



THE EUROPEAN REFERENCE NETWORK FOR RARE NEUROLOGICAL DISEASES – ERN-RND



WHY EUROPEAN REFERENCE NETWORKS FOR RARE AND COMPLEX DISEASES?



- Rareness of patients, but ~30 million patients in Europe
- Rareness of experts and care structures
- Rareness of knowledge
- Rareness of therapies

→ Collaboration on EU-level

→ National expertise centres form European networks

24 ERNs HAVE BEEN ESTABLISHED IN 2017

- ERN Bond
- ERN Cranio
- Endo-ERN
- ERN EpiCARE
- ERKNet
- ERN-RND
- ERNICA
- ERN Lung
- ERN Skin
- ERN EURACAN
- ERN EuroBloodNet
- ERN eUROGEN
- ERN Euro-NMD
- ERN Eye
- ERN Genturis
- ERN Guard-Heart
- ERN ITHACA
- MetabERN
- ERN PaedCan
- ERN Rare-Liver
- ERN ReCONNET
- ERN RITA
- ERN Transplant-Child
- VASCERN

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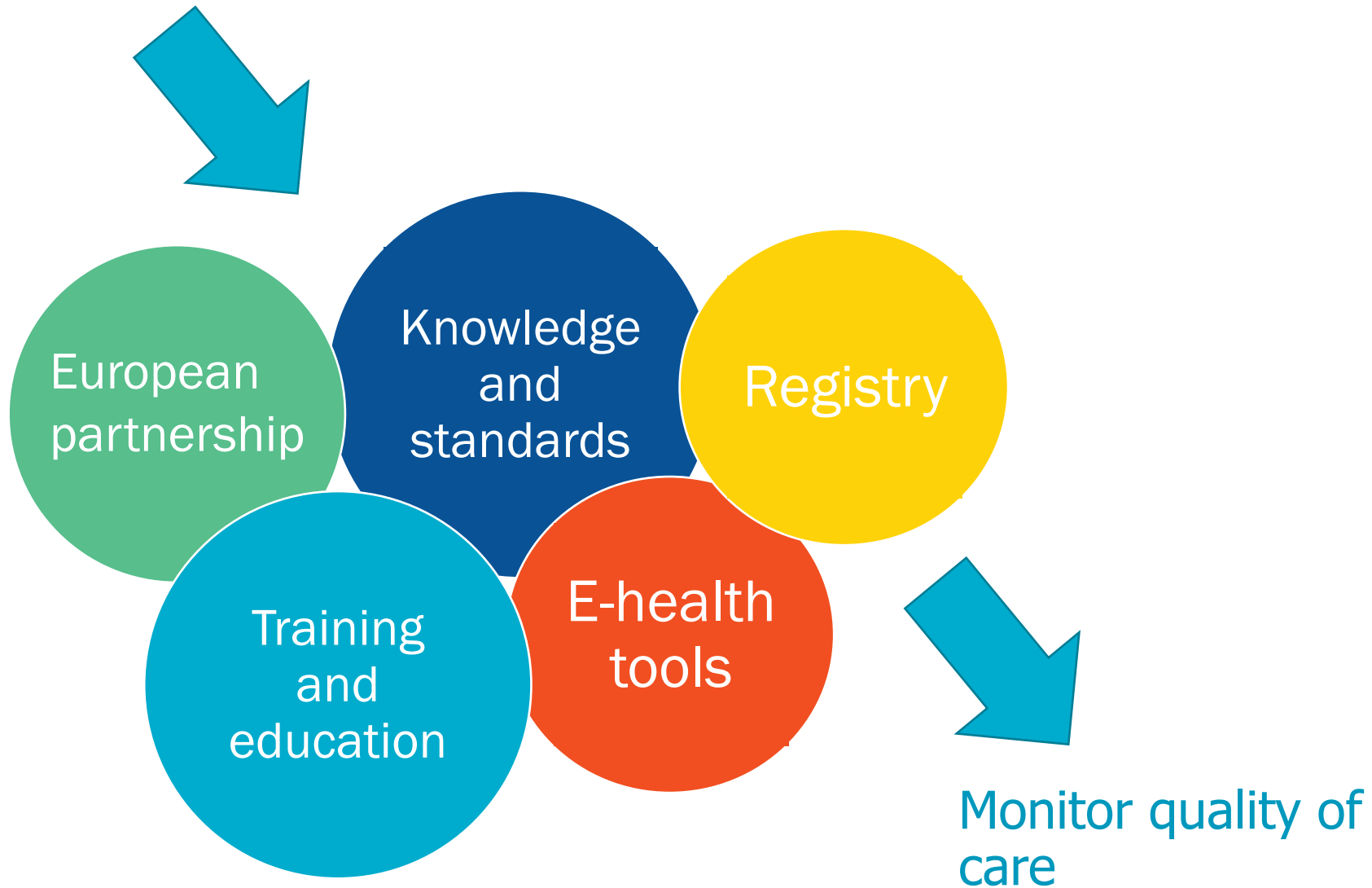
One aim:

Change the way how healthcare is delivered for patients with rare diseases

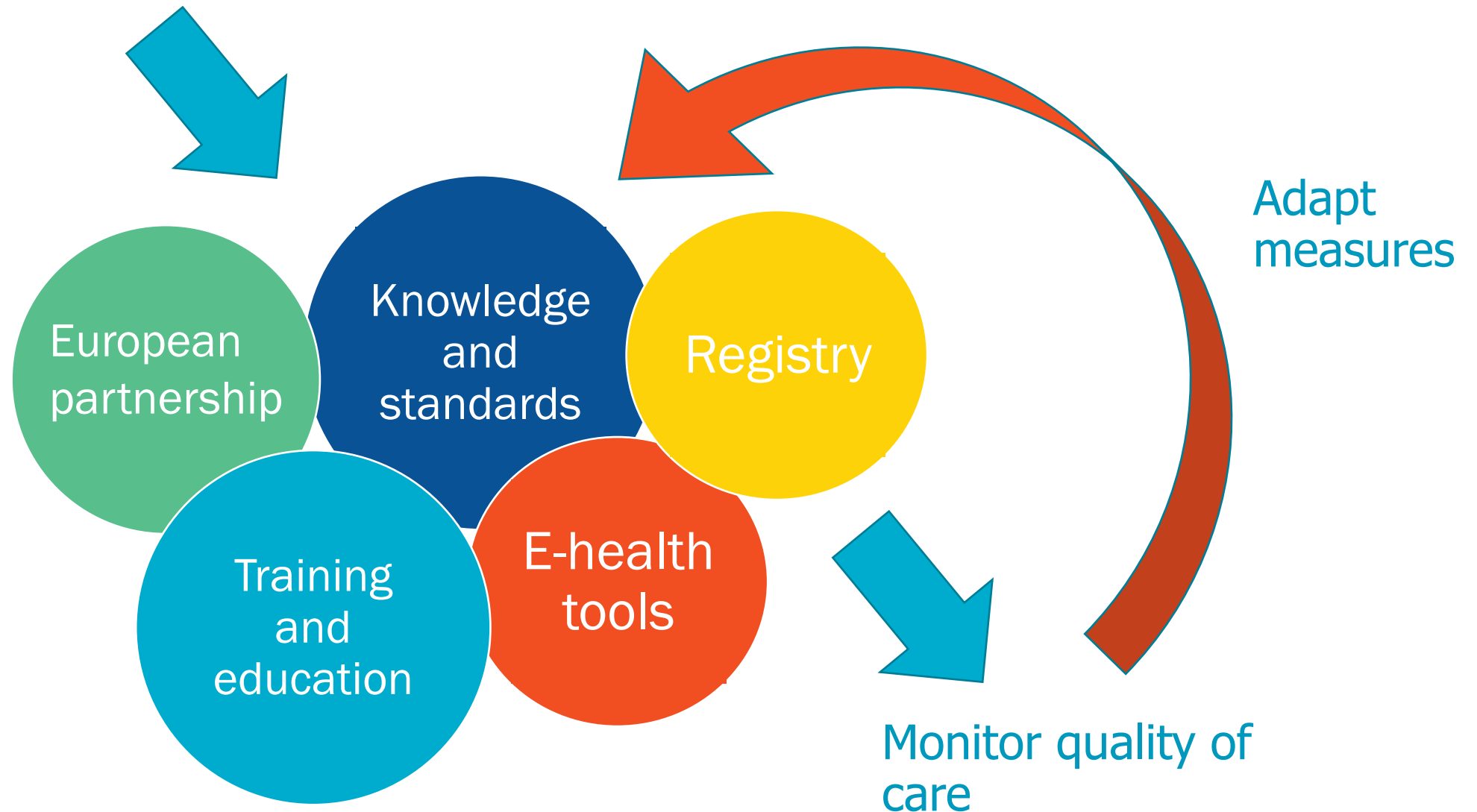
Change the way how healthcare is delivered for patients with rare diseases



Change the way how healthcare is delivered for patients with rare diseases



Change the way how healthcare is delivered for patients with rare diseases



ERN-RND –

EUROPEAN REFERENCE NETWORK FOR RARE NEUROLOGICAL DISEASES

- 68 expert centres in 24 EU countries
- > 50.000 patients per year (adults and paediatric)

Diseases covered

- Ataxia and HSP
- HD and Chorea
- Dystonia, paroxysmal disorders and NBIA
- Leukoencephalopathies
- Atypical parkinsonian syndromes
- Frontotemporal dementia



ERN-RND – EUROPEAN REFERENCE NETWORK FOR RARE NEUROLOGICAL DISEASES

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SETTING THE SCENE

Journal of Neurology

<https://doi.org/10.1007/s00415-022-11412-4>

ORIGINAL COMMUNICATION



Dystonia management across Europe within ERN-RND: current state and future challenges

Liesanne M. Centen^{1,2}  · David Pinter³ · Martje E. van Egmond^{1,2} · Holm Graessner⁴ · Norbert Kovacs³ · Anne Koy⁵ · Belen Perez-Dueñas⁶ · Carola Reinhard⁴ · Marina A. J. Tijssen^{1,2} · Sylvia Boesch⁷

Reported issues:

- Lack of a multidisciplinary approach to patients with dystonia
- Not enough awareness about dystonia as a disease in primary care
- Difficulty in access to genetic testing or specialized care

Suggested measures to be taken:

- National registries of dystonia patients
- A rehabilitation program tailored to fit the needs of dystonia patients
- More educational programmes for general neurologists



PATIENT VIEW ON RND



PATIENT PARTICIPATION IS KEY IN ERN-RND



What is a patient journey?

- Innovative info-graphical overview of patient needs in the different stages of the disease
- Connects professional expert recommendation and guidelines with patient needs

Why patient journeys?

- Working documents jointly developed and used by patients and clinicians → identify gaps in care and respective adaptation of care pathways to better meet the needs of patients → First step toward systematic patient engagement in the design of care pathways
- Useful resource for patients, families, non-specialist clinicians, and the general public to understand the care needs of people living with a rare neurological disease

Benson et al.
Orphanet Journal of Rare Diseases (2022) 17:130
<https://doi.org/10.1186/s13023-022-02270-4>

Orphanet Journal of
Rare Diseases

RESEARCH

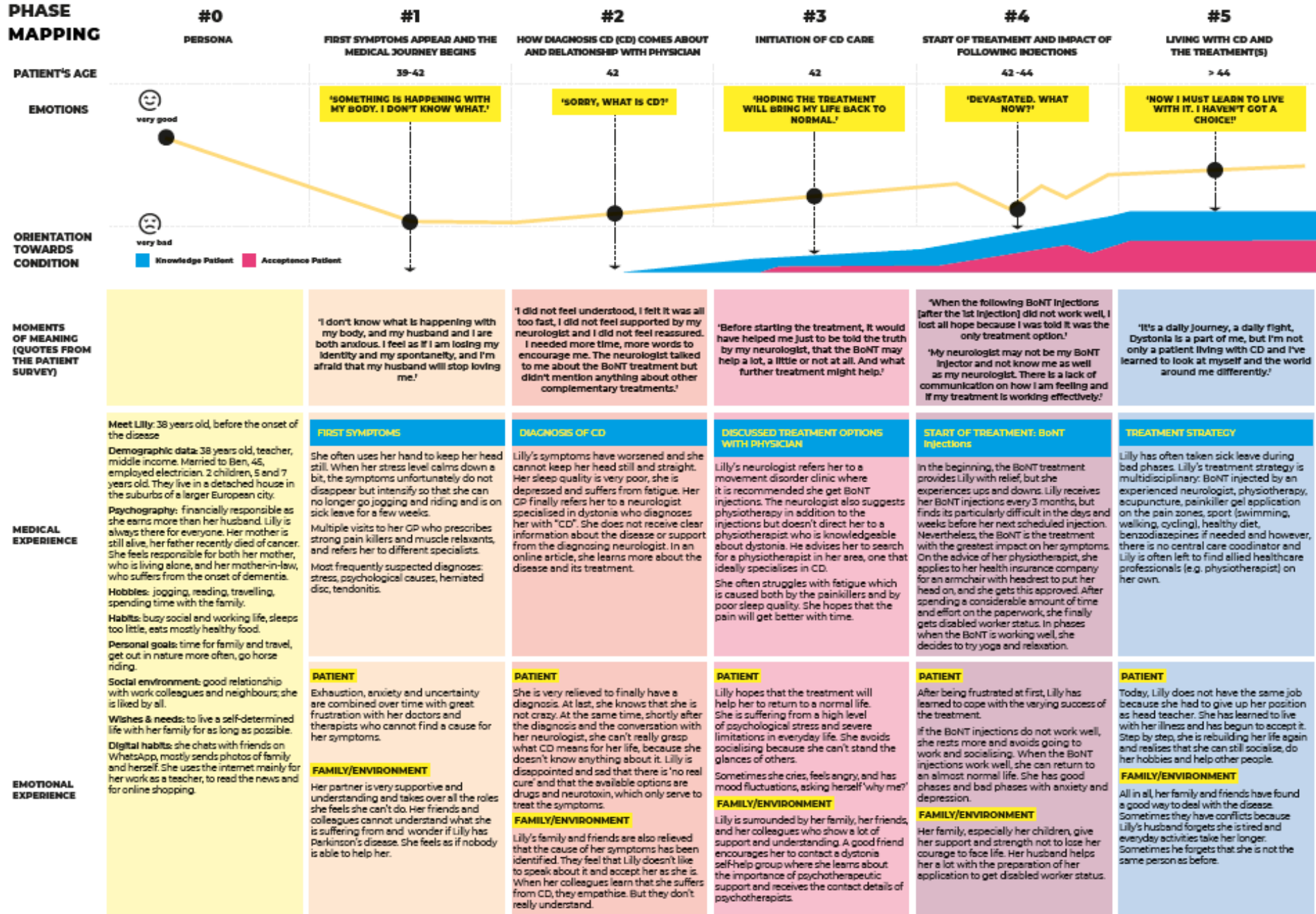
Open Access

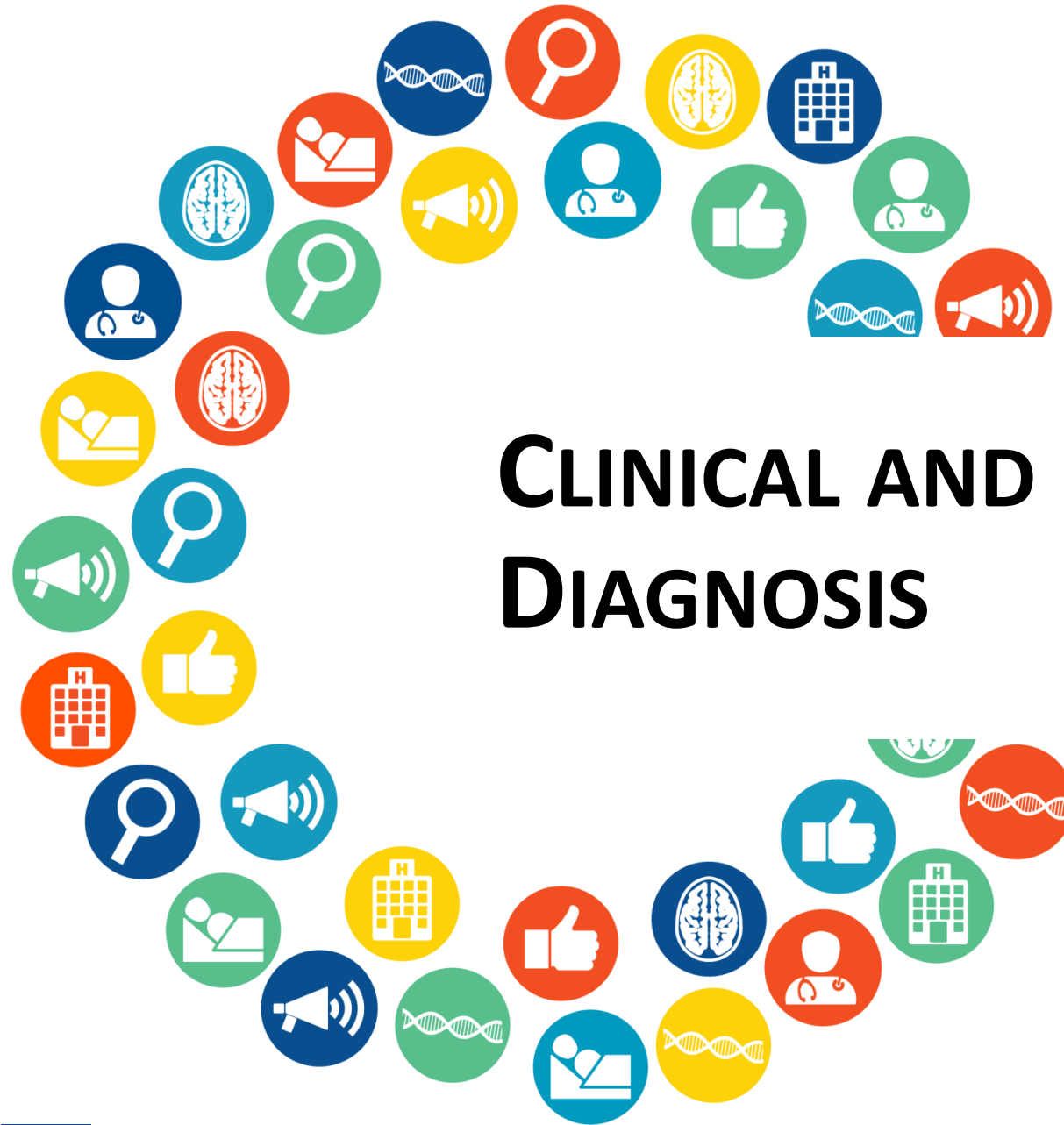


Development of a patient journey map for people living with cervical dystonia

Monika Benson^{1,2}, Alberto Albanese^{2,3}, Kailash P. Bhatia⁴, Pascale Cavillon⁵, Lorraine Cuffe⁶, Kathrin König⁷, Carola Reinhard^{2,8} and Holm Graessner^{2,8*}

PHASE MAPPING





CLINICAL AND GENETIC DIAGNOSIS



DISEASE KNOWLEDGE

- Diagnostic flowcharts
- Disease scales
- Guidelines

DEFINITION

- Full definition of Dystonia
- Full definition of NBIA

ERN-RND CENTRES FOR DYSTONIA, NBIA AND PAROXYSMAL DISORDERS

- Centres and contact details for Dystonias and NBIA

EDUCATIONAL WEBINARS

- Paroxysmal dyskinesias by Giovanna Zorzi
- Genetic dystonia and treatment by Sylvia Boesch
- Treatable dystonias & dystonia in inborn errors of metabolism by Tom de Koning
- Clinical evaluation of dystonia by Kailash Bhatia
- Ultrasound diagnostics for cervical dystonia by Tobias Bäumer
- RCT on intrathecal baclofen in dystonia by Laura Bonouvrié

CARE STANDARDS - DIAGNOSIS

Diagnostic flowchart for Dystonia

 English

Diagnostic flowcharts for Paroxysmal Disorders

 English

CARE STANDARDS - CLINICAL RATING SCALES

Clinical Rating Scales for Dystonia

 English

Clinical Rating Scales for NBIA

 English

CARE STANDARDS - THERAPEUTIC ALGORITHMS

Therapeutic algorithms for dystonia

 English

Therapeutic algorithm for paroxysmal disorders

 English

CARE STANDARDS - GUIDELINES

- Guidelines for Dystonia
- Guidelines for NBIA

PATIENT JOURNEY

Patient Journey - Cervical dystonia

 English

A patient journey map for people living with cervical dystonia

 English

Patient Journey about dystonia here as a flyer

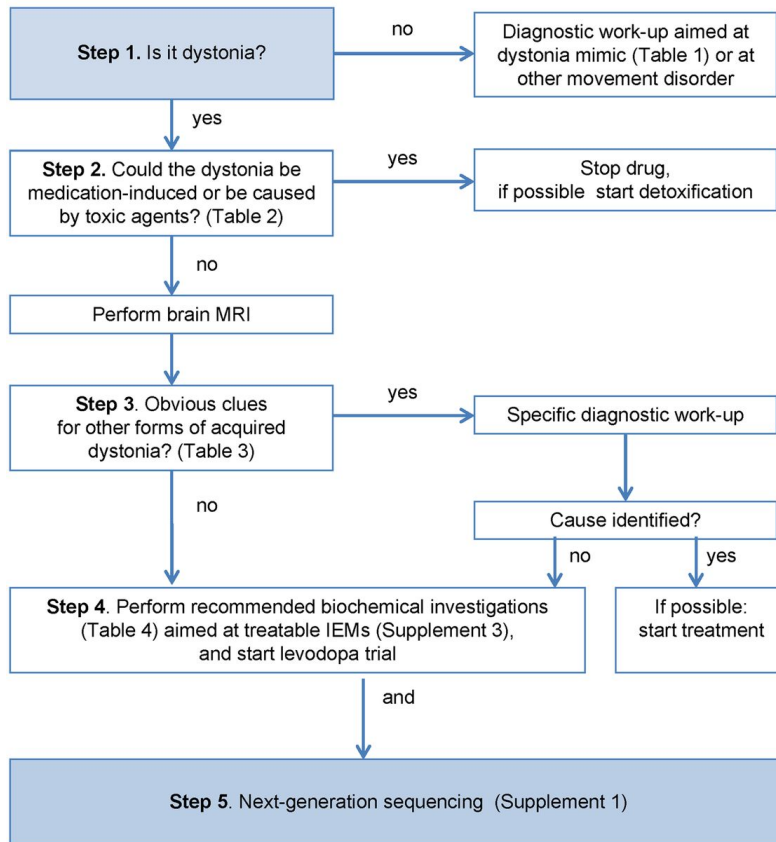
 English

REVIEW

Dystonia in children and adolescents: a systematic review and a new diagnostic algorithm

Martje E van Egmond,¹ Anouk Kuiper,¹ Hendriekje Eggink,¹ Richard J Sinke,² Oebele F Brouwer,¹ Corien C Verschuuren-Bemelmans,² Deborah A Sival,³ Marina A J Tijssen,¹ Tom J de Koning^{2,3}

Diagnostic algorithm of dystonia in children and adolescents



Endorsement

European Reference Network
for rare or low prevalence complex diseases
Network
Neurological Diseases (ERN-RND)

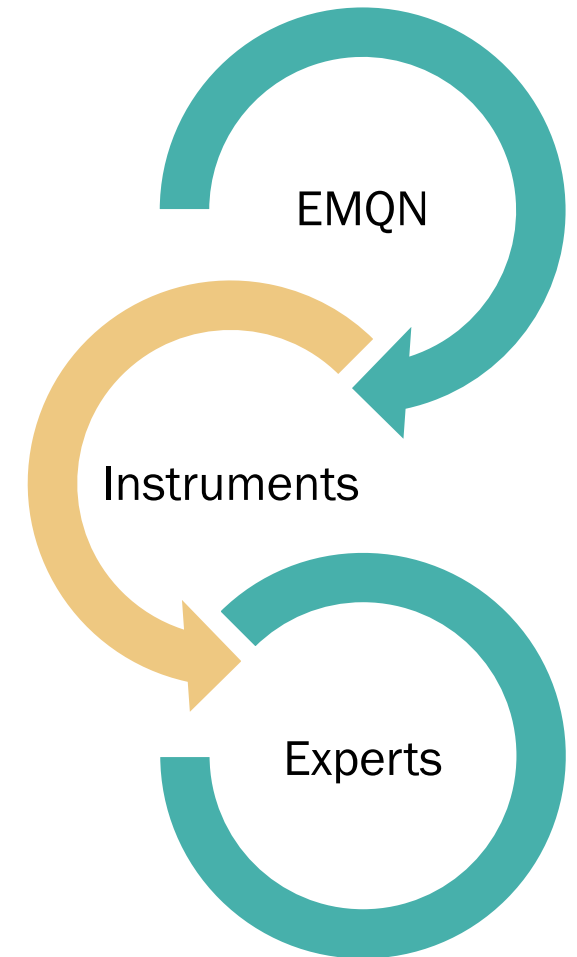
Diagnostic flowcharts for Dystonia:
(1) In adults
(2) In children & adolescents

Published by ERN-RND: 11th February 2019

www.ern-rnd.eu

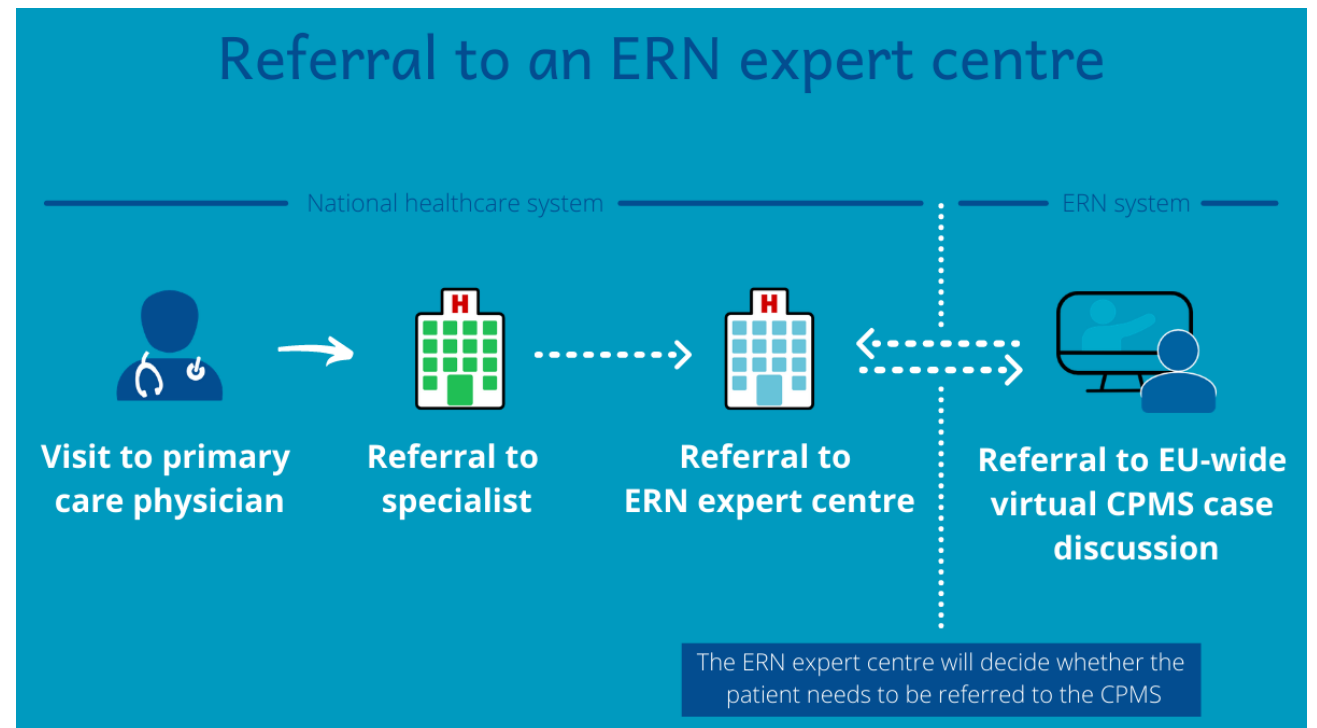
IMPROVED DIAGNOSIS THROUGH BETTER QUALITY OF NGS-BASED DIAGNOSTICS

- Evidence-based process
 - EMQN: iterative check for quality of NGS diagnosis
 - Identify instruments that are used for NGS diagnosis
 - Collect expert opinion
 - Guideline development

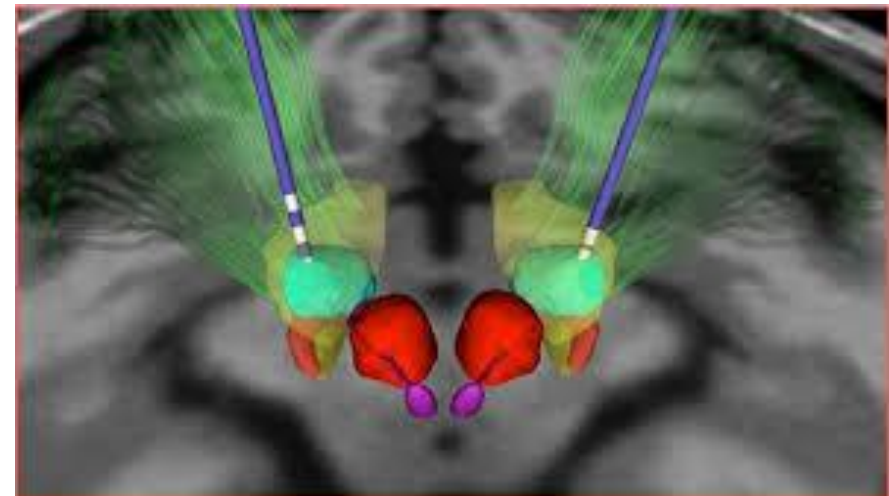
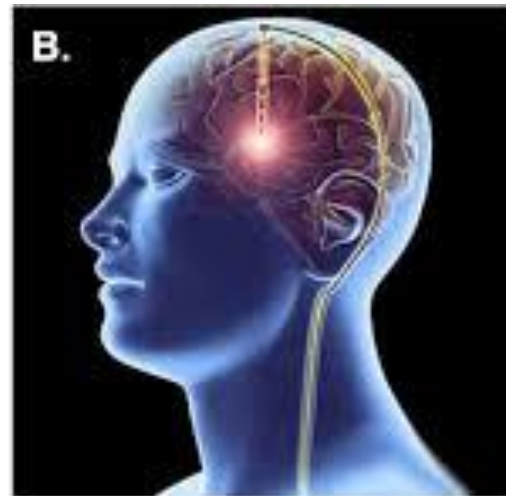
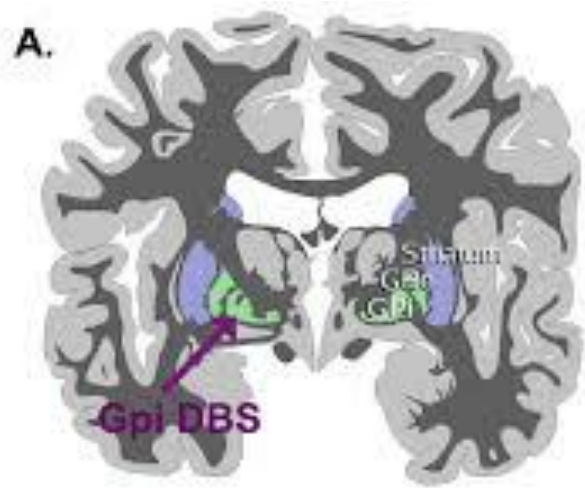


VIRTUAL CASE DISCUSSIONS – THE CLINICAL PATIENT MANAGEMENT SYSTEM - CPMS

- Multidisciplinary online case discussions in a secure online environment
 - Unsolved cases
 - Case management of complex cases
- Care pathway via expert centres



Systematic Review and Expert Recommendations „DBS as a therapeutic option in monogenic dystonia“



ERN-RND REGISTRY

- RND registry
 - In near future: All patients seen in ERN-RND
 - Minimal data set*
 - Availability of patients
 - Disease group specific
 - Diagnosed vs. undiagnosed

*According to the "Set of common data elements for Rare Diseases Registration" aiming at increasing interoperability of RD registries.

https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements_en

- Collaboration with registry for DBS in paediatric Dystonia patients, hosted in Cologne, Germany

Minimal data set
Pseudonym
Year of birth
Sex
Patient status
Date of death
First contact with specialized centre
Age at onset
Age at diagnosis
Orpha code
OMIM code
HPO code
Agreement
Consent for reuse of data
Biological sample
Classification of disability

WORKING GROUP PAEDIATRIC ISSUES

- Paediatric expertise for rare neurological diseases
- Transition
- Paediatric scales: Which? When? Where? What?



Your contribution is highly appreciated, please get in contact with us!

Supporting partner status for individual experts.

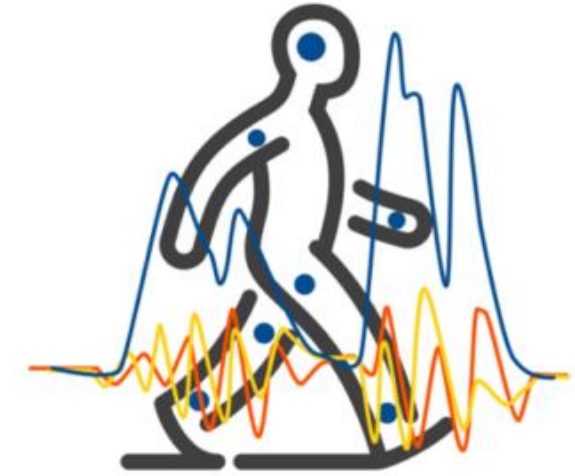


PARAMEDIC TREATMENT



WORKING GROUP NEUROREHABILITATION

- Neurorehab expertise for rare neurological diseases
- ICF... core-set development
- Endorsement of neurorehab guidelines
- Telerehabilitation
- Auditory walking signals as early signs for rare neurological diseases



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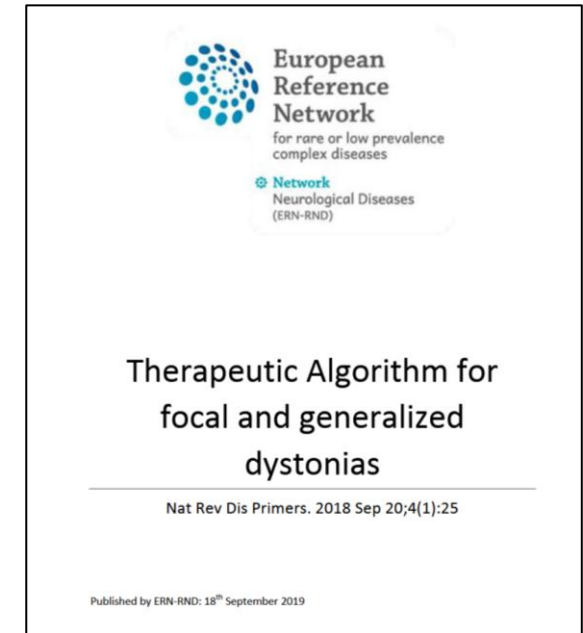


DISEASE MANAGEMENT/ TREATMENT



... AGAIN...

- Disease knowledge – therapeutic algorithms
- Transition
- CPMS case discussions
- Guideline development





DYSTONIA CARE PATHWAY



DBS IN DYSTONIA AS HIGHLY SPECIALISED SERVICE

Journal of Neurology (2023) 270:797–809
https://doi.org/10.1007/s00415-022-11412-4

ORIGINAL COMMUNICATION

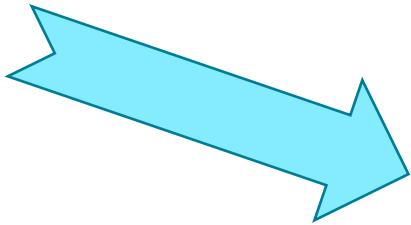
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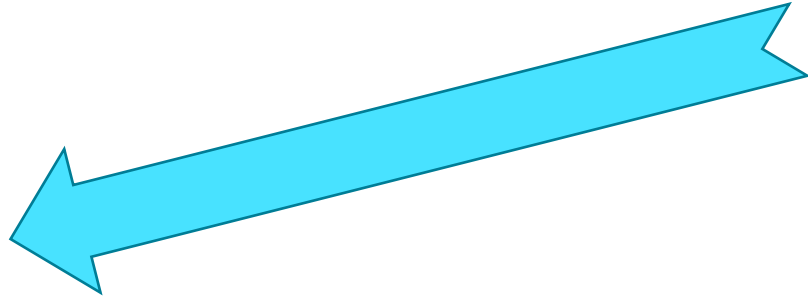
Received: 19 July 2022 / Revised: 28 September 2022 / Accepted: 29 September 2022 / Published online: 6 October 2022
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Abstract
Background Since the first European-wide evaluation of dystonia management in 2016, several efforts have been made to improve dystonia-care. One of these was the development of the Dystonia Disease Group as a part of the European Reference Network for Rare Neurological Diseases (ERN-RND) that implemented several initiatives based on the recommendations made in 2016.

Assessing care

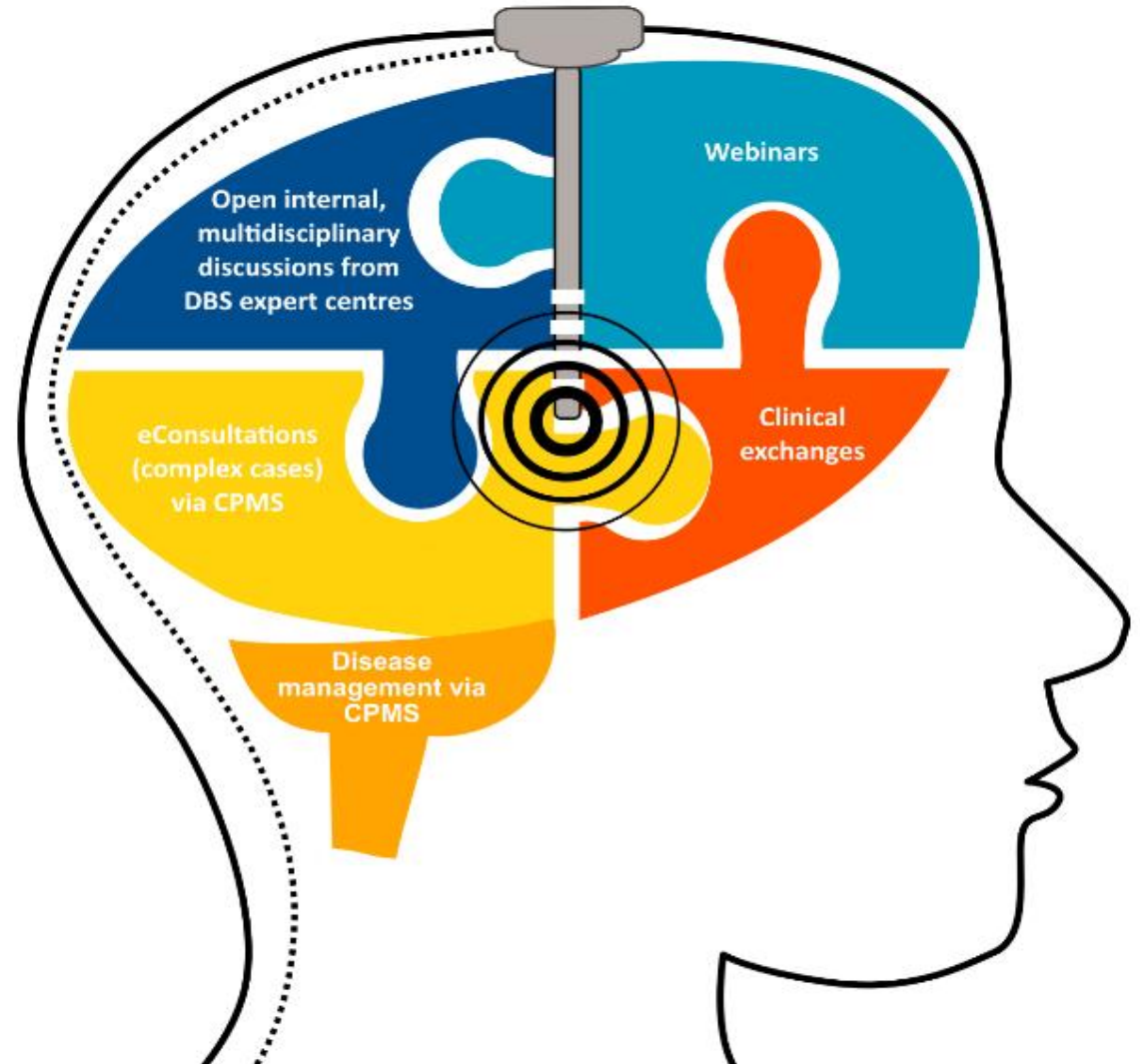


Monitoring

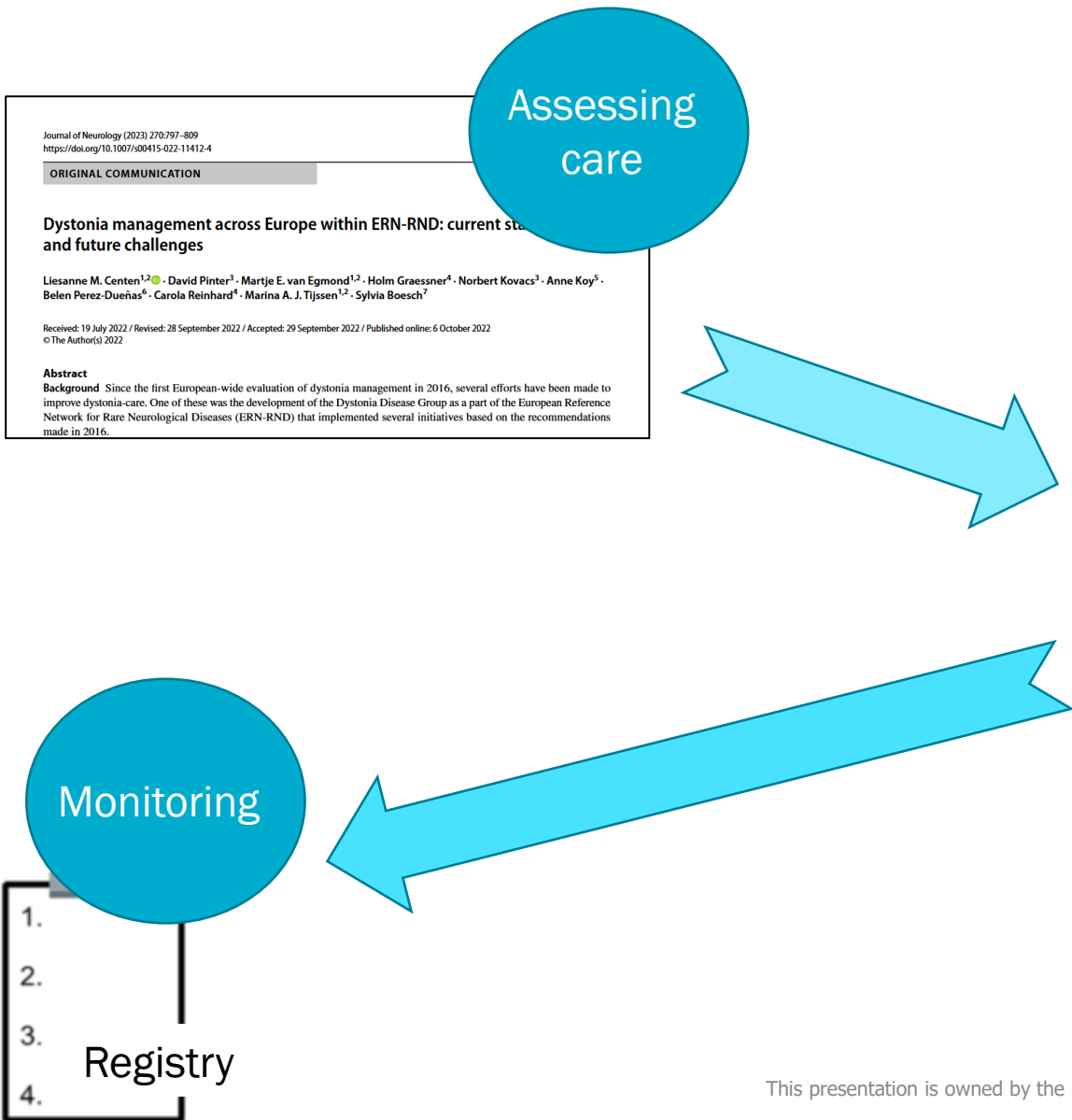


1.
2.
3.
4.
Registry

Deep Brain Stimulation in Dystonia Activities in ERN-RND



DBS IN DYSTONIA AS HIGHLY SPECIALISED SERVICE



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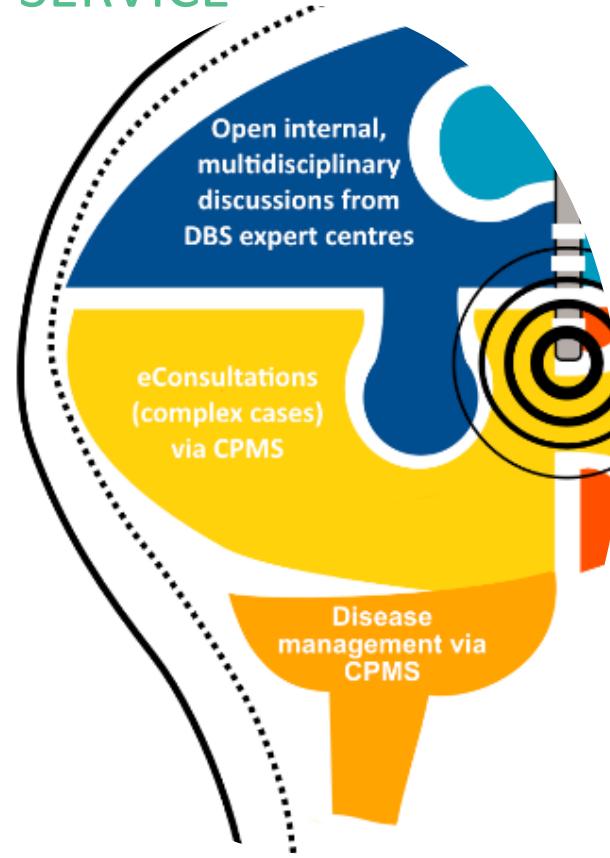
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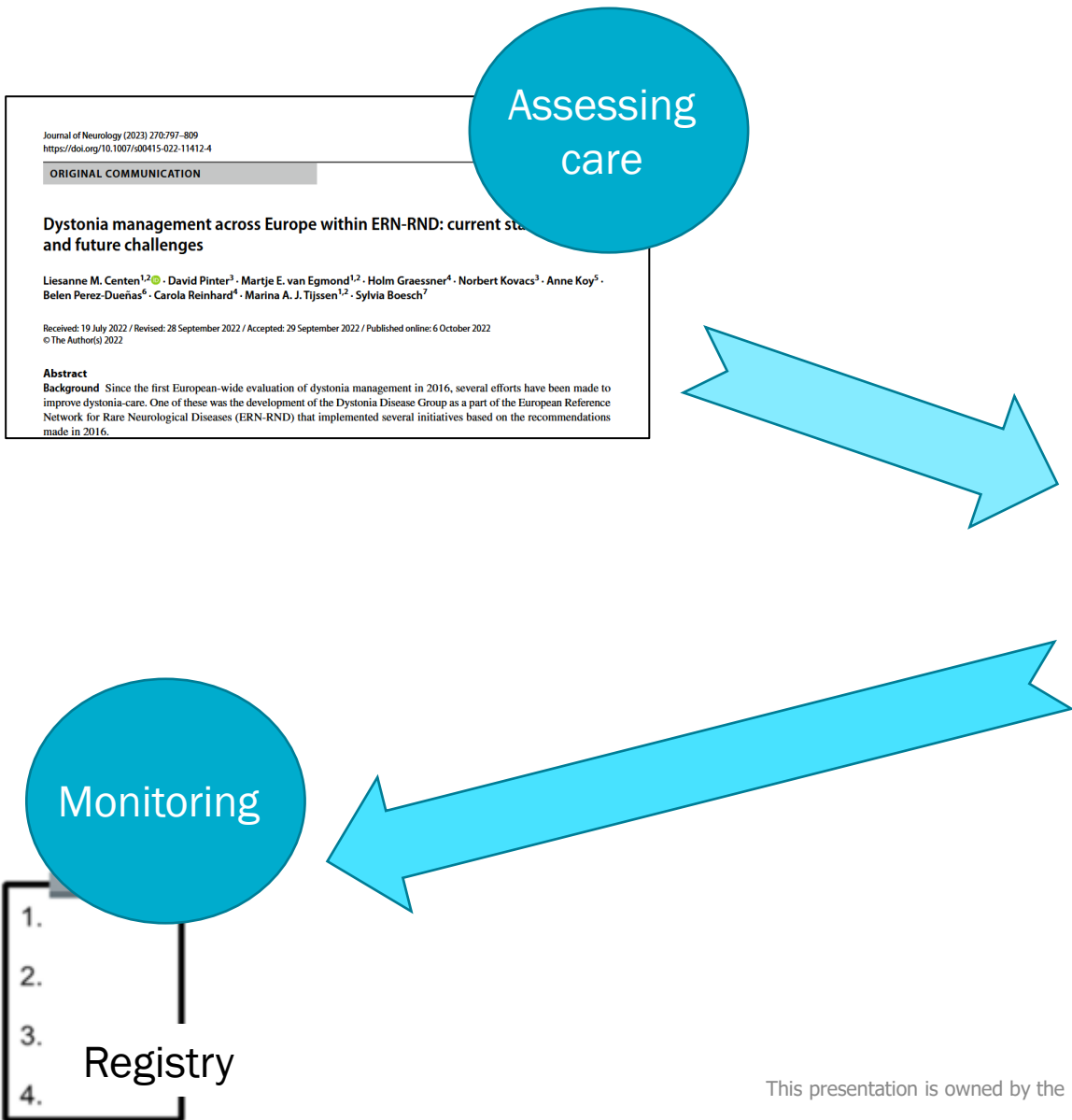
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Virtual online case discussions in CPMS

„The knowledge travels, not the patient“

DBS IN DYSTONIA AS HIGHLY SPECIALISED SERVICE



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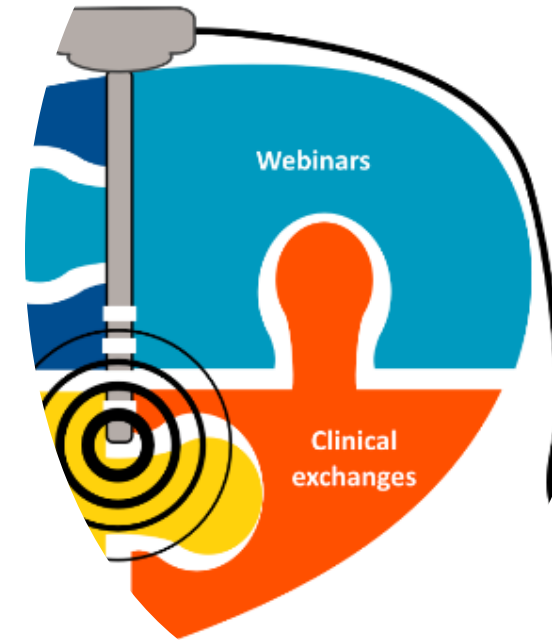
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Training and education activities

ERN-RND WHAT IS IN THERE FOR YOU?

- Free educational webinars on rare neurological and neuromuscular diseases
- Collaboration with Euro-NMD and European Academy of Neurology
- Topics:



Disease-Specific



Neurorehabilitation



Advanced Therapies



Paediatric



Clinical Studies



Deep Brain Stimulation

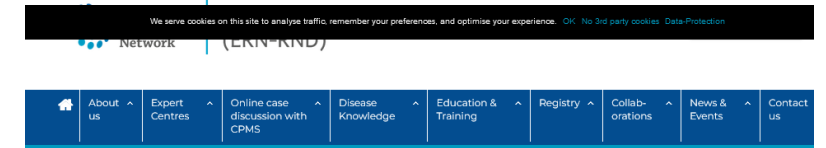
Focus:

Date	Topic	Speaker	Institution	ERN	Focus
23. May 2023, 3 - 4 pm CET	Frontotemporal dementia vs. primary psychiatric disorders	Tim van Langenhove	University Hospital Ghent, Belgium	ERN-RND	
13. June 2023, 3 - 4 pm CET	Dissecting Fahr's disease	Amit Batla and Francesca Magrinelli	Institute of Neurology, University College London, UK	ERN-RND	
20. June 2023, 3 - 4 pm CET	Haematopoietic Stem Cell Transplantation in Leukodystrophies	Jörn-Sven Kühl	University Hospital Leipzig, Germany	ERN-RND	
27. June 2023, 3 - 4 pm CET	Huntington disease: new insights into molecular pathogenesis and therapeutic opportunities	Sarah Tabrizi	UCL Queen Square Institute of Neurology, London, UK	ERN-RND	
6. July 2023, 4 - 5 pm CET	Cardiac Involvement in Neuromuscular Diseases	Karim Wahbi	APHP Hospital Cochin, Paris, France	EURO-NMD	
12. September 2023, 3 - 4 pm CET	Friedreich Ataxia	Kathrin Reetz	RWTH-University Hospital Aachen, Germany	ERN-RND	
26. September 2023, 3 - 4 pm CET	Neuropsychological assessment in primary progressive aphasia – recommendations and pitfalls in clinical practice	Lize Jiskoot	Erasmus Medical Center Rotterdam, Netherlands	ERN-RND	
10. October 2023, 3 - 4 pm CET	Advanced therapies and clinical trials in Leukodystrophies	Caroline Sevin & Nicole Wolf	Reference Centre for Leukodystrophies, University Hospital Bicetre, Paris, France & Amsterdam Leukodystrophy Center, VUMC- Amsterdam University Medical Centers, Netherlands	ERN-RND	
17. October 2023, 3 - 4 pm CET	Neuro-ophthalmological presentations of rare neurometabolic diseases	Tatiana Brémová-Ertl	Center for rare diseases, University Hospital Bern, Switzerland	ERN-RND	
24. October 2023, 3 - 4 pm CET	Newborn screening in leukodystrophies	Lucia Laugwitz	University Hospital Tübingen, Germany	ERN-RND	
9. November 2023, 4 - 5 pm CET	Pathology of Peripheral Nerves	Joachim Weis	University Hospital RWTH Aachen, Germany	EURO-NMD	
14. November 2023, 3 - 4 pm CET	Diagnostic workup and prognostic implication for sleep disorders in atypical parkinsonisms	Giovanna Calandra Buonaura	University of Bologna, IRCCS Institute of Neurological Sciences, Bologna, Italy	ERN-RND	
05. December 2023, 3 - 4 pm CET	Diagnostic workup for genito-urinary dysfunctions in atypical parkinsonism	Pietro Guaraldi	Azienda USL di Bologna, IRCCS Institute of Neurological Sciences, Bologna, Italy	ERN-RND	

www.ern-rnd.eu/education-training/webinars/

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Welcome to ERN-RND

YOUR NETWORK FOR RARE NEUROLOGICAL DISEASES IN EUROPE

The ERN-RND is a European Reference Network established by the EU to support patients and families affected by rare neurological diseases (RND) which requires much specialised knowledge, treatment and resources.

Some diseases are so rare that the necessary knowledge might not be available in the patient's nearest health centre or country. The patient doesn't travel but knowledge does. Specific information about the diseases ERN-RND covers can be found in the [Disease Knowledge](#) section.

European Reference Networks (ERNs) are virtual networks connecting healthcare professionals around Europe with expertise in rare diseases which allows them to discuss a patient's diagnosis and care, with their consent, via an online IT platform called the [Clinical Patient Management System \(CPMS\)](#).

ERN-RND unites 64 of Europe's leading expert centres and 4 affiliated partners in 24 European countries and includes highly active patient organisations. Centres are located in Austria, Belgium, Bulgaria, Croatia, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Luxembourg, Malta, Netherlands, Poland, Slovenia, Spain and Sweden.



Disease Knowledge



Online case discussion



Expert Centres

WEBINAR SCHEDULE



LATEST NEWS

ERN-RND supports Rare Disease Day

November 17, 2024
Rare Disease Day is the official international awareness-raising campaign for rare diseases which takes place on the last day of [...read more >](#)

www.ern-rnd.eu/news/newsletters/

Many thanks!

