization
- The ability to identify issues that are important to patients

Our underlying working principles

- Patients and families are not only „end users” of national strategies and action plans = they are careers, health and social actors, managing complex day to day care all along their life span
- Patient support & advocacy groups are the best allies to take an active role in shaping research, healthcare and social national and regional policies for rare diseases
- Ensure that patients and patient representatives are involved at each step of the policy and decision-making processes in the field of rare diseases
- Support patient groups and rare diseases national alliances: awareness-raising, capacity building & training, exchange of information, networking and outreach

As an ePAGs member

You must commit to adhering to the following core values:

- Respect the mission of the ERN and its governance structure.
- Listen to the opinions and requests of others.
- Show solidarity, mutual respect and support.
- We need to represent all the diseases of a given ERN, not just our „own”.
- Adhere to the principles of equity and social justice.
- Conduct yourself with professionalism in engaging with the clinical, research leads and fellow patient advocates.

EURORDIS
RARE DISEASES EUROPE

European
Patient
Advocacy
Group

The involvement of patient organisations and patients in the European Reference Networks

What are the European Reference Networks?

European Reference Networks (ERNs) are networks of Centres of Expertise that connect experts and researchers, who share the same interests in a specific rare disease or highly specialised treatment, across the European Union and the countries of the European Economic Area region. ERNs are first and foremost healthcare networks that aim to improve access to diagnosis, care and treatment by sharing their expertise, knowledge and resources, guiding care and treatment.

It is therefore of utmost importance that ERNs remain centred on patients' needs. **ERNs give clinicians and patient organisations from different countries the opportunity to co-design better healthcare services for people living with rare diseases.**

What are the European Patient Advocacy Groups?

European Patient Advocacy Groups (ePAGs) are patient

What is the role of ePAG advocates?

They represent the interests and needs of the patient community:

- to ensure that the needs of people living with a rare disease drive the activities of each European Reference Network.

- to champion the diversity of views of the wider patient community relevant for each ERN, and not just of their own disease area.

- to work with their national and European networks in order to facilitate two-way communication between the European Reference Networks and the patient community.

- to work in partnership with the clinicians to support the development of clinical practice guidelines and other clinical decision support tools and contribute to research, education, information and awareness activities.

The position of an ePAG advocate is a voluntary position and

What information should we provide to encourage new applications

- What are the European Reference Networks (ERNs)
- What are the European Patient Advocacy Groups (ePAGs)
- What is the role of an ePAG Advocate
- What do you need to become an ePAG Advocate
- What would be your responsibilities as an ePAG Advocate
- How much time would you need to commit
- How can you apply

1. Capture your community's collective needs and perspective on specific matters

2. Relay these insights to the clinicians involved in the ERN to guide strategic planning and collaborative activities

3. Share ERN's information, results and updates with your community

What can a National Allience do?

- Get informed about
    - What are ERNs?
    - Why do ERNs exist?
    - What do ERNs do?
    - How does it work?
- Raise awareness and help patients find the right information (use the EU Commission's native language guides and videos.)
- Propose concrete measures of interest to patients
- Let's insist that decision makers involve patient organizations as regular partners
- Create (or use Eurordis's) guidelines for patient information
- Feed back experiences to decision makers

Thank you for your attention!

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Ioel Detton, French Noonan Association

A case study within ITHACA from a national to a EU scale: history of our journey in Rasopathies,

A case study within ITHACA from a national to a EU scale : history of our journey in Rasopathie

Ioel Detton,
Board member of the council patient of ERN ITHACA
French President of the Noonan Association

A short history of Rasopathies

- **Rasopathies, a big family**:

The RASopathies are a group of rare genetic conditions caused by mutations in genes of the Ras-MAPK pathway.

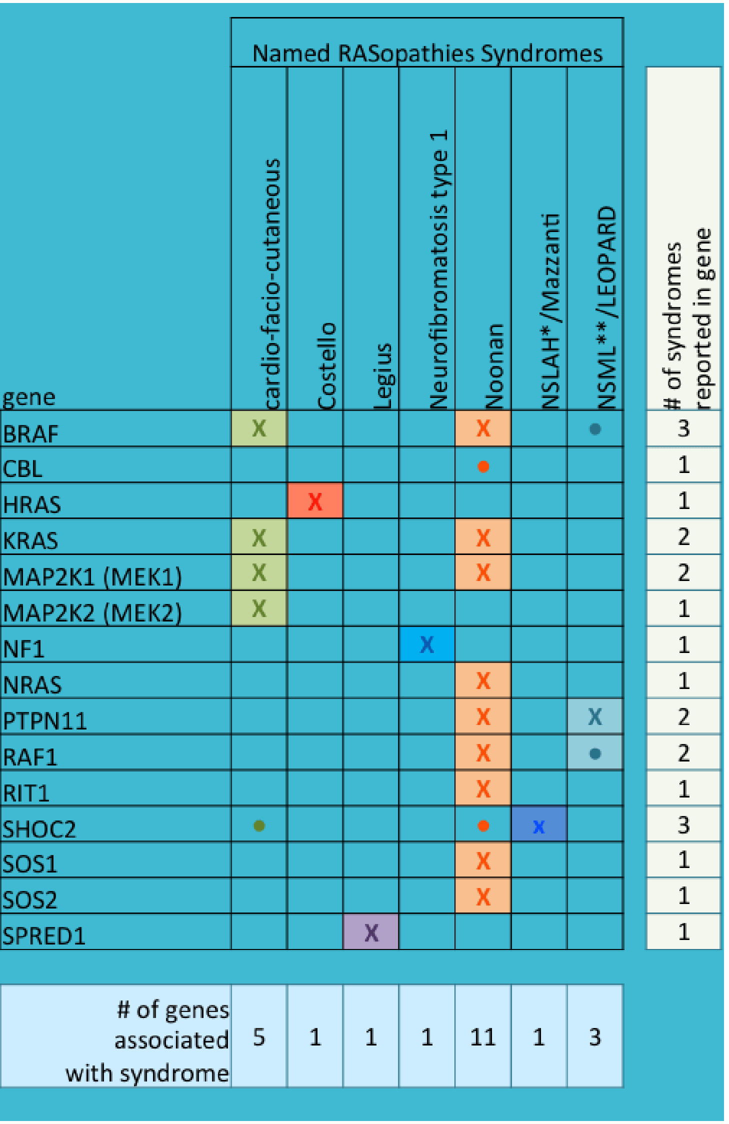
- Several syndromes included like : Costello, Cardio-Facio-Cutaneous, NF1, Legius, Leopard…

- **For SN a rare disease**:

1 in about 2000 / 2500 children.

Discovered in 1968 by Jacqueline Noonan, an American cardio-paediatrician.

Familial autosomal dominant transmission or de novo (genetic accident) for Noonan syndrome.



gene	Named RASopathies Syndromes							
	cardio-facio-cutaneous	Costello	Legius	Neurofibromatosis type 1	Noonan	NSLAH*/Mazzanti	NSML**/LEOPARD	# of syndromes reported in gene
BRAF	X				X		●	3
CBL					●			1
HRAS		X						1
KRAS	X				X			2
MAP2K1 (MEK1)	X				X			2
MAP2K2 (MEK2)	X							1
NF1				X				1
NRAS					X			1
PTPN11					X		X	2
RAF1					X		●	2
RIT1					X			1
SHOC2	●				●	x		3
SOS1					X			1
SOS2					X			1
SPRED1			X					1
# of genes associated with syndrome	5	1	1	1	11	1	3	

A difficult diagnosis:

The expression of a RASopathy is variable according to individuals both in the presence of symptoms and in their severities.

Some syndromes have different possible causal genes.

Various disorders:

- Le visage
- Le coeur
- La croissance
- L'apprentissage
- Le squelette
- Le sang
- La vision
- L'audition
- La fertilité
- La peau
- Divers

* NSLAH = Noonan syndrome with loose anagen hair

** NSML = Noonan syndrome with multiple

From a group of parents…

- Social networks have facilitated the expression and exchanges between people, families concerned in France about their experience, their daily life…
- The isolation of families and patients is often reported as detrimental to the establishment of a diagnosis, the care path and the search for expertise on the disease. Notion of diagnostic wandering.
- The need to understand the syndrome and to know how it fits into the life history of the patient and his family, the guilt sometimes, the doubt and the difficulty in projecting oneself are all themes that impose themselves on us.

From a group of parents…

- Telephone exchanges between parents allowed and allow to put the human aspect back in these exchanges and to perceive the feelings they could feel: anger, sadness, anguish.
- The peer-help has to be built; it is the first step of the organization of an association at a state level.
- The need to formalize an organization in order to be representative in the different milieus: associative, medical and research became more and more important.

ASSOCIATION NOONAN

To the creation of an association at a national level…

- In 2017, under the impetus of Pr Verloes, coordinator of the ERN ITHACA, a national association was created in France.
- Communicating about its existence, developing a territorial network, gathering information and analyzing it with the help of a scientific council, informing, training, popularizing knowledge, supporting research and the medical profession are all actions that we have undertaken.
- Volunteers with their expertise and their practical experiences, are essential as the expression of syndromes is variable.

To the creation of an association at a national level…

It is necessary to **create partnerships at different levels**:

- With associations that gravitate in or around RASopathies or have a convergent interest with ours for a given symptomatology (growth, congenital heart disease for example).
- With the Alliance Maladies Rares, which brings together nearly 200 national associations all concerned with rare diseases.
- With Anddi-Rares health network on developmental anomalies, intellectual disability of rare causes.
- With the rare disease expertise platforms, the centers of reference and competence, the Haute Autorité de Santé and the Ministry of Health.

"Alone we go faster, together we go further".

Up to a European level

Interact at a European level through different plans:

- **The association:** sharing of supports, information, invitations…

- **The policies of the different countries and the European institutions** through Eurordis.

- **Research:** participation in different projects, dissemination of ongoing studies…

It is up to us, as a patient association, to be a committed actor.

Collaborate whit associations up to a European level

Exchanges with associations from different European and international countries

Coordinate actions up to a European level

- Participating in the patient council

as a Board member of the ITHACA ERN 2021/22, I have been able to measure the work done by the epag advocates and add my stone to the building.

- **Actions we have taken through our ERN Patient Council**

- Examining the national plans and strategies of the different countries

- Exchanging with men and women involved in rare diseases

- Finding expertise where it is located

- Creating links to reduce the gap between patients and professionals

- Disseminate experiential knowledge

Coordinate actions up to a European level

- This translates into webinars, representatives' travels, making supports of communication, sharing the activity of the ERN…
- The synergy of actions developed since the creation of the ERN in 2017 continues to bear fruit.
- The synergistic effect is distinct from anything that might have occurred if they had been working in isolation, either each on their own or all together but working independently.
- There is thus the idea of a creative cooperation that is enriched by the action of all.

Research up to a European level

- Europe is also about cross-cutting issues such as registries, genetic testing and research, which are at the heart of our internal scientific council with various projects such as Rasores supported by the French National Research Agency.
- Project coordinated by Pr Verloes and Pr Cavé in France, a multi-site project in different countries with doctors, researchers and patient associations.
- It aims at setting up a unique national reference database. It will support observational studies in populations, as well as translational studies on various aspects of the physiopathology of RASopathies for which access to a large panel of patients is essential.

Research up to a European level

This project will allow the entire ITHACA community to enrich the database (70 academic genetics centers in 25 European countries) and to prepare collaborative work.

The database will be used as a shared clinical resource for several funded projects whose leaders are the French coordinators, notably the eRare NSEuronet network, the PHRC RASTAT, the RAS-Hémato project, and the ANR, whose biobanks will be shared and enriched.

A fabulous action in which we will be involved!

Conclusion

It is by and for the patients that all this is built!

- **This is a necessary evolution because we can no longer reason only in a static manner.**
- **Rare diseases have no borders, expertise is found in different countries.**
- **Working on coordination is bearing fruit, but there is still a lot to do.**
- **Many questions have been asked and will continue to be asked to remove all obstacles**.

Round Table, who are you ?

Epags ITHACA

1.Dorica Dan / dorica.dan@eurordis.org / Prader Willi Association, RO
2.Sue Routledge / sue@pitthopkins.org.uk /Pitt Hopkins UK | Netherlands, NL
3.Ioel Detton / assonoonan@gmail.com /Association Noonan France, FR
4.Tomasz Grybek / tomek@fundacjabb.pl / Fondation Borys the Hero FBB, PL
5.Katarzyna Swieczkowska / katarzyna.swieczkowska@psoni.gda.pl / Rare diseases ID PSONI,PL
6.Gerritjan Koekkoek / gerritjan@cdlsworld.org / Cdls World federation (Cornelia Delange), NL
7.Sylvia Roozen /sylvia.roozen@ifglobal.org /IF Federation for Spina Bifida and Hydrocephalus, EU(BE)
8.Tanja Zdolsek / tanja.zdolsek@ijs.si /Kleefstra syndrome IDefine Europe, SI
9.Maria Palia & Georges Papadopoulos / info@angelman.gr; georgepsy208@gmail.com, Angelman syndrome Greece Associtation, EL
10.Iliana Dimitriu / iuliana.importante@gmail.com /Associtia Sindromul Coffin-Lowry, RO
11.Benoit Fourcroy / benoit.f@spina-bifida.org, /Spina Bifida FR ASBH, FR

- Sweden International , SEGábor Pogány / pogany@rirosz.hu / Hungarian Williams Syndrome Association, HUFERDIS HU
- Ammi Sundqvist-Andersson / ammi.andersson@cordnode.se / Spina Bifida & Hypdrocephalus RBU

Guests

- Bojtor Zsuzsanna, HUFERDIS,HU
- Károlyi Judit, HUFERDIS,HU
- Szili Danileja, Rett Syndrome, EU
- Szabóné Katona Eszter, Disorder of the Corpus Callosum, HU
- Baloghné Liplin Tünde Prader-Willi Syndrome, HU
- Balogh Zsolt , Prader-Willi Syndrome, HU

EPAGS in Ithaca, my plans with you ?

Dorica Dan Prader Willi Syndrome RO

Gábor Pogány Williams Syndrome HU

Sue Routledge Pitt Hopkins Syndrome UK

Ioel Detton Noonan syndrome FR

Tomasz Grybek Fondation Rare diseases PL

Katarzyna Swieczkowska Rare diseases ID PL

Gerritjan Koekkoek Cornelia Delange Syndrome NL

Tanja Zdolsek Kleefstra syndrome, SI

Maria Palia & Georges Papadopoulos Angelman syndrome EL

Iliana Dimitriu Coffin Lowry Syndrome RO

Ammi Sundqvist-Andersson Spina Bifida & Hypdrocephalus SE

Sylvia Roozen Spina Bifida and Hydrocephalus, EU(BE)

Benoit Fourcroy Spina Bifida FR

Bojtor Zsuzsanna, HUFERDIS,HU

Károlyi Judit, HUFERDIS,HU

Szili Danileja, Rett Syndrome, EU

Szabóné Katona Eszter, Disorder of the Corpus Callosum, HU

Baloghné Liplin Tünde Prader-Willi Syndrome, HU

Balogh Zsolt , Prader-Willi Syndrome, HU

Questions /comments

- How do you see your future in our ePAGS group in ITHACA ?
- What do you expect from it ?
- What can you bring to it ?

Dorica Dan

5 years of involvement and evaluation, achievements and challenges

ePAG's chair ITHACA

European
Commission

ITHACA annual meeting 2022

ITHACA 5 years of involvement and evaluation, achievements and challenges

Dorica Dan – ePAG's chair ITHACA

2021, Decembre 08, Budapest

European
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Network
Intellectual Disability
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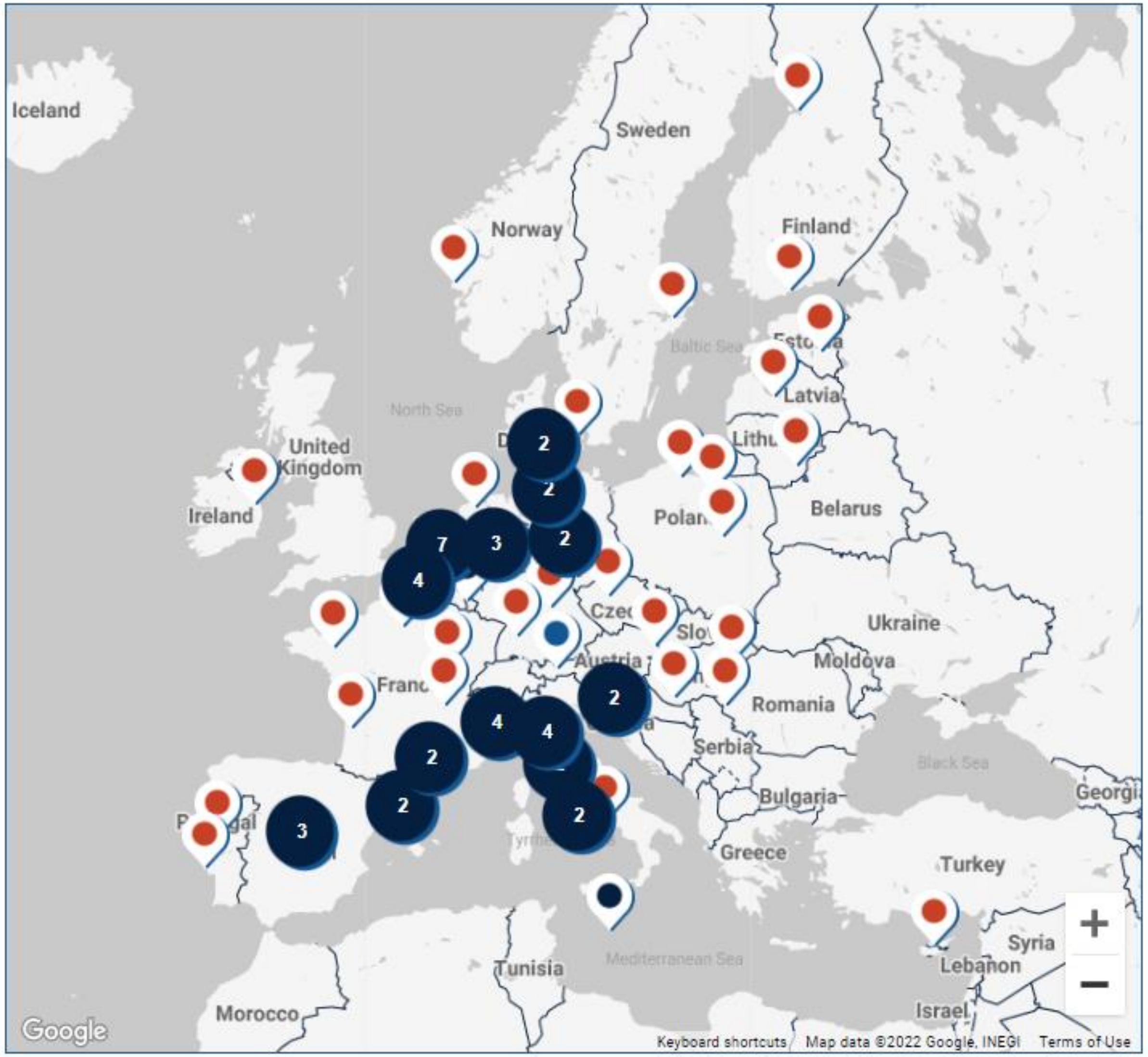
ITHACA "Landscape



- 71 from 23 countries, HCP expert genetic centres in university hospitals
- General Coordination Team of the ERN ITHACA Paris - APHP Hôpital Robert Debré

A big family!

Intellectual disability, telehealth, autism and birth defects

European Reference Networks

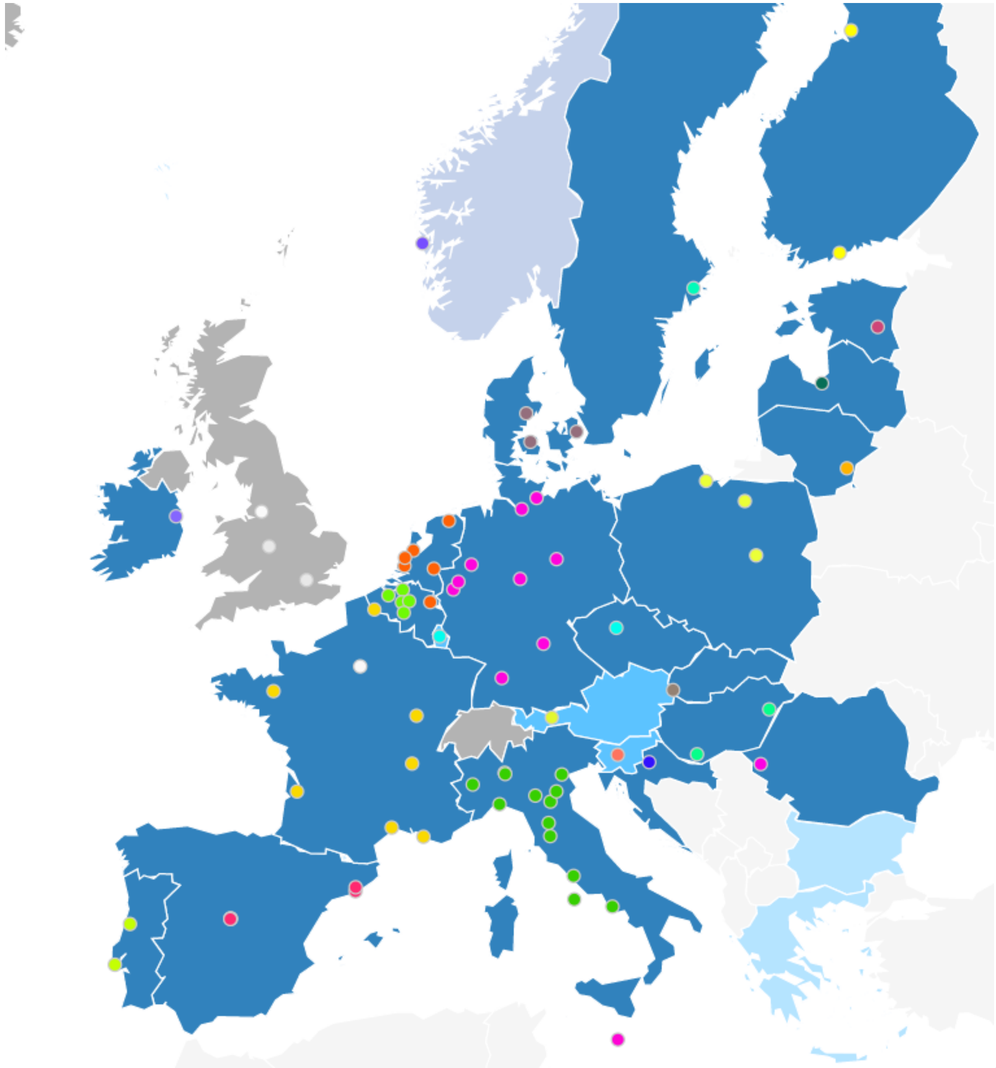
ITHACA

Beginning of ITHACA

- ✓ Organisation and Governance
- ✓ Patients’ engagement in ITHACA (not only in WPs)
- ✓ Team development and knowing each other

Many, diverse & complex needs

- The ERN ITHACA covers more than 5. 000 rare syndromes of genetic origin
  - Empowerment activities and support groups
  - Knowing how to listen to each other
  - Developing close cooperation
  - Ensure that activities include patients
  - families and disease associations as partners



European Reference Networks

ITHACA

INVOLVEMENT OF ePAGs in ITHACA

1. ePAG advocates are involved in all WPs; 2 ePAGs/ each WG;

- *We think that we could be more involved in contacting patients or patient groups for research participation, transfer of best practices into the national health care systems & involving National Alliances on this topic.*
- *ePAG advocates are also involved:*
  - WG NDD Intellectual disability, diagnosis and management
  - Transdiagnosis Guideline WG4 (Transition, Behaviour, Sleep)

2. Started Webinars on transversal themes:

- Integrated Care / **done**
- Transition

3. **Workshops** (ex. Quality of life)

4. **New patient journeys:**

5. **Publications**

https://ern-ithaca.eu/wp-content/uploads/2020/10/ERN-ITHACA-5-Patients-Journey_Corrected-Oct-2020_FINAL.pdf

Who can reach us?

Rare disease associations

Experts, Clinicians
Doctors, all specialists

Patients
With or without diagnosis

National institutions

European and international institutions

Famillies & caregivers

European Reference Networks

ITHACA

GOALS AND ACHIEVEMENTS

- Representing the voice and interests of patients and families
- Ensuring a patient-centred approach
- Identification of needs & priorities
- Provide advice on ethical, social and family support issues
- Support the dissemination and information to the wider patient community
- Participate in working groups
- Work in collaboration with EURORDIS and other partners.

Active working groups:

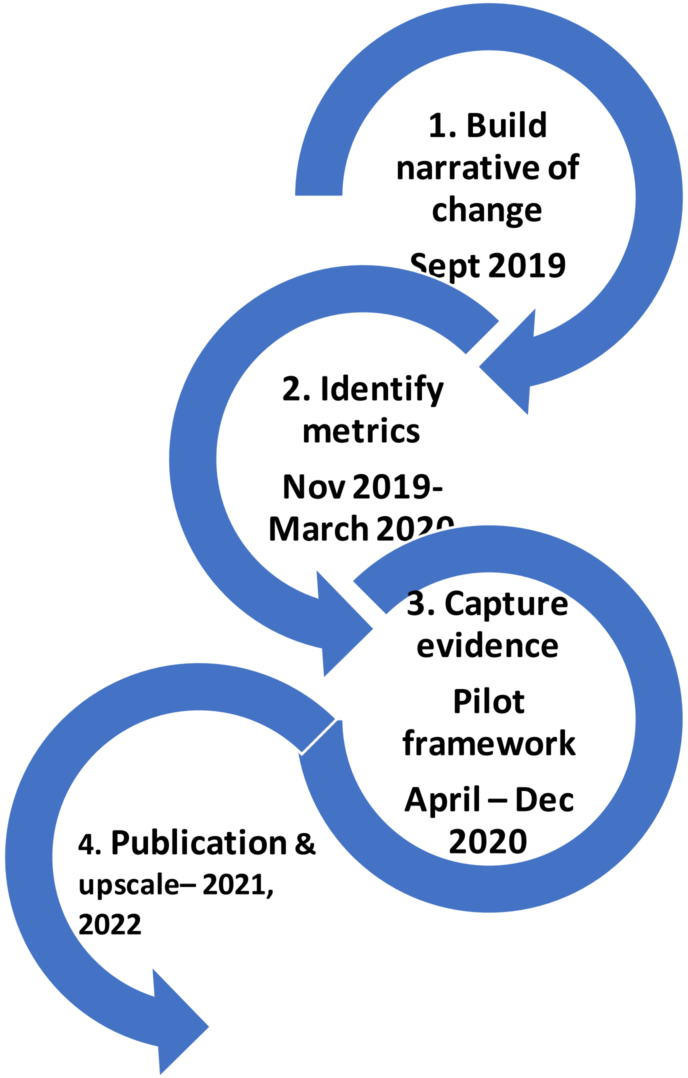
- T&T, NDD, Recommendations
- Achievements

Guidelines, recommendations

- Communication, Resources
- Dedicated seminars
- Training workshops, coaching
- Identification of new needs
- Publications flyers, ePAG ToR ...

Working with other ERNs + other activities

Impact assessment framework: Partnership of Ithaca, ReConnet & RITA;



1. Build narrative of change

Sept 2019

2. Identify metrics

Nov 2019- March 2020

3. Capture evidence

Pilot framework

April – Dec 2020

4. Publication & upscale– 2021, 2022

Our role as being the voice of our community

- Inform on topics related to patient needs, organizing activities and document our needs
- Collaborate with experts to develop a strategic holistic, patient centered approach of our ERN
- Creating and feedback for patient education materials, guidelines, impact assessment of the disease, different ethical issues, from registry to research projects, etc.
- Contributing with patient perspective on each WG activity
- Collaborate and advocate to expand best practices and knowledge at national level and our wider community

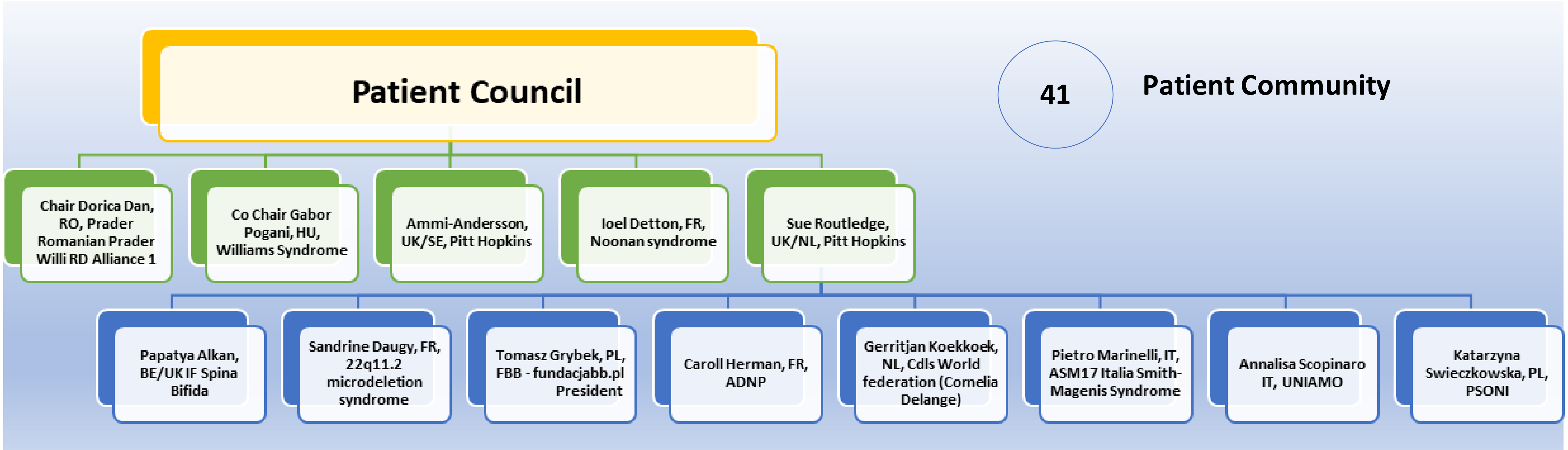
New ePAGs recruited

- ITHACA is covering a wide number of rare diseases and need more ePAGs - New ePAGs recruited !!!

Call for new ePAGs

- European Patient Advocacy Groups are open to rare disease patient organisations (EURORDIS members and non-members) operating in any of these European countries.
- For more information on ERNs and the role of ePAG advocates you may watch this video and read this short guide.
- **Recruitment period: 25th May to 31st July 2022.**

2. Patient Council & Involvement in all workpackages



Patient Council

41 **Patient Community**

Chair Dorica Dan, RO, Prader Romanian Prader Willi RD Alliance 1

Co Chair Gabor Pogani, HU, Williams Syndrome

Ammi-Andersson, UK/SE, Pitt Hopkins

Ioel Detton, FR, Noonan syndrome

Sue Routledge, UK/NL, Pitt Hopkins

Papatya Alkan, BE/UK IF Spina Bifida

Sandrine Daugy, FR, 22q11.2 microdeletion syndrome

Tomasz Grybek, PL, FBB - fundacjabb.pl President

Caroll Herman, FR, ADNP

Gerritjan Koekkoek, NL, Cdls World federation (Cornelia Delange)

Pietro Marinelli, IT, ASM17 Italia Smith-Magenis Syndrome

Annalisa Scopinaro IT, UNIAMO

Katarzyna Swieczkowska, PL, PSONI

13 e-PAG Advocates
13 e-PAG Community
15 Patient Community

1. **1-2 ePAGs are Involved in each workpackage!!!**

3. PC participation in activities - Flyer &welcome package for new ePAGs (including a glossary)

How can I become an ePAG advocate?

Involvement in ePAGs is open to all rare disease patient organisations in Europe.

The recruitment of advocates is ongoing to ensure that patients are fully represented in the governance of each and every ERN.

To apply to become an ePAG advocate in ERN ITHACA please contact:

Project Manager

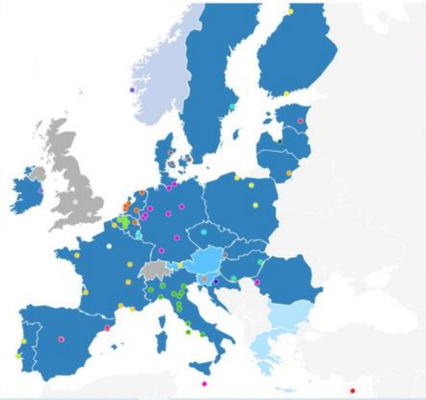
anne.hugon@aphp.fr
coordination@ern-ithaca.eu

Come and join us!

Let's go further together!

Feel free to get some information from the website!

www.ern-ithaca.eu



ITHACA

ITHACA Patient Council

European Patient Advocacy Group

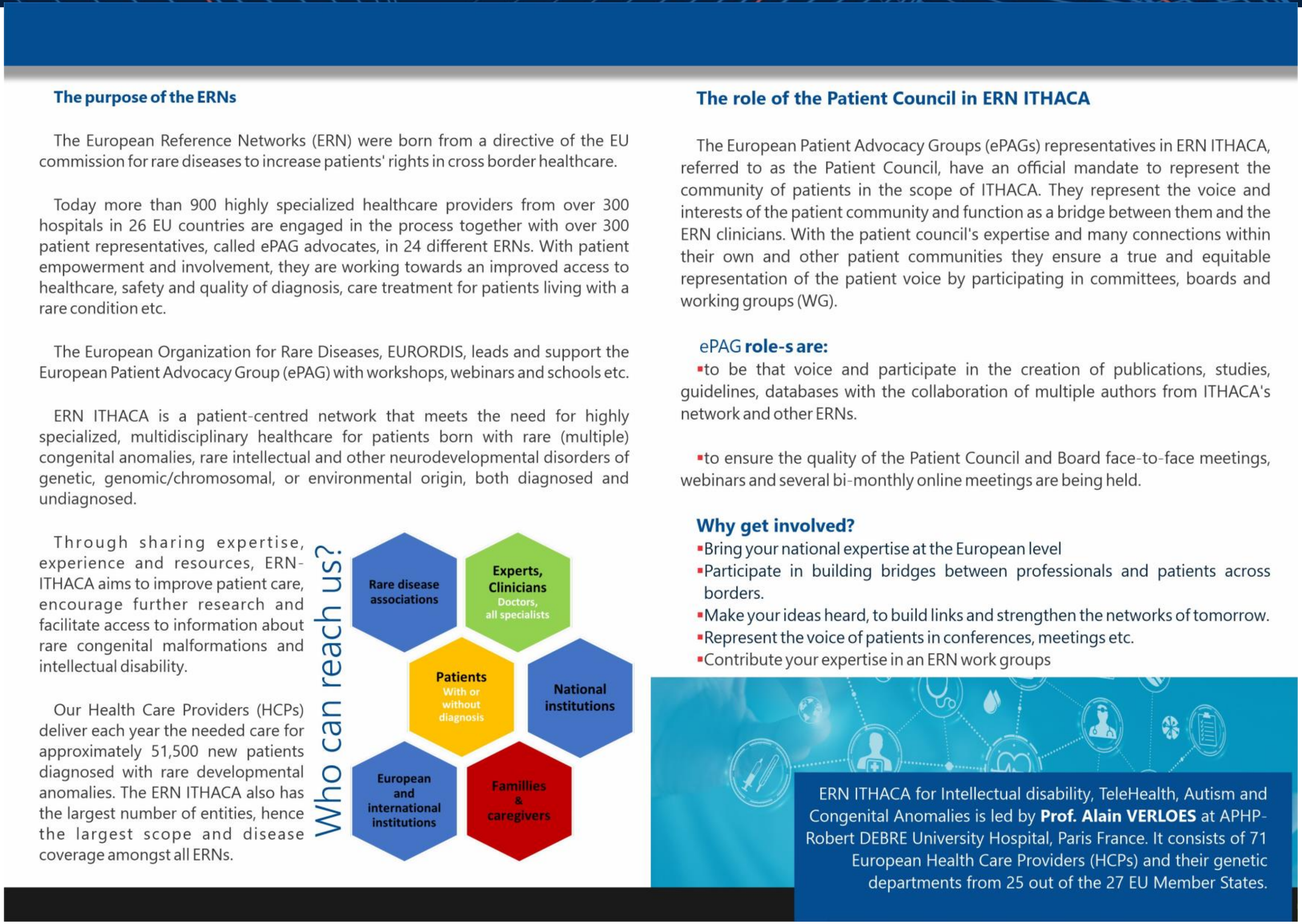
European Reference Network

Co-funded by
the Health Programme
of the European Union

European Reference Networks

ITHACA

Flyer/ welcome pack.



The purpose of the ERNs

The European Reference Networks (ERN) were born from a directive of the EU commission for rare diseases to increase patients' rights in cross border healthcare.

Today more than 900 highly specialized healthcare providers from over 300 hospitals in 26 EU countries are engaged in the process together with over 300 patient representatives, called ePAG advocates, in 24 different ERNs. With patient empowerment and involvement, they are working towards an improved access to healthcare, safety and quality of diagnosis, care treatment for patients living with a rare condition etc.

The European Organization for Rare Diseases, EURORDIS, leads and support the European Patient Advocacy Group (ePAG) with workshops, webinars and schools etc.

ERN ITHACA is a patient-centred network that meets the need for highly specialized, multidisciplinary healthcare for patients born with rare (multiple) congenital anomalies, rare intellectual and other neurodevelopmental disorders of genetic, genomic/chromosomal, or environmental origin, both diagnosed and undiagnosed.

Through sharing expertise, experience and resources, ERN-ITHACA aims to improve patient care, encourage further research and facilitate access to information about rare congenital malformations and intellectual disability.

Our Health Care Providers (HCPs) deliver each year the needed care for approximately 51,500 new patients diagnosed with rare developmental anomalies. The ERN ITHACA also has the largest number of entities, hence the largest scope and disease coverage amongst all ERNs.

Who can reach us?

- Rare disease associations
- Experts, Clinicians – Doctors, all specialists
- Patients – With or without diagnosis
- National institutions
- European and international institutions
- Famillies & caregivers

The role of the Patient Council in ERN ITHACA

The European Patient Advocacy Groups (ePAGs) representatives in ERN ITHACA, referred to as the Patient Council, have an official mandate to represent the community of patients in the scope of ITHACA. They represent the voice and interests of the patient community and function as a bridge between them and the ERN clinicians. With the patient council's expertise and many connections within their own and other patient communities they ensure a true and equitable representation of the patient voice by participating in committees, boards and working groups (WG).

ePAG **role-s are:**

- to be that voice and participate in the creation of publications, studies, guidelines, databases with the collaboration of multiple authors from ITHACA's network and other ERNs.
- to ensure the quality of the Patient Council and Board face-to-face meetings, webinars and several bi-monthly online meetings are being held.

Why get involved?

- Bring your national expertise at the European level
- Participate in building bridges between professionals and patients across borders.
- Make your ideas heard, to build links and strengthen the networks of tomorrow.
- Represent the voice of patients in conferences, meetings etc.
- Contribute your expertise in an ERN work groups

ERN ITHACA for Intellectual disability, TeleHealth, Autism and Congenital Anomalies is led by **Prof. Alain VERLOES** at APHP-Robert DEBRE University Hospital, Paris France. It consists of 71 European Health Care Providers (HCPs) and their genetic departments from 25 out of the 27 EU Member States.

European
Reference
Networks

ITHACA

New Impact Assessment Framework

- The impact assessment framework have been re-launched
- New ERNs are implementing this tool – upscale!!!
- The questionnaire used is the same:
  - *to capture the ePAGs activities in an updated way, the IAF should be a work in progress rather than a fix tool. Relevant activities could be implemented in the next few years, thus new measures/questions should be developed.*

IT IS TIME FOR US TO TAKE THE TEMPERATURE OF PATIENT REPRESENTATIVE INVOLVEMENT

European Reference Networks

ITHACA

WEBINAR T&T - ERN ITHACA Webinar#1 2022

New Proposal : PC Webinars on Tuesday June 21st 17h00 -18h30:

How and why get involve in ITHACA has an EPAG

Welcome and Introduction, and get to know each other

- Pr Laurence Faivre (Dijon Hospital, FR)
- Mrs Dorica Dan (Chair Patient Council, RO)
- *Round table short presentation “Short pitch 1mn”*

- **Patient engagement & Patient Advocacy**
  - Eurordis a“win win” collaboration, Mrs Dorica Dan (Chair Patient Council, RO)
  - Patient orientated action within ITHACA, and resources to rely on, Anne Hugon (APHP Paris, FR
  - Teaching & Education a Work Group experience, Pr Laurence Faivre (Dijon Hospital, FR)
- **Discussion time**
- **How to improve my participation**
  - An ePAG, shared history and experience, M. Ioel Detton (Patient Board Member, FR)
  - How I can be involve, Ammi Andersson (Patient Board Member,SE)
  - What should I spread at my level to inform our RD Community in EU, Gabor Pogany (CoChair Patient Council, HU)
- **Discussion time**
- **Conclusion with speakers and moderator**
  - Mrs Dorica Dan (Chair Patient Council, RO)
  - Pr Laurence Faivre (Dijon Hospital, FR)

Register and Connect !!!!

- deadline registration 13 June!!!

Conferences

2 ESHG and ECRD Posters, accepted at both events + other posters

- ***Abstract Title:*** *Patient Journey Common Needs: Rare congenital malformations syndrome with intellectual and other neurodevelopmental disorders affect one child in a million*

Control Number: 1383

Topic: 23. Genetic Counselling / Services / Education

- ***Abstract Title****: ePAGs' (European Patient Advocacy Groups) role in the European ReferenceNetwork ITHACA on Intellectual disability, TeleHealth, Autism and Congenital Anomalies: How patients’ voices improve care*

Control Number: 1283

Topic: 24. Ethical, Legal and Psychosocial Aspects in Genetics

Eurordis/ITHACA : Capacity building

Coaching sessions to ePAG advocate and clinical leads

- to improve collaboration and teamwork.

- Min 12 and max of 20 participants:

General learning objectives:

- *To develop a common understanding of the role of patient representatives in the ERNs*
- *To develop a shared vision of how to organize patient / clinician collaboration more effectively*
- *To develop soft skills to strengthen team work and shared leadership*

- Specific Ithaca Project / collective impact create Team / common objectives
- Online session in November / 20 partcipants ePAGS + experts
- Budapest meeting - feedback on lessons learns and share the audience at the Board on day 9

European
Reference
Networks

ITHACA

End Agenda 2022 Patients' Council

- **TeamBuilding** session - 16 - 23 - 30 Nov 2022 (ITHACA-EURORDIS)
  - Develop a common understanding - a shared vision of how to organise collaboration between patients and clinicians more effectively.
- **Budapest Satellite Workshop** full day on 8 December from 9am -ITHACA BOARD
  - Collaborative session with 3 workshops with medical teams:
  - WG Expert Consensus Recommendation; WG Teaching and Training; WG Neurodevelopment
- **20-21 April 2023 NDD2023**, Amsterdam, NL "First European Congress on Interdisciplinary Perspectives on Developmental Disabilities".

EX : dedicated webinars

1. How and why to get involved
2. How to optimise patient input into the guidelines
3. Newborn screening and Screen4Care
4. Understanding registers and their relevance to classical studies
5. Organisation of care in the EU

ERN-ITHACA Webinar #4 ePAGS

Published on May 30, 2022

Tuesday June 21st -2022 from 17h -18h30

ERN-ITHACA Webinar #4: “How and why getting involved in ITHACA has an EPAG”

Program

Welcome and Introduction, and get to know each other

- Pr Laurence Faivre (Dijon Hospital, FR)
- Mrs Dorica Dan (Chair Patient Council, RO)
- Round table short presentation "Short pitch 1mn"

Patient engagement & Patient Advocacy

- Eurordis a "win win" collaboration, Mrs Dorica Dan (Chair Patient Council, RO)
- Patient orientated action within ITHACA, and resources to rely on, Anne Hugon (APHP Paris,


Thank You!

Anne Hugon

Activity report & Work Group, What can I do

PM coordination Ithaca

European
Commission

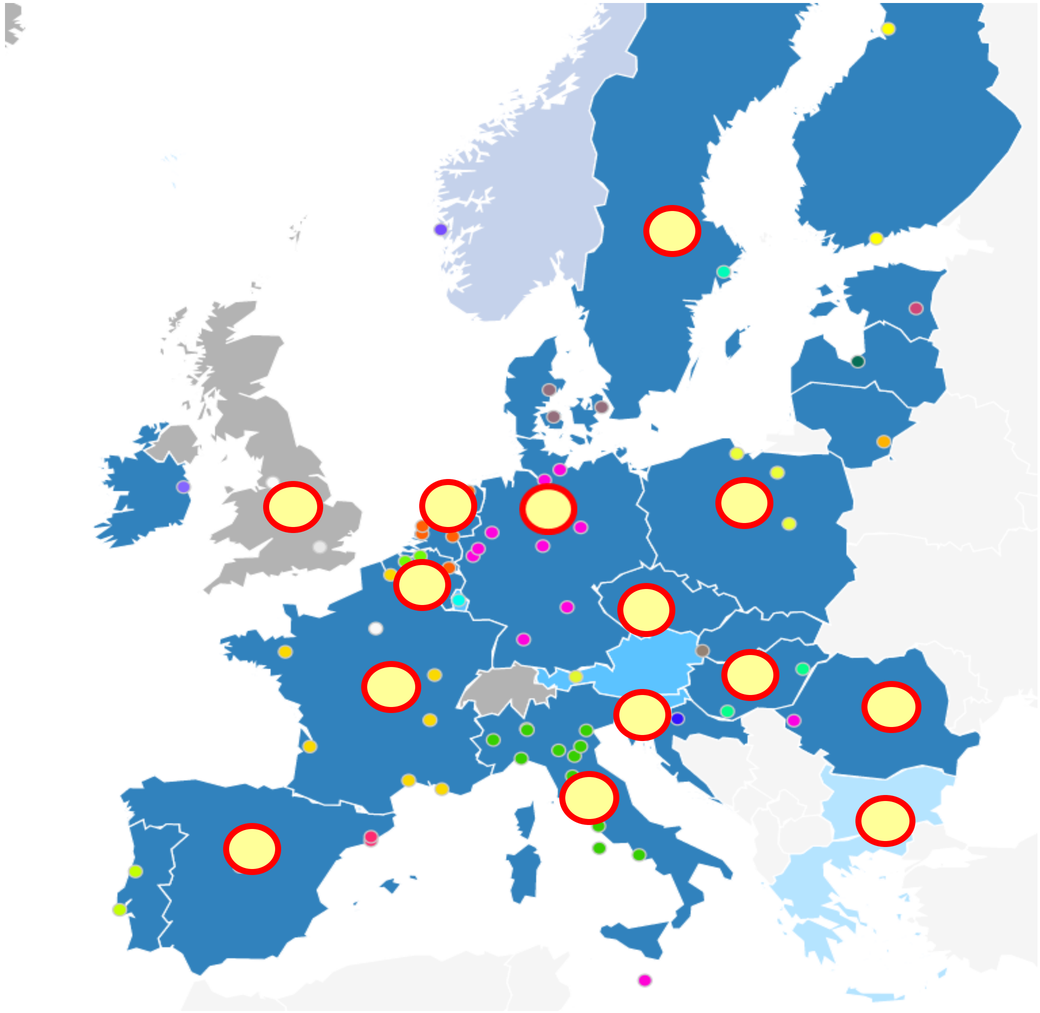
Importance of integrating the patient, the patient, the Associations Built a Collaborative participation and expertise What vision for associations and patients within the ERN ITHACA

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Intellectual Disability
and Congenital
Malformations (ERN ITHACA)

Multiple & growing patient community

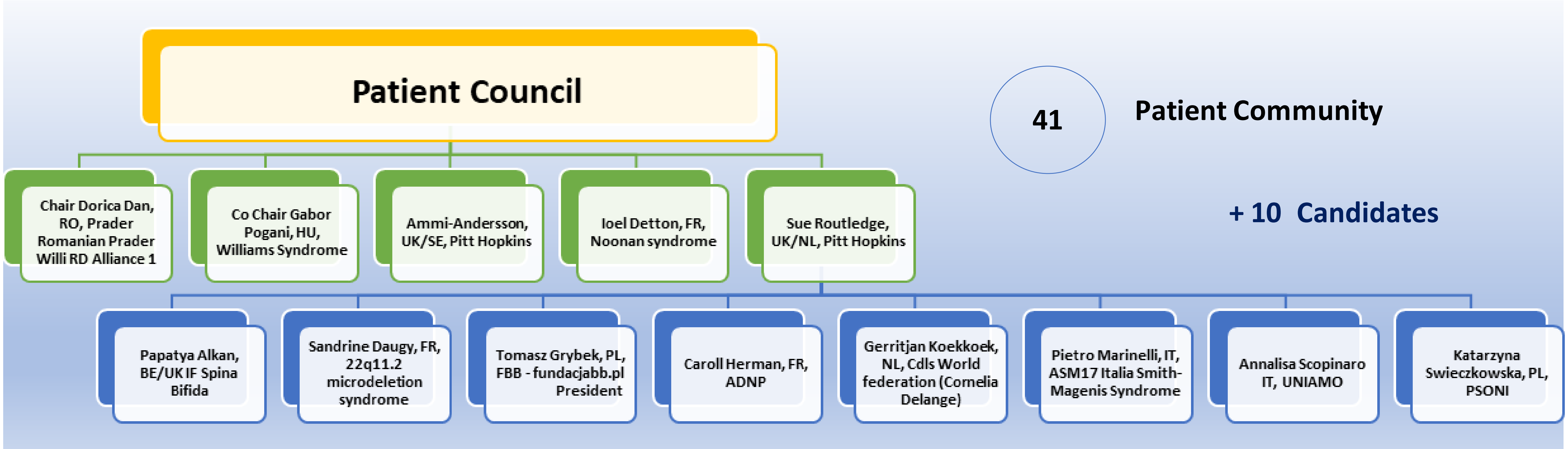
- 22q11.2 syndrome
- ADNP
- Angelman's syndrome
- Chromosomal disorders
- Chromosomal abnormalities
- Costello and cardio-facio-cutaneous syndrome
- Cdls Cornelia Delange
- Goldenhar syndrome
- Noonan syndrome
- PACS1 syndrome
- Pitt Hopkins syndrome
- Prader Willi syndrome
- ID rare diseases
- Rett syndrome
- SAT2B syndrome
- Smith-Magenis syndrome
- Spina Bifida
- Williams syndrome
- White-Sutton syndrome
- RD Borys the Hero Foundation
- Kleefstra syndrome
- MED13L syndrome
- Mowat-Wilson syndrome
- Beckwith-Wiedemann syndrome
- X Fragile
- FraX syndrome
- Dravet syndrome
- RD Borys the Hero Foundation
- RD Alliance IT,ES,CZ,NL,FR,RO
- Etc ……



European Reference Networks

ITHACA

Patient Council & Involvement in all workpackages



Patient Council

Chair Dorica Dan, RO, Prader Romanian Prader Willi RD Alliance 1

Co Chair Gabor Pogani, HU, Williams Syndrome

Ammi-Andersson, UK/SE, Pitt Hopkins

Ioel Detton, FR, Noonan syndrome

Sue Routledge, UK/NL, Pitt Hopkins

Papatya Alkan, BE/UK IF Spina Bifida

Sandrine Daugy, FR, 22q11.2 microdeletion syndrome

Tomasz Grybek, PL, FBB - fundacjabb.pl President

Caroll Herman, FR, ADNP

Gerritjan Koekkoek, NL, Cdls World federation (Comelia Delange)

Pietro Marinelli, IT, ASM17 Italia Smith-Magenis Syndrome

Annalisa Scopinaro IT, UNIAMO

Katarzyna Swieczkowska, PL, PSONI

41 **Patient Community**

+ 10 Candidates

Work as a TEAM / ITHACA is covering a wide number of rare diseases and need more ePAGs to cover all our WG and projects

- 2 ePAGs Involved in each workpackage
- 10 new ePags candidates in 2022

Patient Council - building together

- Representing the voice and interests of patients and families
- Ensuring a patient-centred approach
- Identification of priorities, needs, projects for ITHACA
- Provide advice on ethical, social and family support issues
- Support the network in disseminating its activities and information to the wider patient community
- Participate in working groups
- Work in collaboration with EURORDIS and other partners.

PAB - A Very active working groups 2022 !

Work in constant Progress !!

- Patient Council met 5 times : 31/01; 28/03; 30/05; 26/09; 17/10
  - Budapest Workshop + 2 Days Board
  - Team building UBUNTU 3 Sessions of coaching
  - 2 Dedicated Webinars
  - WG T&T meeting
- Patient Board (5 members) : 21/02; 19/04; 16/05; 09/07; 10/11
  - + Many mails and TC with The chairs
  - Chair/ Co Chairs participation to 4 Steering Committee (EXCOM) and 2 Board of Directors (BOARD)
  - Elections
- A large scale of activitie and back office work
  - Production of various publications and administrative documents
  - 4 public présentations (2 French Networks, University of RD Alliances, French Foudation, RD PO)
  - Participation in EURODIS, EU Program, EUPATIE and RD Alliances meetings
  - Recruitment Process ePAGS // Eurordis goals 50 PO more EU
  - Reception of PO and Presidents
  - Preparation of meetings and minutes and all activities
  - Writing of Posters for congresses (ESHG, RD Foundation, ASHG)
  - Referal of families for care

WG Com, T&T, NDD, Reco, SBoD…

Achievements

- Guidelines,
- Web Site Patient
- Doc Resources
- Dedicated seminars
- Training workshops, coaching
- Publications flyers, ePAG ToR ...
- SC EuroNDD 2023

Full of new Projects +++

- New Goals at EU level + Natiaonal level Patient Advocate ePAGS
- NDD 2023
- Mapping center of expertise and diseases group list
- New guidelines
- New Communication tool, ToR, Welcom Patient guide
- Patient Assessemment
- Patients Journey
- Eurordis Topics and EU Project
- WG ++ Involvment ; Guidelines, Webinar Registry …
- ….etc ***UP to YOU !***

ITHACA WG Leaders, HCP needs collaboration +++

ERN ITHACA - Working Groups

- 3 transversal WGs dedicated to general coordination
  - (WP1 Coordination, WP2 Cover-up, WP3 Evaluation)
- 12 thematic working groups
- 9 specific scientific working groups
- 2 thematic areas
  - Multiple congenital anomalies
  - Neurodevelopment
- 2 advisory boards
  - Patient Council and Advisory Committee
  - Ethical and Legal Advisory Board
- 1 special working group, the APOGeE project
- + Related research projects with EU partners

Working groups 1/2

ERN ITHACA Coordination WP1 Coordination, WP2 Dissemination, WP3 Evaluation / coordination Team

Expert recommendations Guidelines

- Improve patient care using high quality guidelines by evaluating existing national guidelines and publishing a European consensus **ePAGS Participation**

Education and training

- Set up specific teaching and training programmes targeting different populations - future ITHACA specialists and non-specialists on rare ID, congenital malformations **ePAGS Participation**

Neurodevelopmental disorders / 4 SNW

- Deal with rare genetic IDs, improve our knowledge. SySNDD ; 3 SNW on : Mild ID and Adult aging ; PIMD Polyhandicap ; Psychiatric aspects to built Recommandations or expert consensus **ePAGS Participation**

SBoD - SPINA BIFIDA other Dysraphysms / a transnational WG (ITHACA EUROGEN)

- Produce an expert consensus Harmonisation of the clinical practice across the EU and improve patient care – Fetal Surgery – Orpah code classification on Dyraphism **ePAGS Participation**

Patient Council (PC)

Ethical Legal and Social Issues (ELSI)

Working groups 2/2

Research

- Improve collaborative research covered diseases, understanding disease mechanisms Producing of collaborative research studies on a series of patients with unsolved disorders

CPMS

- Clinical Patient Management System (CPMS), an innovative software that allows virtual discussions over challenging clinical cases

Digital activity

- RD syndromes throughout the EU through the innovative use of technology

ILIAD Registry

- set up an interoperable registry dedicated to rare diseases within the scope of ITHACA called ILIAD Rare Diseases patient registry: an International Library of Intellectual disability and Anomalies of Development .

Fetal Medecine

- focuses on generating and sharing knowledge on foetal medicine

Patient Council (PC)

Ethical Legal and Social Issues (ELSI)

Forename	Family Name	PC Board Elected 2021/22	WP 1/2/3 EXCOM	WG4 Expert Consensus	WG5 Digital health (CPMS+IT)	WP6 ILIAD Registry	WG7 Research Innovation	WG8 T&T	WG9 NDD	WG10 Spina Bifida	WG11 Fetal medecine	WG12 APOGEE ?	Ethical Legal AD
Dorica	Dan	**Chair**	1		1			1	1				
Gábor	Pogány	**Co Chair**	1		**1**		1						
Sue	Routledge	**Elected**		1									
Ammi	Sundqvist-Andersson	**Elected**		1		1		1		1?			
Ioel	Detton	**Elected**						1					
Tomasz	Grybek			1	1			1					
Carole	Herman					1			1				
Katarzyna	Swieczkowska			1				1					
Papatya	Alkan									1			
Pietro	Marinelli						1?						
Annalisa	Scopinaro				1?	1	1						
Sandrine	Daugy												
Gerritjan	Koekkoek												
Erika	Stariha												
Nora	Leonardi												
Sylvia	Roozen									1	1?		
Inés	Fernández-Ulibarri			1									
Tanja	Zdolsek												
Maria Palia (Staff)	Georges Papadopoulos (Parent)												
Alejandro	Doval												
Ana	González Hernández												
Sandra	López Cabeza												
Iliana	Dimitriu												
Vesna	Vujičić												
Benoit	Fourcroy									1			

Work group, what should I know

- *where I belong*
- *where I can contribute my knowledge*
- *where am I best to share my experience*

Rita Francisco

EURORDIS in supporting Patient Partnership : A spotlight on ERN ITHACA,

Rita Francisco, EURORDIS

- European Patient Advocacy Groups (ePAG) in numbers
- A spotlight on ITHACA ePAG
- Team support patient partnership
- New patient involvement framework in ERN governance
- New application process + Resources for newcomers
- Empowerment and training (guides) (new EURODIS website)
- ePAG horizontal Working Group: A spotlight on the Patient Partnership Working Group

EURORDIS' ROLE IN SUPPORTING PATIENT PARTNERSHIP IN THE ERNS: A SPOTLIGHT ON ERN ITHACA

8th December 2022

Rita Francisco
Patient Engagement Junior Manager – ERN & Healthcare

EURORDIS.ORG

What will we be covering today?

Time	Topic	Who
20 min	**1.What are Patient Partnerships?**	Rita
	•Mandate and rationale for patient partnership in the ERNs	
	2. How is EURODIS supporting patient partnership in the ERN?	
	•European Patient Advocacy Groups (ePAG) in numbers	
	• Mapping and filling patient representation gaps in the ERNs	
	• A spotlight on ITHACA ePAG	
	•EURORDIS Team support patient partnership	
	•New patient involvement framework in ERN governance	
	•New application process + Resources for newcomers	
	•Empowerment and training (guides) (new EURODIS website)	
	•ePAG horizontal Working Group: A spotlight on the Patient Partnership Working Group	
10 min	•Team-building training: **ITHACA's experience**	Ammi
30 min	• Focus group with Team Building participants	Rita (moderator)

What are Patient Partnerships?

Partnership?

Engagement?

Involvement?

"**Patient partnership** can be defined as a mutual relationship between all stakeholders, including patients, where input from people living with a rare disease or their carers **routinely** and **formally** informs policy reflections and decisions. Patient partnership implies going beyond empowerment and engagement but considering people living with a rare disease and their advocates as **equal partners** and actors in policy and programme design and evaluation."

Source: Rare2030 Recommendations

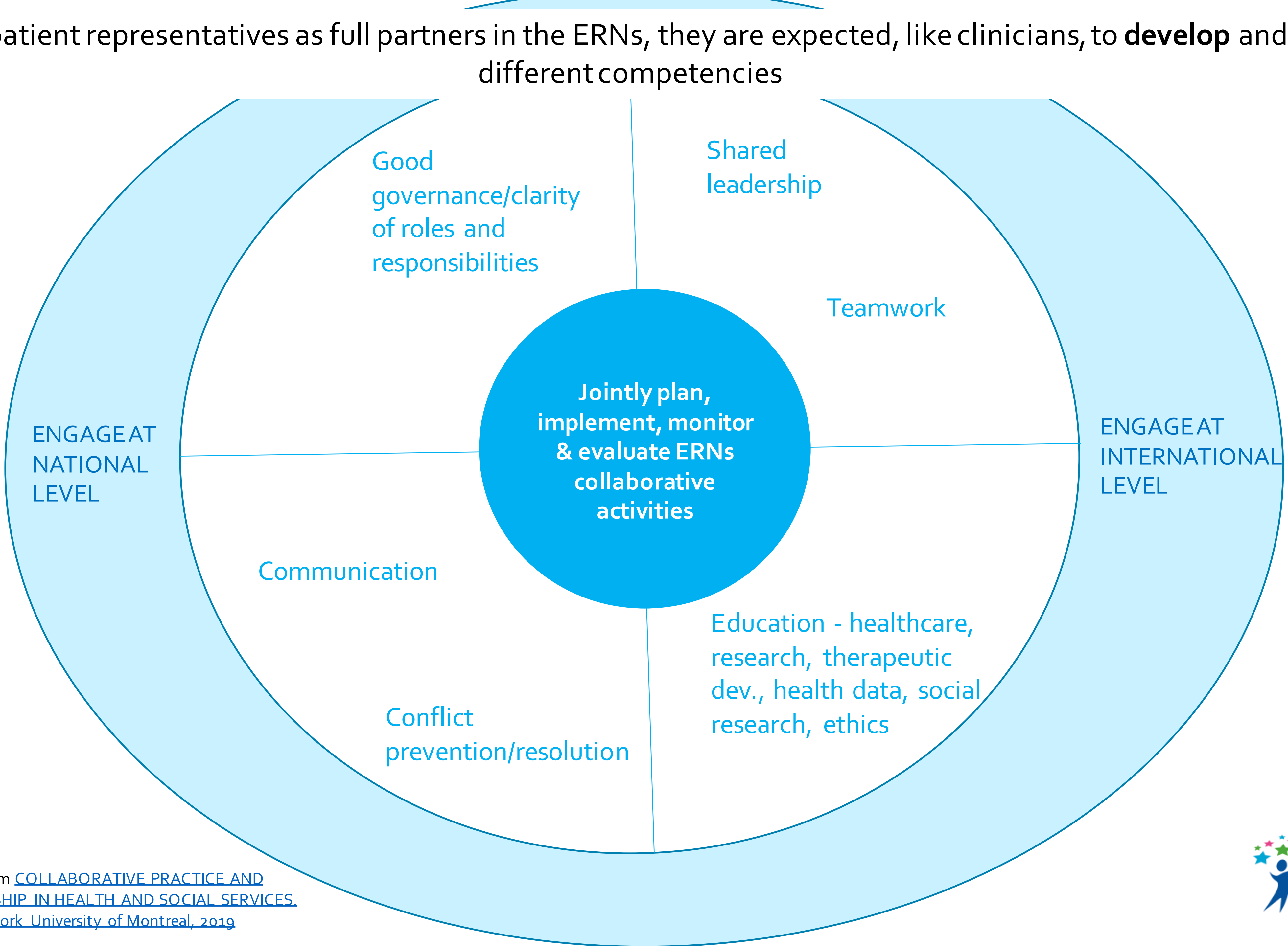
Mandate for Patient Partnership in the ERNs

The mandate to implement patient partnership in the ERNs came from the EU Committee of Experts on RD.

"Patients and patient representatives **should play an integral role in the decision-making process of the ERNs** and be involved in the structural and clinical network activities to meet the legal requirements of the EU Delegated Acts."

Source: Addendum to the EUCERD Recommendations on RD, 2015

To include patient representatives as full partners in the ERNs, they are expected, like clinicians, to **develop** and **maintain** different competencies



Good governance/clarity of roles and responsibilities

Shared leadership

Teamwork

ENGAGE AT NATIONAL LEVEL

Jointly plan, implement, monitor & evaluate ERNs collaborative activities

ENGAGE AT INTERNATIONAL LEVEL

Communication

Education - healthcare, research, therapeutic dev., health data, social research, ethics

Conflict prevention/resolution

Source: Adapted from [COLLABORATIVE PRACTICE AND PATIENT PARTNERSHIP IN HEALTH AND SOCIAL SERVICES. Competency framework University of Montreal, 2019]

How is EURODIS supporting patient partnership in the ERN?

EURORDIS supports and provides patient representatives involved in the ERNs with the information, knowledge and skills that they need **to engage and partner effectively with clinicians** in the Networks' collaborative activities.

EURORDIS potentiate **relationship-building and co-decision-making** between ERN patient representatives and other ERN stakeholders, including ERN Managers and clinicians.

How is EURODIS supporting patient partnership in the ERN?

European Patient Advocacy Groups (ePAG) in numbers

24 European Patient Advocacy Groups (ePAG)

+300 ePAG advocates (volunteers), endorsed by their respective Patient Organisations from 28 European countries working in partnership with clinicians

EURORDIS
RARE DISEASES EUROPE

Mapping and filling patient representation gaps in the ERNs

Recruitment of new ePAG advocates

Mapping and filling patient representation gaps in the ERNs

ERN BOND
ERN CRANIO
Endo-ERN
Neurological Diseases (ERN-RND)
EpiCARE
ERKNet for patients
ERN-EYE
EURACAN
EuroBloodNet
ERN-LUNG
ERN GUARD-HEART
SKIN
eUROGEN Urogenital Diseases
EURO-NMD
MetabERN
rita
ERN RARE-LIVER
TransplantChild
VASCERN
Connective Tissue and Musculoskeletal Diseases (ERN ReCONNET)
ITHACA

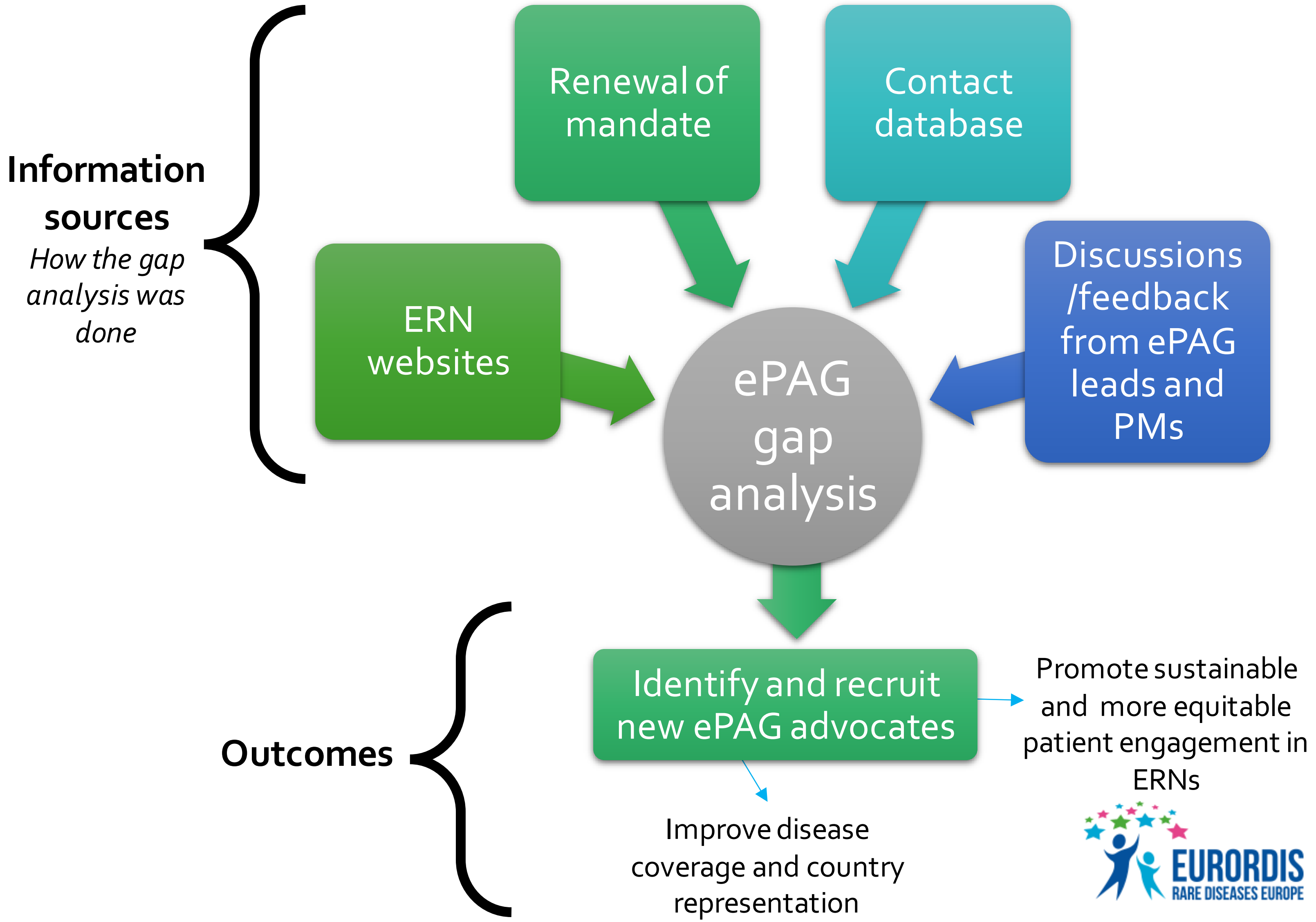
EURORDIS RARE DISEASES EUROPE

Mapping the patient representation gaps
Methodology

Phase 1

Aim: To identify country and/or disease gaps (underrepresented) in the ERNs.

- ERN-specific and global gap analysis were both conducted



Information sources
How the gap analysis was done

Renewal of mandate

Contact database

ERN websites

Discussions /feedback from ePAG leads and PMs

ePAG gap analysis

Outcomes

Identify and recruit new ePAG advocates

Promote sustainable and more equitable patient engagement in ERNs

Improve disease coverage and country representation

Mapping the patient representation gaps

Country representation

48 European countries were considered in our analysis
(EURORDIS reference country list adapted from Council of Europe)

28 (58%) European countries have ePAG representatives

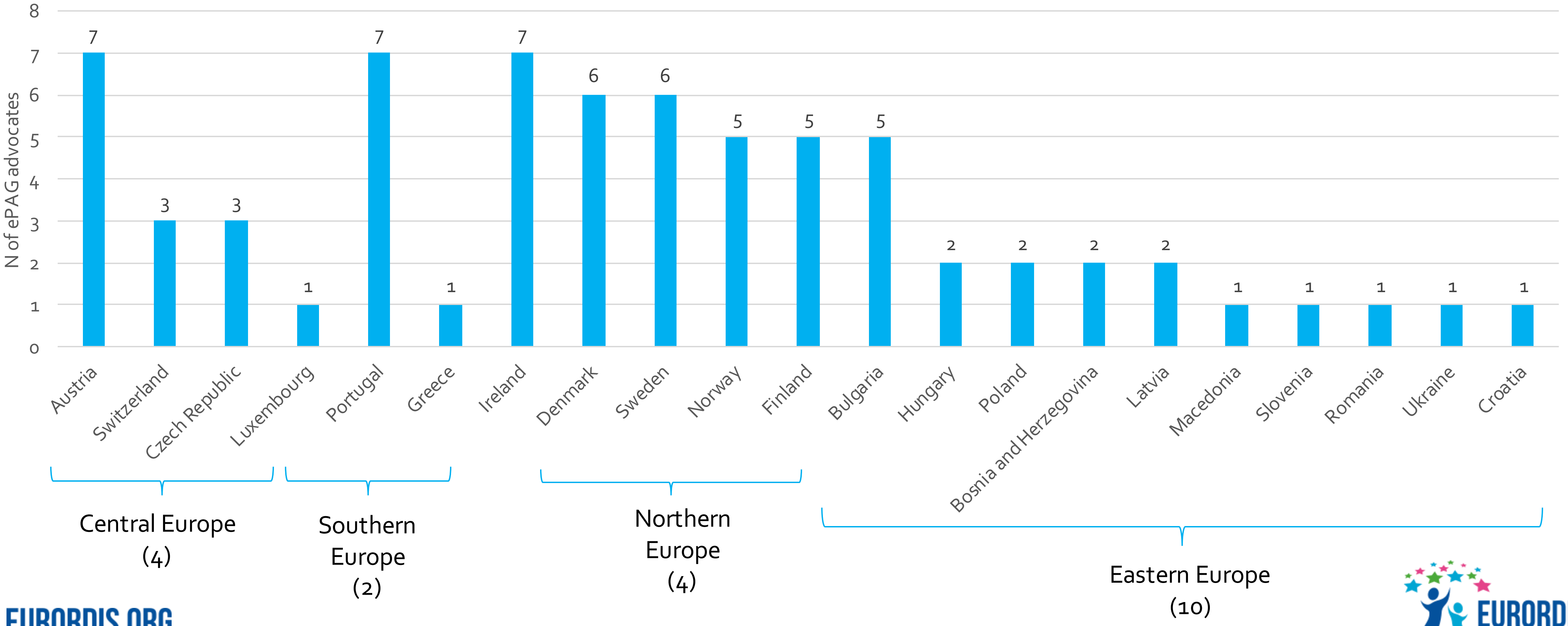
The aim is not to recruit ePAG advocate from all **48** countries all but this was our basis to look at the gaps across the 24 ePAGs.

EURORDIS
RARE DISEASES EUROPE

Mapping the patient representation gaps

Country representation

Among the **28** (58%) European countries with ePAG representatives, the ones with 7 or less are:



Mapping the patient representation gaps

Country representation

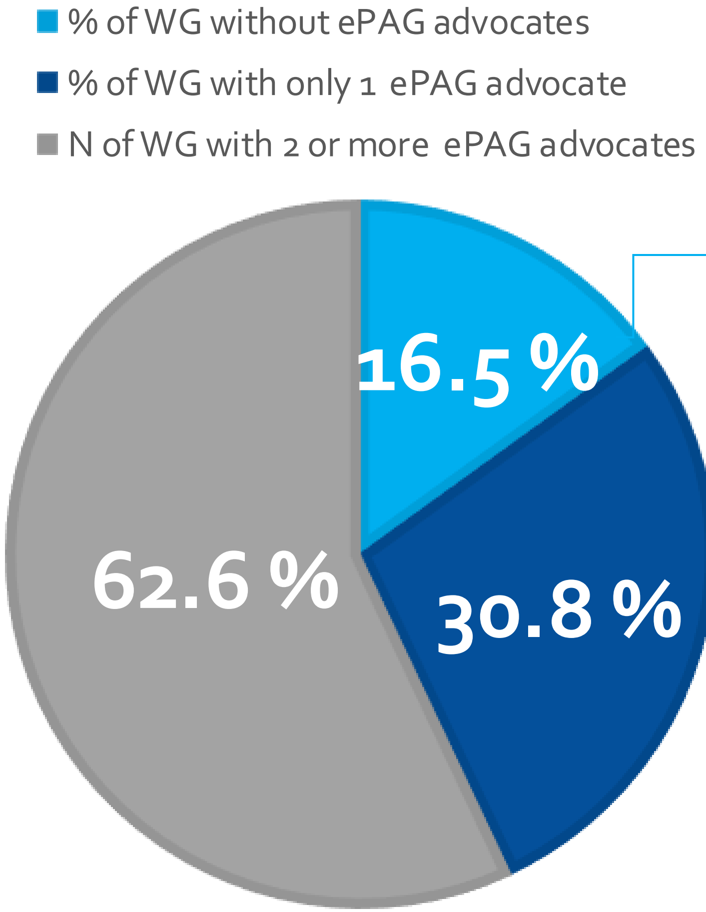
48 European countries were considered in our analysis
(EURORDIS reference country list adapted from Council of Europe)

20 (42%) European countries do not have ePAG representatives

Eastern Europe	Southern Europe	Central Europe	Northern Europe	Western Europe	Other
Albania Armenia Belarus Estonia Georgia Lithuania Montenegro Republic of Moldova Russian Federation Serbia Slovakia	Andorra Cyprus Malta San Marino	Lichtenstein	Iceland	Monaco	Turkey

Mapping the patient representation gaps

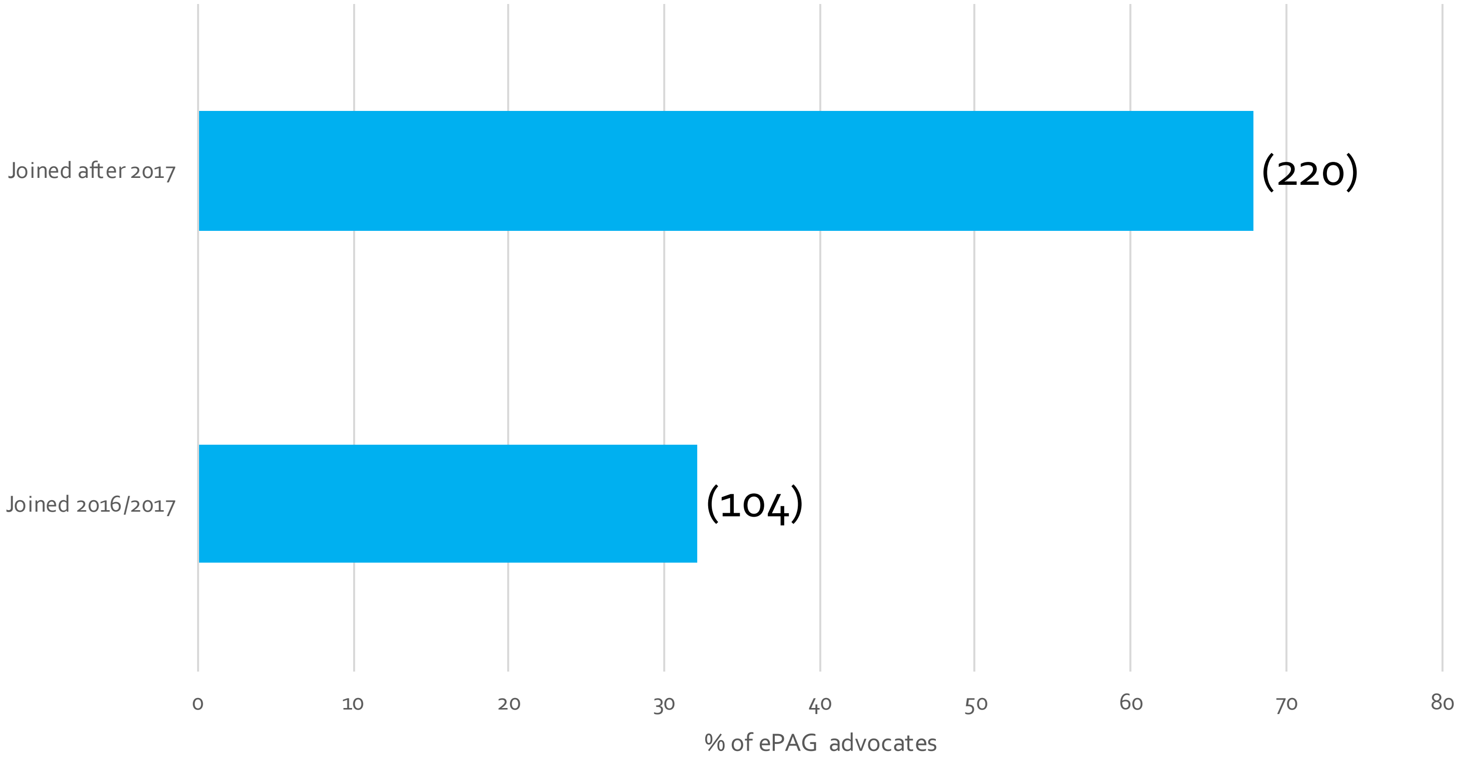
Disease representation



- Chronic Lung Allograft Disfunction (CLAD), Non-CF Bronchiectasis (NCFB) and Primary Cilary Dyskinesia (PCD) (ERN LUNG)
- Frontotemporal dementia (ERN RND)
- Ichthyosis & Palmoplantar Keratoderma, Mendelian Causes of Connective Tissue Disorders and ToxiTEN - Severe cutaneous drug reactions (ERN SKIN)
- Gastroenterological diseases, Intestinal failure and Abdominal wall defects (ERNICA)
- Anterior segment rare eye diseases (ERN EYE)
- Special Electrophysiology Conditions (GUARD HEART)
- Pediatric Rheumatic diseases (PR) (RITA)
- Rare cancer of the male genital organs and of the urinary tract (EURACAN)
- Rare bleeding-coagulation disorders and related diseases and Hemochromatosis and other rare genetic disorders of iron metabolism and heme synthesis (EuroBloodNet)

*ITHACA, EpiCARE, PAEDCAB and ERN BOND do not have disease WG, hence they were not included in this analysis

Mapping the patient representation gaps
Succession planning



Mapping the patient representation gaps
Recruitment numbers (so far)

No of expression of received interest	No of ERNs included	No of countries represented	No of diseases (groups) covered	Have any candidates been rejected or decided not to proceed with a full application?	Have any candidates been officially included in any of the ePAGs?
83	21	24 • 9 from Eastern Europe • 2 from Nordic countries • 4 Southern Europe, and • 9 from Western/central Europe	54	Yes (8).	Yes (14)

Mapping and filling patient representation gaps in the ERNs

A spotlight on ITHACA ePAG

Mapping the patient representation gaps

ITHACA's Recruitment numbers (so far)

ERN ITHACA ePAG numbers in March 2022

- 12 ePAG advocates
- + 10 diseases
- 9 European Countries

Current numbers ERN ITHACA ePAG

- 28 ePAG advocates
- + 21 diseases
- 15 European Countries

No of expression of received interest	No of countries represented	No of diseases (groups) covered	Have any candidates been rejected or decided not to proceed with a full application?	Have any candidates been officially included in any of the ePAGs?
11	9 • 4 from Eastern Europe • 2 Southern Europe, and • 3 from Western/central Europe	10	?	Yes (8)

13 % of all expressions of interest

There have been 7 additional applications

Mapping the patient representation gaps

ITHACA's patient representation

- How do you see the group growing?

- What are your priorities now, in terms of patient representation? Have they changed?

In terms of countries:

- Tandem patient-clinician representation:

1 PO per each country where ITHACA HAS HCPs

In terms of disease coverage:

- 5000 genetic syndromes
- List of most common syndromes and ensure there is Patient representation for all of them

EURORDIS support

MEET THE EURORDIS TEAM

Inés Hernando
ERN & Healthcare Director
ines.hernando@eurordis.org

European
Patient
Advocacy
Group

Matt Johnson
ERN & Healthcare Advisor
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Lenja Wiehe
Patient Engagement Senior Manager,
ERN and Healthcare
lenja.wiehe@eurordis.org

Ariane Weiman
Public Affairs Senior Manager,
PaedCan, EURACAN,
EuroBloodNet, GENTURIS
ariane.weiman@eurordis.org

Rita Francisco
Patient Engagement Junior
Manager, ERN and Healthcare
rita.francisco@eurordis.org

The EURORDIS team is much bigger. There are other colleagues, you might end up collaborating with. You can visit our **Staff webpage** HERE to get to know more about us.

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RARE DISEASES EUROPE

HOW IS EURORDIS SUPPORTING THE ADVOCATES?

1. Supporting the overall governance of ePAGs and recruitment of new advocates and help identify new candidates.
2. Developing tools and processes to help ePAG advocates fulfil their mission and role in the European Reference Networks.
3. Facilitating peer learning and the involvement of advocates in different areas of work: clinical practice guidelines, clinical decision support tools, research, evaluation, training and communication.
4. Promoting and facilitating relationship-building and co-decision-making between ERN patient representatives and other ERN stakeholders, including ERN Managers and clinicians.

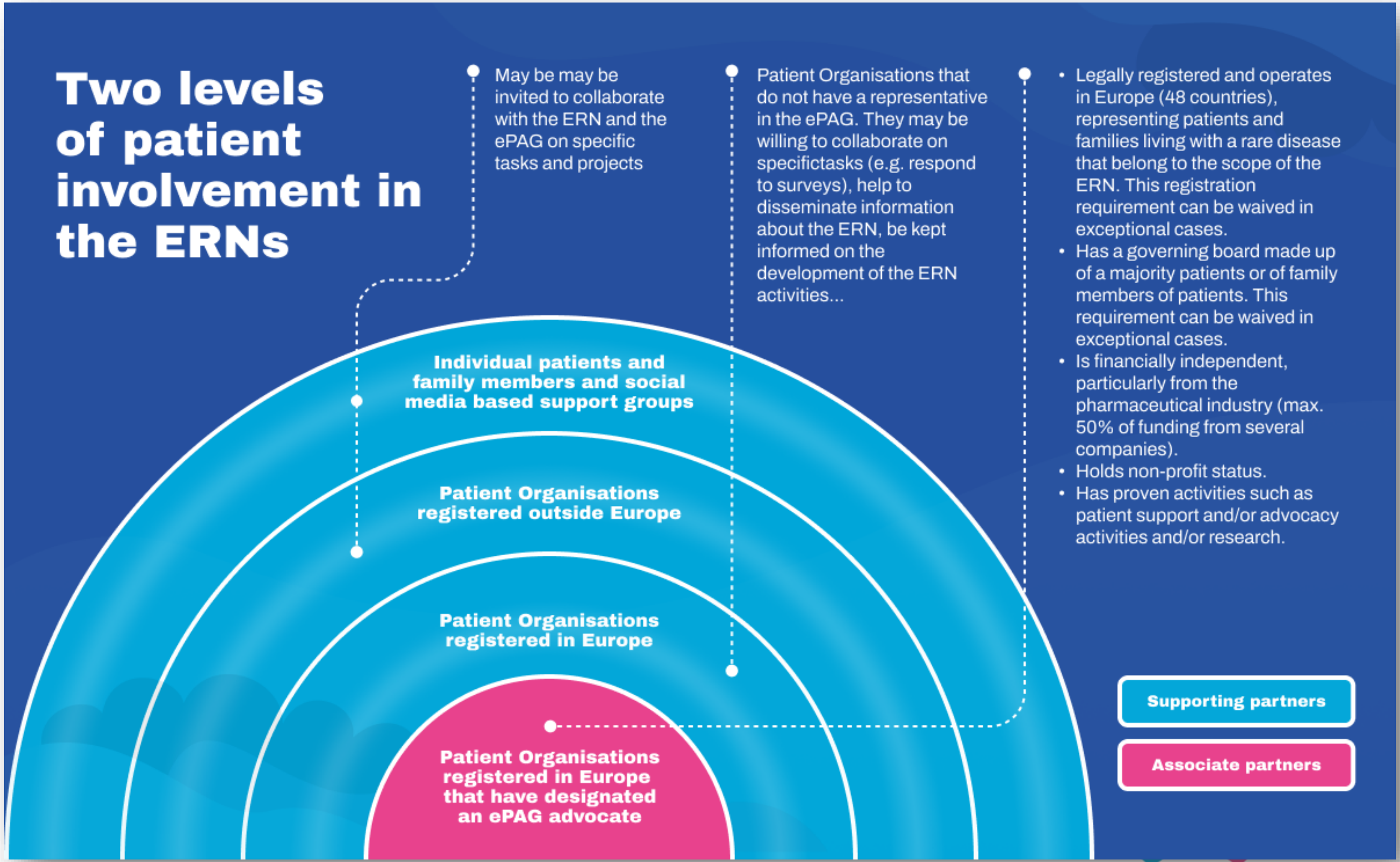
1. RECRUITMENT AND OVERALL GOVERNANCE –
New framework for Patient involvement in the ERNs

What do we want to achieve with this new approach?

1. Establish 2 levels of patient engagement in the ERNs (Associate Partners and Supporting Partners)

1. Formalise the relationship between the ERN and the patient organisations.

1. Establish clear requirements for patient organisations, rights and obligations and include them in the ERN bylaws for greater transparency and clarity.



Two levels of patient involvement in the ERNs

May be may be invited to collaborate with the ERN and the ePAG on specific tasks and projects

Patient Organisations that do not have a representative in the ePAG. They may be willing to collaborate on specifictasks (e.g. respond to surveys), help to disseminate information about the ERN, be kept informed on the development of the ERN activities...

- Legally registered and operates in Europe (48 countries), representing patients and families living with a rare disease that belong to the scope of the ERN. This registration requirement can be waived in exceptional cases.
- Has a governing board made up of a majority patients or of family members of patients. This requirement can be waived in exceptional cases.
- Is financially independent, particularly from the pharmaceutical industry (max. 50% of funding from several companies).
- Holds non-profit status.
- Has proven activities such as patient support and/or advocacy activities and/or research.

Individual patients and family members and social media based support groups

Patient Organisations registered outside Europe

Patient Organisations registered in Europe

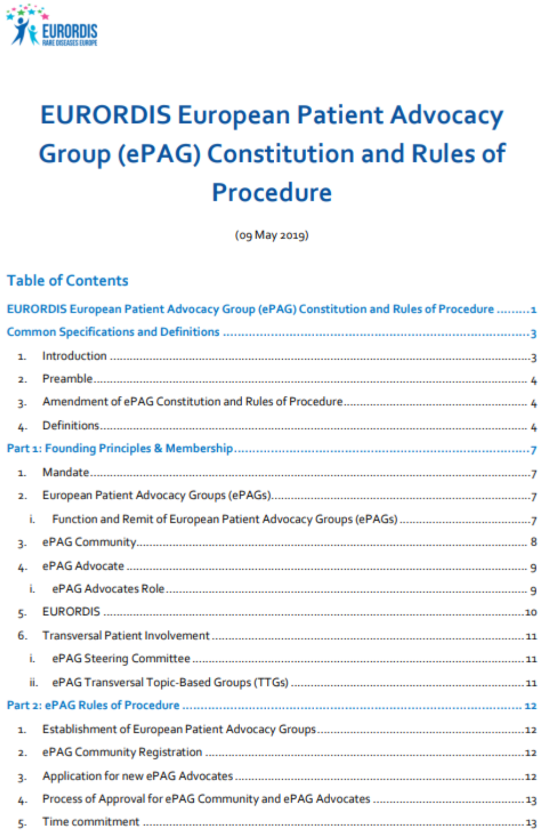
Patient Organisations registered in Europe that have designated an ePAG advocate

Supporting partners

Associate partners

1. RECRUITMENT AND OVERALL GOVERNANCE – New framework for Patient involvement in the ERNs

ePAG Constitution, Rules of Procedure and associated agreements (templates)



EURORDIS European Patient Advocacy Group (ePAG) Constitution and Rules of Procedure

(09 May 2019)

Table of Contents

European Reference Networks

[insert ERN Logo]

ASSOCIATE PARTNER COLLABORATION AGREEMENT

Between the European Reference Network (ERN) for [insert ERN scope] (inset name of ERN) and [insert name of Patient Organisation]

22/11/2021

MISSION STATEMENT

[insert ERN mission statement]

[Insert name of Patient Organisation]
Hereby consents to become an Associate Partner of [Insert name of ERN] and accepts all the rights and obligations foreseen in the ERN bylaws, as well as the terms for ePAG advocates included in [Insert name of ERN] Rules for Patient Engagement.

[Insert name of Patient Organisation] acknowledges that the position of ePAG advocate is a voluntary position and does not involve any financial compensation. Travel and accommodation expenses will be reimbursed according to the [insert name of ERN] policy on reimbursement for travel expenses.

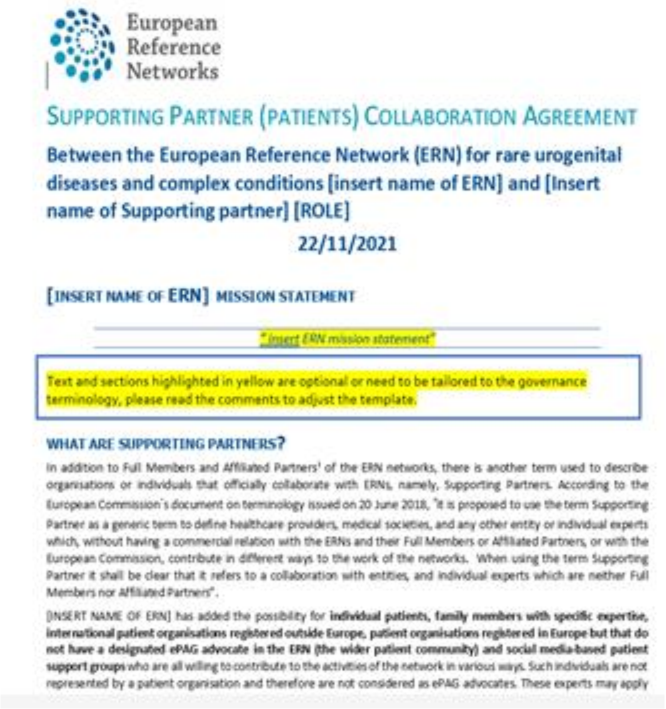
[Insert name of ERN]

Officially recognises [insert name of Patient Organisation] as an Associate Partner and affirms its commitment to work in collaboration with [insert name of Patient Organisation] and its representative for the benefit of patients living with [insert ERN disease area] under the leadership of [insert name of ERN Coordinator] and [insert name of Coordinating Centre] in [insert name of country where the ERN Coordinating centre is located].

[Name of ERN Coordinator]
Hereby certifies that the ERN has accepted the accession of [insert name of Patient Organisation] as an Associate Partner.

Two originals of this Partnership Agreement have been duly signed by the undersigned authorized representatives.

Signature (Associate Partner):	**Signature insert name of ERN:**
[NAME]	Insert name of ERN Coordinator
[ROLE]	[insert name of ERN] Coordinator



European Reference Networks

SUPPORTING PARTNER (PATIENTS) COLLABORATION AGREEMENT

Between the European Reference Network (ERN) for rare urogenital diseases and complex conditions [insert name of ERN] and [Insert name of Supporting partner] [ROLE]

22/11/2021

[INSERT NAME OF ERN] MISSION STATEMENT

"insert ERN mission statement"

Text and sections highlighted in yellow are optional or need to be tailored to the governance terminology, please read the comments to adjust the template.

WHAT ARE SUPPORTING PARTNERS?

In addition to Full Members and Affiliated Partners[1] of the ERN networks, there is another term used to describe organisations or individuals that officially collaborate with ERNs, namely, Supporting Partners. According to the European Commission´s document on terminology issued on 20 June 2018, "it is proposed to use the term Supporting Partner as a generic term to define healthcare providers, medical societies, and any other entity or individual experts which, without having a commercial relation with the ERNs and their Full Members or Affiliated Partners, or with the European Commission, contribute in different ways to the work of the networks. When using the term Supporting Partner it shall be clear that it refers to a collaboration with entities, and individual experts which are neither Full Members nor Affiliated Partners".

[INSERT NAME OF ERN] has added the possibility for **individual patients, family members with specific expertise, international patient organisations registered outside Europe, patient organisations registered in Europe but that do not have a designated ePAG advocate in the ERN (the wider patient community) and social media-based patient support groups** who are all willing to contribute to the activities of the network in various ways. Such individuals are not represented by a patient organisation and therefore are not considered as ePAG advocates. These experts may apply

1. RECRUITMENT AND OVERALL GOVERNANCE –
Are any ERNs already using this new framework?

- **Yes!**

They are (ERNs)
ERNICA
GUARD-Heart
ERN CRANIO
eUROGEN

Considering it (ERNs)
ReCONNET
VASCERN
ERN-SKIN
ERN-RARE-LIVER
ERN RITA
ERKNet

1. RECRUITMENT AND OVERALL GOVERNANCE – Application process

- **Stepwise process**:

- EURORDIS as the process facilitator:

1) EURORDIS kick-starts the applications, sending all the documents to the candidate. BUT **EURORDIS will NOT start an application without the prior approval from the ERN/ePAG**

2) EURORDIS centralizes all the documentation, sharing it with the ERN/ePAG and acknowledging the reception of a full application (when done)

- ERNS/ePAGs as decision-makers:

Application is reviewed and the decision is made by the ERNs/ePAGs

ERNs/ePAG communicate the final decision to the candidate and to EURORDIS

- Based on an online application form:
  - More transparent
  - Standardised
  - Centralised

European Patient Advocacy Group

Application Form for ePAG Advocates

Please read carefully the information below and complete the application form. The application will be assessed against the criteria for Patient Organisations and the candidate's required skills and experience for ePAG advocates described below.

APPLICATION FORM

1. REQUIREMENTS FOR PATIENT ORGANISATIONS

Find the form HERE

1. RECRUITMENT AND OVERALL GOVERNANCE –
Are any ERNs already using this new application process?

Yes!

They are (ERNs)
ERN EYE
ERN CRANIO
ERN ERNICA
ERN GUARD-Heart
eUROGEN
ERN-RND
ERN-LUNG
Endo-ERN
ERN-RITA

Considering it (ERNs)
ERKNet

New ePAG recruitment process

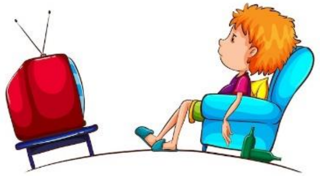
ITHACA's position

- What do you think of the framework for patient involvement in ERN governance?
- What do you think of the new application process?
- Do you want to implement them? If yes, when?
- If yes, which documents do you wish EURORDIS to send to the candidates?

LET'S TALK

EURORDIS
RARE DISEASES EUROPE

2. EPAG TOOLS AND PROCESSES – Resources for newcomers



ERN/ePAG resources		
Read	Short guide on 'Thinking of getting involved in the ERNs as an ePAG advocate?'	HERE
	ePAG factsheet	HERE
Watch	European Reference Networks and European Advocacy Patient Groups - 101	HERE
	Allison Watson	HERE
	Dorica Dan	HERE
	Ilaria Galetti	HERE

BEING AN EPAG ADVOCATE MEANS BRINGING THE UNHEARD VOICE OF A PATIENT COMMUNITY TO THE SPOTLIGHT. MY MAIN ROLE IS TO CAPTURE AND CONVEY MY PATIENT COMMUNITY'S NEEDS AND CONCERNS AT A EUROPEAN LEVEL, PAVING THE WAY FOR FASTER DIAGNOSES AND RESEARCH, WHILE STRIVING TO REDUCE THEIR BURDENS AND THE LACK OF UNDERSTANDING THAT MANY HAVE ENDURED FOR MANY YEARS.

European Patient Advocacy Group

Silvia Aguilera,
ReCONNET ePAG

"The best way to find yourself is to lose yourself in service of others."
- Mahatma Gandhi

I feel that this quote captures my commitment to put my knowledge and energy at the service of patients and willingness to assist with representation of patients' needs in the rare disease community.

Maria Cecilia la Forgia,
New ePAG Advocate ERN-BOND,
Associazione Conto Alla Rovescia

Our organisation acknowledges the importance of keeping up to date with European initiatives around the conditions we support and rare disease as a whole. When the opportunity to play a small part in your work as an ePAG advocate came up, I was keen to be involved and look forward to working with the other ePAG advocates.

Sara Hunt,
New ePAG Advocate ERN-RND,
CEO of Alex, The Leukodystrophy Charity

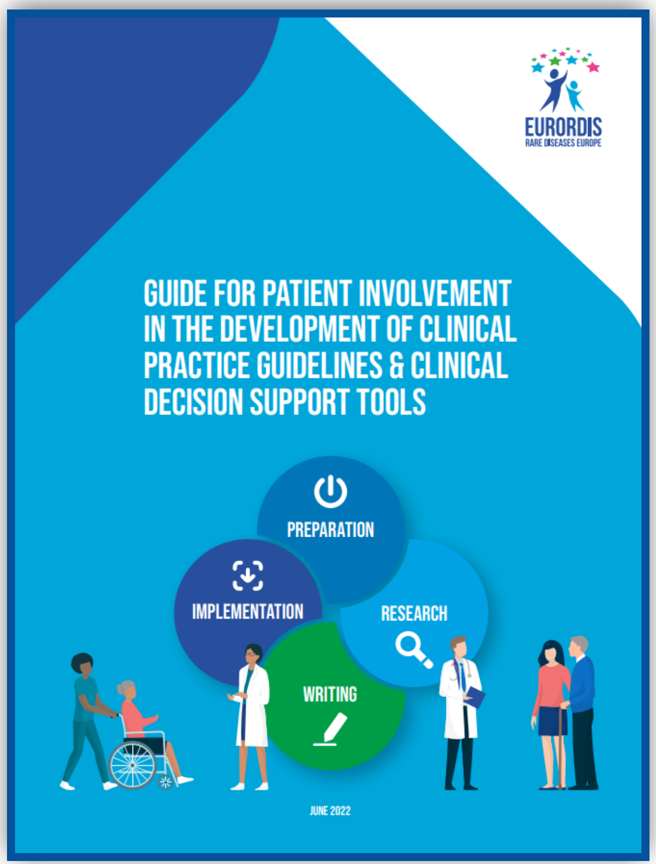
2. EPAG TOOLS AND PROCESSES –
Patient involvement in Clinical Practice Guidelines (CPGs)

1. Why? To promote and facilitate patient involvement in CPG and other clinical decision support tools. CPG development and implementation is a priority for most ERNs.

2. Who with? European Lung Foundation and ePAG Working Group on Clinical Practice Guidelines. Inspired by [ERN Guidelines Methodological Handbook]

3. Who for? Anyone interested in/working on CPGs

- EURORDIS Membership
- ePAGs
- ERN HCPs/Project Managers
- Patient organisations (at large)
- Researchers/medical societies



EURORDIS
RARE DISEASES EUROPE

GUIDE FOR PATIENT INVOLVEMENT IN THE DEVELOPMENT OF CLINICAL PRACTICE GUIDELINES & CLINICAL DECISION SUPPORT TOOLS

JUNE 2022

Access it [here]

4. How is it organized/structured?

In each stage you can find:

- Top tips

TOP TIPS

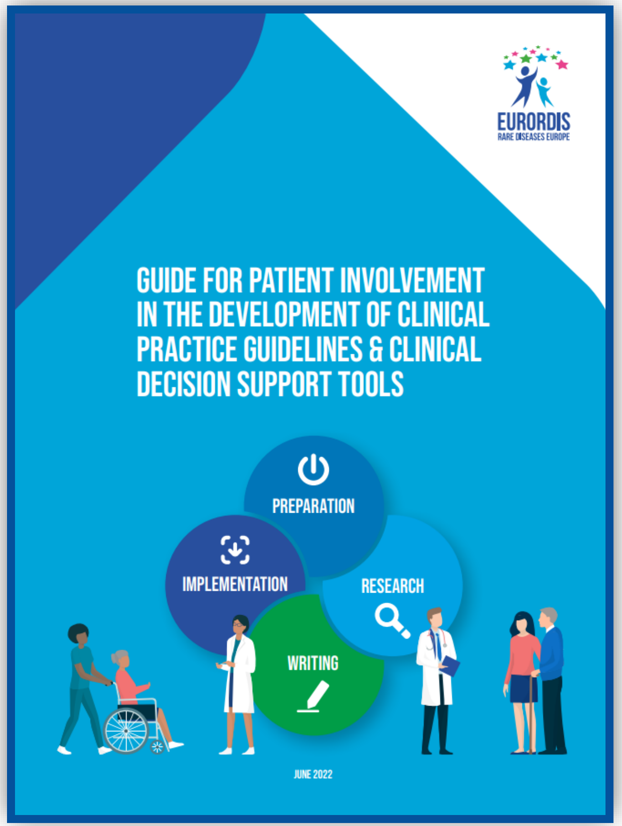
Patients can gather information about patient experiences of the topic to help supplement the scientific literature while the systematic review is underway. This could be through:

- Patient-centred literature review of published research (e.g. interviews, focus groups)
- Review of patient experiences in blogs, online discussion groups and social media
- Consultation with their patient community (e.g. survey, discussion groups

- Summary of main points
- Additional resources

Also, the guide comes with a **project planner** and a **glossary of terms**

2. EPAG TOOLS AND PROCESSES –
Patient involvement in Clinical Practice Guidelines (CPGs)



GUIDE FOR PATIENT INVOLVEMENT IN THE DEVELOPMENT OF CLINICAL PRACTICE GUIDELINES & CLINICAL DECISION SUPPORT TOOLS

JUNE 2022

Toolkit

ELF eau European Association of Urology

ADDED VALUE OF PATIENT INVOLVEMENT IN GUIDELINES

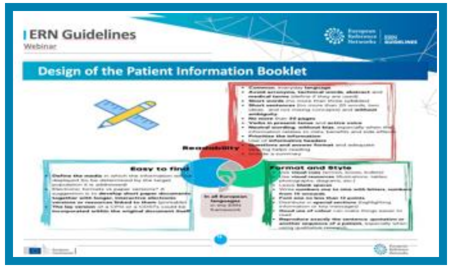
EPAG Webinar - 28 10 2021

1

Webinar on Added Value of Patient Involvement in Guideline Development

2

Webinar on How to Guide on Plain Language Summaries



ERN Guidelines
Webinar

Design of the Patient Information Booklet

ERN Guidelines
Webinar

Consortium members

Coordination
Progress and Health Foundation (FPS)

Health Technology Agencies

1. The Andalusian Agency for Health Technology Assessment (AETSA)
2. The Basque Foundation for Health Innovation and Research (BIOEF)
3. The Aragón Health Sciences Institute (IACS)
4. The Agency for Health Quality and Assessment of Catalonia (AQuAS)
5. The Canary Institute of Health Research (FIISC)

Training Program

1. Andalusian School of Public Health (EASP
2. Open University of Catalonia (UOC)

3

Webinar(s) on Roadmap to Guideline Implementation

4+

Webinar on Appraisal & Adoption of an existing guideline

2. EPAG TOOLS AND PROCESSES –
Evaluations of the ERNs at 5-Year

1. Why? The first e Assessment, Evaluation & Monitoring Quality Improvement System (AMEQUIS) **Evaluation** is about to start. Patient representatives have a strong voice in the process. This guide (+ factsheet) aim to empower them to actively contribute to this key process.

2. Who with? ePAG AMEQUIS task force. Based on ERNs Evaluation Manual & Toolkit

3. Who for? Anyone interested in/working on AMEQUIS

- ePAGs
- ERN Project Managers

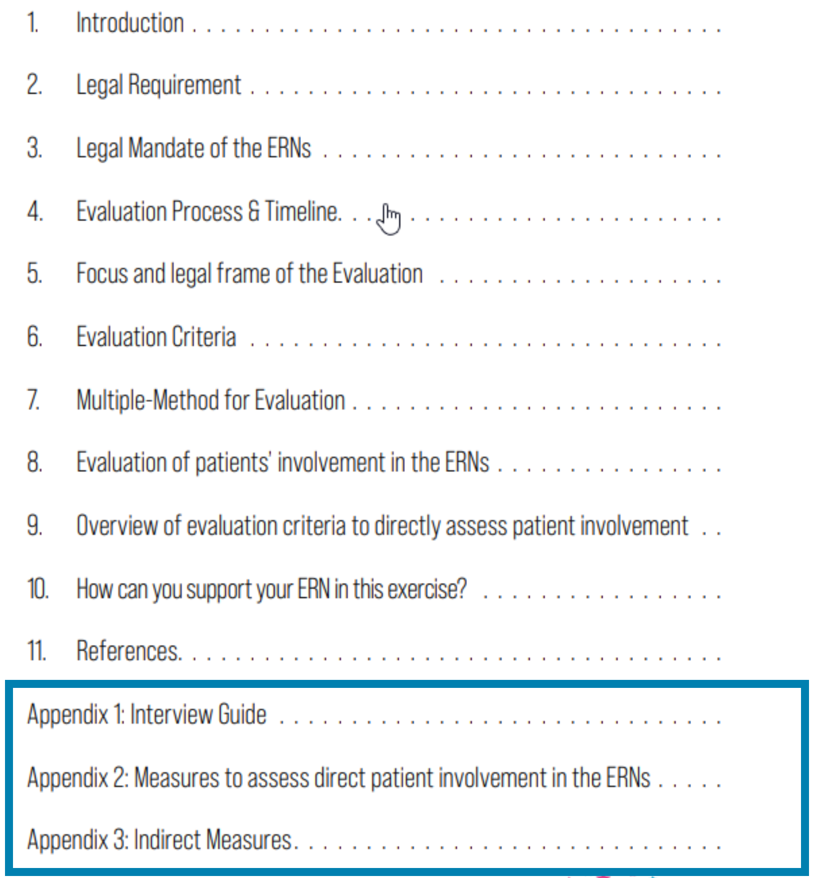
EURORDIS RARE DISEASES EUROPE

PATIENT GUIDE TO THE 5-YEAR EVALUATION OF THE EUROPEAN REFERENCE NETWORKS

SEPTEMBER 2022

Access it here

4. How is it organized/structured?



1. Introduction
2. Legal Requirement
3. Legal Mandate of the ERNs
4. Evaluation Process & Timeline
5. Focus and legal frame of the Evaluation
6. Evaluation Criteria
7. Multiple-Method for Evaluation
8. Evaluation of patients' involvement in the ERNs
9. Overview of evaluation criteria to directly assess patient involvement
10. How can you support your ERN in this exercise?
11. References

Appendix 1: Interview Guide

Appendix 2: Measures to assess direct patient involvement in the ERNs

Appendix 3: Indirect Measures

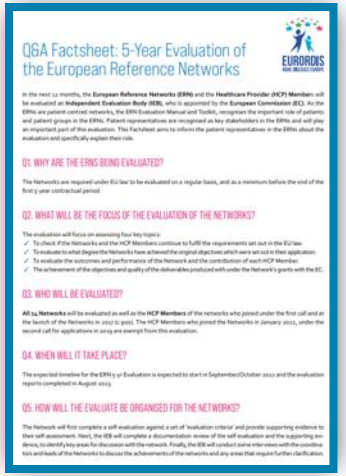
2. EPAG TOOLS AND PROCESSES –
Evaluations of the ERNs at 5-Year


EURORDIS
RARE DISEASES EUROPE

PATIENT GUIDE TO THE 5-YEAR EVALUATION OF THE EUROPEAN REFERENCE NETWORKS

SEPTEMBER 2022

Toolkit


Q&A Factsheet: 5-Year Evaluation of the European Reference Networks

EURORDIS

Q1. WHY ARE THE ERNS BEING EVALUATED?

Q2. WHAT WILL BE THE FOCUS OF THE EVALUATION OF THE NETWORKS?

Q3. WHO WILL BE EVALUATED?

Q4. WHEN WILL IT TAKE PLACE?

Q5. HOW WILL THE EVALUATE BE ORGANISED FOR THE NETWORKS?

1

Short Factsheet compiling the most frequent Q&A on the Evaluation process

2

Webinar on Demystifying AMEQUIS
+ Webinar on 5-year Evaluation through the Patient Lens


ERN 5-Yr Evaluation through the Patient-Lens Webinar

The Why?

3

Individual ePAG Focus Groups planned in Q4 2022

Access them here

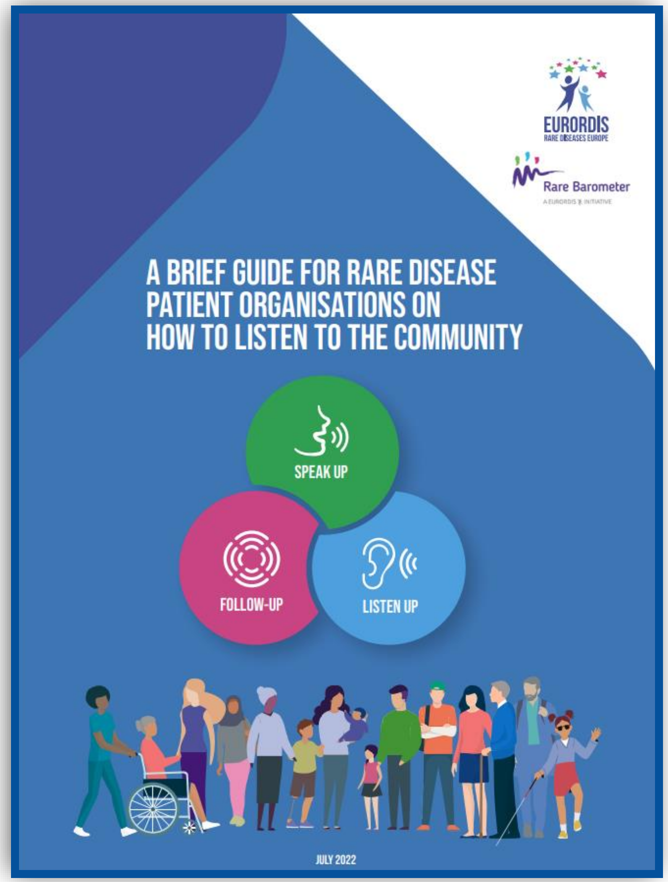
2. EPAG TOOLS AND PROCESSES –
Speak up. Listen up. Follow up

1. **Why?** Representing the views of their wider patient community is one of the central roles of patient representatives, including those involved in the ERNs. However, collecting robust and actionable data on the needs/preferences or experiences of a given population can be a daunting exercise. This practical guides offers tips and a stepwise methodology to guide & empower patient representatives.

2. **Who with? Co-developed by Healthcare/ERN + Rare Barometer teams.** ePAG Steering Committee. Inspired by NHS bite-sized guide: Insight – What is already available?, Writing an effective Questionnaire and Building greater insight through Qualitative Research.

3. **Who for?** Anyone interested in/working on data collection projects

- ePAGs
- ERN Project Managers
- EURORDIS Members
- Patient organisations (at large)
- Researchers



EURORDIS
RARE DISEASES EUROPE

Rare Barometer
A EURORDIS INITIATIVE

A BRIEF GUIDE FOR RARE DISEASE PATIENT ORGANISATIONS ON HOW TO LISTEN TO THE COMMUNITY

SPEAK UP
FOLLOW-UP
LISTEN UP

JULY 2022

Access it here

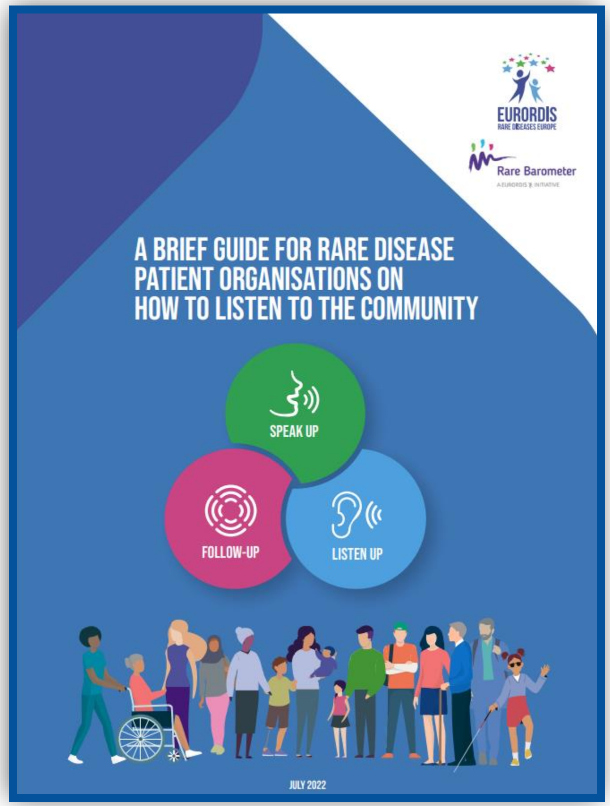
4. How is it organized/structured?

GUIDE STRUCTURE

European
Patient
Advocacy
Group

EURORDIS
RARE DISEASES EUROPE

2. EPAG TOOLS AND PROCESSES –
Speak up. Listen up. Follow up


A BRIEF GUIDE FOR RARE DISEASE PATIENT ORGANISATIONS ON HOW TO LISTEN TO THE COMMUNITY

JULY 2022

Toolkit

1


ePAG exchange of Good Practices: Designing Surveys

ePAG Exchange of Good Practices

Designing Surveys

Agenda

16.00-16.05 - Welcome & Introduction

16.05-16.20 – An Introduction to Designing Surveys

16.20-16.30 - *Questions & Answers*

16.30-17.00 – Co-designing surveys for rare diseases: the ERN ReCONNET journey

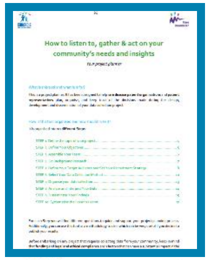
17.00-17.25 - *Questions & Answers*

17.25-17.30 - Wrap-up

ePAG exchange of Good Practices Webinar on Survey Development

2

'Your project planner'



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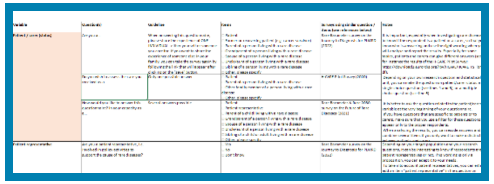


Rare Barometer question repository

4

snapshot version of the guide

2. EPAG TOOLS AND PROCESSES – ERN & EPAG podcast series

Rare on Air

A EURORDIS-Rare Diseases Europe podcast on the experiences, challenges and successes of people living with rare diseases. Julien Poulain, Communications Manager at EURORDIS, meets with people who share their unique experiences of living with a rare disease, those who advocate for them, and experts on rare disease policy. Email the EURORDIS Rare on Air team at: RareOnAir@eurordis.org.

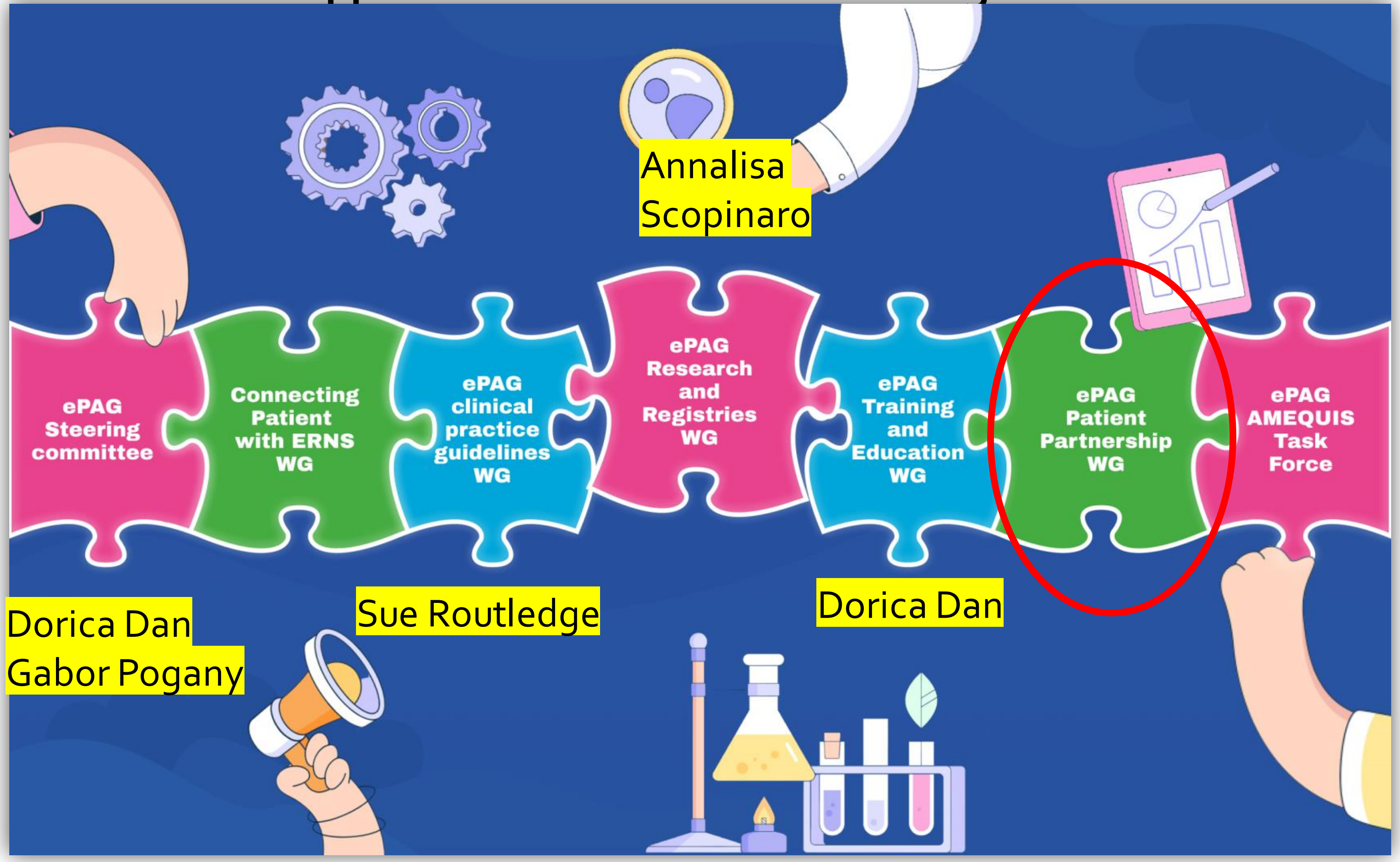
The **first episode**: *The Journey of European Reference Networks*.

In our first episode of Rare on Air, Julien Poulain interviews Yann Le Cam, Chief Executive Officer of EURORDIS-Rare Diseases Europe, as we take a trip down memory lane with the European Reference Networks (ERNs).
Later in the episode, we hear a conversation on how the ERNs began, led by Rita Francisco, EURORDIS' Patient Engagement Junior Manager, and Sarah Weiler from the Luxembourg National Alliance for Rare Diseases/

- Anchor: https://anchor.fm/eurordis
- Spotify: https://open.spotify.com/show/4gpNMGmFMfmsPkIL5Rnsn8
- Amazon Music: https://music.amazon.fr/podcasts/d8ce0e9f-b50b-42b5-add9-b69dcbb40e57/rare-on-air

3. FACILITATING INVOLVEMENT OF ePAG ADVOCATES – ePAG transversal working groups

EURORDIS manages working groups to favour peer learning and to support advocates in the following areas:



ePAG Steering committee

Connecting Patient with ERNS WG

ePAG clinical practice guidelines WG

ePAG Research and Registries WG

ePAG Training and Education WG

ePAG Patient Partnership WG

ePAG AMEQUIS Task Force

Annalisa Scopinaro

Dorica Dan
Gabor Pogany

Sue Routledge

Dorica Dan

3. FACILITATING INVOLVEMENT OF ePAG ADVOCATES – Patient Partnership Working Group

1. Who are the Patient Partnership Working Group?

ePAG advocates + ERN Project Managers

2. What are the objectives of this Working Group?

The group will support the implementation of patient involvement tools and processes to foster a patient-clinician partnership culture that is similar across all ERNs by:

1. Sharing relevant information and updates on resources (tools and processes) and good practices to facilitate patient involvement in the ERNs
2. Providing advise on the implementation of new resources for patient involvement in the ERNs
3. Supporting the development of new resources for patient engagement in the ERNs

3. FACILITATING INVOLVEMENT OF ePAG ADVOCATES – Patient Partnership Working Group

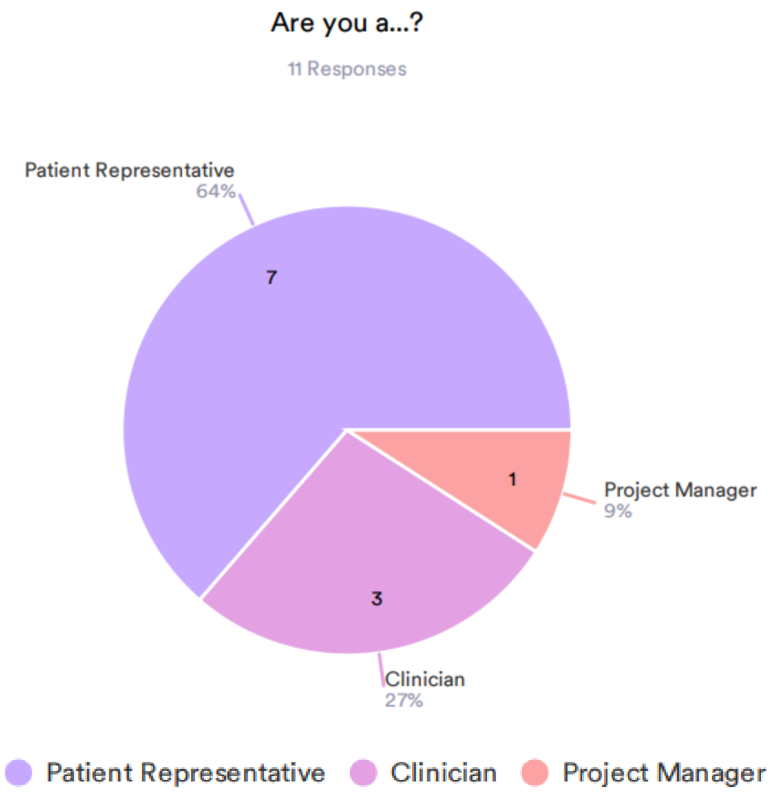
Members

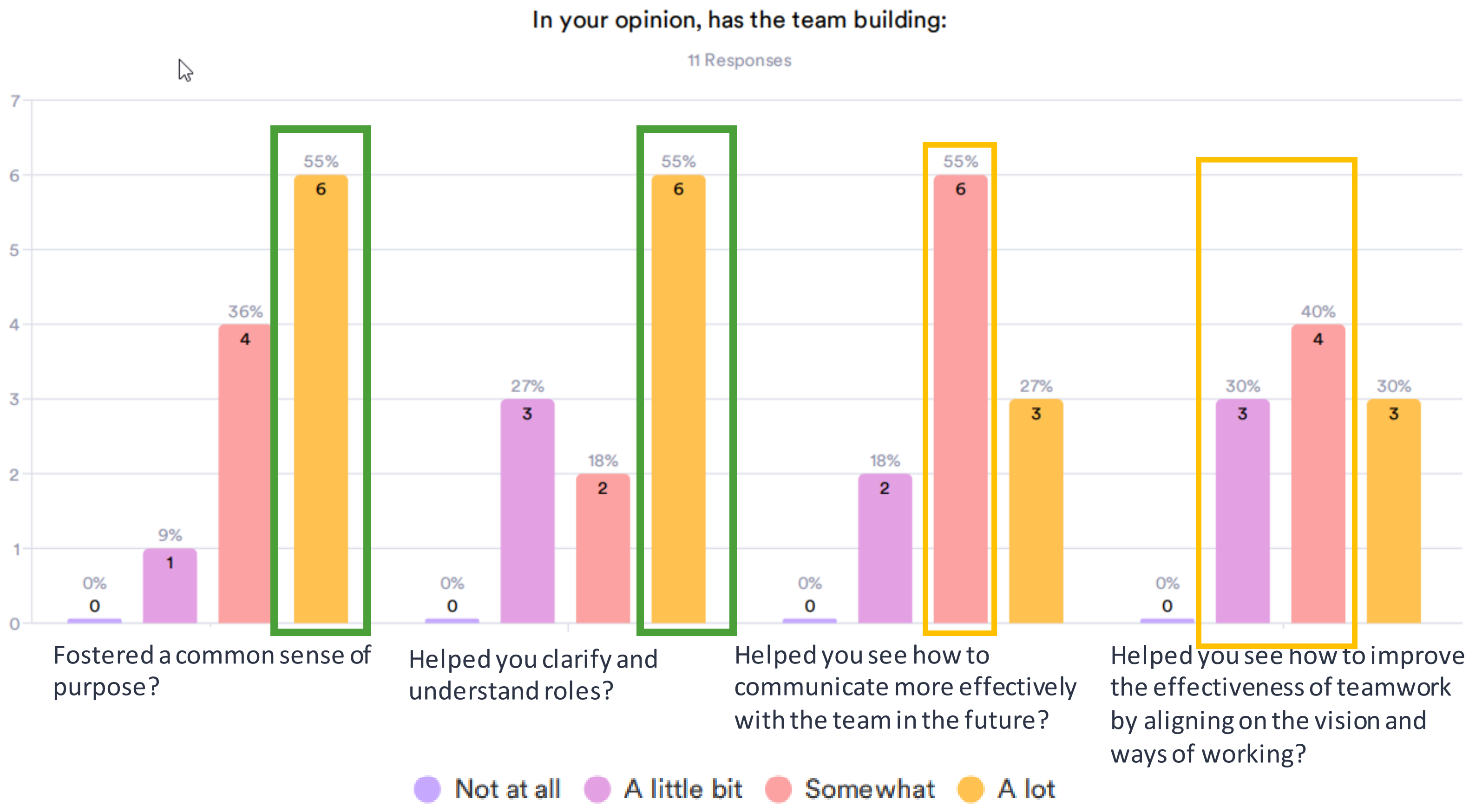
ERN Project Managers (ERN)*	ePAG advocates (ERN)
Olivia Spivak (ERNICA)	Graham Slater (ERNICA)
Diana Marinello (ReCONNET)	Charissa Frank (VASCERN)
Treasure UDECHUKWU (VASCERN)	Ilaria Galetti (ReCONNET)
Mariangela Pellegrini (EuroBloodNet)	**Ammi Anderson (ITHACA)**
Anne Hughon (ITHACA)	Giovanna Campioni (GUARD-Heart)
Jana Steerneman (ERN CRANIO)	Lex van der Heijden (ERN-SKIN)
Caroline Wernert-Iberg (ERN EYE)	Stefano Pavanello (ERN-LUNG)
Ana Merino (TransplantChild)	**Dorica Dan (ITHACA)**
Charlotte van Beuzekom (Endo ERN)	

Total no of full/permanent members = **17**
Total no of represented ERNs= **12**

*Other ERN Project Managers may join on a project basis

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training





4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA's results

What did you hope to get from attending this team-building training?

11 Responses

'Understand better what teamwork means'

'Stronger cooperation'

'Tips how to cooperate together more effectively'

'Improving our mutuals goals'

'Understanding how we can collaborate'

Improve collaboration/co-creation

'Better connections'

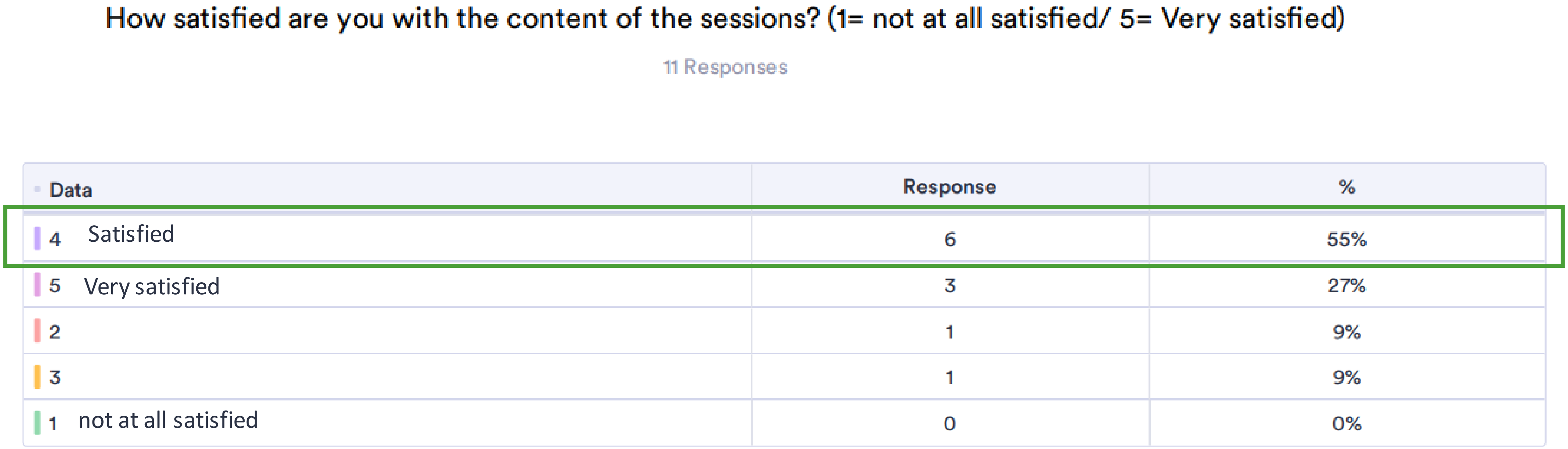
'More connection with ITHACA community'

'Get to know ern ITHACA ePAGs and clinicians'

'Connect with clinicians and researchers from ERN Ithaca'

Community-building

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA's results

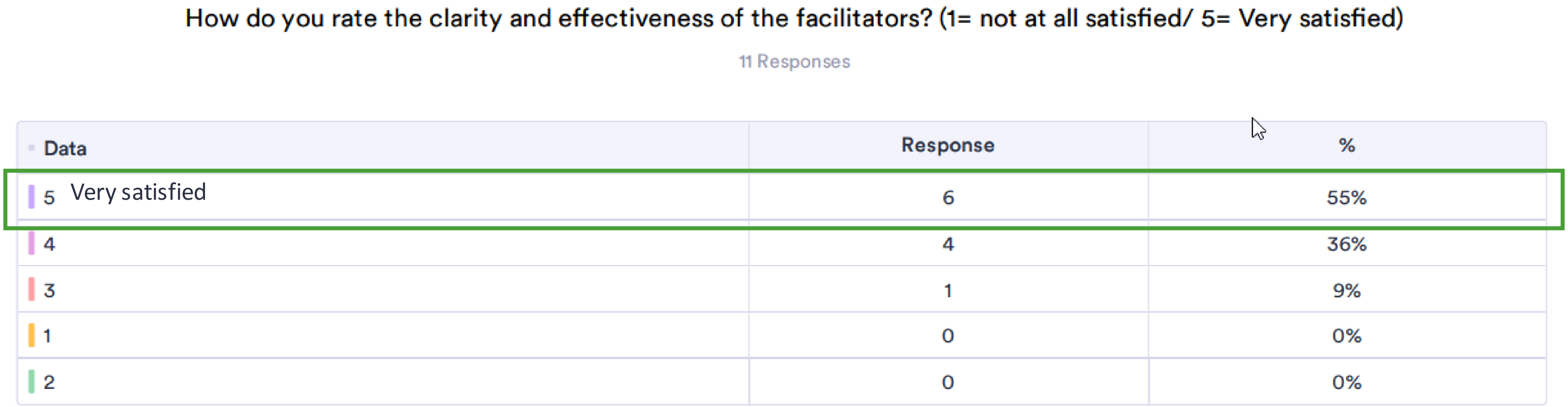


How satisfied are you with the content of the sessions? (1= not at all satisfied/ 5= Very satisfied)

11 Responses

Data	Response	%
4 Satisfied	6	55%
5 Very satisfied	3	27%
2	1	9%
3	1	9%
1 not at all satisfied	0	0%

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES – Team-building training: ITHACA’s results



How do you rate the clarity and effectiveness of the facilitators? (1= not at all satisfied/ 5= Very satisfied)

11 Responses

Data	Response	%
5 Very satisfied	6	55%
4	4	36%
3	1	9%
1	0	0%
2	0	0%

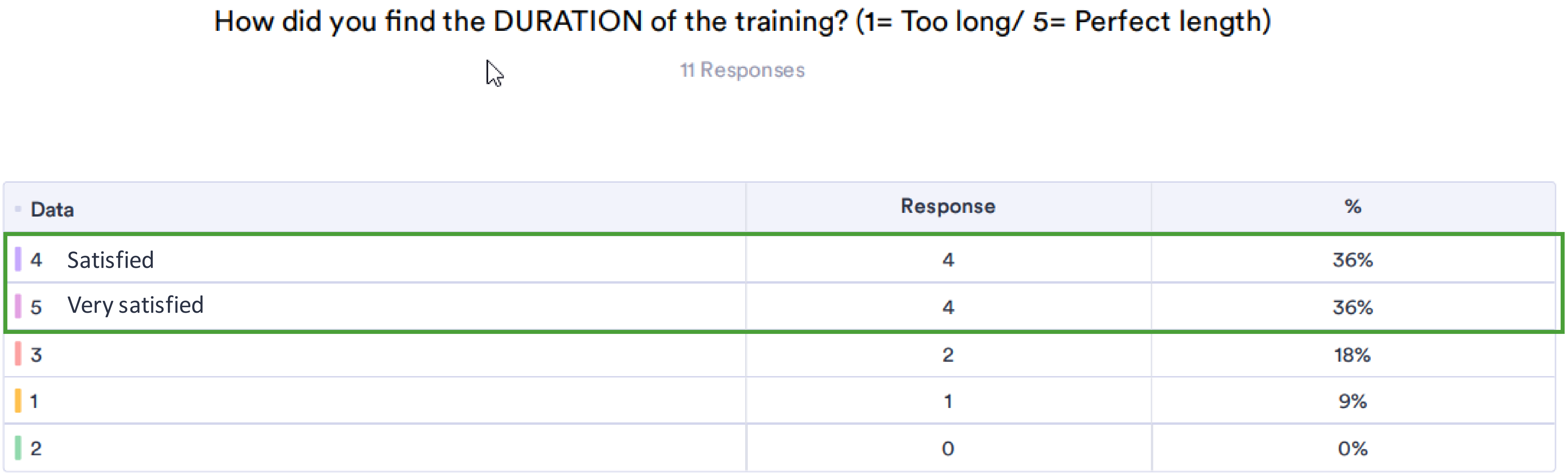
‘*kindness and confident*’

‘Ana was very good!’

‘Ana is very nice and organised person’

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –

Team-building training: ITHACA’s results

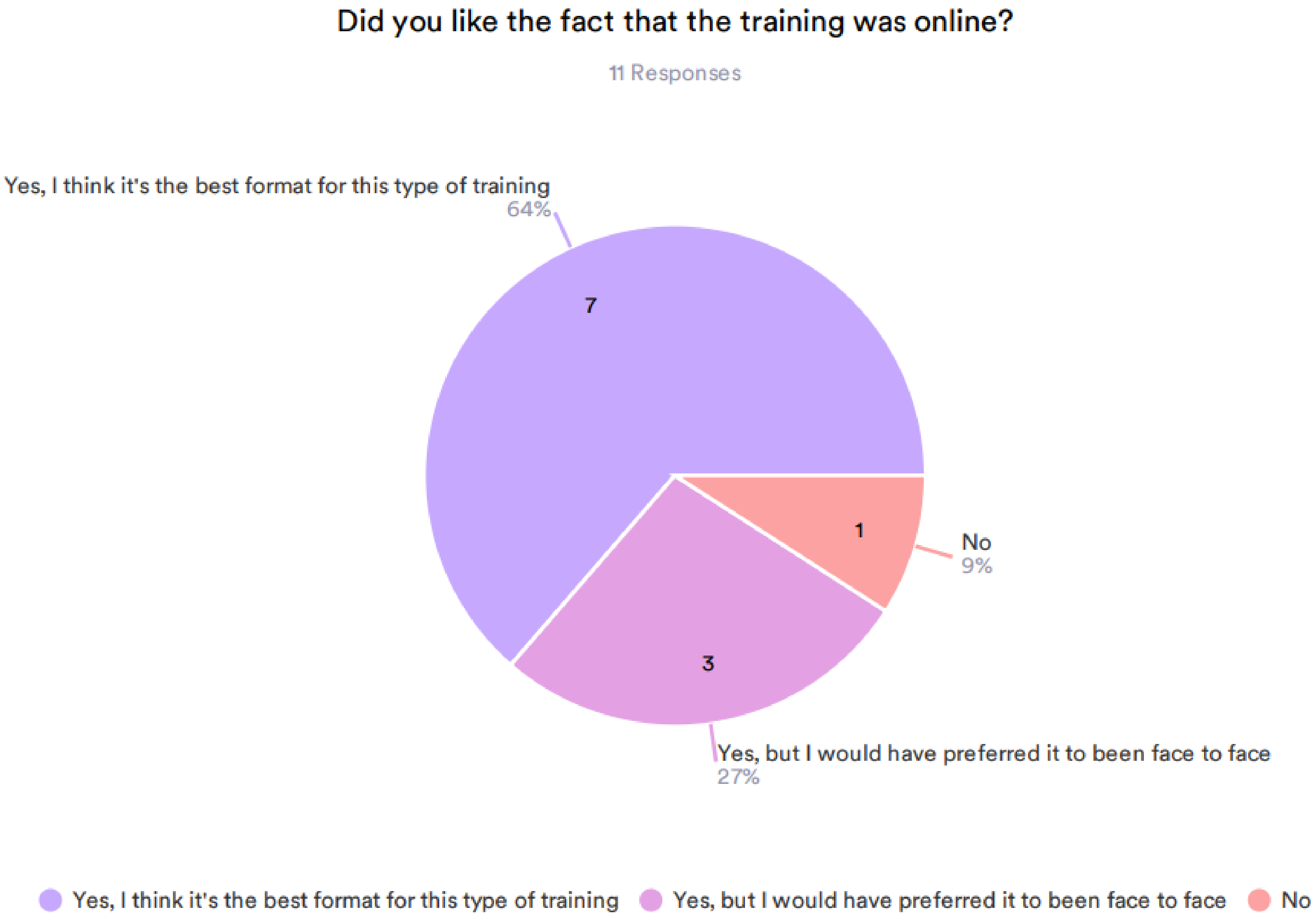


How did you find the DURATION of the training? (1= Too long/ 5= Perfect length)

11 Responses

Data	Response	%
4 Satisfied	4	36%
5 Very satisfied	4	36%
3	2	18%
1	1	9%
2	0	0%

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA's results



'Good pilot 3 x 2h'

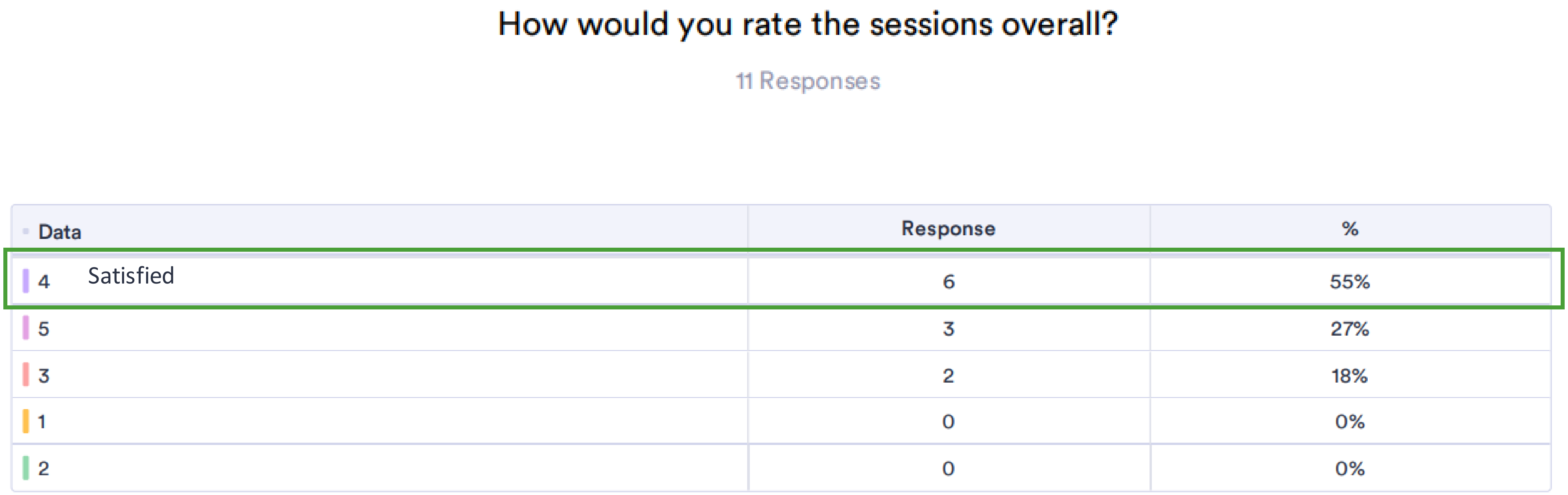
'F2F is great but not practical'

'It is not possible face to face in European organization, but it would be enjoyable'

'It was a very good format'

'Maybe we need some guide for moderating ZOOM small rooms to ensure everyone gives his opinion'

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA’s results



How would you rate the sessions overall?

11 Responses

Data	Response	%
4 Satisfied	6	55%
5	3	27%
3	2	18%
1	0	0%
2	0	0%

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –

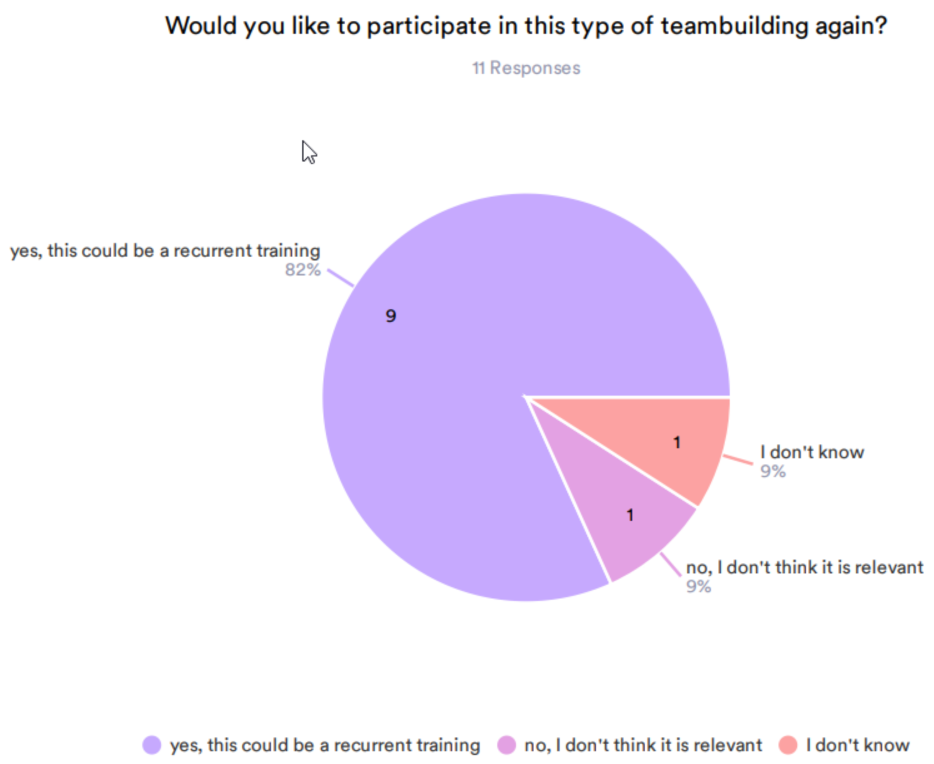
Team-building training: ITHACA's results

What was great about this team building training? (9 responses)	What could be improved? (And is there anything you would drop to make room for it?) (8 responses)
As I am a new epag, this training got me a good insight of the other epags - what are they focused on or interested in (who is more research oriented, who is working on policy, also who is devoting time to discuss what should the rare diseases been named	More efficient information how to cooperate better, more sessions regarding getting to know each other, more sessions in small groups
Positive atmosphere	More people could attend.
The connections made	number and origin of people
People	more balanced group of participants
Meeting new people and learning about other ePAGs and clinicians	Too long and too general in some parts
meeting other ePAG and clinicians	technical issues. To start on time
mutual understanding and honesty	The format, more specific guidelines and a clear roadmap about what this ERN is aimed to.
the recaps that Ana did	

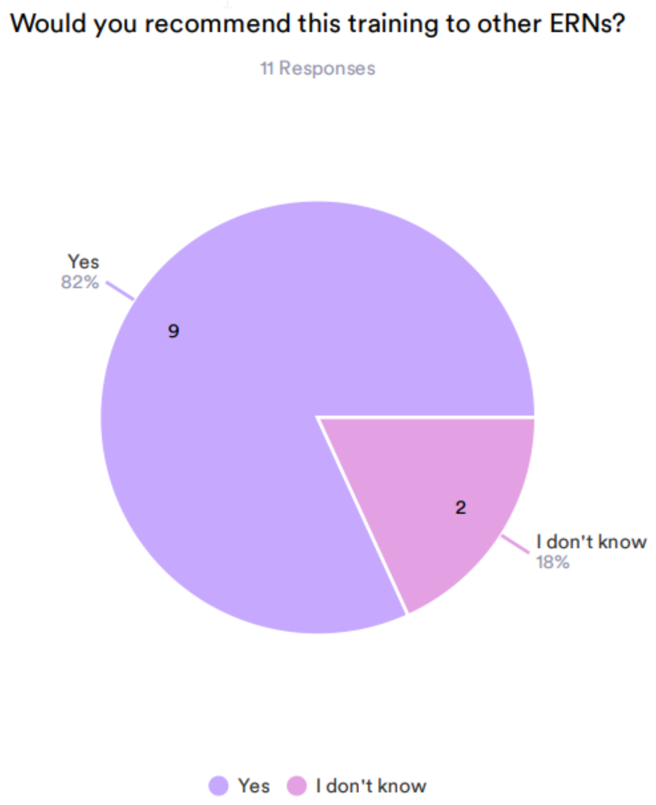
Community-building

Increase participation: in numbers and diversity

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA's results


Would you like to participate in this type of teambuilding again?

11 Responses


Would you recommend this training to other ERNs?

11 Responses

'It is very useful'

'Connects people'

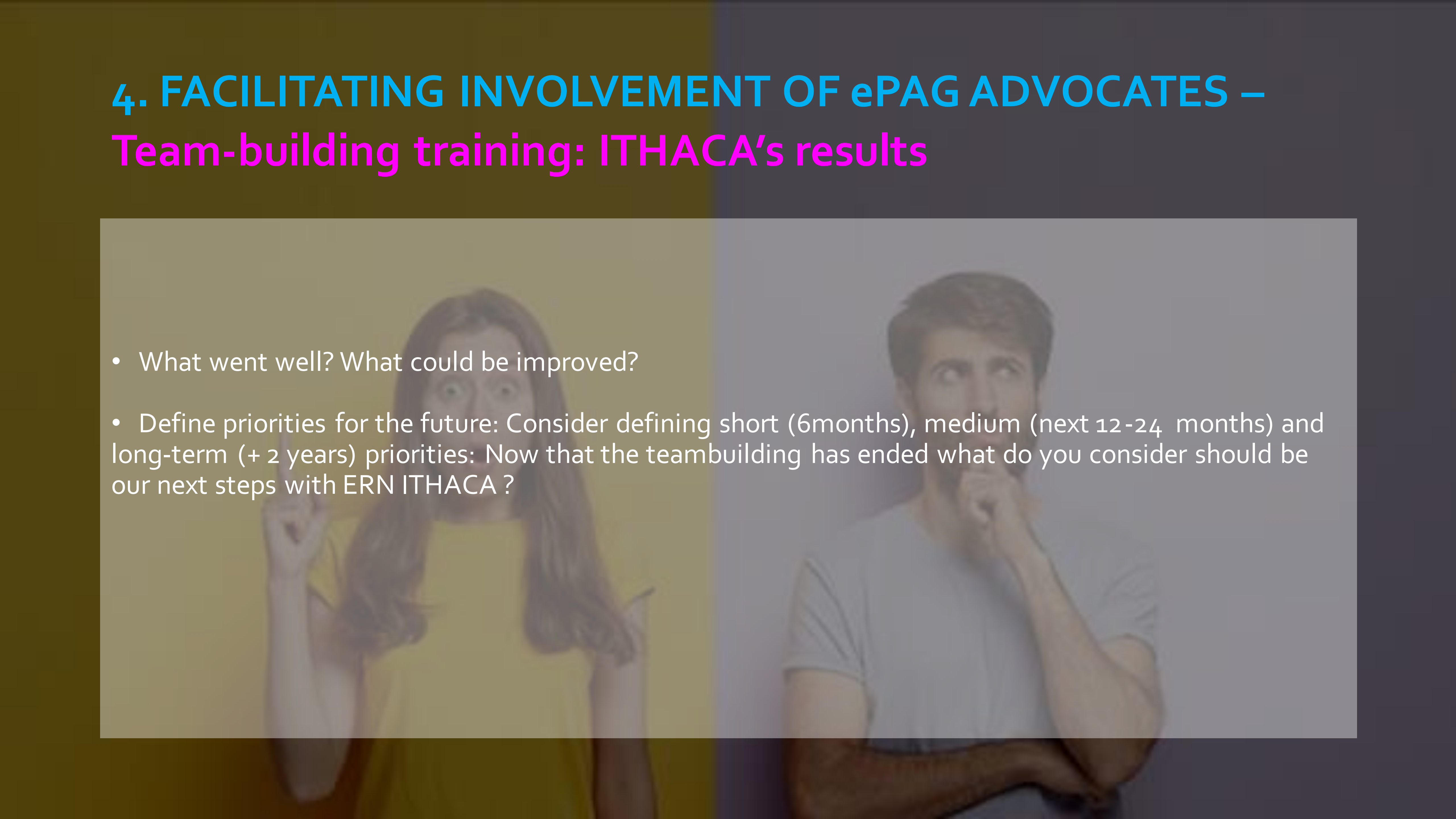
'It was fun and worthwhile.'

'I would say: you would understand, in a safe environment, how is to be in the clinicians' view. It is an opportunity to understand yourself and each other'

'Very difficult for clinicians to take 3x 2 hours off in the middle of the day. Difficult to get enough clinicians to participate. Announce it well in advance.'

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –

Team-building training: ITHACA’s results



- What went well? What could be improved?
- Define priorities for the future: Consider defining short (6months), medium (next 12-24 months) and long-term (+ 2 years) priorities: Now that the teambuilding has ended what do you consider should be our next steps with ERN ITHACA ?

4. FACILITATING INVOLVEMENT OF ePAG ADVOCATES –
Team-building training: ITHACA's results

Now that the teambuilding has ended what do you consider should be our next steps with ERN ITHACA ?

'Work together to achieve the goals'

'Going deeper in our goals and work topics'

Get down to work

'Given the lower participation rate of clinicians, they should be helped to understand the importance of communicating with ePAGs.'

'To build on the connections.'

'We would like to re-connect few times per year 2-3 times'

Continue community-building

'Show what we as ePAGs are intended for.'

'We would like to understand each other and share the vision'

Better clarify roles

I think team building focused on a specific groups who will be working together (like training, research or guideline) or focused on specific syndromes instead of all clinicians and ePAGs would also be useful, result in more clear approach/changes in working styles and also motivate more people to join compared to a general meeting

'I would recommend more training about how to organize your time more efficiently, how to work more efficiently'

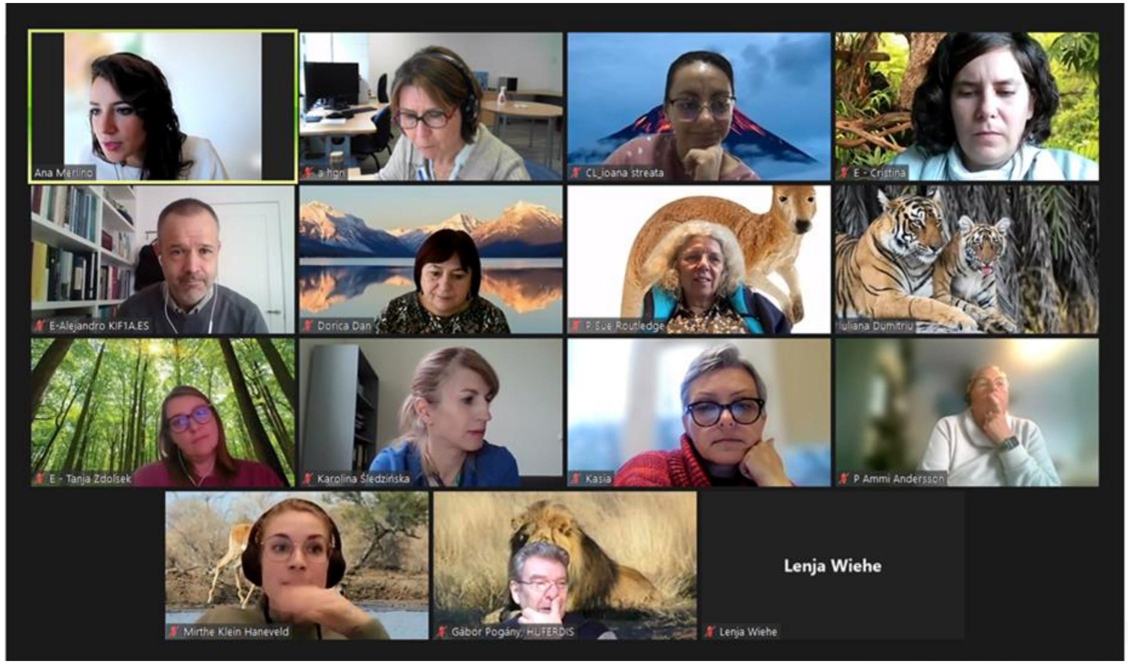
More trainings

The team building session "UBUNTU”

Discussion

Team building session "UBUNTU

- Team Building
  - ITHACA Patient Council in partnership with Eurordis, has to propose patient-clinician 3 teams building sessions with clinical and patients leads working group in Ithaca on nov 16th, nov 23rd, nov 30th, withy Ana Merlino a professional coach
  - 3 sessions nov 16th (11.30am - 1.30pm CET) nov 23rd (10am - 12.pm CET) nov 30th (10am - 12.pm CET)
  - 16 participants (7 prof / 9 ePags)
- The overall objective was of these sessions UMBUTU
  - improve our work perception, to develop interactions and working methods with different team members actively involve in Ithaca .
  - How to Identify the strengths and areas for improvement in our current collaboration. Adding value to our work
- Ubuntu, an African word that means "I'am because we are " and Sawubana : "I see you"



- Active listening
- Lots of patience
- Looking you under the premise that I don't want to change you
- Be open for change
- Reframe what the other person is saying

Thank you!

Ana Merlino
Coach, trainer y speaker

www.anamerlino.com

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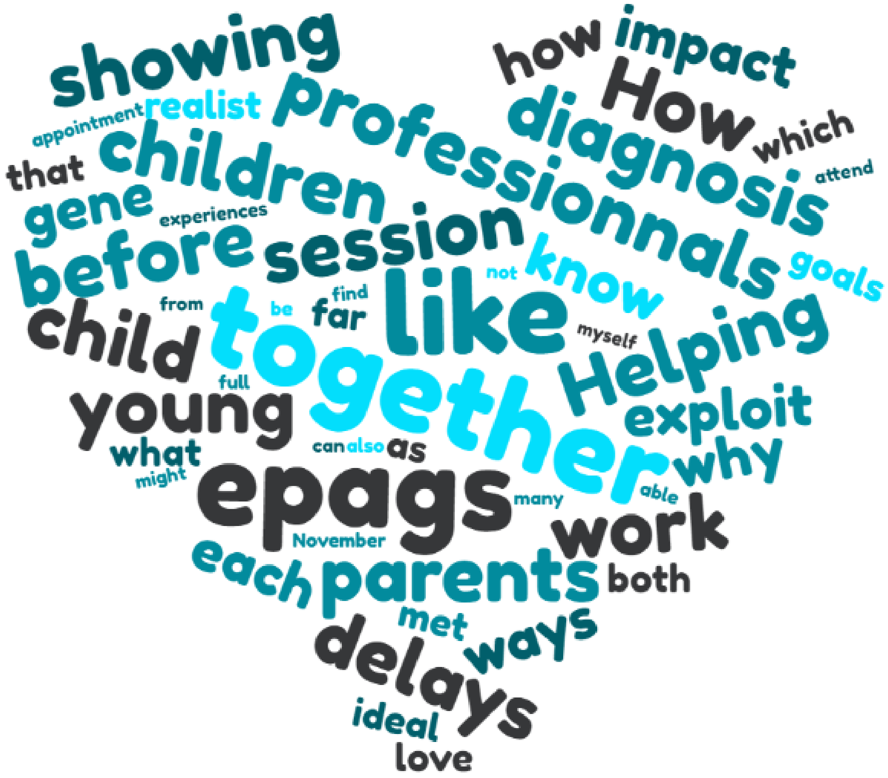
Team building an ongoing process ! 2023 ?



What do we do next ??

A Win Win relationship

Improve our work perception,
develop interactions and better collaboration
Be heroes and not victims
Evolve into a cohesive unit

Gratitude & Understanding

Building resilience
Trust Each Other
Willingness to Share Expertise
Complement One Another
Be Open to Suggestion
Appreciation of each other's expertise
Identify the strengths and weaknesses

Patient Board Election

Patient Board 2023 - 2024

Chairs of Patient Board 2023 – 2024

Candidates to the Patient Board 2022 – 2023


Dorica Dan
Prader Willi
Association
Eurordis, RO


Sue Routledge
Pitt Hopkins UK |
Netherlands, NL


Ioel Detton
Association
Noonan
France, FR


Ammi Sundqvist-
Andersson Spina
Bifida &
Hypdrocephalus
RBU Sweden
International, SE

Gábor Pogány
Hungarian
Williams Syndrome
Association,
HUFERDIS, HU

?

Election / mentimeter

Lunch time

Focus on Work Group with clinicians, sharing expertise and expectations

Afternoon

Agenda – pm Focus on Work Group with clinicians, sharing expertise and expectations

- 14h00 Introduction
- 14h15 Focus WG Guidelines
  - When and how should be Patients be involved in guidelines, asking your opinion

  Agnies van Eeghen, Charlotte Gaasterland, Mirthe Klein Haneveld
- 15h45 Focus WG Teaching & Education
  - What resources can I rely on, what can we develop as educational support needed, asking your opinion

  Laurence Faivre, Giuzeppe Zampino, Anne Hugon

16h15 Coffee Break

- 16h45 Focus WG NDD Neuro Developmental Disorders
  - Over view on Neuro-developmental Disorders work Group, Tjitske Kleefstra, Anne Hugon
  - World Café "Clinical genetics & Psychiatry (Psychiatric phenotyping) from a multidisciplinary approach, including the perspective of the expert by lived experience"

  Tjitske Kleefstra, Claudine Laurent-Levinson, Jolanda Van Golde
- 18h00 Final discussion, Wrap up and conclusion Dorica Dan, Gabor Pogany

1 - Focus WG Guidelines
When and how should be Patients be involved in guidelines, asking your opinion

Agnies van Eeghen, Charlotte Gaasterland, Mirthe Klein Haneveld

European Reference Networks

European
Commission

ITHACA: Update on guideline projects

Agnies van Eeghen

December 2022

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Intellectual Disability
and Congenital
Malformations (ERN ITHACA)

WG Guideline and Experts consensus

- Chair: Agnies van Eeghen (AUMC), co-chair: Katalin Szakszon
- Methodological support and research: Charlotte Gaasterland (postdoc), Mirthe Klein Haneveld (PhD-candidate)
- Project management: Klea Vyshka, Anne Hugon

Agnies van Eeghen, Charlotte Gaasterland, Mirthe Klein Haneveld, Anne Hugon, Klea Vyshka, Katalin Szakszon

Guideline projects: transdiagnostic

Syndrome specific

Kleefstra guideline	End of 2023
Phelan McDermid guideline	Fall 2022
Rubinstein Taybi guideline	End of 2022
Williams guideline	End of 2022
Noonan guideline	End of 2022
Kabuki guideline	Summer 2023
Spina bifida	*Summer 2023*
Just started: Fragile X guideline	

Transdiagnostic

Transition of care	Summer 2023
PIMD/Polyhandicap	Summer 2023
Sleeping problems	End 2023
Just started Challenging behaviour	

Our guideline projects in more detail

Charlotte Gaasterland, PhD

Post-doc for ITHACA at the AUMC

European Reference Network

for rare or low prevalence complex diseases

Network

Intellectual Disability and Congenital Malformations (ERN ITHACA)

European
Commission

Our guideline projects in more detail

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for rare or low prevalence complex diseases

Network
Intellectual Disability and Congenital Malformations (ERN ITHACA)

Kleefstra guideline

- Overall search: +/- 80 original clinical research articles
- Prioritization of 45 clinical topics to 12 clinical topics
- 5 working groups working on these topics
- Consensus meeting to be planned – autumn 2023

Phelan McDermid guideline

- Consensus meeting in June 2022
- Documents in preparation: publication in EJMG in 2023
- Patient leaflet in preparation

Fragile X guideline

- Project just starting up
- Many national guidelines on this topic; we are now analyzing if we can use any of these existing guidelines

Transition of care

- Systematic search has been done on transition of care: still many research articles included
- Surveys are translated and sent out to patient groups all over Europe
- Themes of the guideline are to be determined

PIMD/Polyhandicap

- 5 questions are systematically seached and summarized by the Osteba group
- A Delphi has already been performedfor recommendations
- Consensus meeting will be planned in summer/fall 2023

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Challenging behaviour

- Project just starting up
- Core working group is established
- Focus on 10-12 clinical topics, that are determined based on patient surveys and cinical expertise
- Literature search may be performed by the Osteba group

Support by Osteba

- Translations
- Patient leaflet format (example PMS)
- Literature searches, selections and summaries
- Analysis of patient surveys
- AGREE-analysis of existing guidelines (see PhD project Mirthe)

Osteba

European Reference Networks ITHACA

Our ambitions for the next few years

- Mirthe Klein Haneveld: PhD-candidate at the AUMC
- Develop methodological framework
- Test and improve guideline methodology as we go

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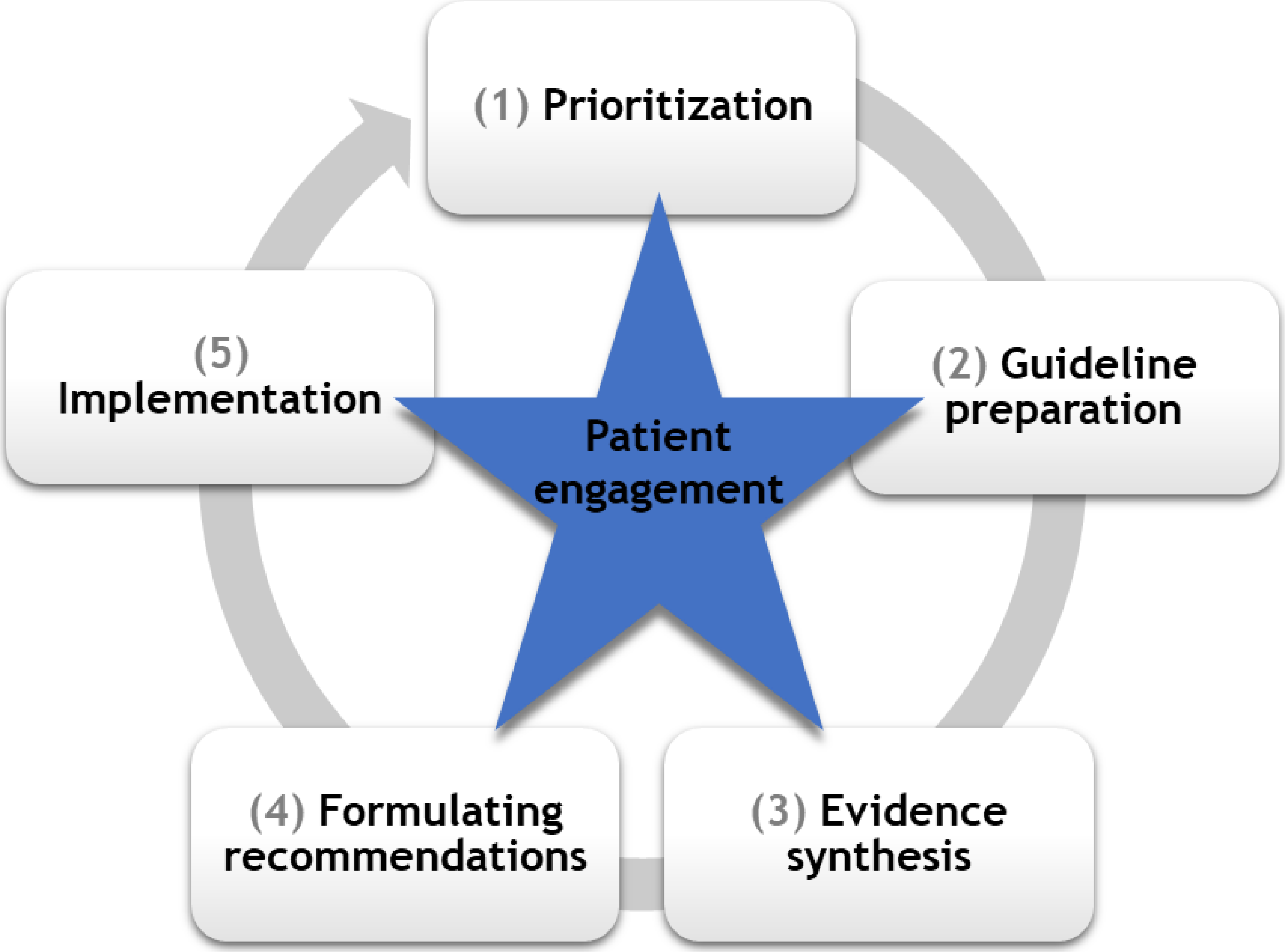
Methodological framework



(1) Prioritization

(2) Guideline preparation

(3) Evidence synthesis

(4) Formulating recommendations

(5) Implementation

Patient engagement

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or low prevalence
complex diseases

Network
Intellectual Disability
and Congenital
Malformations (ERN ITHACA)

Thank you!

2 - Focus WG Teaching & Education

What resources can I rely on, what can we develop as educational support needed, asking your opinion

Laurence Faivre, Giuzeppe Zampino, Anne Hugon

European
Reference
Networks

ITHACA

Introduction, Objectives WG T& T

- Chair: Laurence Faivre (Dijon – France), Co-chair: Giuseppe Zampino (Roma – Italy) PM in charge Anne HUGON
- Perspectives 2023, Different level of Training
  - More active participation from the Patient Council
  - How to increase the interest on rare disease?
  - How to increase the knowledge on genetic, biology and treatment prospective of RD?
  - How to increase the knowledge on different aspects of disability?
  - How to increase the medical competence?
  - How to better understand RD for children and parents?

High expertise training, professionnals

Training and deepening, all audiences, topics

Educational Ressources & Tools

Information and awareness,

all audiences

Patient awareness training & shared experiences,

all audiences

Activities 2022

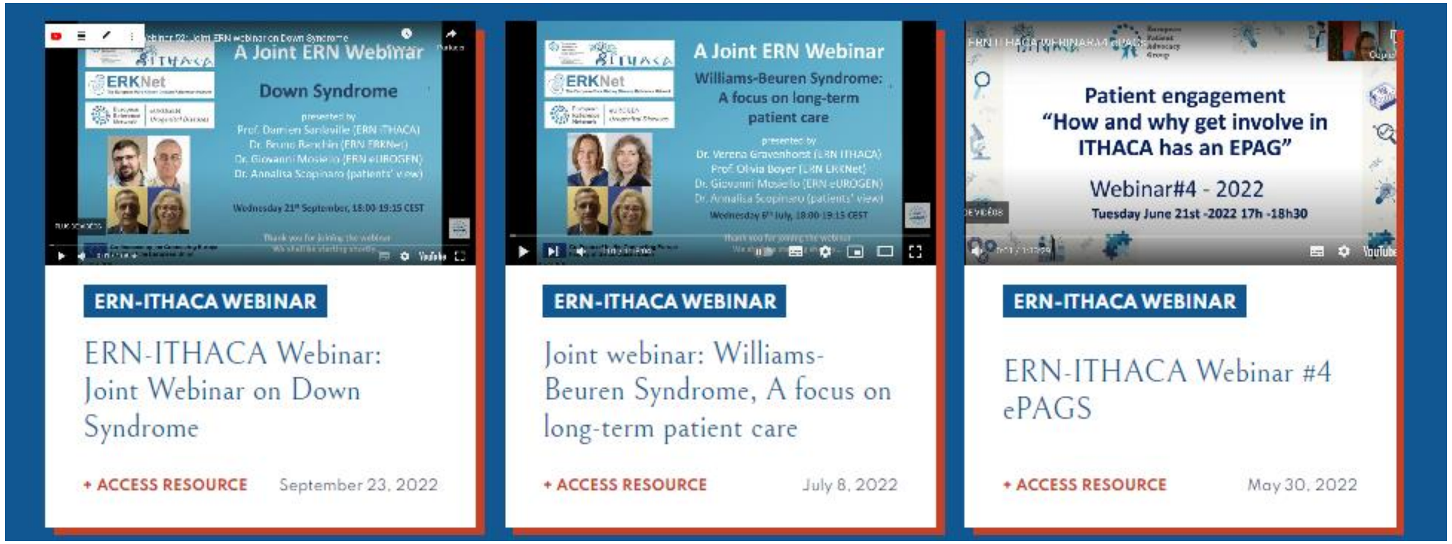
- EuroDysmorpho 2022 - The 32nd held in Barcelona
  - 14 to 17 September 2022 : 91 participants actifs (110 total)
  - Save the date 2023 Lisbonne : 13-16 septembre 2023 (Ljubliana 2024)
- MOOC BiG / EJP RD - "Diagnosing RD: from the Clinic to Research and back
- European Master in Genetics - Pilot 2023 P LAPUNZINA (Madrid, Marseille, Rome)
- Webinars series to be continued
- Syndromic movies and movie on fetopathological examination (2022- WG)
- Production of Orphanet files
- Resources available ITHACA WEB SITE
- Collaboration with - ESHG Young Geneticists ESHG-Y (EuroDysmorpho, UNIQUE)
- From WG Fetal Medecine - Winter School dec 2022 Multidisciplinary Fetal Diagnostics

Encourage mixed training

- Professionals/patients/relatives (strengthening the knowledge of patients and families)
  - Educational Ressources on web site (Syndrome moovies syndromes, Genetics researchs ….and vidéo; Cartoons; Iconographies https://ern-ithaca.eu/educational-resources/
  - Patients/publication : PO & YGN Unique Proofreading Rare Chromo; PO & Patient Journey and other (validated by WGTT publications)
  - Webinars from the Patient Council : #1 Integrated Care at NoRo Center; #2 10 years of actions in Integrated NORO center
  - Webinars, developed by the PC : Transition of care (Thomas and Kasia); Quality of Life (Dorica), Mature ERNs (Eurordis)
  - Promoting the inclusion of patient’s history in the educational process

Webinars series # 2022

https://ern-ithaca.eu/documentation/educational-resources



A Joint ERN Webinar

Down Syndrome

presented by
Prof. Damien Sanlaville (ERN ITHACA)
Dr. Bruno Ranchin (ERN ERKNet)
Dr. Giovanni Mosiello (ERN eUROGEN)
Dr. Annalisa Scopinaro (patients' view)

Wednesday 21st September, 18:00-19:15 CEST

Thank you for joining the webinar
We shall be starting shortly

ERN-ITHACA WEBINAR

ERN-ITHACA Webinar: Joint Webinar on Down Syndrome

+ ACCESS RESOURCE September 23, 2022

A Joint ERN Webinar

Williams-Beuren Syndrome: A focus on long-term patient care

presented by
Dr. Verena Gravenhorst (ERN ITHACA)
Prof. Olivia Boyer (ERN ERKNet)
Dr. Giovanni Mosiello (ERN eUROGEN)
Dr. Annalisa Scopinaro (patients' view)

Wednesday 6th July, 18:00-19:15 CEST

ERN-ITHACA WEBINAR

Joint webinar: Williams-Beuren Syndrome, A focus on long-term patient care

+ ACCESS RESOURCE July 8, 2022

Patient engagement
"How and why get involve in ITHACA has an EPAG"

Webinar#4 - 2022
Tuesday June 21st -2022 17h -18h30

ERN-ITHACA WEBINAR

ERN-ITHACA Webinar #4 ePAGS

+ ACCESS RESOURCE May 30, 2022

- Tuesday from 17h – 18h30
- 20 - 70 participants
- From 15/23 countries
- Very Good feed back
- 4 x years from 5 to 6.30 pm

Webinars 2022

- Webinar#4: "How and why getting involved in ITHACA as an EPAG" / Laurence Faivre, Dorica Dan
- V1 eUROGEN " Williams Syndrome and Down Syndrome "(ITHACA, eUROGEN) / Giovanni Mosiello
- V2 eUROGEN " Williams Syndrome and Down Syndrome "(ITHACA, eUROGEN) / Giovanni Mosiello
- Webinar#5: Ithaca Guidelines development: From patients to guidelines and back again" / Agnies van Eeghen

Webinars series # 2023 to be continued

- Webinars For 2023
  - Mars 23rd, 2023 – Webinar : European Initiatives to extend newborn screening (Screen4Care, NBS, NGS) / Laurence Faivre
  - Fall, 2023 - Webinar: Fœtal Therapies and dysraphym surgery / Jean Marie Jouannic
  - Dec 9th, 2023 – Webinar : IA genetic use Best practice / Sofia Douzgou Houge
- Call for proposals ??? Topics + Speakers + Agenda
  - Understanding registers and their relevance to classical studies / Fœtal Therapies and dysraphym surgery / Organisation of Care in EU / Genetic data - Secondary data issued from NGS / IRDIC Task Force / Therapeutic progress, Mini brain model / Guidelines within ERN ITHACA / Care givers / Paramedic webinars
- New Topics, such USE Tools for expert's for pro : ILIAD / Sysndd …

WG T&T perspectives 2023 ? / mentimeter

- New Topics
- Call for proposals ??? Topics + Speakers + Agenda

We need more participation, Welcome aboard

Contact : anne.hugon@aphp.fr

3.1 - Focus WG NDD

Neuro Developmental Disorders Over view on Neuro-developmental Disorders work Group

Tjitske Kleefstra, Anne Hugon

Key Objectives WG NDD - Neurodevelopmental disorders

- Chairs Tjitske Kleefstra, Christiane Zweier
- This workgroup is dedicated to rare genetic intellectual disabilities (ID)
  - To deal with rare genetic NDDs, improve our knowledge and understanding of these disorders, which number in the thousands but are poorly known for the most part.
  - Among the workgroup's deliverables are the development of a web-based rare ID gene database to provide access to information on genetic causes of ID and the organisation of a European congress on neurodevelopmental disorders.

Coordination contact : Anne Hugon (WGNDD, SNW 1,2,3); Jolanda van Golde (EuroNDD 2023, SNW 4)

European Reference Networks

4 SNW + EuroNDD 2023

SNW 1: Sys-NDD database : Create and make accessible an NDD gene database connected to clinical information and interacting with Orphanet.

Dr **Christiane Zweier**

SNW 2: Recommendations of good practice, for the management of children with profound intellectual impairment and multiple disabilities (**PIMD/POLYHANDICAP**)

Dr S Huisman and Dr MC Rousseau

Euro NDD 2023

SNW3: Best practices in management for ID older adults, management of mentally disabled adults and old, launch in june 2021,

Dr S. Miot and Dr L de Graaf

SNW4 NDD WG Clinical genetics & Psychiatry, dealing with ASD, early psychosis and other neuropsychiatric conditions.

PrT Kleefstra, Dr F Degenhardt and Dr C Laurent-Levinson

3.2 World Café

“Clinical genetics & Psychiatry from a multidisciplinary approach, including the perspective of the expert by lived experience”

Tjitske Kleefstra, Claudine Laurent-Levinson, Jolanda Van Golde

Diner

- ePAGs dinner on Thursday night, 19:00,

at the Ypsilon café, located a 20 minutes walk away

Ypsilon Café

Budapest, Stefánia út 11143