



# ITHACA Board Meeting 2020

# 2020 December 10 - 12

# Patient Council Meeting : December 10<sup>th</sup>

14h00 - 17h00

# Patient Council Meeting : December 10th



## Agenda

Time Slot	THEME	SPEAKER	SUBJECT
14h00 -14h05	<b>Organisation</b>	Anne Hugon	Welcome, e-Pag ITHACA's Internal meeting
14h05 -14h30	<b>Patient Council</b>	Dorica Dan, Ammi Sundqvist-Anderson	Agenda & e-Pag Internal business
14h30 -14h45	<b>Eurordis trans - activities</b>	Anne Laure Aslanian	Eurordis activities connected to Ithaca
14h45 -15h00	<b>Work plan for 2021</b>	Dorica Dan	Work priorities for tomorrow
15h00 -15h10	BREAK time / HCPs and Clinicians are invited to join the afternoon session at 15h00		
15h10 -15h30*	<b>ePAG Assessment Framework</b>	Ammi Sundqvist-Andersson	Assessment Framework ePAGs and clinician*
15h30 -15h45	<b>Brainstroming</b>	All attendees	Brainstorm and needs gathering from all ePAGs and clinicians
15h45 -16h00	<b>Transition</b>	Katarzyna Świeczkowska, Tomaz Grybek	Transition on quality of life
16h00 -16h05	<b>Quality of life</b>	Dorica Dan	Introduction on quality of life
16h05 -16h30	<b>Quality of life</b>	Chris Underhill (Guest)	Presentation « the importance of Gardening for disabled people »
16H30 - 16h45	<b>Quality of life</b>	All attendees	Open discussion
16h45 -17h00	<b>What next and Wrap up</b>	Anne Laure Aslanian Anne Hugon	Conclusion

# Welcome e-Pag ITHACA's Internal meeting



- ✓ A "tour de table" for participants
- ✓ Presentation of the ERN ITHACA

ITHACA aims to maintain a high degree of cooperation with European Patient Advocacy Groups (ePAGs) and other National or European lay public or Patients' Organizations

The Patient Council is elected through the process proposed with **Eurordis**

The Patient Council is empowered through formal roles in the governance of ITHACA

*ITHACA Project Manager contact : Anne Hugon [anne.hugon@aphp.fr](mailto:anne.hugon@aphp.fr)*

Coordinating a European-wide network in growth is a challenging task

*Go to our Web site*

*Get the NL, Follow us on twitter....*

## ERN ITHACA WEBSITE

Visit our website: <https://ern-ithaca.eu/>

Please Register here: <https://ern-ithaca.eu/register/> and do specify that you come from a new member HCP, in order to have access to the exclusive Members Area.

This registration also allows you to receive the ERN ITHACA newsletter, and many others news

## ZOOM or TEAMS

**Twitter** Follow us on @ERNIthaca!

## EUROPA WEBSITE

Visit: [https://ec.europa.eu/health/ern\\_en](https://ec.europa.eu/health/ern_en) for general information on ERNs.

## ECP (ERN Collaborative Platform)

Please visit: <https://webgate.ec.europa.eu/ern/>.

You will need an EU login to first access the platform. Then, please **request access** to ERN ITHACA network.

# e-PAG internal bussines



- ✓ Organization and Gouvernance
- ✓ Patients engagement in WGs
- ✓ Leader of HCP
- ✓ WG Leader



# ITHACA organization and governance



**The ExCom** facilitate dissemination of information from EU to the HCP network, updates about Workpackage progress and communication between workgroups.

The members of the ExCom are:

- All Members of the Coordination Team
- Chair and co-chair of all Workgroups
- Chair and co-chair of the Patient Council.

The Executive Committee has 6 bimestrial meetings per year:

## Board of Members

- The Board of ITHACA consists of
- the local Coordinators of full member HCPs,
- the local Coordinators of affiliated members and affiliated hubs,
- the members of the ExCom that are not local coordinators
- 5 representatives of the Patient Council (including the 2 who are members of the ExCom).

In case of a vote, all members of the ExCom, all representatives of full members and the 5 representatives of the Patient Council have a vote.

- ⇒ **The Patient Council is involved in the Governance of all Workgroups**
- ⇒ **The Patient Council hold an online bi monthly meeting**
- ⇒ **The Patient Council hold one annual face-to-face Or WebEx meetings**

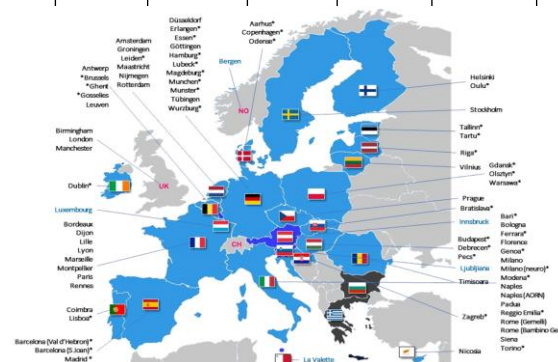




# Patient engagement in WGs



Family Name	Forename	PC /Eurordis	PC Board Elected	WP1	WP2	WP3	WP4	WP5	WP6	WP7	WP8	WP9	WP10	Disorder(s)
Dan	Dorica	ePAG Advocate	Co- Chair	1	1	1		1			1	1		Prader Willi Syndrome & RD
Herman	Carole	ePAG Advocate							1			1		ADNP
Jopp	Renée	ePAG Advocate											1	Spina Bifida
Milne	Yvonne*	ePAG Advocate					1		1					Rett Syndrome
Marinelli	Pietro	ePAG Advocate								1				Smith-Magenis Syndrome
Pogány	Gábor	ePAG Advocate	Elected							1				Williams Syndrome
Routledge	Sue	ePAG Advocate	Elected				1							Pitt Hopkins Syndrome
Sundqvist-Andersson	Ammi	ePAG Advocate	Chair	1	1	1							1	Spina Bifida
Grybek	Tomasz	ePAG Advocate						1			1			Rare diseases
Katarzyna	Swieczkowska	ePAG Advocate									1	1		Rare diseases ID
Daugy	Sandrine	ePAG Advocate												22q11.2 microdeletion syndrome
Koekkoek	Gerritjan	ePAG Advocate												Cornelia de Lange Syndrome
Scopinaro	Annalisa	ePAG Advocate												Williams Syndrome



# WG 1; 2; 3 (Management, Dissemination and Evaluation)



## The Coordination Team / EXCOM Committee

ERN ITHACA Coordinator , Alain Verloes, MD, PhD, medical geneticist

WG 1 Management Coordination of ITHACA	WG 2 Dissemination & communication	WG 3 Evaluation, monitoring process, evaluate, activity reports
Team Coordination Anne	Team Coordination Sarra	Team Coordination Kléa
<b>e-PAG from PC</b> Chair Ammi Sundqvist-Andersson Co Chair Dorica Dan		

### The members of the ExCom

- All Members of the Coordination Team
- Chair and co-chair of all Workgroups
- Chair and co-chair of the Patient Council.



# WG 4; 5; 6 (Guidelines, Telehealth and Registry)



## WG 4 Expert Recommendations

Improve patient care using high-quality guidelines by evaluating existing national guidelines and issuing EU-wide consensus statements

<b>Leading Team</b>	<b>Chair: R. Hennekam (Amsterdam)</b> <b>Co-chair: tbd</b>
<b>e-PAG from PC</b>	Yvonne Milne Sue Routledge

Team Coordination Klea Vyshka

## WG 5 Telehealth and Diagnosis

Increase use of CPMS by ITHACA members to share and discuss patient cases, propose diagnoses and identify ultrarare disorders. Develop ITHACA-specific fields and data-sharing tools in collaboration with CPMS developers.

<b>Leading Team</b>	<b>Chair: S. Dougzou (Manchester)</b> <b>Co-Chair: A. Verloes (Paris)</b>
<b>e-PAG from PC</b>	Dorica Dan Tomasz Grybek

Team Coordination Anne

## WG 6 Registries, Solve RD linked to the ITHACA-specific ILIAD registry

Identify and list biobank-linked registries with potential for collaborative research, and create an ITHACA-wide FAIR registry of pseudonymized patients with rare syndromal and non-syndromal ID disorders to fill gaps in available information

<b>Leading Team</b>	<b>Chair: A. Renieri (Siena)</b> <b>Co-Chair: tbd</b>
<b>e-PAG from PC</b>	Carole Herman Yvonne Milne

Team Coordination Klea Vyshka

# WG 7; 8; 9; 10 (Research, T&T, ID, Spina Bifida)



## WG 7 Research

Set up exchanges of research fellows (trainees are partially funded by ITHACA) to promote sharing of knowledge and research skills across ITHACA, and engage in collaborative research projects between interested HCPs to identify disease genes and mechanisms for molecularly uncharacterized cohorts of patients with an established clinical diagnosis or patients with unclassified phenotypes.

<b>Leading Team</b>	<b>Chair: M. Tartaglia (Rome)</b> <b>Co-chair: tbd</b>
<b>e-PAG from PC</b>	Pietro Marinelli Gábor Pogány
Team Coordination Anne	

**WG 9 Intellectual disabilities diagnosis and management** to deal with rare genetic IDs, improve our knowledge and understanding of these disorders, which number in the thousands but are poorly known for the most part. 2 SNW are created on on Mild ID and one on Profound ID

<b>Leading Team</b>	<b>Chair: T. Kleefstra (Nijmegen)</b> <b>Co-chair: D. Wiczorek (Dusseldorf)</b>
<b>e-PAG from PC</b>	Dorica Dan Carole Herman Swieczkowska Katarzyna
Team Coordination Anne	

## WG 8 Teaching and Training

Set up specific teaching and training programmes targeting different populations - future ITHACA specialists (young geneticists) and non-specialists to teach them about ITHACA-specific disorders (rare ID, congenital malformations) and how to handle such patients. Improve knowledge of disease biology and genetics of rare IDs and developmental anomalies

<b>Leading Team</b>	<b>Chair: L. Faivre (Dijon)</b> <b>Co-chair: J. Clayton-Smith (Manchester)</b>
<b>e-PAG from PC</b>	Dorica Dan Tomasz Grybek Swieczkowska Katarzyna
Team Coordination Anne	

**WG 10 Spina Bifida** a cross-ERN WG on Spina bifida to produce an expert consensus statement for this multisystemic disorder. Harmonisation of the clinical practice across the EU and improve patient care.

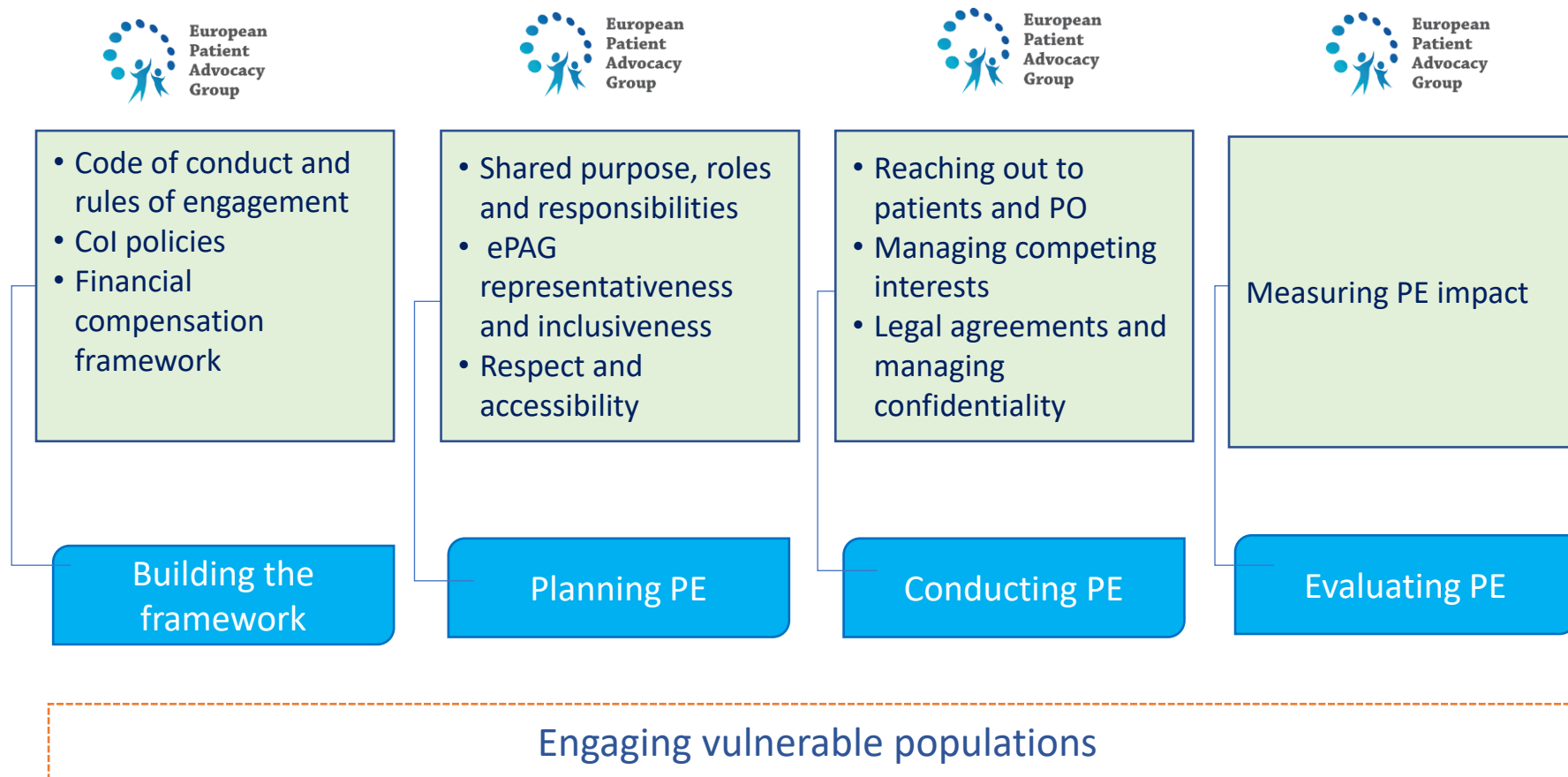
<b>Leading Team</b>	<b>Chair: A. Manunta (Rennes)</b> <b>Co-chair: G. Mosiello (Rome)</b>
<b>e-PAG from PC and lead</b>	Ammi Sundqvist-Andersson Renée Jopp
Team Coordination Anne	

# EURORDIS TRANSVERSAL ACTIVITIES

## Patient Engagement in ERNs



### Patient engagement framework



Source: Paradigm – adapted: <https://imi-paradigm.eu/petoolbox/>



1. Revise/amend the **ITHACA ePAG ToR**
2. **Organise quarterly induction calls on ERNs and ePAGs** and cover governance aspects (individual ePAGs should organise their own to complement)

Building the framework

# Activities 2021 – Planning PE



1. **Continue developing/updating Patient Journeys** as a tool to engage with clinicians on **needs**
2. **Existing 5 Patient Journeys/Common needs:** map the resources developed by ERN ITHACA to cover the gaps in your patient journey
3. Support ePAGs and ERN Project Coordinators to **map the wider ePAG community by disease grouping** (in liaison with the National Alliances)



- 1. Set up transversal ePAG WG on Communiation (Q1 2020)** to organise regular exchanges so that each ePAG can define its own communication strategy. The group will also develop templates, tools and methods to support them in engaging with the wider patient community.
- 2. ERICA, new H2020 project on ERNs Research and Innovation capacity. Coordinate participation in ERICA WPs** where EURORDIS participates through the ePAG Research WG + individual ePAGs  
↳ structuring the engagement of patients in clinical trial design.

Conducting PE

Supporting ePAGs to reach out to patients and PO



1. Finalise the **report on the ePAG Impact Ass Framework pilot**, incl. recommendations (ITHACA ePAG Ammi & Dorica)
2. **Refine the ePAG Impact Ass Framework measures** in collaboration with clinicians (with input from the 3 pilots).
3. Extend to other ePAGs with the ones who are willing and ready to implement it (very few groups)



Measuring PE

# Activities 2021 – Capacity building



## Soft skills

- Launch of the 3rd edition of the **EURORDIS Leadership School** (Q2 2021)

## Conducting and Measuring Patient engagement in ERNs

- All-ePAG annual meeting 2021
- ePAG SC annual meeting
- Quarterly exchange of good practices

Conducting PE  
Capacities/Training

European Union

# PC & WORKPLAN 2021



WP1 - Management Coordination	EXCOM/ board elected members	Proposals Dorica	Proposals Ammi
WP3 - Evaluation	EXCOM/ board elected members		
WP2 - Dissemination	Improve support tools Flyers/booklet/Statement /tailored information /improve orientation to the right services/patients rights/ shorts video	To work together to make available Flyers/booklet/Statement /tailored information /improve orientation to the right services/patients' rights/ short videos.	Yes, all information we can spread is good. Last year I had the opportunity to present the work of the ERN and mostly the work of ITHACA to the leading HCP of Spina Bifida in Sweden. To be able to do so I had to look for info from several web pages. Now when we (the ePAGs) gets a signal to create national groups with both ePAGs and HCP it would be nice to have printed information and maybe a power presentation to use.
WP4 - Expert Recommendations	Provide specific information adapted to patients / in their own language / CDG CDTs reviewing and elaborated with clinician/ lay publications	<p>WP4 - ePAGs could assess if the needs for their patients are fulfilled and search what resources already exist and let ITHACA know about it.</p> <p>Ex. IPWSO has guidelines for PWS and I can ask to use them and mention IPWSO contributions (experts from all over the world contributed to this and some are also translated in many languages). I think that we can also assess if the PO has different other types of guidelines to support patient care and might be useful for others, too. Ex. We have guidelines for integrated care for several diseases (in Romanian), guide of services, etc. ...</p>	I think it is crucial to have a lay version of recommendations/guidelines. Often the patients are experts on their own disease so maybe it needs to be different versions of it. One more simplified for "newcomers" and a second one who has the same info as to the HCP but in a lay version. I'm a strong believer that parents/patients should have all information needed so they can be in charge of their own disease and treatment and the parent/patient is the one who decide what info they need. Maybe we could have a parent/patient for each recommendation who describes their life with a certain diagnosis. We should aim for having info in their own language but I think that will be hard but we could at least have it in the main languages such as English, French, German and Spanish....
WP5 - TeleHealth and Diagnosis CPMS	Empowerment input through CPMS / indicate patient case to be discussed /application to help care	WP5 - For sure the most important thing is to develop the communication with CoE and experts to propose cases for CPMS. Maybe to stimulate Helplines where they exist. Training and induction of new ePAGs.	I have a feeling that the CPMS is a little bit over our head today. I don't think it is enough that the communication is only between the HCP's. I hear of so many cases where the patient don't get the help needed because a HCP is reluctant to ask for feedback from someone else. I think a patient should have a possibility to contact an expert themselves but that is not possible through CPMS, they say. Maybe that would be possible though TeleHealth.

# WORKPLAN 2021



WP6 - Registries	Improve data collect on RD (Génida)/ patient enrolment	WP6 - WP7- Patients can contribute to data collection and survey (not only for implementation but also to design the questionnaire) but I guess that they can also contribute to the Ethics committee for Registry and Research, etc.	
WP7 - Research	Patient expertise through survey to be published	WP6 - WP7- patients can contribute to data collection and survey, ethics committee for Registry and Research, etc.	
WP8 - Teaching and Training YGG	Therapeutic education / workshop or seminaire for patients / transition / Quality of life QoL/ Patients needs	Therapeutic education / workshop or seminars for patients / transition / Quality of life QoL/ Patients needs. I would also propose webinars for sharing best practices of ePAGs' organizations they belong to and how they directly support their patient community, how do they spread the information about ITHACA & ERN at national level or in their community.	Adding Patient lead workshops/webinars for HCP to all comments.
WP9 - Intellectual Disability	Tools for care following / Polyhandicap/ ID / reviewing documents guide leaflet	WP9 - As many of our diseases have no specific treatment yet and produce polyhandicap and ID maybe a webinar on rehabilitation/ habilitation programs available would be interesting and we could invite RareResourceNet _ European Network of Resource Centers to present their services. For sure these services could support us in the process of reviewing documents guide leaflets.	No comments

# WORKPLAN 2021



WP9 - Intellectual Disability	Tools for care following / Polyhandicap/ ID / reviewing documents guide leaflet	WP9 - As many of our diseases have no specific treatment yet and produce polyhandicap and ID maybe <b>a webinar on rehabilitation/ habilitation programs</b> available would be interesting and we could invite <b>RareResourceNet _ European Network of Resource Centers to present their services</b> . For sure these services could support us in the process of reviewing documents guide leaflets.	No comments
WP10 - Spina Bifida	co develop Expert consensus statement on spina bifida / interERN	SB	Yes. Ongoing work. <b>Needs more structure in my opinion</b> (but I'm a Swede and we like our kind of structure)
WG PC Transversal activities	Management of PC/ Projects develop with EURORDIS + others /patient journey/commons needs/ethic's/ Impact Assess Framework/ monitoring	WP11 - I guess that the ideas mentioned here cover most of what we do and should do.	

# WORKPLAN 2021 – priority propositions



1. **Make available tailored information /improve orientation to the right services/patients' rights/ short videos**
2. **Create national groups** with both ePAGs and HCP and share ERN expertise at national level
3. **Map the existed resources to achieve our goals**
4. **Continue work on Patient Journeys** for other diseases – translate
5. **Facilitate a better communication between CoE and patients** and stimulate Helplines where they exist and propose difficult cases for CPMS;
6. **Contribute to data collection and survey**, ethics committee for Registry and Research, etc.
7. **Organize webinars to share best practices:** therapeutic education / therapies/ transition / Quality of life QoL/ Patients needs/ rehabilitation/ habilitation programs available;
8. **Share best practices between** ePAGs' organizations; Management of PC/ Projects develop, ethic's/ Impact Assess Framework/ monitoring
9. **Invite RareResourceNet** - European Network of Resource Centers to present their services;
10. **Training on different topics:** collective impact & shared leadership (both for ePAGs and experts)



# WORKPLAN 2021 – ePAG priorities & Forward-looking projects



## • Future objectives & projects

- What's needed? Responsibilities, involvement in WG
  1. Priority n° 1
  2. Priority n° 2
  3. Priority n° 3
  4. ....

ePAG Activities 2021	Content	ePAG Leads	Timeline

*Do not hesitate to send your feed back  
Thankyou for your participation – PM contact [anne.hugon@aphp.fr](mailto:anne.hugon@aphp.fr)*

# ITHACA Board Meeting 2020

## Patient Council meeting



Time Slot	THEME	SPEAKER	SUBJECT
<b>15h00 15h10</b>	<b>BREAK time / HCPs and Clinicians are invited to join the afternoon session at 15h00</b>		
<b>15h10 -15h30*</b>	<b>ePAG Assessment Framework</b>	Ammi Sundqvist-Andersson	Assessment Framework ePAGs and clinician*
<b>15h30 -15h45</b>	<b>Brainstroming</b>	All attendees	Brainstorm and needs gathering from all ePAGs and clinicians
<b>15h45 -16h00</b>	<b>Transition</b>	Katarzyna Świeczkowska, Tomaz Grybek	Transition on quality of life
<b>16h00 -16h05</b>	<b>Quality of life</b>	Dorica Dan	Introduction on quality of life
<b>16h05 -16h30</b>	<b>Quality of life</b>	Chris Underhill (Guest)	Presentation « the importance of Gardening for disabled people »
<b>16H30 - 16h45</b>	<b>Quality of life</b>	All attendees	Open discussion
<b>16h45 -17h00</b>	<b>What next and Wrap up</b>	Anne Laure Aslanian Anne Hugon	Conclusion

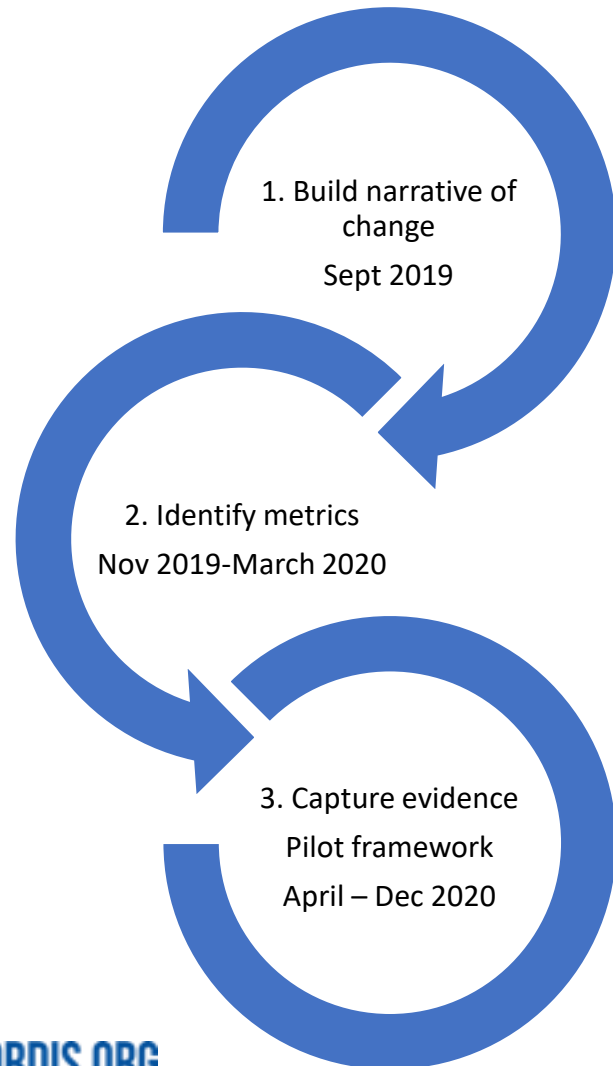
Why do we want to measure the impact of patient engagement in ERNs?

1. Understand **WHAT** our role is and what are our goals. To measure impact, we need first to define very clearly what are we are trying to achieve (narrative of change)
2. Identify **HOW** to achieve our goals and focus on what's important. Once our goals are clear, we can think of the activities that we need to undertake to progress towards these goals.
3. Explain the **VALUE** of our involvement in the ERNs and provide measures to assess the progress overtime.

Learning  
(internal  
audience/ePAG  
advocates, Network  
Coordinators and  
clinical leads)

Accountability  
(external  
audience/EC  
BoMS and other  
ERN  
stakeholders)

# What have we done?



## 1. Build narrative of change

### F2F workshop with ePAG Steering Committee

- Workshop with 20 participants from different ERNs (all patient representatives)
- Following the methodology developed by NHS England to measure impact of health programmes, we covered in the workshop step 1 out of the 4-steps included in this methodology:
  - **Step 1: Articulate what will change, how and why**
  - Step 2: Capture evidence
  - Step 3: Review the evidence
  - Step 4: Share your impact with others

## 2. Logic model used to capture Narrative of Change

### 3. Results of the workshop

1. Equity of access to quality care across Europe for rare disease patients
2. Improve quality of care, diagnosis and treatment of rare diseases
3. Ensure patient-centric ERNs that meet our community needs – the needs of patient community steer ERNs activities and their structure
4. Ensure a good level of awareness among the patient community about ERNs and their work (so that all can benefit)

\*Based on the [NHS Impact Framework](#) and the PARADIGM Monitoring and Evaluation of Patient Engagement in the medicines development lifecycle

# Patient perspective

## Assessment framework measures



	Num.	Measures	Questions that is asked
<b>Goal 1: Improve quality of care, diagnosis and treatment</b>	<b>1</b>	Listed as co-authors of consensus statements and Clinical Practice Guidelines	Title of the paper? Nr of meetings in which ePAG participated? Nr of documents reviewed? How many documents were published with your name?
<b>Goal 2: Ensure patient-centric ERNs that meet our community needs - Needs of patient community inform all ERN activities and their structure (for example, ERN covering in equal terms adult and paediatric care, MDT, etc)</b>	<b>2</b>	Percentage of ERN WGs with ePAG advocates participating as members	Are you participating in any ERN WG? Are you involved in the ITHACA board? How many meetings have you been invited to and how many have you attended?
	<b>3</b>	Patient representatives are members of the ERN Registry governance structure and/or ERN Registry Working Group (Yes/No)	Are you a member of the ERN Registry group? Were you involved in the Registry governance structure?
	<b>4.1</b>	Number of patient journeys or surveys to capture patients' needs that have been discussed with ERN clinicians	Did you conduct patient journeys and/or surveys to capture patients needs AND discussed it with ERN clinicians?
	<b>4.2</b>	Percentage of sub-thematic areas where patient needs have been captured through a Patient Journey (or a survey on patients') and results have been discussed with ERN clinicians	
	<b>5</b>	Perceptions or level of satisfaction on how ePAG advocates and clinicians are working together as a team to advance the ERN goals	Subjective question. Are you satisfied working as a team with the clinicians?
	<b>6</b>	Percentage of outcome measures identified with the input of ePAG advocates	Have you participated in the development of outcome measures? (PREMS or PROMS)
	<b>7</b>	Number of posters/presentations on ERNs delivered in national or international meetings (conferences and workshops)	How many posters/presentations on ERNs did you deliver in national or international meetings?
<b>Goal 3. Ensure a good level of awareness among the patient community about ERNs and their work (so that all can benefit)</b>	<b>8.1</b>	Perception on how the ePAG has organised the outreach to its wider patient community (DISSEMINATION)	How often do you disseminate information on your ERN activities to your wider patient community? If never or rarely, Why?
	<b>8.2</b>	Perception on how the ePAG has organised the outreach to its wider patient community (CAPTURING FEEDBACK AND PATIENTS NEEDS)	How often do you reach out to your wider patient community to capture feedback and patients' needs to report back to your ERN? If never or rarely, Why?

# ASSESSMENT FRAMEWORK SURVEY



QUESTIONS	YES	NO	COMMENTS
Are you participating in any ERN WG in your role as an ePAG advocate?	5	2	<p>3 working groups has invited ePAGs to their meetings, in 1 of them an ePAG sends out the invitations</p> <p>4 working groups are NOT inviting ePAGs to their meetings</p> <ul style="list-style-type: none"> <li>- Would have been interested to attend meetings but I assume they did not feel I could make a contribution.</li> <li>- We were not invited to any workgroup meetings.</li> </ul>
Are there any Clinical Practice Guidelines or Clinical Decision Making Support Tools were developed by the ERN and published with your name?	1	6	<p>1 is acknowledge as co-author.</p> <p>1 is under development and not yet published.</p>
Are you involved in the ERN ITHACA Board?	4	3	
Are you a member of the ERN ITHACA Registry working group?	1	6	-Most patients/parent groups I know of would be grateful if we could have a better idea of prevalence even if that was the only thing we could have.
Are you a member of the ERN ITHACA Registry governance structure?	2	5	-I have contributed on the work for application for funding of the ERN Registry with the patients perspective
Did you conduct patient journeys and surveys to capture patients needs and perspectives?	5	2	-We might consider of creating as many patient journeys as possible, involving patients and wider community and maybe, to reassess the common needs and integrate/update them in the WG



# ASSESSMENT FRAMEWORK SURVEY



Subjective questions	Never/ Strongly disagree	Rarely/ disagree	Often/ Agree	Every time/ Strongly agree	Comments
I'm satisfied working as a team with the clinicians of my ERN to advance the ERN goals?		2	4	1	-I would like to know more about what ways we can help with "patient" input. -Improvement on communication with respect to expectations and setting goals forward.
How often do you disseminate information on your ERN activities to you wider patient community?		4	3		-I use to present the ERN at all our events at national level; Centers of expertise, National Alliance members, Europlan Conference, RDD and the RD school. -Very difficult to organise a wide communication, most of the associations do not know nothing about ERN.
How often do you reach out to your wider community to capture feedback?		3	2	2	-Capture feedback and patient needs is a continues process for our organisation

*Results of the survey / discussion*

**Questions?**

**Comments?**

**Recommendations?**

## **Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop.**

KATARZYNA ŚWIECZKOWSKA, TOMASZ GRYBEK

# Exploring transition



**Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop.**

## **32nd European Academy of Childhood Disability Annual Meeting.**

- The EACD is an academic association of professionals working with children with disability throughout Europe.
- At present the academy has over 500 members who are paediatricians, neuropaediatricians, physiotherapists, occupational therapists, speech therapists, psychologists, nurses, social workers and special-needs teachers.
- Over 35 countries are represented.
- The main goal of EACD is to promote teaching and facilitate sharing of information, research and good practice.
- 32nd European Academy of Childhood Disability Annual Meeting - Poland
- Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop

# Exploring transition



**Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop.**

**32nd European Academy of Childhood Disability Annual Meeting.**

## **Goals:**

1. Allow families to gain and share information in relation to transition.
2. Allow professionals to share experience about transition.
3. Allow parents and the people concerned to share and contrast their views with those of professionals reciprocally to improve practice and understanding of the transition process.
4. Improve knowledge and favor appropriate decisions for the welfare of people living with disabilities and facilitate the transition into adulthood

# Exploring transition



**Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop.**

**32nd European Academy of Childhood Disability Annual Meeting.**

## **PART 1**

Users and professionals separately:

Most common concerns of

- people in transition
- family of people in transition
- professionals involved with transition
- Transition from children's to adults health care system ( lack of path for smooth transition, little knowledge on rare diseases)
- Services are not family centred
- Discrimination and lack of participation and inclusion
- Lack of respect for individual autonomy including the freedom to make one's own choices, and independence of persons
- Lack of money
- Sexuality
- Lack of proper support in COVID-19 pandemic



# Exploring transition



**Exploring transition from childhood to adulthood through co-working between families and professionals to enhance practice and participation – workshop.**

**32nd European Academy of Childhood Disability Annual Meeting.**

## **PART 2**

Users and professionals together

Solutions:

- Preparing a path for smooth transition from children's to adults health care system ( patients' passports, coordinated health care)
- Family/person centred approach in providing services, building users – professionals partnership
- Promoting personal assistance
- Promoting inclusion and participation
- Circles of support approach
- Educating users and professionals on persons with disability sexuality
- Sharing of information, research and good practice in supporting people with disability in COVID-19 pandemic

# Exploring transition



Rare Disease Centre of Gdansk – Conference „Rare Disease – Together into adulthood”

## Basics:

2 Days online event: **1st Day:** Conference (7 presentation & Speakers ) | **2nd Day:** Workshop „From Childhood 2 Adulthood”

## Workshop stats:

**26 participants:**

- **13 Users** (RD Patients & Caregivers representing **8 different RDs**).
- **13 Professionals** (HCP, Psychologists, Pedagogics, Physiotherapists, Authorities Officers).

**6 Moderators** (ePAG Adv., HCP, Therapist, Special-needs teachers, Med. Univ. Student) of **6 Workshop Groups**.

**4 hours via Zoom**

## Rare Disease Centre of Gdansk – Conference „Rare Disease – Together into adulthood”

### Main Challenges:

- Deficit of autonomy in everyday life
  - Limits of disabilities - self acceptance and empowerment
- Growing with age lack of understanding in social, educational, professional environment
  - Nascent sexuality of persons with disabilities
- Transition of care – independence & subjectivity vs. limits & challenges of HCS (shortage of time, lack of care coordination, limited knowledge and access to expertise network outside big metropolis)
  - Disease–Life Balance (pursuit of reaching golden mean in daily life)

## Rare Disease Centre of Gdansk – Conference „Rare Disease – Together into adulthood”

### Key Solutions:

- Lifelong assistance & support of RD patients
  - Psychological systemic support for persons and his family in the concept from the diagnosis till adulthood
- Social Awareness & Inclusion Policies – campaigns, education on all levels, small scale initiatives „me & my ecosystem”
  - Sexual Awareness Educational Programs & Campaigns
- Coordinated, networked, IT supported, patient-centred specialised health care implemented by evolution not revolution
  - Holistic empowerment & life companionship – cooperation of all stakeholders on all levels

# Exploring transition

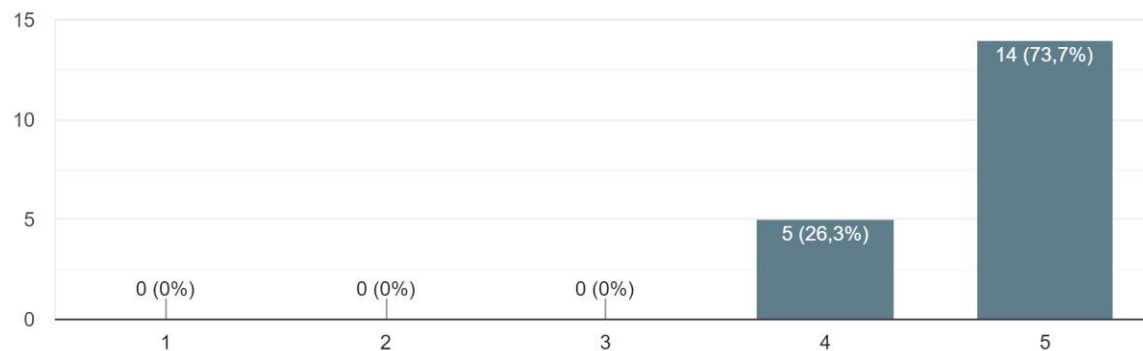


Rare Disease Centre of Gdansk – Conference „Rare Disease – Together into adulthood”

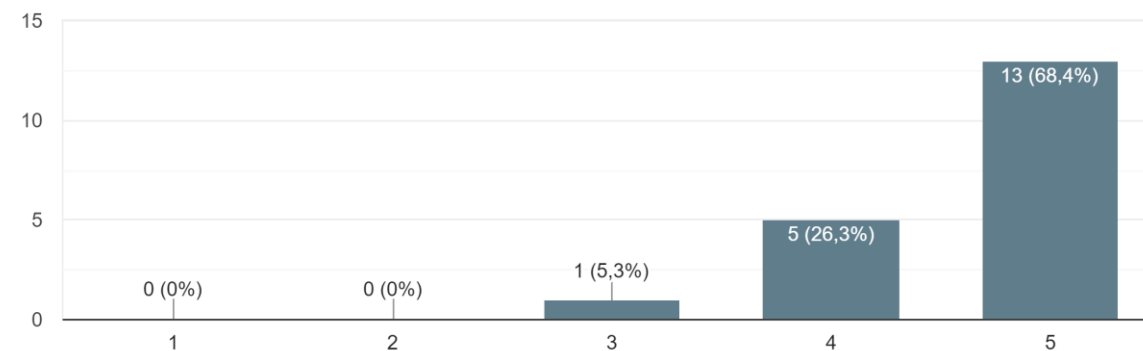
## Evaluation – early conclusions:

**Basics:** anonymous online survey, response rate: 73% (47% Professionals, 53% Users):

Rating of organizational level

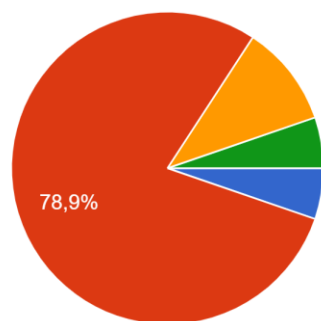


Rating of substantive level



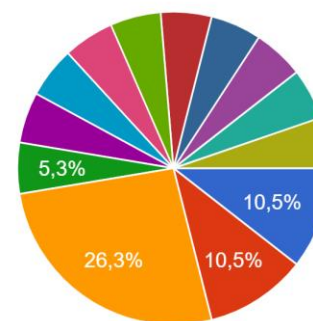
### Evaluation – early conclusions:

Which part of workshop was the most valuable?



- Praca w grupie jednorodnej (tylko OzN/ Rodzice lub tylko Profesjonaliści)
- Praca w grupie mieszanej (połączone grupy tylko OzN/Rodzice i Profesjonalistów)
- Sesja plenarna podsumowująca pracę w grupach
- Wszystkie części były dla mnie wartościowe

Which part of workshop was the least valuable?



- Praca w grupie jednorodnej (tylko Uży...
- Praca w grupie mieszanej (połączone...
- Sesja plenarna podsumowująca pracę...
- brak
- brak najmniej wartościowych
- wszystkie sesje były bardzo cenne
- trudno powiedzieć
- nie było takiej części warsztatów

▲ 1/2 ▼

Rare Disease Centre of Gdansk – Conference „Rare Disease – Together into adulthood”

## Evaluation – early conclusions:

### Substantive aspects:

- Important & current topic
- Great concept of mixed groups: bilateral perspective on challenges & solutions
  - Next step challenge – how effectively and efficiently implement solutions?
    - conference & workshop concept should be continued and repeated
- Key success factor lays in comprehensive, diverse selection & allocation of participants

### Technical aspects:

- Shortage of time
  - Moderation and taking notes – room for improvement
- Limits of allocation – the less diverse the less comprehensive approach

# ITHACA Board Meeting 2020



**Thank you for your attention!**

**Katarzyna Świeczkowska | [katarzyna.swieczkowska@psoni.gda.pl](mailto:katarzyna.swieczkowska@psoni.gda.pl)**

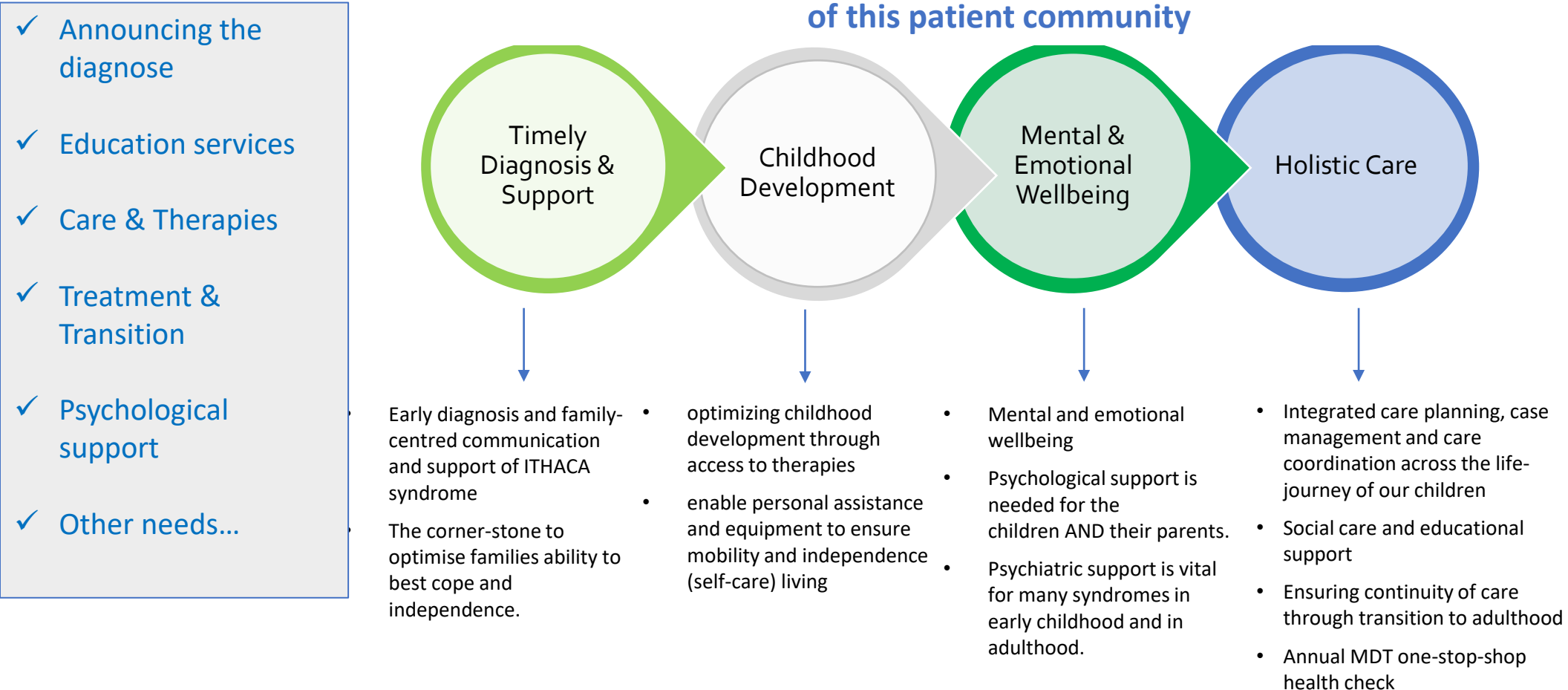
**Tomek Grybek | [tomek@fundacjabb.pl](mailto:tomek@fundacjabb.pl)**



# Quality of life ➡ Quality of life



Genetics is the key for many ITHACA syndromes however it is only one aspect of child and ERN ITHACA needs to go beyond the genetic diagnosis and think about the holistic needs of this patient community

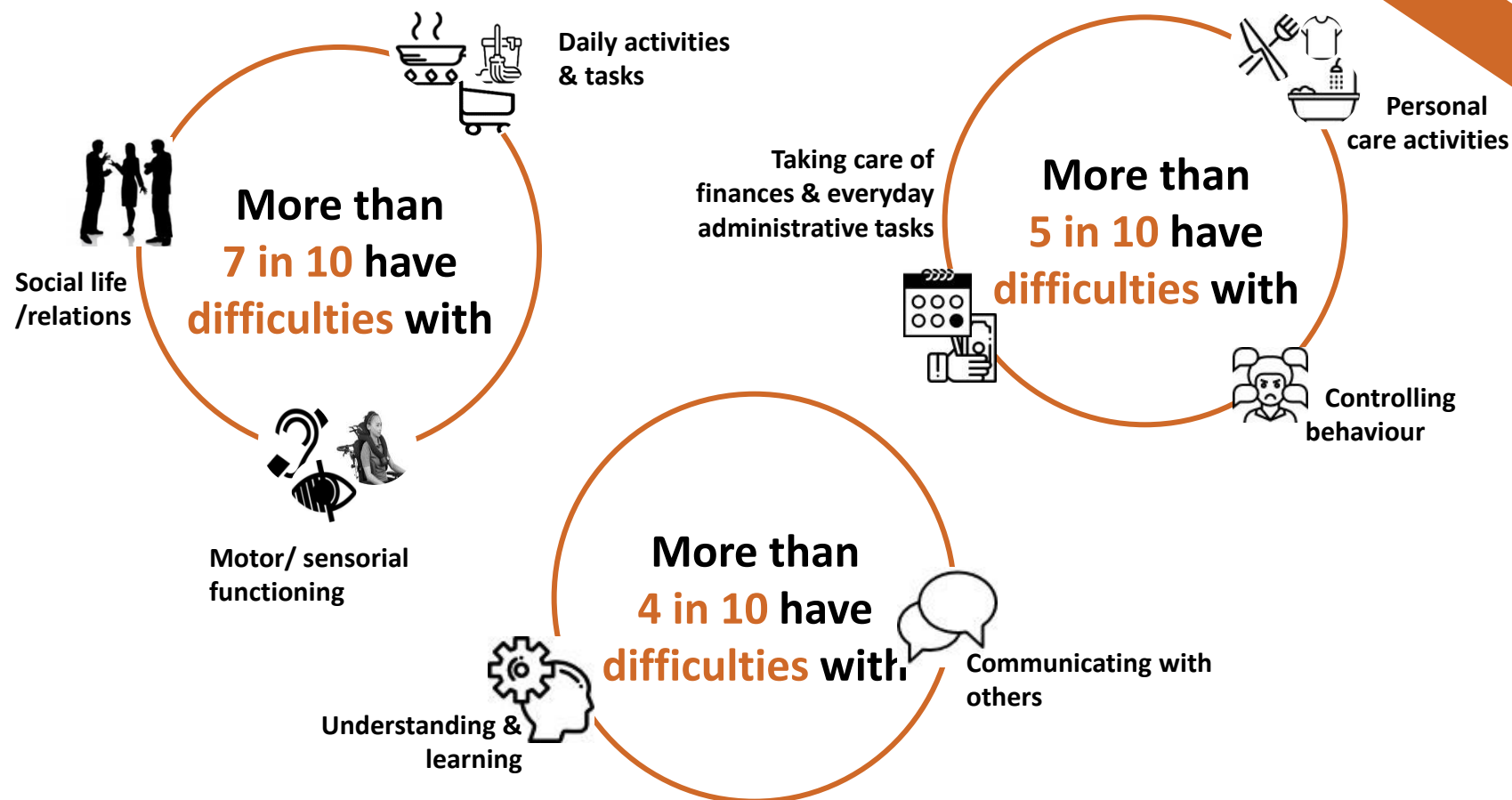


# QUALITY OF LIFE – DEFINITIONS AND REALITY



**WHO:** The degree to which an individual is healthy, comfortable, and able to participate in or enjoy life events.

**Collins Dictionary:** “A state of complete physical, mental and social well-being, not merely the absence of disease or infirmity.”



# INNOVCare project & QoL for RD patients



**Goal:** *to explore the social needs of people living with a rare disease and their carers, specifically by:*

1. Assessing **the impact of the rare disease** on mental, social and physical functions;
2. Evaluating rare disease **patients access to social and healthcare services;**
3. Exploring **challenges and difficulties surrounding coordination of care;**
4. Evaluate the **economic burden related to the rare disease;**
5. Assessing **the impact of the rare disease on employment and job careers;**
6. Exploring **the impact of the disease on family life and well-being.**

Quality of life is highly subjective!!!

*The concept of quality of life is very subjective to the extent to which one believes they are living a good life. Many things, including how someone perceives things or how they see the world, colour this belief.*

# Health is wealth



*RD patients need a lot of help and support, so they need professional assistance (physical, mental, emotional, financial, etc.).*

*QoL is like a summary of your entire life:*

- *Is it good or bad?*
- *Do you have enough money?*
- *Are you eating well and healthy?*
- *Can you afford to buy what you need?*
- *Are you happy?*
- *Do you have people to support you?*
- *How stressed are you?*
- *Do you ever feel depressed or unable to engage in daily activities of life?*
- ...







# How do we measure quality of life?



- **Quality of life measures** are designed to enable *patients' perspectives on the impact of health and healthcare interventions* on their lives to be assessed and taken into account in clinical decision making and research.
- **Some patients cannot complete quality of life measures** because they have cognitive impairments, communication deficits, are in severe distress, or because the measures are too burdensome.
- The list of indicators that was set up by Eurostat with the help of this Expert Group can be found on the [dedicated section on Quality of life](#).  
([The 8+1 dimensions of quality of life](#))
- Based on academic research and several initiatives, the following 8+1 dimensions/domains have been defined as an overarching framework for the measurement of well-being.
- [Material living conditions \(income, consumption and material conditions\)](#)
- [Productive or main activity](#)
- [Health](#)
- [Education](#)
- [Leisure and social interactions](#)
- [Economic security and physical safety](#)
- [Governance and basic rights](#)
- [Natural and living environment](#)
- [Overall experience of life](#)



# Thank you!



# Quality of Life / Presentation « the importance of Gardening for disabled people »



## *Little Bio*

Chris Underhill

Works as a social entrepreneur in the field of quality of life and health, he is a mentor and a thinker in the field of community and development.

He has developed a number of social enterprises, for example Thrive, an organisation working in gardening, disability and community ([www.thrive.org.uk](http://www.thrive.org.uk)) and ADD International ([www.add.org.uk](http://www.add.org.uk)), working in the developing world with disabled people creating systems of representation, advocacy and policy creation. He was recognised by the Skoll Foundation and Schwab Foundation for his work in global mental and he is a Senior Fellow of Ashoka.





# Quality of Life

Chris Underhill MBE  
10<sup>th</sup> December 2020

# Contents



- Thrive
- The importance of gardening for disabled people
- Being in touch with nature
- Being productive
- Helping to create a special safe environment
- The importance of gardening in being still (a type of meditation)

- Started in 1978
- The first customers were long stay hospitals
- Occupational therapists were the first professional customers

# Potting up/physical dexterity



# The importance of gardening for disabled people



- Socialisation
- Satisfaction
- Integration



# Teamwork



# Being in touch with nature



- How plants work
- How the earth works
- Nature – a spiritual experience



# Being in touch with nature





# Being productive



- The story of Robert
- Early days
- Work in the horticultural industry

# Helping to create a special safe environment



- Safety
- Co-operation between various bodies (eg, local authority parks and gardens)
- Keeping clean / enjoying the mud

# The importance of gardening in being still



- A type of meditation
- Weeding
- Integration: being social, teamwork, nature



# HRH Princess Alexandra Patron of Thrive





# A special award



**European  
Reference  
Network**  
for rare or low prevalence  
complex diseases

**Network**  
Intellectual Disability  
and Congenital  
Malformations (ERN ITHACA)

- [chris@chrisunderhill.com](mailto:chris@chrisunderhill.com)
- [Thrive](#)
- [Thrive: an interview 40 years on](#)

Open discussion and next steps?

## *What next and Wrap up*

- Next meeting
- Points to summarize for the plenary session



**Rare Barometer survey** on the future of your rare disease (closes on 3 January 2021) [You can access the online survey here](#)

*Thank you for your participation, do not hesitate to send your feed back*  
PM contact Anne.hugon@aphp.fr