Towards a Rare Brain Disease Ecosystem

Rare Disease Day 2025 "More Than You Can Imagine"

University Foundation, Brussels

20 February 2025 | 09:30 - 14:00

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European Brain Council

Session 1

Welcome & Opening

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European Brain Council

Welcome

Suzanne Dickson
President, European Brain Council

Elena Moro
President, European Academy of Neurology

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European Brain Council

Welcome

Objectives:

- Refer to Rare Disease Day 2024 that introduced the concept of a **Rare Brain Disease Ecosystem**; based on positive feedback, decision was taken to proceed with the concept and crystallize the idea in a programmatic paper that during the 20 February 2025 will be discussed;
- **Host discussions on RBDs such as Myasthenia Gravis, Rare Epilepsies, Neurofibromatosis type 1, stroke and rare cerebrovascular diseases** drawing on insights from ongoing research and clinical practice;
- Discuss the consultation paper on **Towards a Rare Brain Disease Ecosystem & Knowledge Hub**.

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Welcome

Agenda

9:30 - 10:00 Registration & Welcome Coffee
10:00 - 10:10 Welcome & Opening
10:10 - 10:30 Telling the Story
10:30 - 11:00 Session 1: Policy Commitment to Tackling Rare Diseases
11:00 - 11:30 Coffee Break
11:30 - 12:20 Session 2: Unmet Needs and Optimising Patient Care Pathways for Rare Diseases in Europe
12:20 - 12:50 Session 3: Towards a Rare Brain Disease Ecosystem & Knowledge Hub: Open Discussion
12:50 - 13:00 Wrap Up and Closing Remarks
13:00 - 14:00 Networking Lunch

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Welcome

- **Our ambition for Rare Disease**
- **Why do we need a Rare Brain Disease Ecosystem?**

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Welcome

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President, European Brain Council

Elena Moro
President, European Academy of Neurology

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Telling the Story

"Waiting for Zorro", the story of a child with a rare brain disease and related severe disabilities

Sarah Moon Howe
Filmmaker

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European Brain Council

Telling the Story

"Waiting for Zorro", the story of a child with a rare brain disease and related severe disabilities

Sarah Moon Howe
Filmmaker
Contact: sarah@sarahmoonhowe.com

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Policy Commitment to Tackling Rare Disease

Alexandra Heumber Perry, *Chief Executive Officer, Rare Disease International (RDI)*

Marzena Nelken, *Director, Polish National Forum for the Treatment of Rare Diseases ORPHAN*

Valentina Bottarelli, *Public Affairs Director & Head of European Advocacy, EURORDIS*

Enrique Terol, *Health Counsellor, Permanent Representation of Spain to the EU*

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Policy and Action on Rare Diseases: The Patients Angle

European Brain Council

Towards a Rare Brain Disease Ecosystem
Rare Disease Day 2025 Theme
"More Than You Can Imagine"

Scale of the Challenge

30 million people in Europe
300 million globally

National, European, and Global Plans

- Operational
- Funded
- Clear, measurable objectives

Collaboration Involvement of:

- Institutions / Policymakers
- Experts
- Healthcare managers
- Patients

Coordinated Governance

- Regional, national, European, global alignment
- Cross-border cooperation
- Sustained political commitment

Performance Indicators

- Measurable targets
- Progress tracking
- Accountability

- ✓ **Alexandra Heumber Perry**, Rare Diseases International
- ✓ **Valentina Bottarelli,** EURORDIS
- ✓ **Marzena Nelken,** Alliance RD Poland

Reflection Questions:

- ➢ How do we drive ambitious and realistic policy commitments?
- ➢ How do we ensure effective implementation of strategies?

Policy Commitment to Tackling Rare Disease

Alexandra Heumber Perry, *Chief Executive Officer, Rare Disease International (RDI)*

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Coffee Break

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Session 2

Unmet Needs and Optimising Patient Care Pathways for Rare Brain Diseases in Europe

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Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

The Patient Perspective

Lutgarde Allard,
President, European Myasthenia Gravis Association (EuMGA)

Vera Lipkovskaya,
Public Policy and Project Manager, Neurofibromatosis (NF) Patients United

Vinciane Quoidbach,
Research Project Manager, European Brain Council

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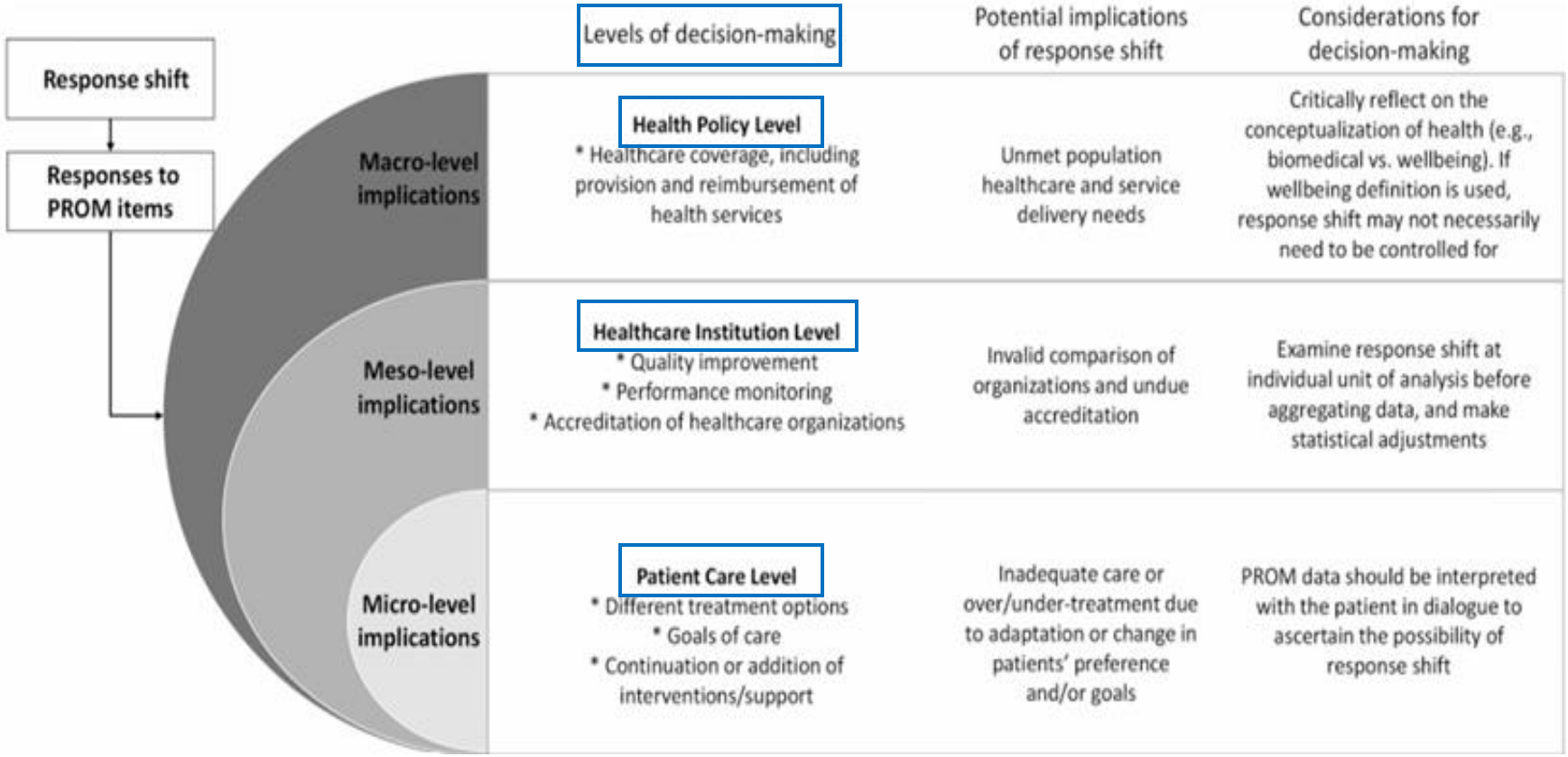
Unmet Needs and Optimising Patient Care Pathways

Main Topics for Discussion:

- Common Challenges across Rare Brain Diseases
- Key Unmet Needs in Patient Care
- The Importance of Optimizing Care Pathways (Use of the RarERN Path Methodology and PROMs)
- Solutions and Recommendations



Qual Life Res. 2021 Mar 2;30(12):3343–3357. doi: 10.1007/s11136-021-02766-9

Panel Contributions:

- Insights from patient organizations, healthcare providers and industry representatives
- Sharing experiences and recommendations to optimize patient care pathways

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Rethinking Myasthenia Gravis Launch

Focus Areas

- The Socio-Economic Burden of Myasthenia Gravis
- Challenges & Gaps in Current Care Pathway of Myasthenia Gravis
- Policy Recommendations to Improve the Management of Myasthenia Gravis

Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

The Patient Perspective

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EuMGA
European Myasthenia Gravis Association

An Auto-immune Neuro-Muscular rare disease

Pathway of an MG-patient :

Misdiagnosis or a delayed diagnosis (due to lack of knowledge or misunderstanding the patient, especially female patients)

Difficulties with access to :

-Information (not available or not given by the specialist)

-Care (long waiting times to have an appoitment with a specialist)

Addressing patient unmet needs along the care pathway

-Treatments/ therapies (especially innovative treatments)
-Lack of psychological support (not foreseen in the guidelines for treatment)

No recognition (due to the invisible nature of the disease)

What does the MG-community need?

-Inclusive care (better guidelines and easier access to the innovative treatments)

-Cross-border care (especially for patients in countries with a lower standard of care)

-**Equal standard of care** in all countries of Europe

Unmet Needs and Patient Care Pathways for Neurofibromatosis

Vera Lipkovskaya, NF Patients United

20 February 2025

Many needs are unmet...

01 Patients

Diagnostic journey, lack of reliable information, access to innovative treatments, mental health, access to employment, fertility, fragmented healthcare pathways, transition to adulthood...

02 Caregivers

Physical & financial strain, employment challenges, emotional well-being, patient–caregiver treatment preference discordance, lack of recognition...

03 Patient communities

Cross-border support, diversity & inclusion, public policy support, recognition of Patient Voice, digital literacy, unified standards of care, combined patient services...

...but some are beyond unmet

Prolonged Diagnostic Journeys leading to disease progression and complications (also higher costs for the system)

Fragmented Care Pathways: lack of coordinated care, disjointed healthcare systems

Transition to Adulthood: teens and adults' unmet needs are often neglected, high drop-out rates

Access to Innovative Treatments: lack of equitable and timely access to innovative treatments and technologies

Cross-border Standards of Care: lack of consistency and unification

Public Support & Patient Community Recognition: policy support is vital and patient voice is a *sine qua non*

Different Conditions, Same Battles

- Wider public policy support
- Shared decision-making
- Consistent & uninterrupted knowledge building

Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

The Patient Care Pathway Methodology: the Organizational and Economic Perspective

Marialuisa Zedde,
Neurologist, Local Health Authority of Reggio Emilia, Italy

Giuseppe Turchetti,
Health Economist, Scuola Superiore Sant'Anna, Pisa, Italy

Vinciane Quoidbach,
Research Project Manager, European Brain Council

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The Patient Care Pathway methodology: the organizational and economic perspective

- Stroke is the second leading cause of death, the third leading cause of disability-adjusted life-years worldwide, and a major cause of dementia
- Stroke is a time-dependent disease and in the chain of care every second count (time is brain)

Treatment	NNT
Stroke Unit	16
IVT	5-9
EVT	3

Stroke care pathway

Stroke Unit
A specialized ward designated for acute stroke patients with continuous monitoring of vital parameters with a multidisciplinary team approach including specialist nursing staff.

Eur J Neurol. 2021 Feb;28(2):717-725; Eur Stroke J. 2023 Sep;8(3):618-628; Eur Stroke J. 2018 Dec;3(4):309-336

Coordinating Panel on Rare Neurological Diseases

Società italiana di neurologia

Coordinating panel on Rare Diseases
Working group on rare neurovascular diseases

The Patient Care Pathway methodology: the organizational and economic perspective

- Acute stroke is a common manifestation for both frequent and rare cerebrovascular diseases
- Acute care pathway should be managed taking into account the special features of patients with rare vascular diseases (e.g. Moyamoya, CADASIL, Ehlers Danlos, Pseudoxanthoma elasticum, etc.

- Several rare cerebrovascular diseases
- Genetic and sporadic diseases
- Small vessels vs large-medium vessel involvement
- Some diseases are covered by an ERN (moyamoya, CADASIL)
- Several phenocopies
- Underdiagnosed diseases have the same unmet need as rare diseases (e.g. Fibromuscular Dysplasia)

FMD-Be

Association belge de patients atteints de dysplasie-fibromusculaire artérielle
FMD Groep België

EURO FMD

Common unmet needs

- **Early diagnosis and regular monitoring** are crucial to **prevent complications**
- **Multidisciplinary specialized management**
- **Periodic assessment**
- **Management of associated symptoms** is critical and need **personalized treatment**.
- **Adequate patient education** is vital for timely recognizing signs of complications.
- **Emotional and Psychological Support** is essential to **help patients cope with the stress and uncertainty** associated with the disease.
- **Systematic recognition of the disease by healthcare systems**

Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

The Patient Care Pathway Methodology: the Organizational and Economic Perspective

Marialuisa Zedde,
Neurologist, Local Health Authority of Reggio Emilia, Italy

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Patient's Care Pathway

The **patient's care pathway** is the route that a patient follows from the first contact with the healthcare system through referral to the completion of treatment and over.

Patient's care pathway represents a health management tool including the spatial and temporal sequence of the activities to be carried out based on scientific-technical knowledge and organizational, professional and technological resources.

A method to implement, make transparent, standardise, optimise and organise the continuous activities of patient-centered care processes.

Patient's Care Pathway: which are the expected benefits?

- Coordination of the care processes
- Integration among different teams, organisational units and different Organizations
- Identification of possible bottlenecks and unmet expectations, useless activities, duplication of activities, organizational-managerial criticalities from the healthcare facility and the patient's point of view
- Efficient use of resources
- Culture of monitoring and continuous improvement

It helps healthcare organizations to create and/or implement a safe, effective, patient-centered, fair, integrated and continuous care process.

Proposed methodology: 1. «Optimised» patient's care pathway

RarERN Path

Methodology specificially designed for the development of a common and shared **organisational reference patient care pathway model** for rare and complex diseases

Rosaria et al. Orphanet J Rare Dis (2020) 15:347
https://doi.org/10.1186/s13023-020-01631-1

Orphanet Journal of Rare Diseases

RESEARCH Open Access

RarERN Path: a methodology towards the optimisation of patients' care pathways in rare and complex diseases developed within the European Reference Networks

Talarico Rosaria[1], Cannizzo Sara[2], Lorenzoni Valentina[2], Marinello Diana[1], Palla Ilaria[2], Pirri Salvatore[2], Ticciati Simone[1], Trieste Leopoldo[2], Triulzi Isotta[2], Terol Enrique[3], Bucher Anna[3] and Turchetti Giuseppe[2*]

Abstract

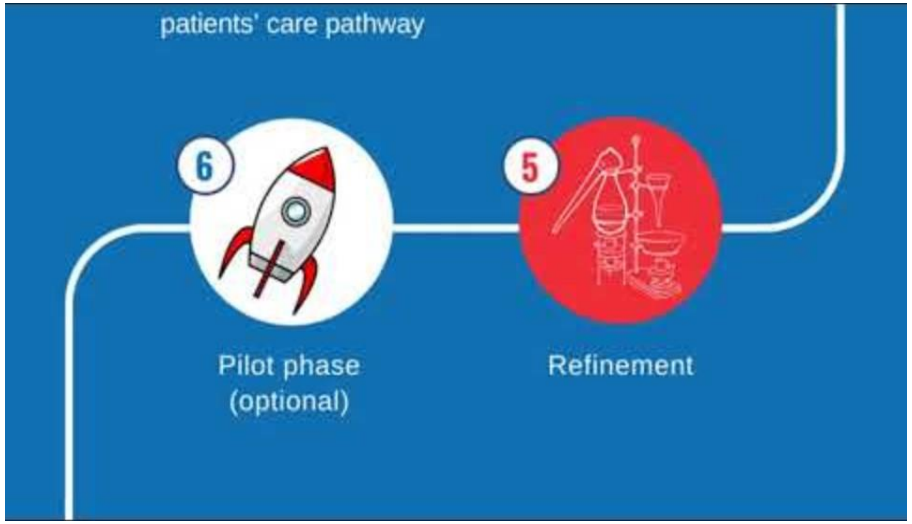
Background: In 2017, the European Commission has launched the European Reference Networks (ERNs), virtual networks involving healthcare providers across Europe. The aim of the ERNs is to tackle complex and rare diseases and conditions that require highly specialized treatment and a concentration of knowledge and resources. The ERN on rare and complex connective tissue and musculoskeletal diseases (ERN ReCONNET) is one of the 24 ERNs approved that aims to improve the management of Rare and Complex Connective Tissue and Musculoskeletal Diseases.

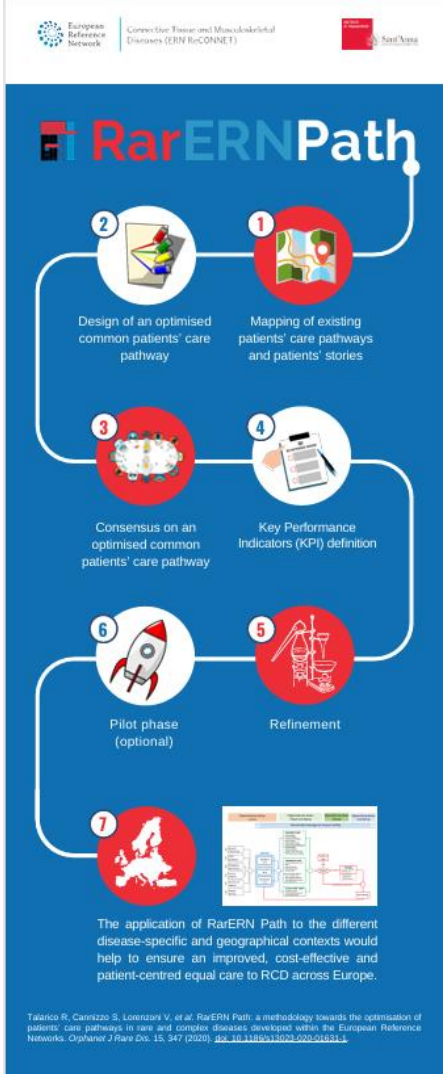
Objective: The RarERN Path methodology aims to create a single reference organisational model for patients' care pathways which, if applied in different contexts, helps to ensure an improved, cost-effective and patient-centred equal care to rare and complex diseases.

Methods: Starting from existing standard methods for the creation and elaboration of patients' care pathways, a specific methodology was created in order to take advantage of the distinctive and peculiar characteristics of the ERNs. Specifically, the development of the RarERN Path methodology involved different stakeholders: health economists, clinicians and researchers expert in rare and complex diseases, communication experts, experts in patients' involvement and narrative medicine and policy-makers.

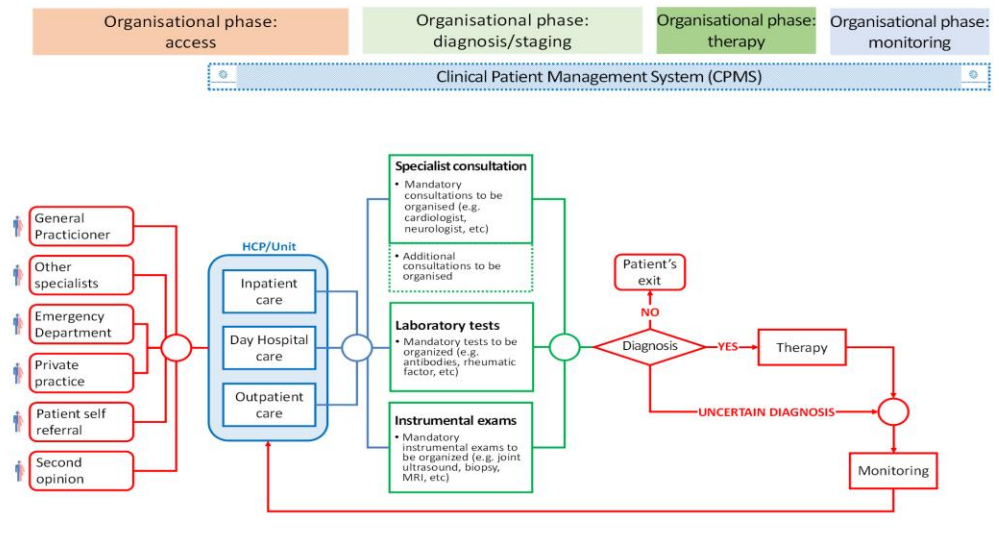
Results: The RarERN Path methodology foresees six consecutive phases, each with different and specific aims. Specifically, the six phases are represented by: Phase 1—mapping of existing patients' care pathways and patients' stories; Phase 2—design of an optimised common patients' care pathway; Phase 3—consensus on an optimised common patients' care pathway; Phase 4—key performance indicators definition; Phase 5—refinement; Phase 6—pilot phase (optional).

Conclusion: The application of RarERN Path to the different disease-specific and geographical contexts would help to ensure an improved, cost-effective and patient-centred equal care to rare and complex diseases across Europe as well as a possible tangible action towards the integration of ERNs into the different European healthcare systems.





RarERN Path: organisational phases



Benefits from the proposed approach

It is INNOVATIVE because:

- **It is really based on the CO-DESIGN approach**
  - **Healthcare professionals**
  - **Patients**
  - **Hospital managers**

- **Multistakeholder**
- **Generalisable**
- **Contextualisable**

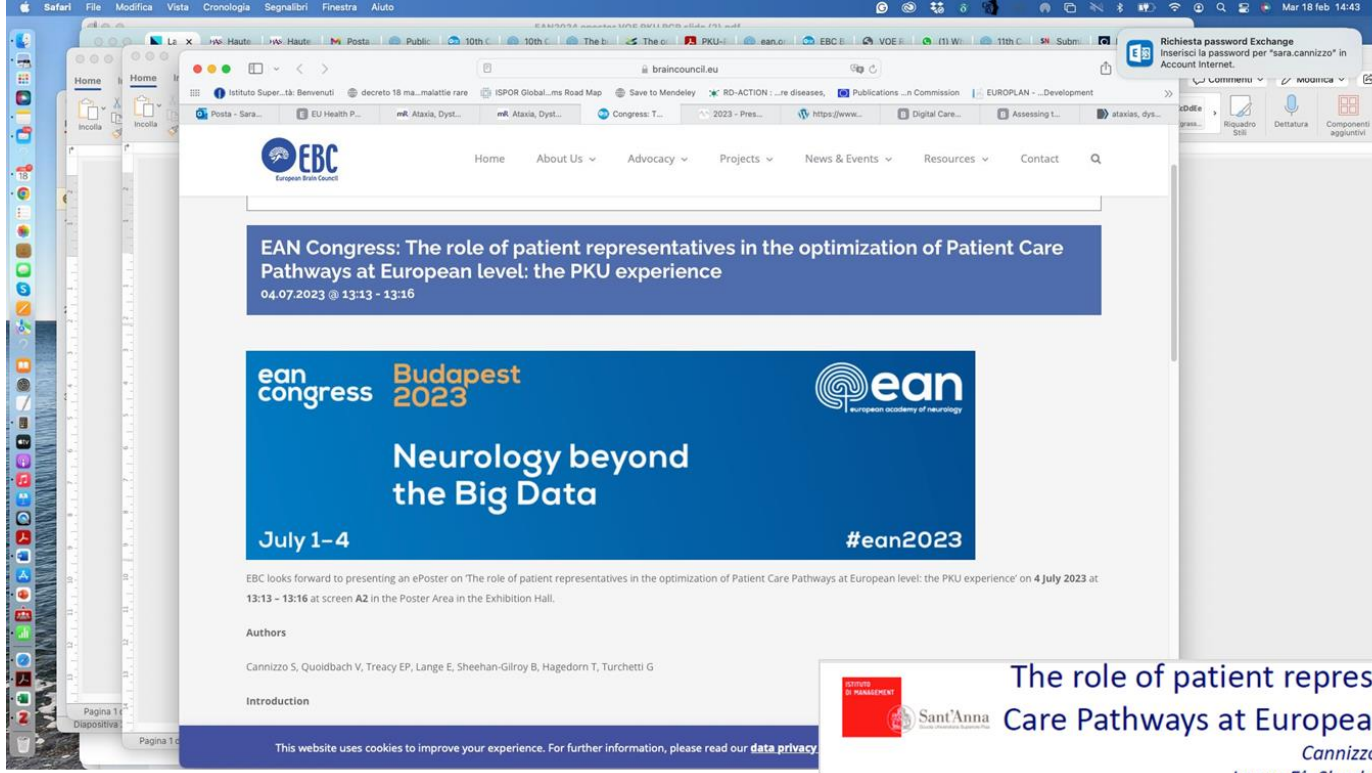
- **It provides operative/concrete organisational INSTRUCTIONS/SUGGESTIONS**

- **It is FLEXIBLE/ADAPTABLE to different national/regional/local contexts**

Benefits from the proposed approach

It allows to provide concrete **recommendations** to the health policy makers both at the European and the national level on how to:

- Increase the ***homogeneity*** in the management of patients affected RDs
- Promote the ***efficient use*** of resources (economic sustainability)
- Pursue a more **EQUITABLE ACCESS** to care all around Europe!



The role of patient representatives in the optimization of Patient Care Pathways at European level: the Phenylketonuria experience

Cannizzo S[1], Quoidbach V[2], Treacy EP[3], Lange E[4], Sheehan-Gilroy B[5], Hagedorn T[6], Turchetti G[1]

1, Institute of Management, Scuola Superiore Sant'Anna, Pisa, Italy; 2, The European brain Council, Brussels, Belgium; 3, National Centre for inherited Metabolic Disorders, Mater Misericordiae University Hospital, Dublin, Ireland; 4, The European Society for Phenylketonuria and allied disorders; 5, Munster Technological University Kerry Campus, Tralee, Ireland); 6, German PKU and Allied Disorders Patients Association DIG PKU, European Society for Phenylketonuria & Allied Disorders.

VOT by EBC European Brain Council

Background

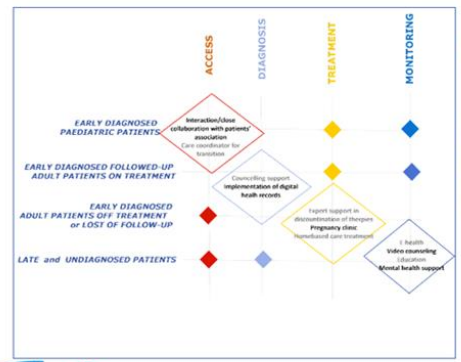
Phenylketonuria (PKU) is a rare autosomal recessive inborn error of phenylalanine (Phe) metabolism caused by pathogenic variants in the gene encoding phenylalanine hydroxylase (PAH). PAH deficiency causes abnormal accumulation of Phe in the blood and in the brain. High blood Phe levels are strongly linked to neurocognitive dysfunction, and if not treated PKU causes intellectual disabilities, motor deficits, microcephaly, autism, eczematous rash, seizures, developmental problems, aberrant behavior, and psychiatric symptoms. In Europe PKU prevalence is about 10:100,000 newborns with higher rate in Turkey and Ireland, and a very low rate in Finland. In most European countries, the national newborn screening (NBS) programs include Phe measurement. The aim of the NBS is to discover hyperphenylalaninemia (HPA), and this is defined as any blood Phe >120 µmol/L. The early detection of HPA and its treatment can prevent neurological damages. Despite the high diffusion of NBS, there are still late diagnosed patients or undiagnosed patients such as immigrant children from countries where NBS is lacking, or adults born before the introduction of the NBS.

Methods

In defining and optimising patient care pathways (PCPs) the role of patient representatives (PRs) is very important; it is crucial in the contest of rare diseases where the complexity of the disorders is higher, co-morbidity and multi-organ involvement are present, multidisciplinary care is needed, and patients may experience inequality in the access to specialised diagnostic/treatment procedures. In this work we have analysed the role of PRs in the design and optimization of Phenylketonuria's (PKU) PCP. We applied RarERN Path© methodology to PKU PCP within the Value of Treatment (VOT) for Rare Brain Disorders project (European Brain Council). PRs of PKU Associations of Ireland and Germany were involved. The PCPs in place in centers of excellence (COE) were analysed, and patients provided input through a semi-structured questions survey exploring organization of care and perception of criticalities when receiving healthcare services. A first draft of the optimized PCP for PKU was discussed in a plenary meeting attended by neurologists and PRs. Finally, PRs were requested to provide additional suggestions through a second ad-hoc survey.

Results

Patients Representatives contributed to the design of an optimized PCP, providing unique information on the main organizational challenges in COEs and on the coordination of care between COE and non-hospital care at European level.



Patients said about patient care pathways, how to improve the care for their PKU:

"Many patients were born before newborn screening. Other than their intellectual and physical disabilities, the underlying condition PKU often has not been adequately looked after. It is a significant milestone that the Patient Care Pathways shed a light on them and explicitly highlight their need to having access to treatment and better quality of life."

„Too often, we PKU patients have been told we should be grateful for the value of early diagnosis and an effective nutritional therapy. However, PKU is not a solved problem and too much gratefulness is delaying progress. It is so important that the Patient Care Pathways outline many of the residual unmet needs in PKU care."

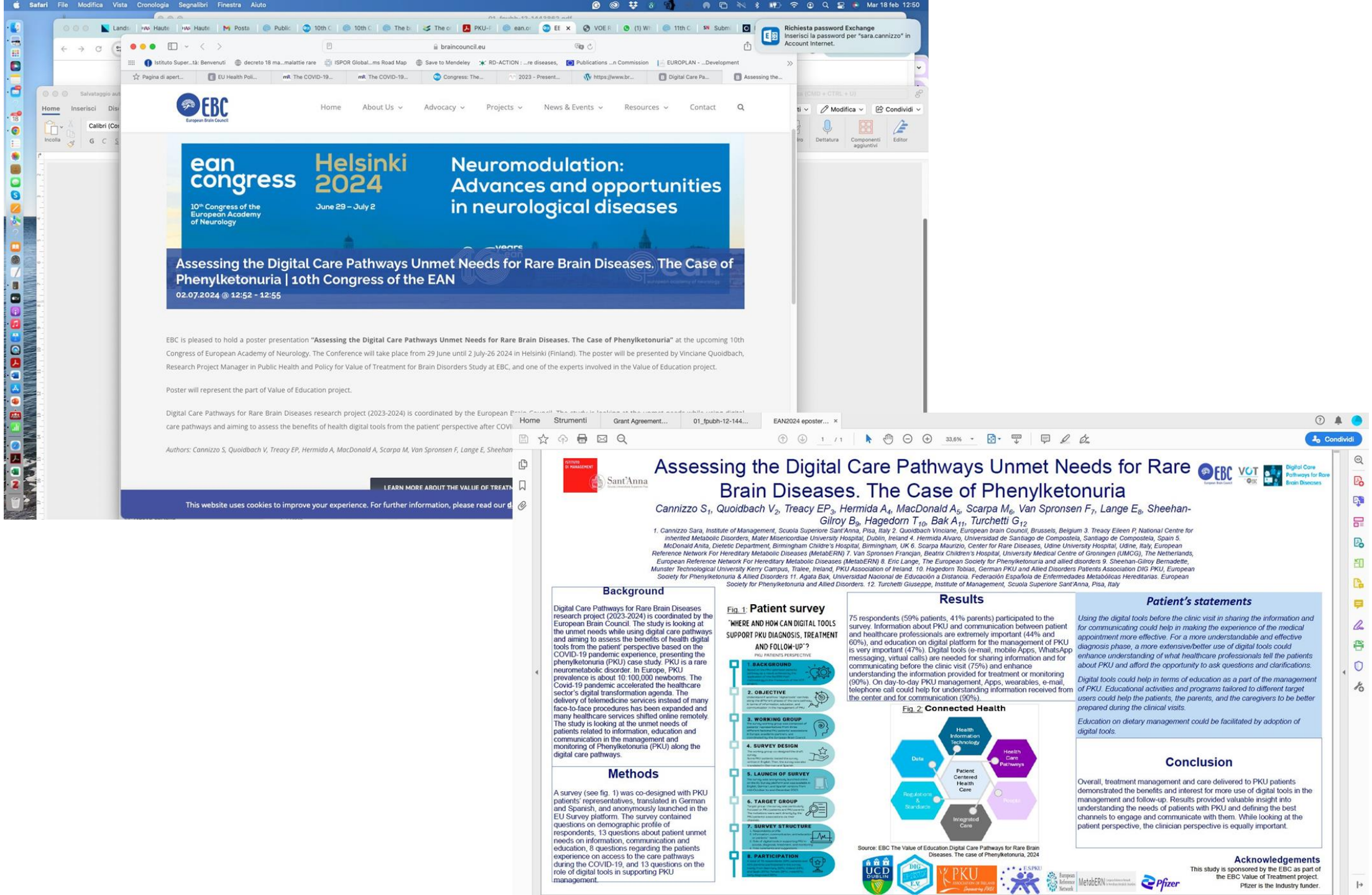
„For years I did not recognize how I was mentally chocked by the diagnosis. I functioned, however was unable to cope. Thanks to trauma screening and mental health support I learned to accept my sons' disease and the treatment as something normal. It means the world to me that the Care Pathways acknowledge the individual needs of both patients and caregivers."

Conclusion

To formally involve PRs in the co-design of the PCP is necessary because it allows to complement clinicians' perspective about "ranking" and "weight" of what really matters throughout the PCP. PKU case is particularly interesting in this respect.

Acknowledgements

This study is sponsored by the EBC as part of the Value of Treatment project.
Biomarin is the Industry funder.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Helsinki 2024
10th Congress of the European Academy of Neurology
June 29 – July 2
Neuromodulation: Advances and opportunities in neurological diseases
Assessing the Digital Care Pathways Unmet Needs for Rare Brain Diseases. The Case of Phenylketonuria | 10th Congress of the EAN
02.07.2024 @ 12:52 - 12:55
EBC is pleased to hold a poster presentation "Assessing the Digital Care Pathways Unmet Needs for Rare Brain Diseases. The Case of Phenylketonuria" at the upcoming 10th Congress of European Academy of Neurology. The Conference will take place from 29 June until 2 July-26 2024 in Helsinki (Finland). The poster will be presented by Vinciane Quoidbach, Research Project Manager in Public Health and Policy for Value of Treatment for Brain Disorders Study at EBC, and one of the experts involved in the Value of Education project.
Poster will represent the part of Value of Education project.
Digital Care Pathways for Rare Brain Diseases research project (2023-2024) is coordinated by the European Brain Council.
care pathways and aiming to assess the benefits of health digital tools from the patient' perspective after COVI
Authors: Cannizzo S, Quoidbach V, Treacy EP, Hermida A, MacDonald A, Scarpa M, Van Spronsen F, Lange E, Sheehan
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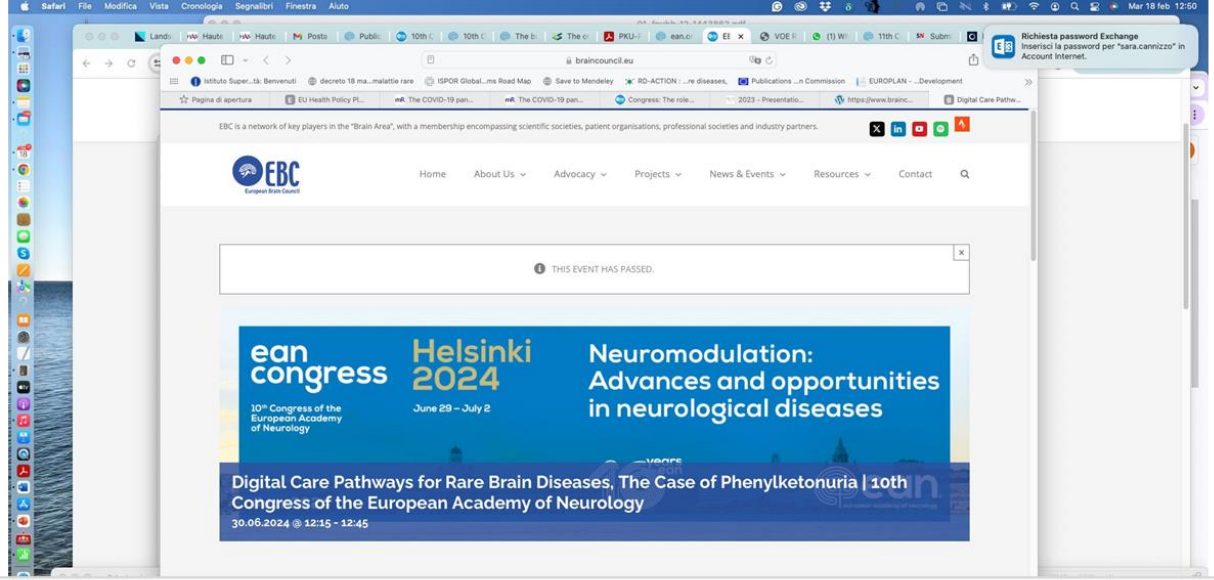
Condividi
Assessing the Digital Care Pathways Unmet Needs for Rare Brain Diseases. The Case of Phenylketonuria
Cannizzo S1, Quoidbach V2, Treacy EP3, Hermida A4, MacDonald A5, Scarpa M6, Van Spronsen F7, Lange E8, Sheehan-Gilroy B9, Hagedorn T10, Bak A11, Turchetti G12
1. Cannizzo Sara, Institute of Management, Scuola Superiore Sant'Anna, Pisa, Italy 2. Quoidbach Vinciane, European brain Council, Brussels, Belgium 3. Treacy Eileen P, National Centre for inherited Metabolic Disorders, Mater Misericordiae University Hospital, Dublin, Ireland 4. Hermida Alvaro, Universidad de Santiago de Compostela, Santiago de Compostela, Spain 5. McDonald Anita, Dietetic Department, Birmingham Childre's Hospital, Birmingham, UK 6. Scarpa Maurizio, Center for Rare Diseases, Udine University Hospital, Udine, Italy, European Reference Network For Hereditary Metabolic Diseases (MetabERN) 7. Van Spronsen Francjan, Beatrix Children's Hospital, University Medical Centre of Groningen (UMCG), The Netherlands, European Reference Network For Hereditary Metabolic Diseases (MetabERN) 8. Eric Lange, The European Society for Phenylketonuria and allied disorders 9. Sheehan-Gilroy Bernadette, Munster Technological University Kerry Campus, Tralee, Ireland, PKU Association of Ireland. 10. Hagedorn Tobias, German PKU and Allied Disorders Patients Association DIG PKU, European Society for Phenylketonuria & Allied Disorders 11. Agata Bak, Universidad Nacional de Educación a Distancia. Federación Española de Enfermedades Metabólicas Hereditarias. European Society for Phenylketonuria and Allied Disorders. 12. Turchetti Giuseppe, Institute of Management, Scuola Superiore Sant'Anna, Pisa, Italy
Background
Digital Care Pathways for Rare Brain Diseases research project (2023-2024) is coordinated by the European Brain Council. The study is looking at the unmet needs while using digital care pathways and aiming to assess the benefits of health digital tools from the patient' perspective based on the COVID-19 pandemic experience, presenting the phenylketonuria (PKU) case study. PKU is a rare neurometabolic disorder. In Europe, PKU prevalence is about 10:100,000 newborns. The Covid-19 pandemic accelerated the healthcare sector's digital transformation agenda. The delivery of telemedicine services instead of many face-to-face procedures has been expanded and many healthcare services shifted online remotely. The study is looking at the unmet needs of patients related to information, education and communication in the management and monitoring of Phenylketonuria (PKU) along the digital care pathways.
Methods
A survey (see fig. 1) was co-designed with PKU patients' representatives, translated in German and Spanish, and anonymously launched in the EU Survey platform. The survey contained questions on demographic profile of respondents, 13 questions about patient unmet needs on information, communication and education, 8 questions regarding the patients experience on access to the care pathways during the COVID-19, and 13 questions on the role of digital tools in supporting PKU management.
Fig. 1: Patient survey
"WHERE AND HOW CAN DIGITAL TOOLS SUPPORT PKU DIAGNOSIS, TREATMENT AND FOLLOW-UP"?
PKU PATIENTS PERSPECTIVE
1. BACKGROUND
2. OBJECTIVE
3. WORKING GROUP
4. SURVEY DESIGN
5. LAUNCH OF SURVEY
6. TARGET GROUP
7. SURVEY STRUCTURE
8. PARTICIPATION
Results
75 respondents (59% patients, 41% parents) participated to the survey. Information about PKU and communication between patient and healthcare professionals are extremely important (44% and 60%), and education on digital platform for the management of PKU is very important (47%). Digital tools (e-mail, mobile Apps, WhatsApp messaging, virtual calls) are needed for sharing information and for communicating before the clinic visit (75%) and enhance understanding the information provided for treatment or monitoring (90%). On day-to-day PKU management, Apps, wearables, e-mail, telephone call could help for understanding information received from the center and for communication (90%).
Fig. 2: Connected Health
Health Information Technology
Health Care Pathways
Data
Patient Centered Health Care
People
Regulations & Standards
Integrated Care
Source: EBC The Value of Education.Digital Care Pathways for Rare Brain Diseases. The case of Phenylketonuria, 2024
Patient's statements
Using the digital tools before the clinic visit in sharing the information and for communicating could help in making the experience of the medical appointment more effective. For a more understandable and effective diagnosis phase, a more extensive/better use of digital tools could enhance understanding of what healthcare professionals tell the patients about PKU and afford the opportunity to ask questions and clarifications.
Digital tools could help in terms of education as a part of the management of PKU. Educational activities and programs tailored to different target users could help the patients, the parents, and the caregivers to be better prepared during the clinical visits.
Education on dietary management could be facilitated by adoption of digital tools.
Conclusion
Overall, treatment management and care delivered to PKU patients demonstrated the benefits and interest for more use of digital tools in the management and follow-up. Results provided valuable insight into understanding the needs of patients with PKU and defining the best channels to engage and communicate with them. While looking at the patient perspective, the clinician perspective is equally important.
Acknowledgements
This study is sponsored by the EBC as part of the EBC Value of Treatment project.
Pfizer is the Industry funder.



Digital Care Pathways for Rare Brain Diseases, The Case of Phenylketonuria

30 June 2024 – Scientific Theatre

12:15 to 12:45 CEST

ean congress Helsinki 2024

Nähdään Helsingissä

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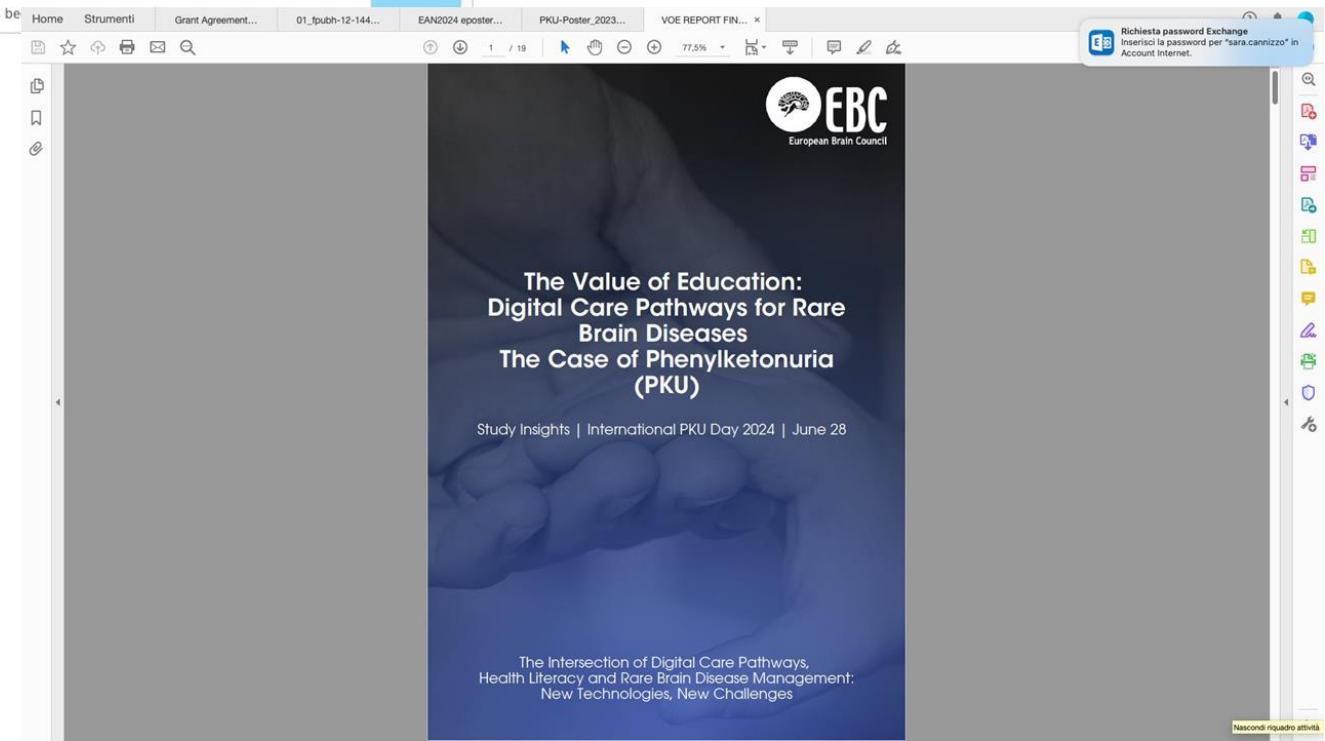
Digital Care Pathways for Rare Brain Diseases

The Value of Education: Digital Care Pathways for Rare Brain Diseases. The case of Phenylketonuria

One of the most relevant challenges for healthcare providers and patients during the COVID – 19 pandemics has be



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Orphanet Journal of Rare Diseases

REVIEW Open Access

The COVID-19 pandemic impact on continuity of care provision on rare brain diseases and on ataxias, dystonia and PKU. A scoping review

Sara Cannizzo[1], Vinciane Quoidbach[2], Paola Giunti[3], Wolfgang Oertel[4], Gregory Pastores[5], Maja Relja[6] and Giuseppe Turchetti[1,7*]

Abstract

One of the most relevant challenges for healthcare providers during the COVID– 19 pandemic has been assuring the continuity of care to patients with complex health needs such as people living with rare diseases (RDs). The COVID–19 pandemic accelerated the healthcare sector's digital transformation agenda. The delivery of telemedicine services instead of many face-to-face procedures has been expanded and, many healthcare services not directly related to COVID-19 treatments shifted online remotely. Many hospitals, specialist centres, patients and families started to use telemedicine because they were forced to. This trend could directly represent a good practice on how care services could be organized and continuity of care could be ensured for patients. If done properly, it could boast improved patient outcomes and become a post COVID-19 major shift in the care paradigm. There is a fragmented stakeholders spectrum, as many questions arise on: how is e-health interacting with 'traditional' healthcare providers; about the role of the European Reference Networks (ERNs); if remote care can retain a human touch and stay patient centric. The manuscript is one of the results of the European Brain Council (EBC) Value of Treatment research project on rare brain disorders focusing on progressive ataxias, dystonia and phenylketonuria with the support of Academic Partners and in collaboration with European Reference Networks (ERNs) experts, applying empirical evidence from different European countries. The main purpose of this work is to investigate the impact of the COVID-19 pandemic on the continuity of care for ataxias, dystonia and phenylketonuria (PKU) in Europe. The analysis carried out makes it possible to highlight the critical points encountered and to learn from the best experiences. Here, we propose a scoping review that investigates this topic, focusing on continuity of care and novel methods (e.g., digital approaches) used to reduce the care disruption. This scoping review was designed according to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses extension for scoping reviews (PRISMA-ScR) standards. This work showed that the implementation of telemedicine services was the main measure that healthcare providers (HCPs) put in place and adopted for mitigating the effects of disruption or discontinuity of the healthcare services of people with rare neurological diseases and with neurometabolic disorders in Europe.

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Title: The organizational dimension in rare and complex diseases care management: an application of RarERN Path© methodology in Ataxias, Dystonia and Phenylketonuria

Authors:

Sara Cannizzo, Vinciane Quoidbach, Leopoldo Trieste, Monika Benson, Antonio Federico, Alessandro Filla, Bernadette Sheehan Gilroy, Paola Giunti, Holm Graeßner, Julie Greenfield, Tobias Hagedorn, Alvaro Hermida, Barry Hunt, Anita McDonald, Francesca Morgante, Wolfgang Oertel, Gregory Pastores, Martje G Pauly, Carola Reinhard, Maja Relja, Eileen Treacy, Francjan Van Spronsen, Julie Vallortigara and Giuseppe Turchetti.

SUBMITTED – UNDER REVIEW

Title: The role of digital tools and their implementation within Patient Care Pathways for Rare Brain Disorders: the case of Phenylketonuria

Authors:

Sara Cannizzo, Vinciane Quoidbach, Bernadette Sheehan-Gilroy, Tobias Hagedorn, Agata Bak, Suzanne Dickson, Eileen Treacy, Alvaro Hermida, Anita McDonald, Eva Venegas, James O'Byrne, Maurizio Scarpa, Francjan Van Spronsen, Eric Lange, Tim Buckinx, Ahmad Monavari, Stephan vom Dahl, Leopoldo Trieste, Giuseppe Turchetti

Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

The Clinical Perspective

Alexis Arzimanoglou,
Neurologist, Spain: The case of Rare Epilepsies (ERN EpiCARE)

Lorenzo Maggi,
Neurologist, Italy: The case of MG (ERN-NMD)

Vinciane Quoidbach,
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Unmet Needs and Optimising Patient Care Pathways for Rare Brain Disease in Europe

Industry Perspective on Myasthenia Gravis

Walter Atzori,
Global Patient Advocacy Strategic Lead, Alexion

Anna Kole,
Global Patient Engagement Lead MG, UCB

Vinciane Quoidbach,
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Session 3

Towards a Rare Brain Disease Ecosystem & Knowledge Hub:

Open Discussion

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Towards a Rare Brain Disease Ecosystem & Knowledge Hub: Open Discussion

A Multistakeholder Perspective

Julian Grosskreutz,
ALS Coalition

Frédéric Destrebecq,
Executive Director,
European Brain Council

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European Brain Council

Towards a Rare Brain Disease Ecosystem & Knowledge Hub: Open Discussion

Panel Discussion

Sameer Zuberi,
Past President, European Paediatric Neurology Society (EPNS)

Astri Arnesen,
President, European Federation of Neurological Associations (EFNA)

Kailash Bhatia,
President-Elect, European Academy of Neurology (EAN)

Frédéric Destrebecq,
Executive Director,
European Brain Council

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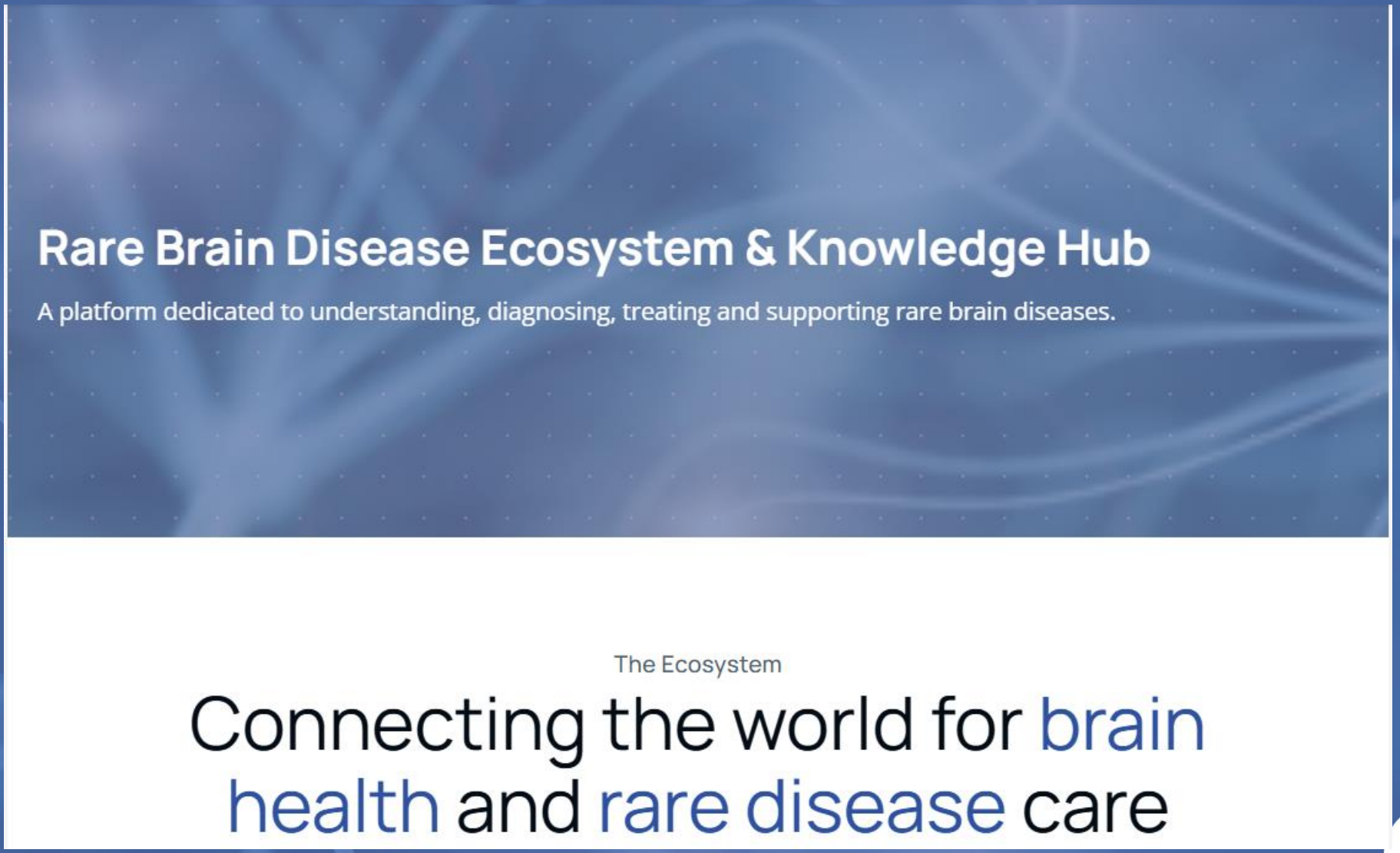

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The Rare Brain disease Ecosystem and Knowledge Hub - Objectives

Build a scientifically based, transparent and independent Rare Brain Disease Ecosystem and Knowledge Hub that:

- Brings together, connects the community that share a common interest in delivering effective, equitable and sustainable solutions to facilitate the prevention, diagnosis, treatment and management of RBDs, especially in areas where there is an unmet public health need.

- Promotes research projects and dissemination of study results led by EBC with its members and partners as well as any study on rare brain disease led by all the stakeholders in the field (patient associations, European Reference Networks, scientific societies, industry, etc).

- Prioritizes brain health: bridging science to society and informing policy through the set-up of a knowledge hub using a shared platform.

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The Rare Brain disease Ecosystem and Knowledge Hub - Objectives

The Rare Brain Disease Ecosystem, more specifically, aim will be:

- to **translate** to scientific results and issues in a more understandable and readable way;
- To **discuss** current and potential challenges and opportunities (transfer of innovation, organizational and technological, regulatory, economic sustainability, equity of access,…) that, if unblocked, would be game-changers in the development of healthcare solutions;
- To **articulate** research areas towards these opportunities that could be addressed in EBC's Ecosystem as a cross-sectoral public-private partnership;
- To **maximize** policy impact of EBC's and any other stakeholders' projects in the Rare Brain Disease Ecosystem.

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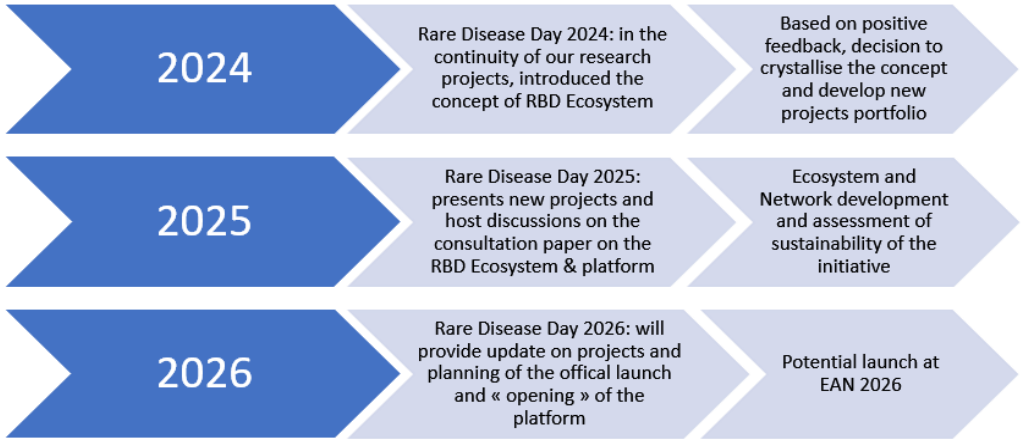
Proposed timeline:



EBC will invite you to contribute to define and build the Ecosystem & Knowledge Hub further.

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Towards a Rare Brain Disease Ecosystem & Knowledge Hub

CONSULTATION PAPER (FEBRUARY 2025)

This document has been prepared in the framework of the EBC Rare Brain Disease Event and will be discussed during the meeting session 3.

1. Background

Our ambition for rare disease

Over the past few decades, significant progress has been made in rare disease research, healthcare, and policymaking. Landmark developments, such as the UN Resolution on the Challenges of Persons Living with a Rare Disease and Their Families (adopted in December 2021) and a forthcoming WHA Resolution on Rare Diseases (2025) have brought rare diseases to the forefront of public health. Advances in the discovery of new medicines, the establishment of clinical centers (e.g., European Reference Networks [ERNs]), and policy frameworks have improved many lives. However, the journey toward comprehensive and equitable care for rare disease patients remains unfinished.

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Towards a Rare Brain Disease Ecosystem & Knowledge Hub: Open Discussion

Q&A

Sameer Zuberi,
Past President,
European Paediatric Neurology Society (EPNS)

Astri Arnesen,
President, European Federation of Neurological Associations (EFNA)

Kailash Bhatia,
President-Elect,
European Academy of Neurology (EAN)

Frédéric Destrebecq,
Executive Director,
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Wrap Up & Closing Remarks

Suzanne Dickson
President, European Brain Council

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Networking Lunch

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