ERN-RND Board Meeting

Minutes

Location: Zoom meeting
Date: 29 November 2021, 14:00 - 17:00 (Berlin time)

Attendees:

Samih Almudafar

Angelo Antonini

Enrico Bertini

Kailash Bhatia

Sylvia Boesch

Odile Boespflug-Tanguy

Klara Bozova

Pietro Cortelli

David Crosiers

Susanne de Bot

Alexandra Durr

Antonio Federico

AlessandroFilla

Michael Freilinger

Matthias Gerberding

Holm Graessner

Francisco Grandas

Dimitri Hemelsoet

Sanja Hermanns

Günter Höglinger

Jon Infante

Alisa Jemelka

Mary Kearney

Thomas Klockgether

Thomas Klopstock

Katja Kollewe

Marina Konig-Thijssen

Lenka Krajcovicova

Bernhard Landwehrmeyer

KristaLazdovska

Michelangelo Mancuso

Caterina Mariotti

Maria Marti

Tamara Martin

Judit Molnar

Alexander Münchau

Sinead Murphy

Dario Ortigoza

Mayke Osterloo

Marit Otto

Celia Painous

Johanna Pera

Klivenyi Peter

Annemarie Post

Kathrin Reetz

Carola Reinhard

Carsten Saft

Dario Saracino

Ludger Schoels

Harro Seelaar

Caroline Sevin

Sandy Siegert

Anna Sulek

Algirdas Utkus

Johanna Uusimaa

Bartvan de Warrenburg

Rik Vandenberghe

Martin Vyhnalek

Nicole Wolf

Ginevra Zanni

1. AGENDA ITEMS

- Update network
- Patient journeys
- Update disease group activities
    - Ataxia & HSP (incl. cross-cutting activity Transition)
    - Choreas & HD
    - Dystonia, paroxysmal disorders & NBIA
    - FTD
    - Leukodystrophies
    - Atypical parkinsonian syndromes
- Update cross-cutting activities
    - Guidelines and pathways
    - Care coordination
    - Education & Training
    - Communication
    - Registry
    - Pediatric issues
    - Neurorehabilitation
- Challenges
- AOB

2. ACTION POINTS

Who?	What?	By when?
CPMS		
Management team, coordination office	Come up with a modified proposal for CPMS implementation, board decision via e-mail	15 January
Patient journeys		
ePAG, management team	Present patient journeys to management team	30 January

ePAG, coordination office	Disseminate patient journeys to professional societies and patient organisations	30 January
Challenges		
All	Contact coordination office if they should get in contact with the hospital management again to make clear that activities don't come without funding	N/a

3. MINUTES

- see attached slides

3.1. UPDATE NETWORK

Supporting partner integration

DECISION:

- Process and document for inclusion of supporting partners has been agreed unanimously

CPMS

- Proposal
  - each HCP to assign a CPMS manager to help with local training and case management
  - each HCP to contribute to one case per year per disease group covered by the respective HCP
- Discussions
  - concerns were raised that the proposed procedure is not possible without funding
  - concerns were raised that the CPMS is difficult to use, cases are discussed outside the CPMS
  - it was clarified that a new and easier to use CPMS has been launched, those participants that have already worked with it are happy with it
  - wording might be changed to "aim for"

DECISION and ACTION POINT:
The proposal should be given back to the management team to come up with a modified proposal. Decision by the board should be done e-mail.

3.2. PATIENT JOURNEYS

ACTION POINT:
- To be presented and discussed in the management team, should be disseminated to professional societies and patient organisations

3.3. REGISTRY

- Purpose of the registry

1. Number of patients that are seen within ERN-RND
    - might replace monitoring measure If numbers are equal to numbers counted locally
    - might be too low as not all patients consent
2. Care quality indicator, e.g. number of patients with confirmed genetic diagnosis and scores from disease scales
3. Trial and clinical study cohort formation

NEXT STEPS:
- The registry project manager Dorotea Koepper is on Illness/paternal leave
- Holm Graessner will contact each HCP separately about the registry implementation (ethics and Data Sharing Agreement) and
- Tübingen will support where and how much we can

3.4. CHALLENGES

- Challenge for next 5 years: Integration into national healthcare systems, including funding of activities

ACTION POINT:
- Please contact coordination office if they should get in contact with the hospital management again to make clear that activities don't come without funding.

https://ec.europa.eu/health/ern_en

European Reference Networks

European Reference Network
for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)

www.ern-rnd.eu

Co-funded by the European Union

European Reference Networks

BOARD MEETING

29TH NOVEMBER 2021

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

Co-funded by the EU

Agenda

Network

- Update application for new membership
- Supporting partners collaboration agreement - VOTE
- CPMS use - VOTE
- Collaboration with EpiCARE and Neuro-NMD
- Monitoring/reporting/AMEQUIS
- Status preparation and timelines for next 5 years
- Annual meeting 2022

ePAGs: Patient Journeys

Update of disease group activities

- Ataxia/HSP (incl. Transition)
- Choreas/HD
- Dystonia, paroxysmal disorders, NBIA
- FTD
- Leukodystrophies
- Atypical PD

Update on cross-cutting activities

- Guideline development and Value of treatment
- Compostion of the multidisciplinary team for Movement Didsorders
- Case discussions/Training and education
- Communication
- Registry
- Pediatric issues
- Neurorehabilitation

Upcoming challenges

AOB

European Reference Networks

APPLICATIONS FOR NEW MEMBERSHIP

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

Co-funded by the EU

ENLARGEMENT OF ERNS

IAB assessment process

19th April – 30th April

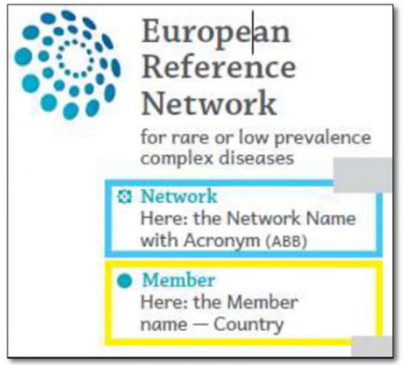
12th May-09th June

August 2021

October 2021

Confirmations expected to be sent by end of November

Official entry date: 01/01/2022



European Reference Networks

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

ENLARGEMENT OF ERN-RND

- 33 applicants from 16 countries
  - 20 new ataxia centres
  - 24 new HD/chorea centres
  - 17 new dystonias/paroxysmal disorders/NBIA centres
  - 16 new FTD centres
  - 9 new leukoencephalopathies centres
  - 22 new atypical Parkinsonian syndromes centres

Status	Country	Applicant	Ataxia/HSP	Choreas/HD	Dystonias/...	FTD	Leukodystrophies	Atypical PD
Applicant	Austria	University Hospital Innsbruck	x	x	x	x		x
Applicant	Belgium	Antwerp University Hospital				x		x
Applicant	Belgium	Ghent University Hospital				x	x	
Applicant	Croatia	Klinicki bolnički centar Zagreb	x	x	x	x	x	x
Applicant	Cyprus	Cyprus Foundation for Muscular Dystony Research	x	x	x	x	x	x
Applicant	Czech Republic	Facultni nemocnice U Sv Anny Brne		x	x	x		x
Applicant	Czech Republic	Thomayer Hospital Prague					x	
Applicant	Denmark	Aarhus Universitets Hospital	x	x	x	x		x
Applicant	Denmark	Rigshospitalet Copenhagen	x	x	x	x	x	x
Applicant	Finland	Northern Ostrobothnia Hospital District Oulu	x	x		x	x	x
Applicant	Germany	Hannover Medical School	x	x	x		x	x
Applicant	Germany	Katholisches Klinikum Bochum		x				
Applicant	Germany	Universitätsklinikum Aachen	x	x	x	x		x
Applicant	Germany	Universitätsklinikum Würzburg		x	x			x
Applicant	Greece	Eginito Hospital Athens	x	x	x	x		x
Applicant	Hungary	Szent-Györgyi Albert Medical School	x					
Applicant	Ireland	Tallaght University Hospital	x					x
Applicant	Italy	AO di Padova	x	x	x	x	x	x
Applicant	Italy	AOU Pisana Pisa	x					
Applicant	Italy	AOU Policlinco Bari		x				
Applicant	Italy	Azienda Ospedaliera Universitaria Federico II Napoli	x	x				x
Applicant	Italy	UOCD Clinica Neurologica Rete Metropolitana NEUROMET Bologna	x	x	x	x		x
Applicant	Netherlands	Leiden University Medical Center		x				
Applicant	Netherlands	Maastricht University Medical Center		x				
Applicant	Poland	Institute of Psychiatry and Psychology Warsaw	x					
Applicant	Spain	Complejo ospitaliario Regional Virgen del Roci		x	x			x
Applicant	Spain	Hospital Clinic San Carlos Madrid			x			x
Applicant	Spain	Hospital General Universitario Gregorio Maranon		x	x			x
Applicant	Spain	Hospital Universitario Central de Asturias Oviedo	x	x	x	x		x
Applicant	Sapin	Hospital Universitario La Paz Madrid	x	x	x	x	x	x
Applicant	Spain	Hospital Universitario Narques de Valdecilla Santander	x			x		
Applicant	Sweden	Centre for Huntington's Disease Sweden		x				
Applicant	Sweden	Karolinska Universitetssjukhuset	x	x				x
Full member	Belgium	Université libre de Bruxelles	X					
Full member	Belgium	University Hospitals Leuven	X	X		X		X
Full member	Bulgaria	University Neurological Hospital "St. Naum" Sofia			X			
Full member	Czech Republic	Charles University Prague		X	X			X
Full member	Czech Republic	Charles University in Prague Faculty Hospital Motol	X					
Full member	France	Reference Center (associated center) for Rare Multiple System Atrophy (Bordeaux)						X
Full member	France	Reference Centre for Rare Diseases 'Neurogenetics', Paris	X		X			
Full member	France	Reference centre for rare dementias (Paris)				X		
Full member	France	Reference centre for Huntington's disease (Paris)		X				
Full member	France	Reference Center for Rare MultipleSystem Atrophy diseases (Toulouse)						X
Full member	France	APHP - Reference Centre for Leukodystrophies					X	
Full member	Germany	Universitätsklinikum Tübingen	X	X	X	X	X	X
Full member	Germany	Universitätsklinikum Bonn	X		X			X
Full member	Germany	University of Lübeck	X	X	X			X
Full member	Germany	Klinikum der Ludwig- Maximilians-Universität München	X	X	X	X	X	X
Full member	Germany	University Hospital Ulm		X	X	X		X
Full member	Hungary	Semmelweis University Budapest	X	X	X	X	X	X
Full member	Hungary	University of Pécs, Medical school	X	X	X	X		X
Full member	Italy	Istituto Neurologico Carlo Besta, Milan	X	X	X	X	X	X
Full member	Italy	Istituto Clinico Humanitas di Rozzano, Milan		X	X	X		X
Full member	Italy	Azienda ospedaliera Universitaria Senese, Siena	X	X	X	X	X	X
Full member	Italy	Bambino Gesù Children's Research, Rome	X	X	X		X	
Full member	Lithania	Vilnius University Hospital		X				
Full member	Netherlands	VU University Medical Center, Amsterdam					X	
Full member	Netherlands	University Medical Center Groningen	X	X	X		X	
Full member	Netherlands	Erasmus MC: Universitair Medisch Centrum Rotterdam				X		
Full member	Netherlands	Radboud University, Nijmegen Medical Center	X		X			X
Full member	Poland	University Hospital Krakow	X		X			X
Full member	Slovenia	University Medical Centre Ljublijana	X	X	X	X		X
Full member	Spain	Hospital Sant Joan de Deu, Barcelona	X	X	X			X
Full member	Spain	Val d'Hebron	X					
Guest	UK	University College London Hospitals	X	X	X	X	X	X

IAB - Eligible Applications% – Final IAB Assessment - Favourable Assessments	AT	BE	BG	CY	CZ	DE	DK	EE	EL	ES	FI	FR	HR	HU	IE	IT	LT	LV	NL	NO	PL	PT	RO	SE	SI	SK	Total:
BOND	1					3	3			4	1	1			1	4			2	1		1					22
CRANIO	1	3				1				1		1		1	1	1	1		1	2							14
Endo-ERN		1			1	4	1		3	2	1	2			1	10			1		2	2	2	1			34
EpiCARE	2	1				2					1	3		1		2				1					1		14
ERKNet		1			1	5	3		1	4		2		1	1	4			3		2	1	1		1		31
ERN-EYE		2				5	1	1		7	1	1			1	5			1				1	1	1		28
ERNICA		2			1	4				3		1				5			1		2			2			21
ERN-LUNG		3		1	1	1				1	1			2	1	8		1	2	1		2		1	1	1	28
ERN-RND	1	2		1	2	4	2		1	6	1		1	1	1	5			2		1			2			33
ERN-SKIN	1															6			1								8
EURACAN		2			2	2			1	3	1	7			1	12	1		1		2	1	1	1			38
EuroBloodNet		1			2	7	2		4	3	1	1		1		13					1						36
eUROGEN	1	1			2	5				7	1	1	1			3			4		1			2			29
EURO-NMD						2	2		1	2	1	2			1	6				1		1					19
GENTURIS		1			2	3	2		1		1					9	1	1	2			2					25
GUARD-HEART		1			1	1	1			4		1	1	1	1	4			3		1	3					23
ITHACA		2				6	3	1		3	1		1	2	1	5		1	1		3	1				1	32
MetabERN						1		1	1	2	1	2			1	7		1	1		2	1	1			1	23
PaedCan		1				10	1		1	2	1			1	1	3			1	1	3	1		1		1	29
RARE-LIVER		1			1	5	1		1	1	1	2		2		10	1		1		1	1					29
ReCONNET		1			1	1	1		1	3	1	3				12			3		1	1		1			30
RITA		1			1	8	2		2	3	1	3	1	1	1	9			2	1	3	2		1		1	43
TRANSPLANTCHILD		1				1	2			5	1				1	1			2	1				1			16
VASCERN						3	2			2		1				1	1		1	2		1		1			15
Total:	**7**	**28**		**2**	**18**	**84**	**29**	**3**	**18**	**68**	**18**	**34**	**5**	**14**	**15**	**145**	**5**	**4**	**36**	**11**	**25**	**21**	**6**	**15**	**4**	**5**	**620**

AFFILIATED PARTNERS – CURRENT STATE OF PLAY

	COUNTRIES	AT	BE	BG	CY	CZ	DE	DK	EE	EL	ES	FI	FR	HR	HU	IE	IT	LT	LU	LV	MT	NL	NO	PL	PT	RO	SE	SI	SK
	BOND	1						3			5	1		1	1			2	1	1	1							1	
	CRANIO	1						2										1	1	1	1			1				1	
	Endo-ERN	4			1							1		2						3	1		2						2
	EpiCARE	2			1			2	2					2	1			2	1	1	1							1	
	ERKNet	2						3	2						1				1	2	1		1					2	
	ERN-EYE	4									7			1	1				1		1							1	1
N	ERNICA	4							1		4			1	1			2	1	2	1							1	
E	ERN-LUNG	4			1				1			1		1	1			2	1	3	1							1	2
T	ERN-RND	2						2	2			1		1					1	1	1								
W	ERN-SKIN																		1	1	1							1	
O	EURACAN	1			1				2					2					1	1	1								
R	EuroBloodNet	2						2	1						1				1		1							1	1
K	eUROGEN	1									9			1	1				1	1	1							1	
	EURO-NMD	2			1			2	1					1				1	1	2	1								
N	GENTURIS	2			1			3	1										1	1	1		1						
A	GUARD-HEART	2							1					1	1			2	1	1	1							1	1
M	ITHACA	1						3			5				1				1	2	1		1					1	
E	MetabERN	4																	1	2	1								1
	PaedCan			1	1				2					3					1		1					2			1
	RARE-LIVER	1							1					2	1			1	1	2	1		1					1	
	ReCONNET	1						2	1		2			1	1			1	1	3	1								
	RITA	3						2	1					3	1			2	1	1	1								1
	TransplantChild	1						2	1					1	1				1	1	1							1	
	VASCERN	3			1						2								1		1							1	

Legend: Associated National Center | National Coordination Hub

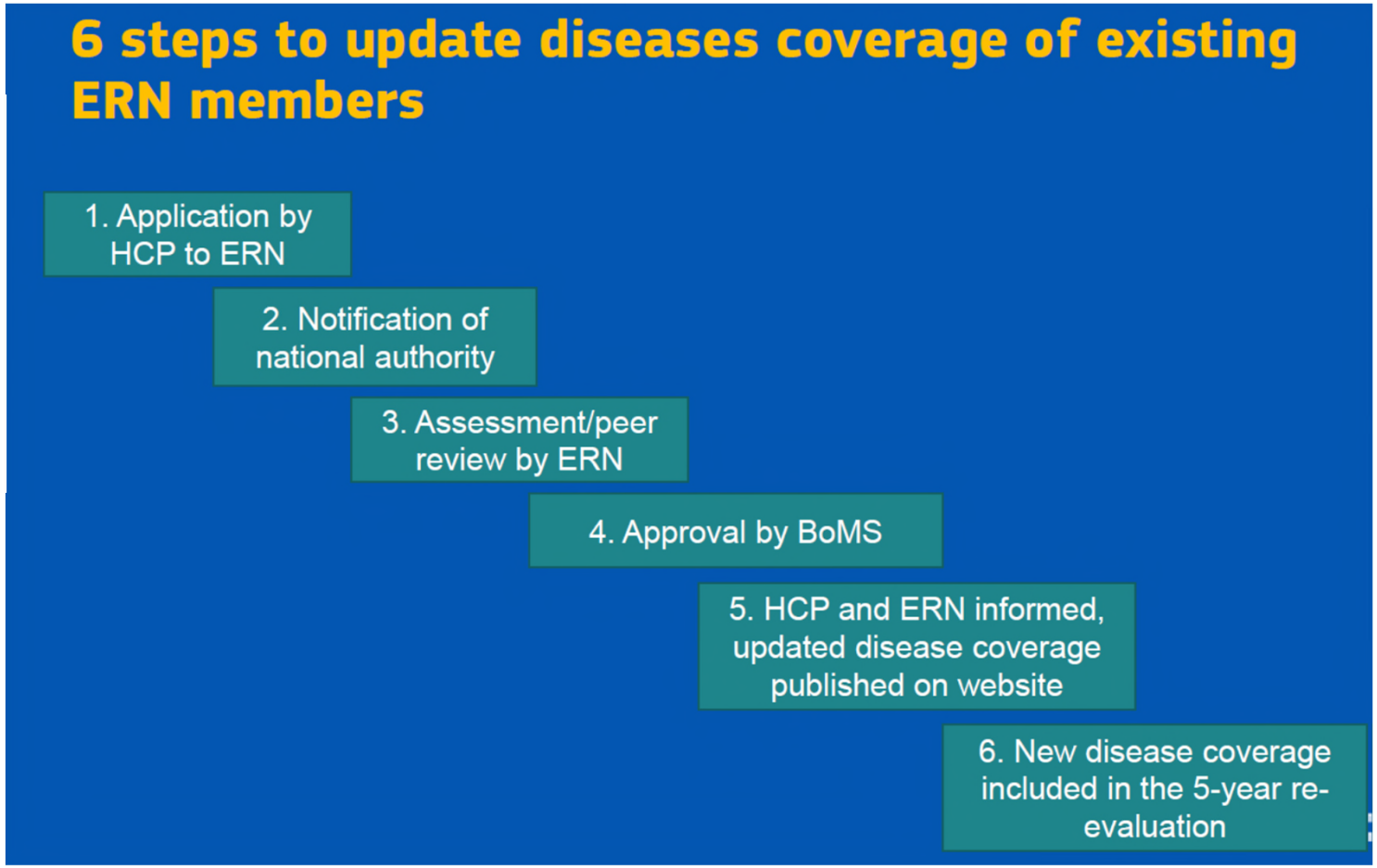
AFFILIATED PARTNERS - TERMINATION

COUNTRIES		AT	BE	BG	CY	CZ	DE	DK	EE	EL	ES	FI	FR	HR	HU	IE	IT	LT	LU	LV	MT	NL	NO	PL	PT	RO	SE	SI	SK
NETWORK NAME	BOND	1						3			5	1		1	1			2	1	1	1							1	
	CRANIO	1						2										1	1	1	1			1				1	
	Endo-ERN	4			1							1		2						3	1		2						2
	EpiCARE	2			1			2	2					2	1			2	1	1	1							1	
	ERKNet	2						3	2						1				1	2	1		1					2	
	ERN-EYE	4									7			1	1				1		1							1	1
	ERNICA	4							1		4			1	1			2	1	2	1							1	
	ERN-LUNG	4			1				1			1		1	1			2	1	3	1							1	2
	ERN-RND	2						2	2			1		1					1	1	1								
	ERN-SKIN																		1	1	1							1	
	EURACAN	1			1				2					2					1	1	1								
	EuroBloodNet	2						2	1						1				1		1							1	1
	eUROGEN	1									9			1	1				1	1	1							1	
	EURO-NMD	2			1			2	1					1				1	1	2	1								
	GENTURIS	2			1			3	1										1	1	1		1						
	GUARD-HEART	2							1					1	1			2	1	1	1							1	1
	ITHACA	1						3			5				1				1	2	1		1					1	
	MetabERN	4																	1	2	1								1
	PaedCan			1	1				2					3					1		1					2		1
	RARE-LIVER	1							1					2	1			1	1	2	1		1					1	
	ReCONNET	1						2	1		2			1	1			1	1	3	1								
	RITA	3						2	1					3	1			2	1	1	1								1
	TransplantChild	1						2	1					1	1				1	1	1							1	
	VASCERN	3			1						2								1		1							1	

MANAGEMENT OF DISEASE AREAS OF ERN-RND MEMBERS – ANNUAL CYCLE

Practical steps

- Standardised template for application to be circulated by EC to ERNs
- ERNs to continuously collect applications and assess them according to their internal arrangements (including the requirement of notification by HCP to the Member States' competent authority)
- ERNs to submit all applications with positive assessment received during this period to EC by 31 January 2022 (one joint submission)
- EC transmits the consolidated package of applications from all ERNs to BoMS: February 2022
- Approval of applications with positive assessment by BoMS: Spring 2022 BoMS meeting (March/April)



European Reference Networks

SUPPORTING PARTNERS

ERN-RND – DEFINITION OF SUPPORTING PARTNERS

- In addition to Full Members and Affiliated Partners of the ERN networks, there is another term used to describe organisations or individual experts which officially collaborate with ERNs, namely, Supporting Partners. Definitions of Affiliated Partners and the process of their designation by the Member States to work with ERNs can be found here: https://ec.europa.eu/health/ern/board_member_states_en
- HCP or expert

ERN-RND – DEFINITION OF SUPPORTING PARTNERS

- HCP:
  - If [INSERT] is a healthcare provider, [INSERT] will have to be situated in a European Country from which no full or associated members of ERN-RND is situated in and will also be asked to provide evidence that they fulfill the specific criteria defined for ERN-RND.
  - [INSERT] will receive a data collection spreadsheet corresponding to the ERN monitoring exercise twice a year from the Coordination Team for the Disease Area applied for and asked to provide their numbers and information
  - Same obligations as ERN-RND member with regard to CPMS and ERN-RND registry
  - Enquire why respective HCP is not in ERN-RND and as to whether it plans to do so

ERN-RND – DEFINITION OF SUPPORTING PARTNERS

- Expert:
  - If [INSERT] is a single clinician or expert they will have to be nominated by a disease or working group to contribute to a certain activity
  - Enquire why the respective HCP is not in ERN-RND and as to whether it plans to do so

VOTE TO IMPLEMENT SUPPORTING PARTNERS

Vote of ERN-RND board to implement supporting partners

European Reference Networks

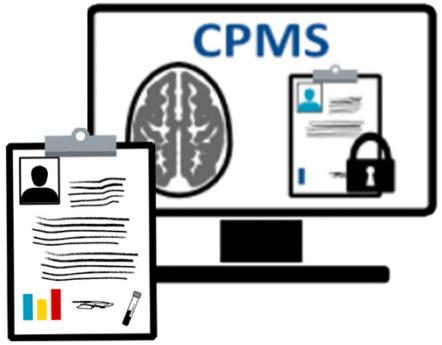
CPMS

CPMS AND CASE DISCUSSIONS

Commiting to cross-border healthcare in 2022:

Nominate CPMS manager for your HCP

- Contact point for ERN-RND CPMS helpdesk
- Multipliers for CPMS training
- Manage centre‘s panels



Submit cases for Online Case discussions

Offer expert advice for Online Case Discussions

→ Contribute to one Case Discussion per DG covered by your HCP /year

→ Soon you will receive more information via mail

CPMS VOTE

- Install CPMS manager per HCP: primary contact point for ERN-RND CPMS helpdesks (coordination office

- Contribution to one case per disease group in Online Case Discussions (either advising or case submission) per year

European Reference Networks

COLLABORATION WITH EPICARE AND EURO-NMD

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

Co-funded by the EU

Collaboration with EpiCare and Euro-NMD

1. ***Joint training curriculum – lead ERN-RND***
2. ***Next Generation Sequencing***
3. *Registry and European Health Data Space*
4. *Gene therapy and stem-cell transplantation*
5. *Surgical therapies*
6. ***Mitochondrial diseases***
7. *Channelopathies*
8. *Neurophysiology / myoclonus – lead ERN-RND*
9. *Neurometabolic diseases – lead ERN-RND*

European Reference Networks

AMEQUIS/ MONITORING

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

Co-funded by the EU

AMEQUIS – WHAT IS IT?

- AMEQUIS = Assessment, Monitoring, Evaluation Quality Improvement System
  EC project run by Nivel Foundation, NL, and Donabedian Foundation, SP, ends II/2022

- Methodology: AMEQUIS organises workshops, expert interviews, review of material
- Workshops 1st round Spring 2021, 2nd round Fall 2021, one person per ERN

General Objective

- To develop an integrated assessment, monitoring, evaluation and quality improvement system (AMEQUIS) of the ERNs, in order to:
  i. assess any new ERN or HCP application;
  ii. monitor the activities carried out and the produced deliverables developed by the approved ERNs and their members;
  iii. evaluate the ERNs; and
  iv. improve the ERN system on the basis of a continuous quality improvement system approach (PDCA) and develop an integrated AMEQUIS model.

AMEQUIS – WHAT HAS BEEN DONE SO FAR?

- Suggestions from ERNs and from AMEQUIS team so far
  - Monitoring should be simplified and collect quality indicators and aim for targets
  - Evaluation should use the locally stored records of indicators of the funding period
  - Evaluation could go by criteria used during self-assessment

- For the new rules of assessment, monitoring and evaluation: **Many criteria and parameters are still not specified** in terms of their definition, scope, application, validation, goal, responsibility to be collected, counted, evaluated

MONITORING OF Q1 + Q2 OF 2021

- **New:** (1) Indicators have to be validated by documentation accessible on the ERN website (2) Clinical and observational trials still count as ERN-RND affiliated when ERN-RND is not mentioned in clinicaltrials.gov or EUDRACT entries and when trials are run locally in only one center

- **Positive**: ERN-RND achieved a collection of 31 of 31 HCPs or of 30 of 31 HCPs which is great.

- **Neutral**: We find fluctuating responses from centers reporting many items in one period, few items in the next – looks highly unlikely.

- **Reminder**: The indicator „research" – „publications acknowledging ERN-RND" – counts **if ERN-RND centers, clinicians and consortia acknowledge ERN-RND in publications**

European Reference Networks

STATUS PREPARATION AND TIMELINES FOR NEXT 5 YEARS

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

Co-funded by the EU

Timelines and Structure

Two phases of next five years

- Phase 1: 18 months – network work plus evaluation
- Phase 2: 36 months – network work
- No concrete deadlines yet

More homogenous work structure of all ERNs

- All ERNs WPs on monitoring, training, CPMS, guidelines, ERN registry and communication
- All ERNs WPs on bespoke topics

STATUS PREPARATION

A. Continuation and consolidation

- Disease Group Activities
- Disease Group Activities – done by all DG
- Disease Group close WG activities (pediatric issues, transition and neurorehabilitation)
- Cross-cutting structural and supporting activities
  - CPMS – at least one case per HCP per disease group per year (panel participation will be recognized, too)
  - ERN-RND registry
  - Guideline development
  - Training and education
  - (Care pathway development)
  - Communication

B. Multi-pillar activities for highly specialized services provided in ERN-RND

- Continue, implement and develop multi-pillar activities (CPMS, training, guideline, policy, etc.) for
  - DBS
  - NGS
  - Advanced therapies (Stem Cell Transplantation, Genetic Therapies)
  - Neuroimaging

STATUS PREPARATION

C. Collaboration with "Neuro" ERNs EuroNMD and EpiCare – complementing and informing ERN-RND specific activities in A. and B.

- WGs
  - Training curriculum – RND training curriculum
  - Next generation sequencing
  - Registries and European Health (Brain) Data Space
  - Gene therapy and stem cell transplantation
  - Surgical therapies (e.g. epilepsy surgery, DBS)
  - Overlapping disease groups: myoclonus, channelopathies, mitochondrial disorders, neurometabolic disorders

D. Collaboration with European and international professional organisations and networks

- EAN, EPNS, MDS Europe and EACD
- Disease networks (EHDN, Ataxia Global Initiative, European Dystonia Network, etc.)
  - Objectives for all collaborations:
    - Promoting RND
    - Leveraging activities in A., B., C. and respective cooperation

E. Equal care and equal access to diagnostics and therapy in ERN-RND

European Reference Networks

ANNUAL MEETING 2022

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

Co-funded by the EU

Annual Meeting 2022

- To be held in Leuven, Belgium
- Early summer 2022

European Reference Networks

PATIENT JOURNEYS

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

Co-funded by the EU

What is a parient Journey?

- Opportunity for systematic patient involvement in design of their care
- Makes the needs of patients visible in their specific rare disease
- It is part text and graphics – show care needs and pathway (at different times of disease)

→Patient journeys are always created by a group of patients, reviewed by expert clinician

What are patient representatives hoping to achieve?

- To identify gaps in care so can pathways can be adopted for the patients benefit

- Information for:

  1) Patients, families,

  2) Clinicians with limited expertise in rare disease i.e. people with Friedreich's Ataxia often present to cardiologist, orthopedic surgeons with scoliosis,

  3) General Practitioners & other Health care professional (OT, physio, Speech therapist)

  4) Public information for rare disease awareness

➔European reference networks (ERN) has tried from its inception to integrate patient and patient organization opinion into patient care

Patient Jopurney - Method

Stages/ over time

	First sign(s)	Symptoms	Diagnosis	Treatment	Monitoring
Disease					
Clinic					
Challenges / needs					
Ideal situation / Goal					

Review needed at this step by clinicians

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

Methology for compiling Patient journey for Friedreich's Ataxia

European Patient Advocacy Group

- Structured questionnaires – which had been validated by Endocrinology - ERN
- 10 patients & family members: 4 women, 6 men; 8 - 40 years old
- Lithuania, Austria, Montenegro, Ireland, Norway, UK

ATAXIA

Friedreich Ataxie Förderverein e.V.

afi Ataxia Foundation Ireland

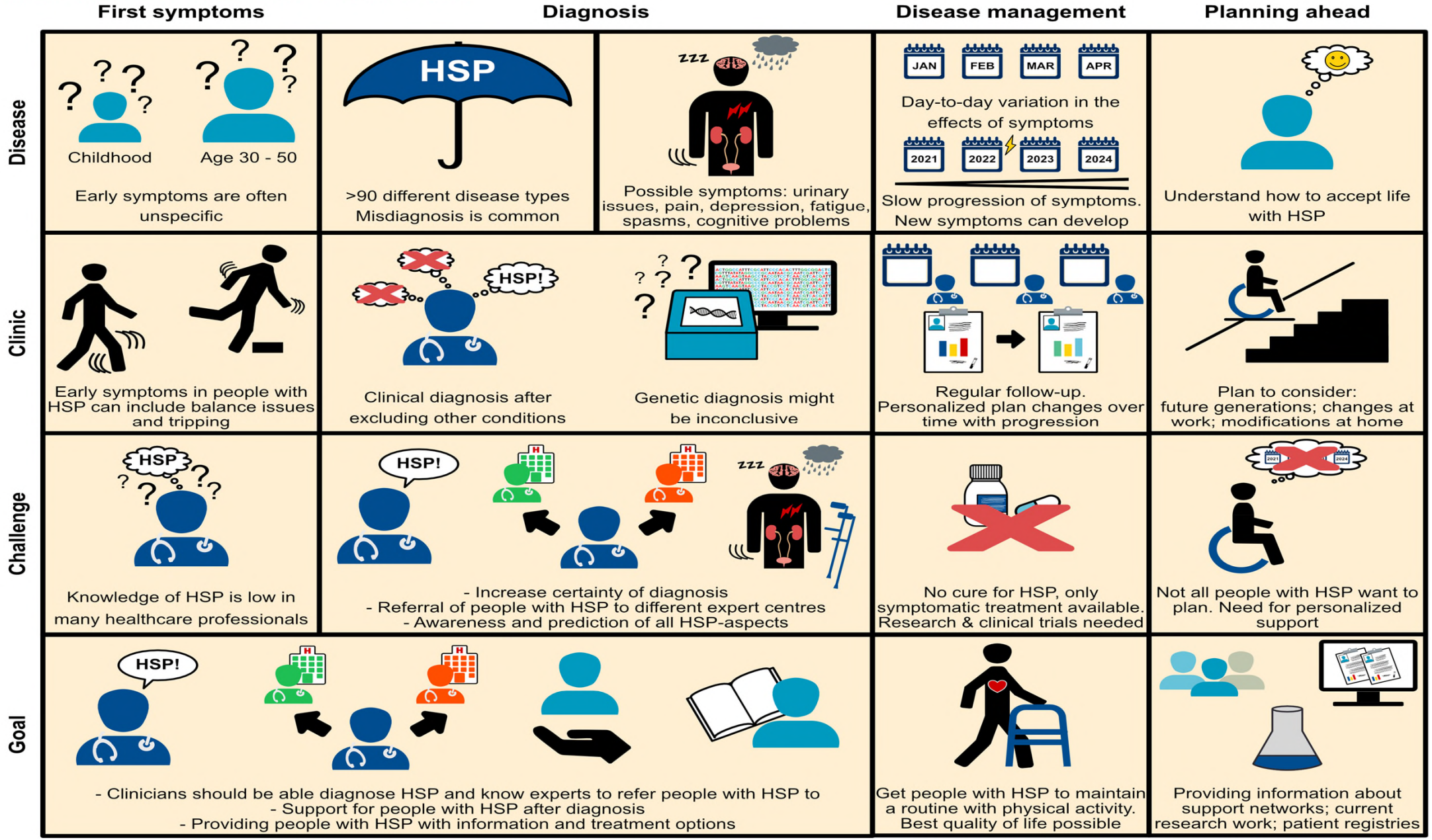
Friedreich's Ataxia Research Alliance IRELAND

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

Patient Journey – word format for Friedreich's Ataxia

Phases	First symptoms	First Symptoms	Genetic Diagnosis	Treatment	Follow-up
Disease	Difficulty walking in the dark, unsteadiness in standing or walking, followed by progressive limb and gait clumsiness	91% of people present with falls, poor balance 9% present with non-neurological symptoms i.e., scoliosis, heart trouble	Genetic testing can be done since 1996	There are no effective disease-modifying therapies available yet	Referral to expert centre with involvement of multi-disciplinary teams for life-long monitoring of the heart and risk of diabetes mellitus is necessary
Clinic	Assessment of symptoms and referral to relevant specialists	Multidisciplinary teams that include neurologists, psychologists, psychiatrists, physiotherapists, speech therapists, social workers, occupational therapists, and nutritionists	Counselling of parents regarding future pregnancies Siblings unless symptomatic are usually not tested before the age of 18 years	1) Full neurological assessment 2) lumbar sacral spine to assess if scoliosis is present 3) ECG and ECHO to rule out cardiac involvement 4) Psychological support for all family members for this life altering condition	Mental Health support as the diagnosis of FA is a life altering disorder
Challenges	Delay in diagnosis: Easy to confuse clumsiness associated with a growth spurt in girls who are 10 years old and boy at 12 years with FA.	Changes are insidious – getting a diagnosis may take many years FA symptom complexity leads to frequent misdiagnosis. 1% of all cases of FA is in a person who has an onset after 60 years of age	1% of all those with FA present are over 60 years of age. Consider the diagnosis in all age groups	No FA clinical trial Encourage use of posterior walker to try and prolong their ability to walk Encourage participation in social activities with peers Encourage parents to avail of outside help if available	The child may not be able to compete with their peers and may retreat into themselves The parents are often traumatised and unsure how to treat the individual with FA. The individual in their 20s' may feel that life is not worth living
Goals	"Clumsiness in a child" should be taken seriously when present with another symptoms Get a 2nd opinion in those with vague complaints of loss of balance	Take parents and child complaints seriously even if they seem trivial and especially if there are multi-system complaints, i.e. poor balance, fatigue, irritability and anxiety	Aunts & uncles, grand-parents of the person with ataxia should be offered genetic counselling and testing to avoid FA presenting in cousins	Care guidelines are available and share them with the person if asked.	Maximise the person's potential to live as normal a life as possible

GRAPHIC REPRESENTATION FOR HSP



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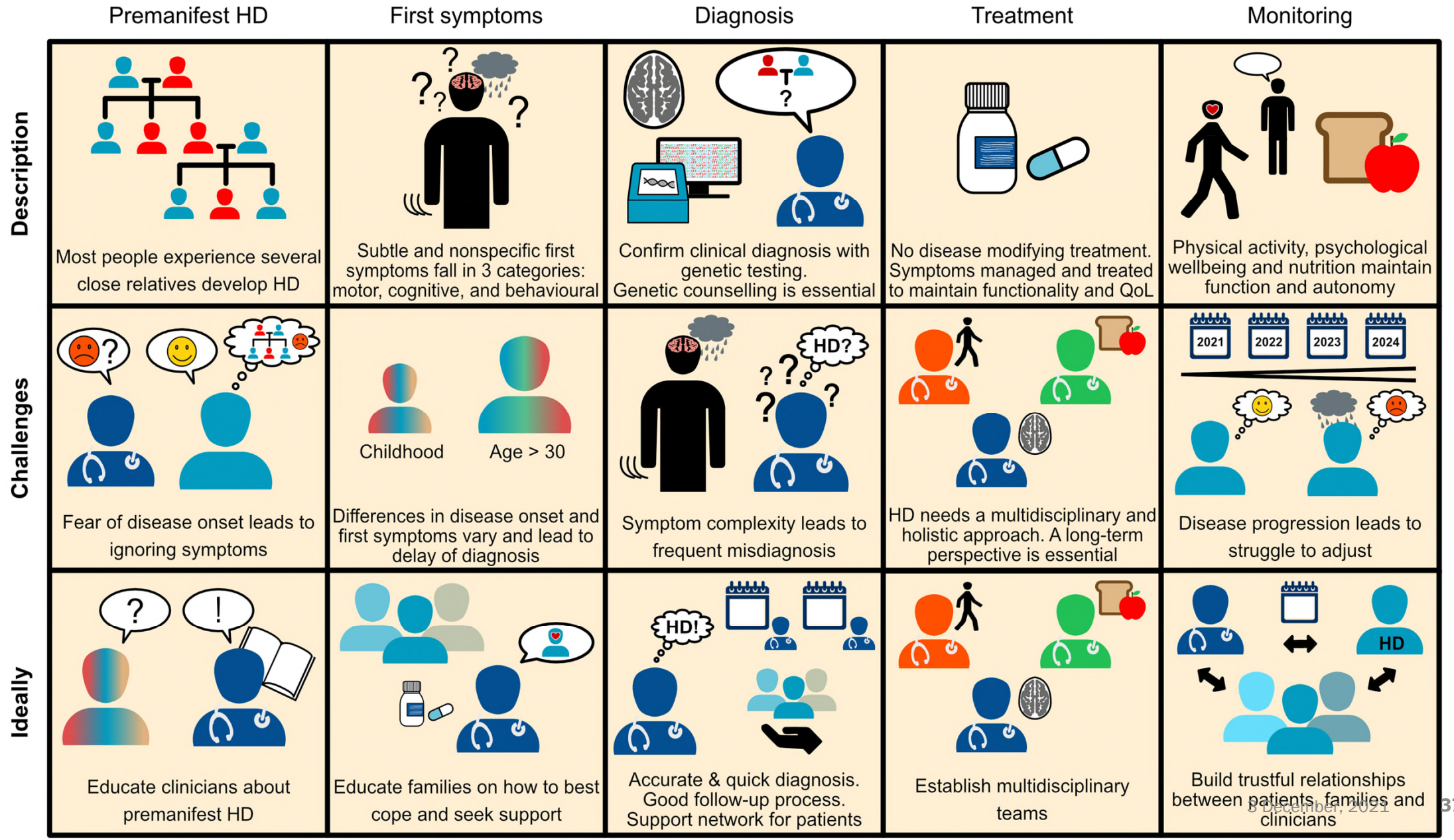
Network Neurological Diseases (ERN-RND)

Graphics representation of Huntington's disease



STATUS PATIENT JOURNEYS IN RND

* DENOTES AWAITING CLINIAN REVIEW

ERN-RND Disease group	Patient Journey
Ataxia and HSP	- Friedreich's Ataxia* - Hereditary Spastic Paraplegia*
Dystonia Paroxysmal Disorders and NBIA	-In development for Cervical Dystonia (by Dystonia Europe/Ipsen) - Alternating Hemiplegia of Childhood (developed by ERN epi-CARE – awaiting launch)
Choreas and HD	- Huntington's disease*
Fronto-temporal Dementia	- No patient representative – suggestions invited
Atypical parkinsonian syndromes	- MSA: completed and reviewed
Leukodystrophies	- In development – questionnaire stage

Patient Journey Hereditary Breast and Ovarian Cancer Syndrome (HBOC)

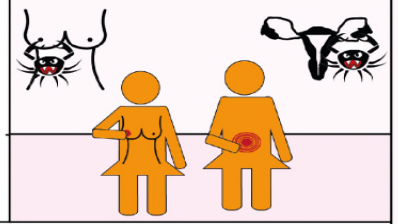
Disease

Clinic

Challenge

Goal

1st symptoms and Cancer diagnosis

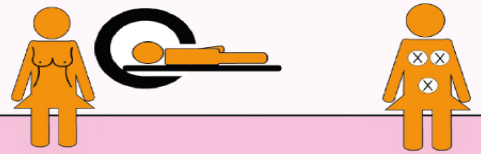


Investigation by medical specialists

Genetic diagnosis

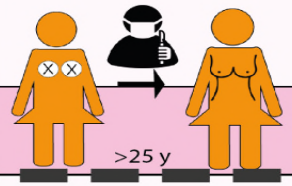
For example:
BRCA1
BRCA2

Consequences for healthy relatives: after genetic diagnosis at risk to develop cancer



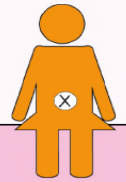
Early detection versus preventive surgery

Breast cancer: surveillance or preventive surgery (and reconstruction)



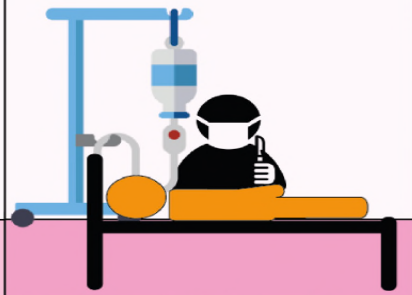
>25 y

Ovarian cancer: preventive surgery advised: 35-45y

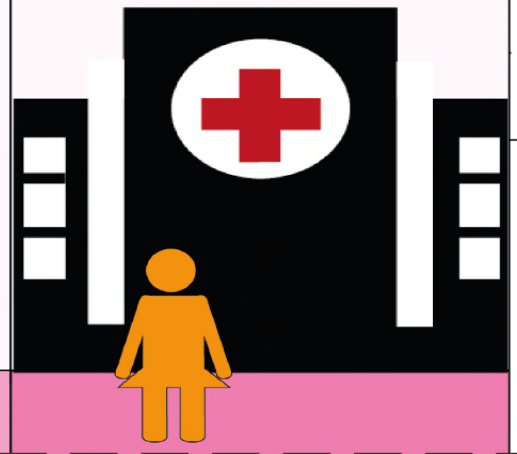


35-45y

Cancer treatment and (preventive) surgery

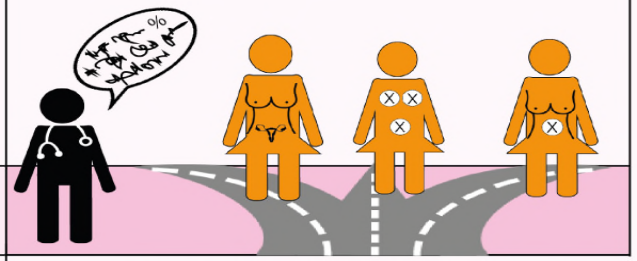


Follow-up



Early detection

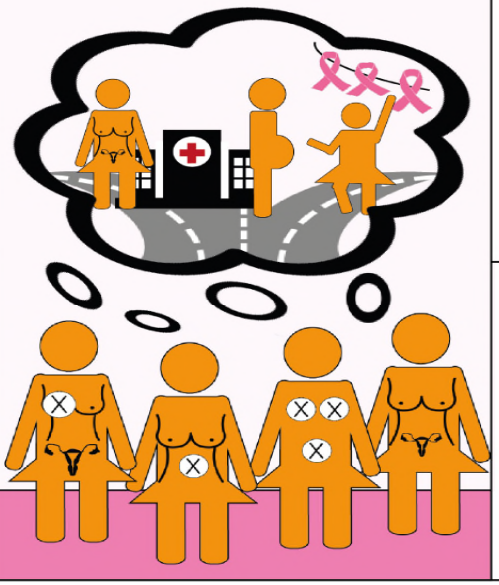
Informed decision making and counselling



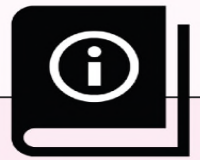
Personalized treatment

BRCA

Personalized follow-up

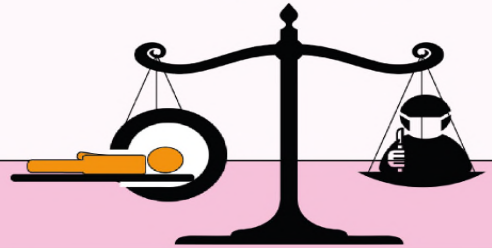


Awareness and education for GPs and clinicians



Psychological support

Options for screening and preventive surgery for proven carriers



European Reference Network

Genetic Tumour Risk Syndromes (ERN GENTURIS)

www.genturis.eu

©Jelena Meek

07-05-2019 Version 7.0

COMMON TEAMS IN PATIENT JOURNEYS?

- Patients' personal experiences may vary depending on the person, clinic, country, but over different diseases in RND - the patient journey can be similar particularly in RND i.e. – not seen much in other ERNs'

- Similarities
  - late diagnosis,
  - poor prognosis due to no specific treatment for rare disease
  - little medical information on the disease

    i.e. patient often attends for physio, A&E and have to tell their story/Journey several times to different people
  - Minimal specific rehab services

Difficult subjects for clinicians to discuss with person who has rare disease in patient Journeys?

- Poor prognosis
- Quality of life versus to quantity of life
- Psychological effects of progressive disease with no cure while
- Neurologists “are all about symptoms and signs and may not see the person”

- Exercise usually prolongs the person’s ability to keep active; It is expensive for National Health care systems, usually limited with variable access even within the same country

- Discussing social and psychological effects of living with the constant fear for the future if coming from a family with dominant illness:

  Huntington’s, Fronto-temporal dementia or HSP, Dominant ataxia

NEXT STEPS: DISSEMINATION

- ERN-RND website with graphics & text format
- Newsletter & social media, National Rare disease group,
- General Practitioners, General Physicians, Paediatricians

- But **can you, the expert Neurologists, use them in their work** for other medical specialists & health care professionalls to demonstrate:
  - A) the difficult path to diagnosis
  - B) the things that are important to people who have a rare disease progressive disease which has no specific treatment
  - C) the needs of the extended family

- Thank you

  for your attention

  for the opportunity to present patient journey

  Matthias, Carola in helping with the presentation

  Annemarie for her patience and perseverance in doing the graphics

- References

- Botz-Johnson M, Meek J, Hoogerbrugge N; “Patient Journeys”: improving care by patient involvement; European Journal of Human Genetics October 2019
- https://genturis.eu/l=eng/For-patients/Patient-Journeys.html

European Reference Networks

DG Ataxia and HSP

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

Co-funded by the EU

Board Meeting 29 Nov 2021

DG Ataxia/HSP

1. Diagnostic Flowchart early ataxias (Macaya)
2. Treatabolome for early-onset ataxias (Gómez)
3. Clinical rating scales for children with HSP (Schüle, Gómez)
4. Management of transition between pediatric and adult care (Mariotti)
5. Organisation of DG Ataxias and HSP after the enlargement of experts (Mariotti)

COORDINATORS ELECTED FOR THE NEXT 2 YEARS

- Alfons Macaya
- Enrico Bertini
- Caterina Mariotti
- Rebecca Schuele

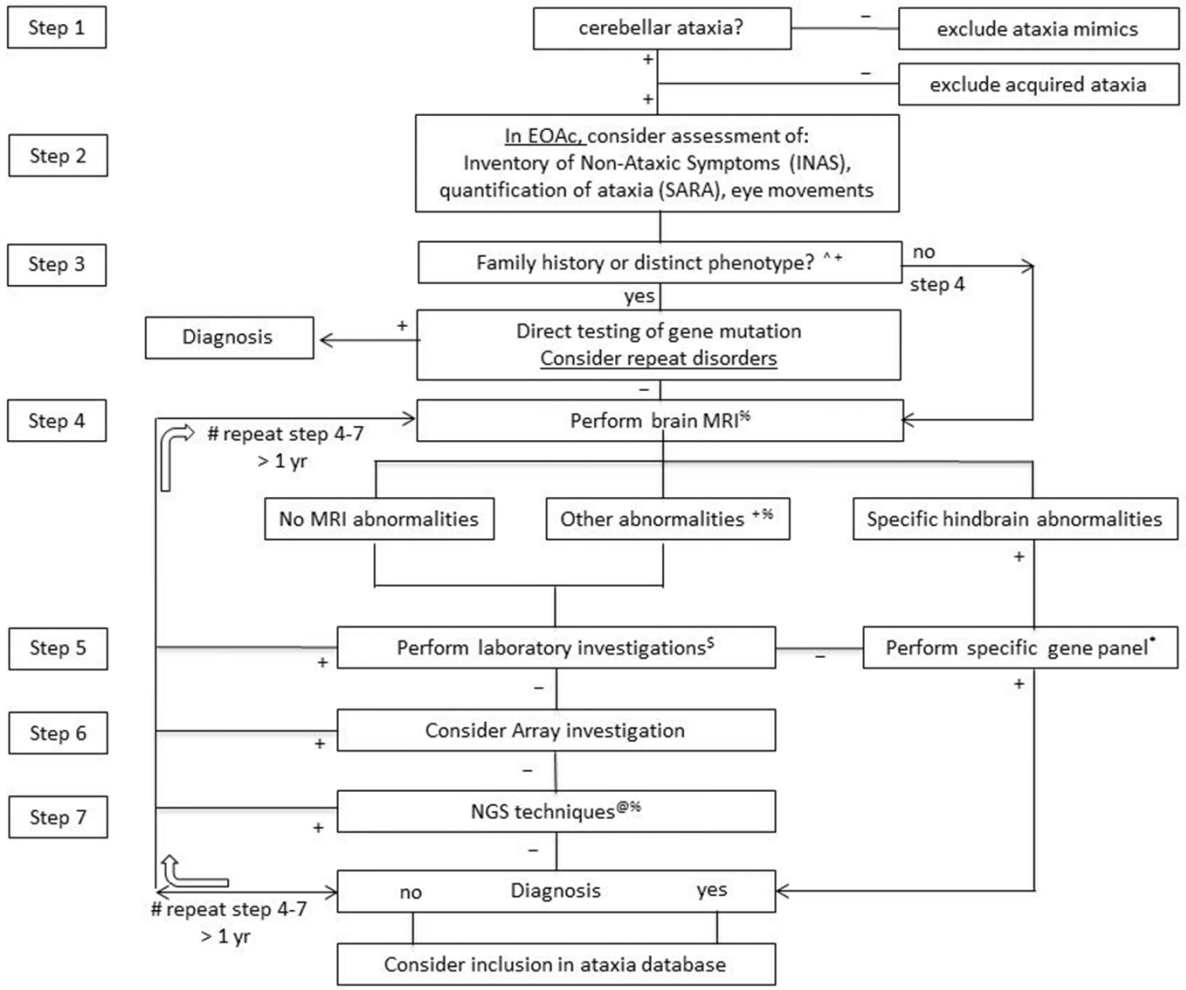
WEBINARS DG ATAXIA AND HSP

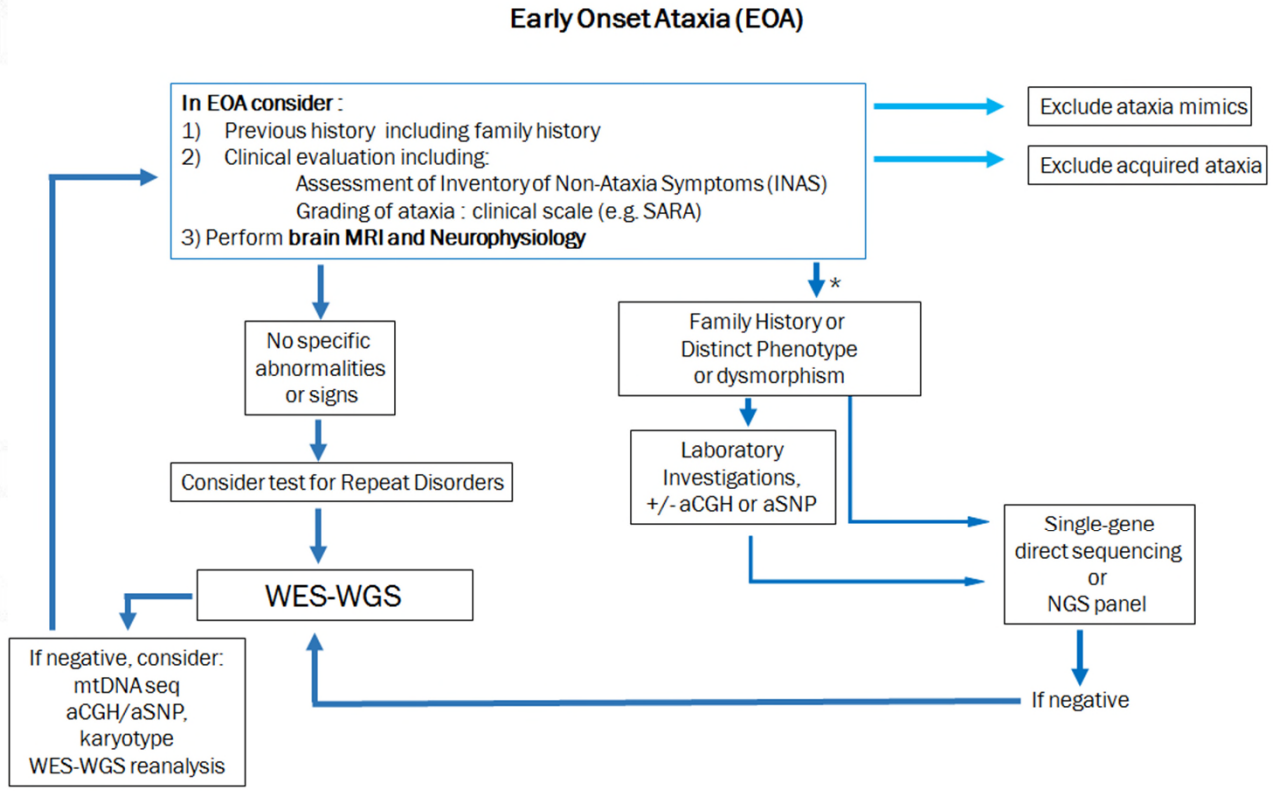
53 : total number of webinars,
7 in this DG
7 with focus on Neurorehabilitation in Ataxia and HSP

Date	Topic	Speaker	Focus	Proposed topics
5. 11. 2019	Clinical features of ataxia	Bart van de Warrenburg		
17.12.2019	Non-progressive congenital ataxia	Alfons Macaya	pediatric	
14.01.2020	The inherited ataxias	Paola Giunti		
01.10.2020	Hereditary Spastic Paraplegia - Clinical Disease Course	Rebecca Schüle		
13.04.2021	MR biomarkers in Ataxia /Imaging	Gülin Öz		
06.07.2021	Speech as a biomarker in ataxia: What can it tell us and how should we use it?	Adam Vogel		
09.11.2021	Clinical outcome assessments	Thomas Klockgether		

Topics and Speakers for next webinars in 2022, in particular for HSP are welcome!
Contact: Sanja.Hermanns@med.uni-tuebingen.de

DIAGNOSIS OF ATAXIA IN CHILDREN: ERN-RND/EPNS-CACG SURVEY (MACAYA)





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

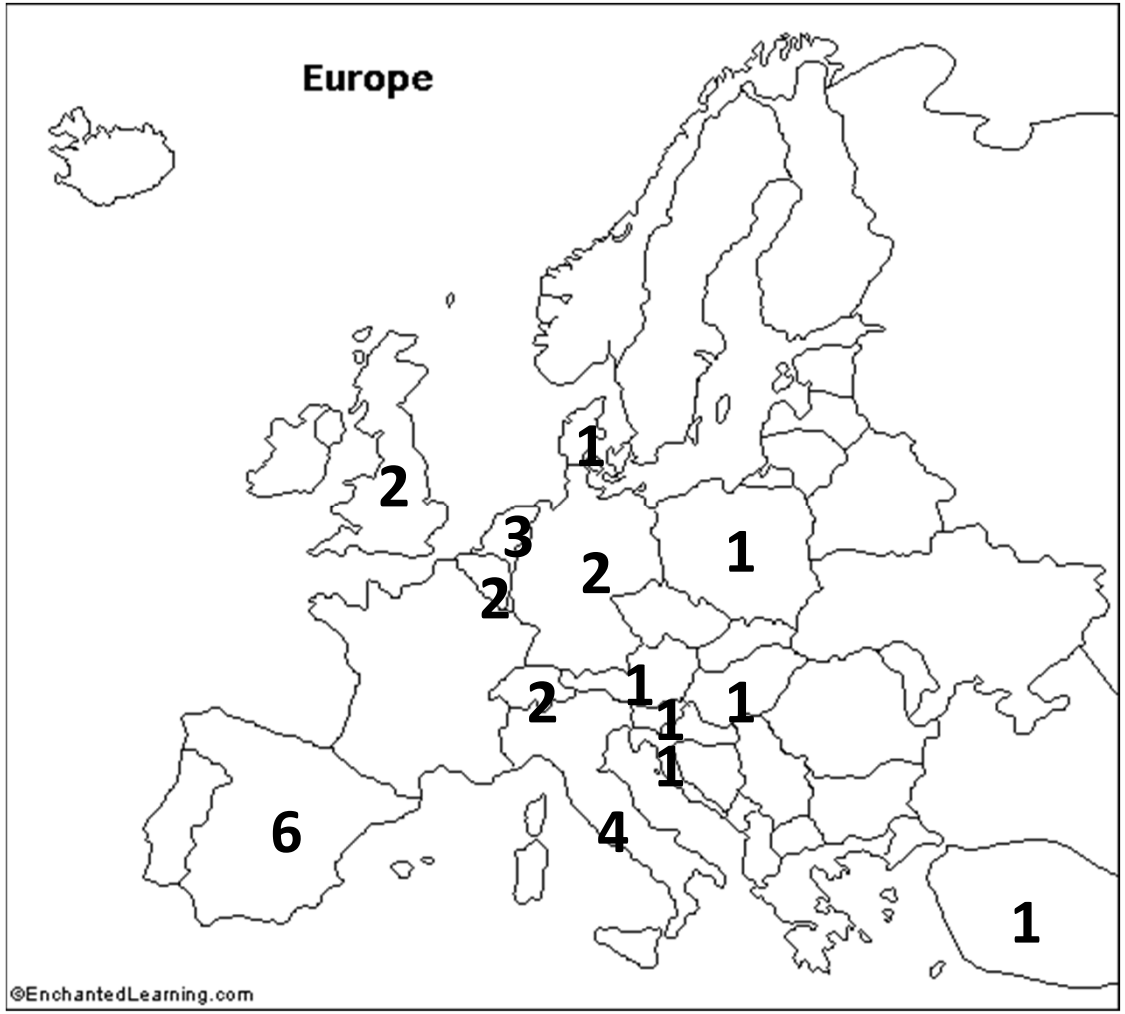
EUROPEAN PAEDIATRIC NEUROLOGY SOCIETY

European Reference Networks

ERN-RND Newsletter
European Reference Network for Rare Neurological Diseases

Survey Respondents

	N=31 (%)
Training background	
Child Neurology	21 (68%)
Adult Neurology	10 (32%)
Current practice	
Mainly child neurology	19 (61%)
Both child and adult neurology	3 (9.7%)
Mainly adult neurology	9 (29%)
Years of clinical practice	
Less than 10 years	4 (13%)
More than 10 years	27 (87%)
Age	
Under 35	1 (3%)
35-44	8 (26%)
45-55	6 (19%)
More than 55	16 (52%)
No. of children with ataxia under follow-up	
Less than 10	9 (29%)
10-25 children	11 (35%)
> 25 children	10 (32%)



2 Israel

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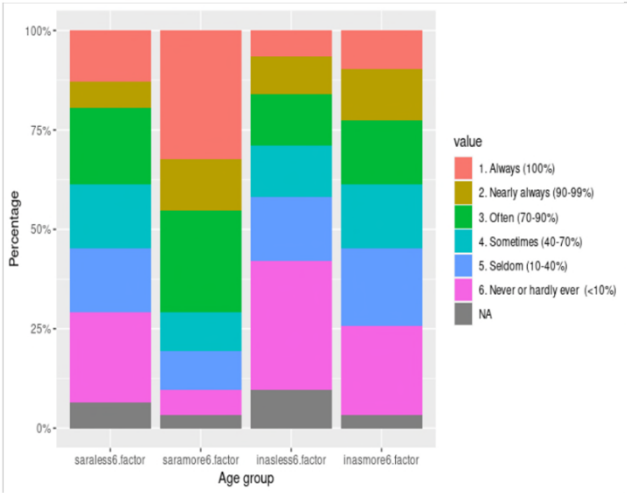
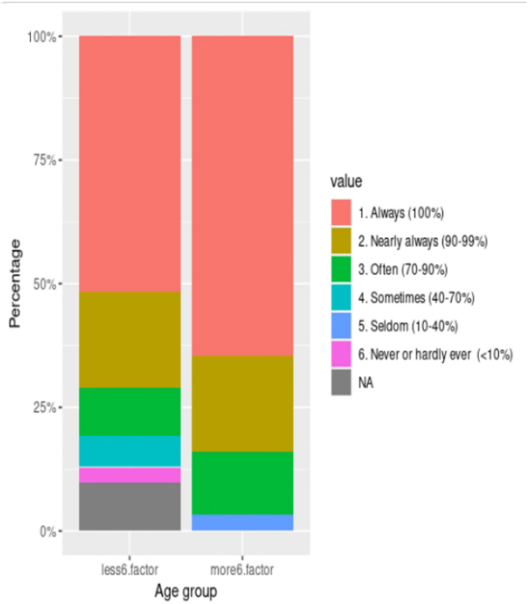
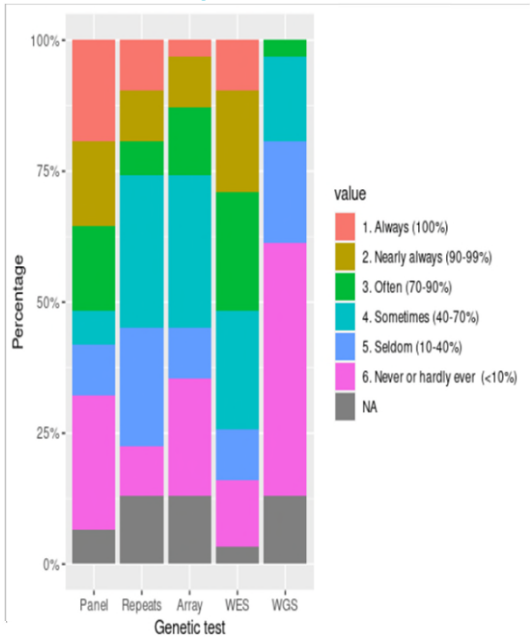
Scales at 1st visit



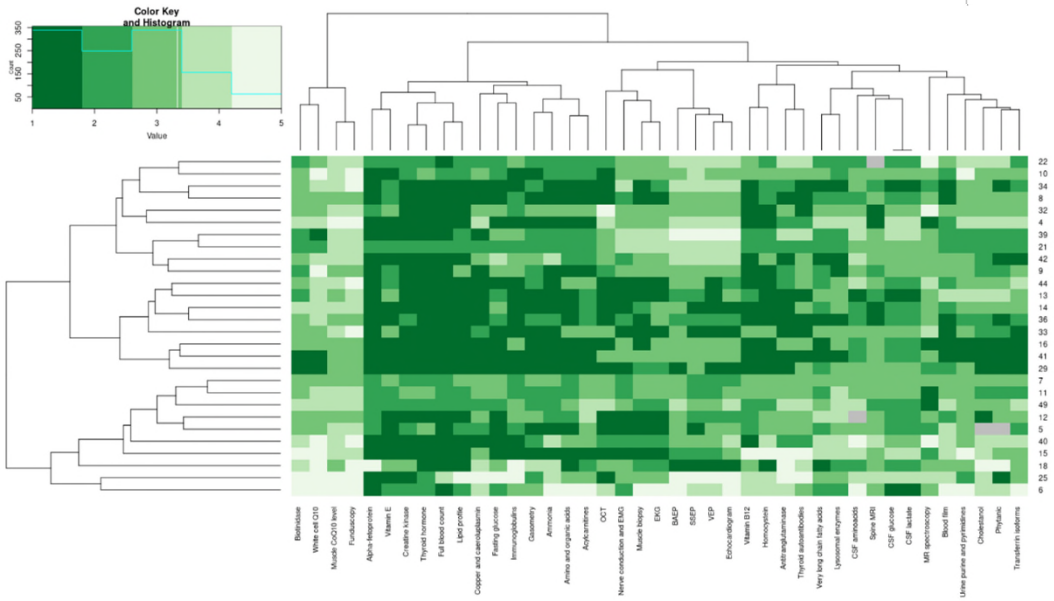
Use of MRI



First Genetic test (>6 yo)



Diagnosis of Ataxia in Children: Survey Round 1 - Results



Other Dx tests

European Reference Network

DELPHI SURVEY ON EARLY-ONSET ATAXIA DIAGNOSTIC FLOWCHART

Round 1

Respondent's profile

Current clinical practice

Round 2

Evaluate diagnostic tools

- diagnostic yield
- actionability

Strategies after negative NGS results

.....

Algorithm consensus

Oct-Nov 2021

Dec 2021

More participants welcome! → david_gomez@vhebron.net

European Reference Networks

TREATABOLOME FOR EARLY-ONSET ATAXIAS GÓMEZ

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

Co-funded by the EU

STEPS TOWARDS AN ATAXIA TREATABOLOME: SYSTEMATIC REVIEW OF TARGETED THERAPIES FOR AUTOSOMAL DOMINANT AND EPISODIC EARLY ONSET-ATAXIAS (AD/E-EOA)

A. Salazar
Supported by the Spanish Pediatric Neurology Society

SYSTEMATIC REVIEW

Gene selection: *CACNB4 ; ATP1A3 ; PRRT2 ; KCNA1 ; CACNA1A ; SLC1A3 ; TGM6 ; CAMTA-1 ; EBF3 ; VAMP1 ; KCNC3 ; CTBP1; TMEM240 ; KCND3 ; SPTBN2 ; IFRD1 ; TUBB4A ; PIGN ; POLG ; ITPR1 ; AFG3L2 ; PRKCG ; OPA1 ; CTNNB1 ; SCN8A ; FGF14 ; SLC2A1 ; GFAP*

21 genes for AD EO-ataxias
7 genes for episodic ataxias

Search strategy example
(GENE +/- Ataxia)
AND
(("therapy/broad"[filter] OR drug OR therap* OR treat*))

Pubmed initial search
n = 4299

Duplications (n=309)

Filter
(2 independent, blinded reviewers selection based on title and abstract) *Rayyan QCRI*

Inclusion criteria:
-English, Spanish, French or German
-Therapy applied to patients with confirmed genetic diagnosis
-Case, case series, cohort, clinical trials,... Prospective or retrospective
-Peer reviewed
-1980-2020

Exclusion criteria
-Basic or animal studies
-Genes that are not included in the list
-Insufficient data about therapy
-Access to complete text is not available online

Full article reading
n= 607

Excluded
n= 3383

Rayyan QCRI

Secondary search n= 42

Included
n= 280 *

Excluded
n= 327*

Exclusion criteria: Previous + unspecific therapies

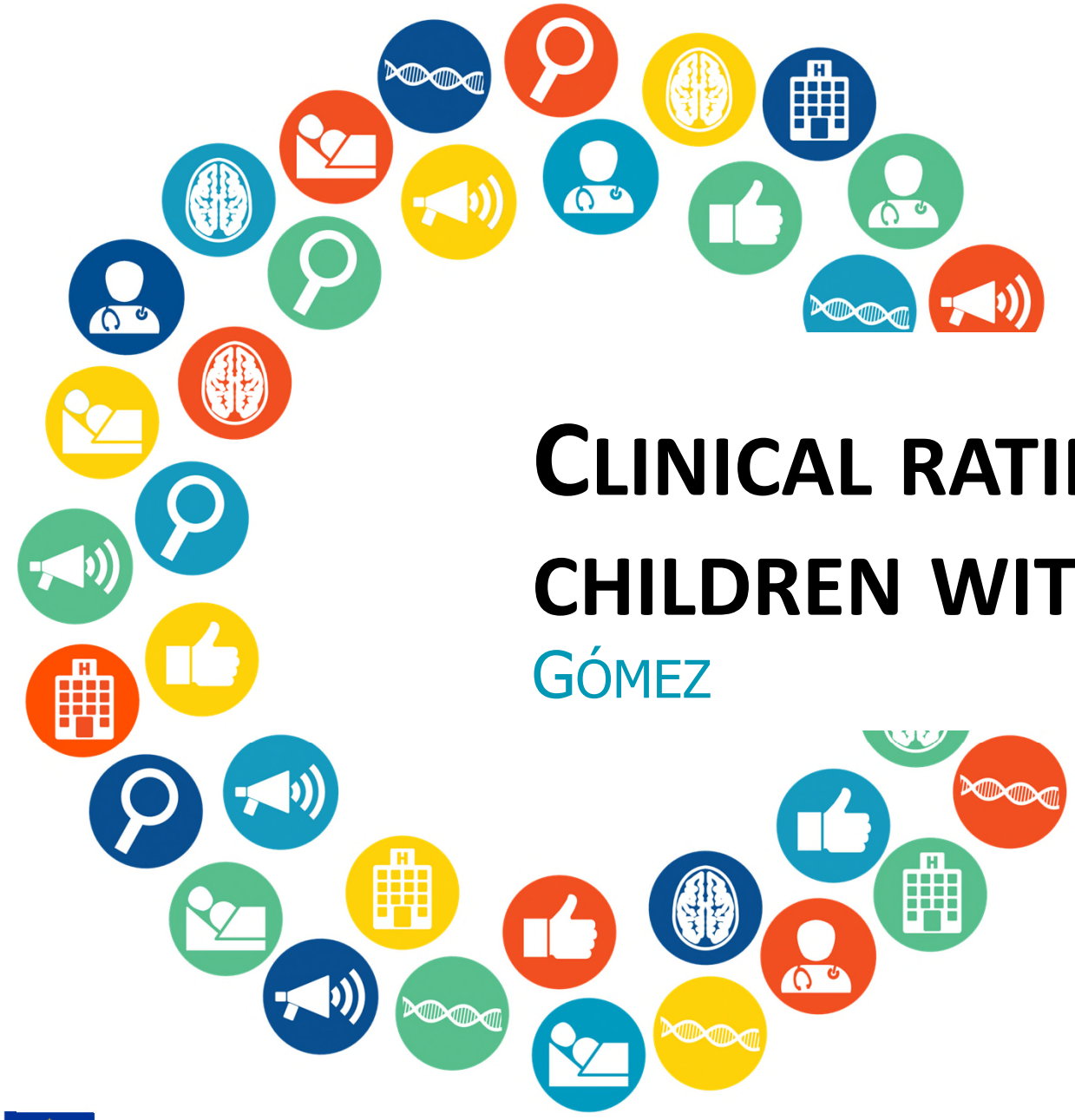
Total 332*

Rare disease general phenotype	Early onset episodic ataxias
Specific phenotype	**Episodic ataxia type 2** (ORPHA 92, OMIM 108500)
Gene	*CACNA1A*
1st line treatment (Evidence)	Acetazolamide (OCEBM 4)

Genetic Variants reported

	Effective	Variable	Non effective
Missense	c.742T>A (p.Tyr248Asn) ; c.889G>A (p.Gly297Arg) ; c.1096G>A (C287Y) ; c.2023G>A (p.Gln583Arg) ; c.2030G>A (p.Gly677Glu) ; c.2992G>C (p.Glu998Gln) ; c.4453 T>G (F1406C) ; R1434Q	c.2233C>T (T666M) ; c.4881C>T (R1549X)	c.1113G>A (G293R) ; R1668W ; c.6284G>A (p.Arg2095Gln)
Truncating	Exon deletion 31 ; IV36-2A>G ; c.4054C>T (p.Arg1352*) ; c.3575delA (p.Asp1192AlafsTer49) ; c.1747C>T (STOP codon) ; c.3244+1insG (cambio 1004, STOP 1070) ; c.4636+1G>T (Splicing) ; c.4110C>T (STOP codon 1279) ; c.3871_3873delGAG (p.Glu1924DEL) ; c.5005T>C (p.Arg1669*) ; c.4588G>A (W1451X) ; c.4963C>T (Q1561X) ; c.4077 C>T (R1281X)		c.5743+14 A>G ; R1820STOP ; c.2278-9delAG (cambio681, STOP783) ; c.5589C>T (R1785X) ; c.4645C>T (p.ARg1549Ter) ; c.3321dupC (p.Gly1108ArgfsTer40) ; c.6605_6616del (p.Asp2202_Arg2205del)

Alternative treatment (Evidence)	4- aminopyridine (OCEBM 2 (10 patients in a randomised clinical trial)) -4)
Other treatments	Flunarizine ; Dalfampyridine ; Sodium Valproate ; Zonisamide
Treatable comorbidities reported	Epilepsy. 1st line treatment: No specific. Depending on epilepsy subtype Dystonia. 1st line treatment: Botulinum toxin, DBS. Migraine. 1st line treatment: Acetazolamide, Topiramate
Probably ineffective	Verapamil, Gabapentin, nortriptyline, sumatriptan, baclofen,...
Number of publications	40
Key references (PMID)	21734179, 18718350, 23183922, 30713867, 29891059, 24222635
OCEBM/GRADE	GRADE: Very low OCEBM: 4 → 93% ; GRADE: low OCEBM: 3 → 5 % ; GRADE: Moderate OCEBM: 2 → 2%



CLINICAL RATING SCALES IN CHILDREN WITH HSP SCHÜLE - GÓMEZ

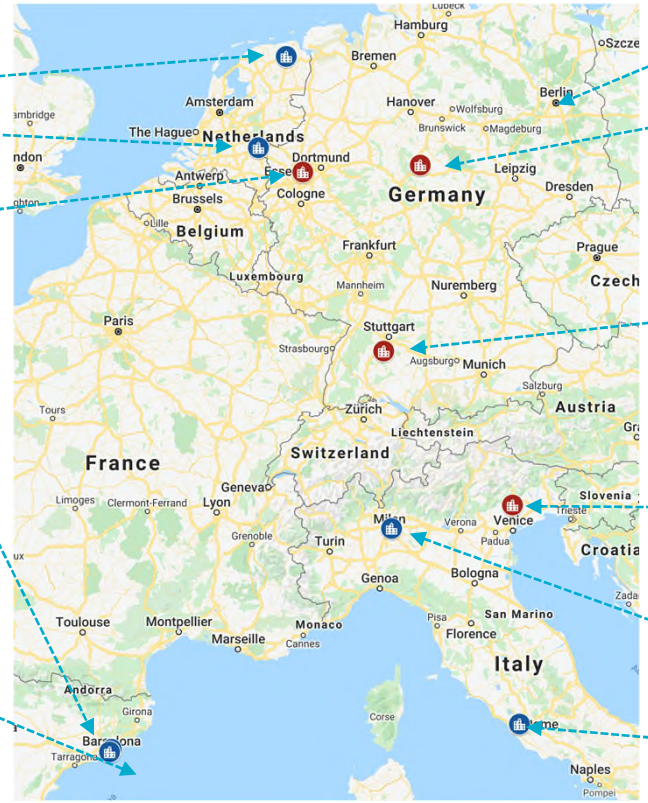
Co-funded by the EU

HSP Childrens Rating Scale Schüle, Gómez

collaboration with TreatHSP network

aims:

1. define phenotypic standards to clinically characterize children with HSP
2. identify, develop and validate measures to select and stratify children with HSP for clinical trials
3. validate outcome measures for clinical trials (progression outcomes, treatment response outcomes)



Groningen: Deborah Sival

Nijmegen: Charlotte Haaxma

Essen: Ulrike Schara, Frederik Braun

Barcelona (Val d'Hebron): Alfons Macaya, David Gomez

Barcelona (Sant Joan de Déu): Carlos Ortez

Berlin: Annette George

Göttingen: Knut Brockmann, Anne van Riesen

Tübingen: Inge Krägeloh-Mann, Marion Döbler-Neumann, Rebecca Schüle, Peter Martus (Statistics)

Conegliano/Pieve di Soligo: Andrea Martinuzzi

Milano: Nardo Nardocci, Isabella Moroni, Giovanna Zorzi

Rome: Enrico Bertini, Francesco Nicita, Gessica Vasco

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

HSP Childrens Rating Scale

- separate approaches for
  - young children (≤ age 5), lead: David Gomez
  - older children andd adolescents (≥ age 6), lead: Rebecca Schüle
- monthly alternating meetings for young / older children
- validation study in healthy and affected (HSP) children
  - to determine age-dependent reference values
  - to validate novel / modified scales
  - to determine progression characteristics for selected outcomes and select outcomes with highest sensitivity to change

European Reference Networks

MANAGEMENT OF TRANSITION BETWEEN PEDIATRIC AND ADULT CARE MARIOTTI

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

Co-funded by the EU

MANAGEMENT OF TRANSITION BETWEEN PEDIATRIC AND ADULT CARE (MARIOTTI)

Emerging need for pediatric issues for ataxia and HSP: diagnostic flowchart; scales etc.

↓

Cross-cutting pediatric issues in collaboration with other DG –WG- ERNs

↓

Idea of investigating existing protocol or clinical practice for the transition from adult to children in different Centers /HCPs and Countries within the ERN-RND

We started a collection of papers / documents from different HCP and countries to document the existing literature and protocols

↓

Survey paper
and /or ERN-RND recommendations

Survey

among the HCPs of the ERN-RND (existing procedures or needs)

Decide about neurological diseases to be considered

- CEREBELLAR ATAXIAS ?
- ATAXIAS & HSP ?
- NEUROLOGICAL DISEASES OF ERN-RND ?
- OTHER NEUROLOGICAL DISEASES ?

Preparation of the questionnaire

Literature search (methodology)

Decide for publication (survey/ recommendations/ reviews…)

Disease – Ataxia and HSP- Specific PROJECTS

Planned:

- Revision paper "physical therapy"
- Collaboration with Working group on NeuroRehabilitation
- Updating existing Guidelines
- Management Guideline for HSP
- Define MDT for the disease group
- Patient information (ATAXIA LEAFLETS et al.)
- Translation of disease knowledge

--- Decide on planned projects

--- Discuss Organization of Disease Group for future activities

European Reference Networks

DG CHOREAS AND HD

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

Co-funded by the EU

COORDINATORS

- Anne-Catherine Bachoud-Levi
- Juan Darío Ortigoza-Escobar
- G. Bernhard Landwehrmeyer

Overview

- Differential Diagnosis of Chorea Syndromes - combining a systematic literature review with a ground truth approach
- NKX2-1-related disorders
  - A review of the literature, a survey on management, and case series from ERN-RND partners
  - Development of a clinical practice guide
- Pharmacological treatment of Chorea Syndromes – a survey of current practice & thinking (a ground truth approach) and a scoping review
- Eliminating barriers: providing validated assessment tools for HD and Chorea Syndromes – language and cultural validation in the languages/regions represented in ERN-RND

DG CHOREAS AND HD PUBLICATION

Combining literature review with a ground truth approach for Huntington disease phenocopy diagnosis

Authors: **Quang Tuan Rémy Nguyen**, Juan Dario Ortigoza Escobar, Jean-Marc Burgunder, Caterina Mariotti, Carsten Saft, Lena Elisabeth Hjermind, Katia Youssov, G. Bernhard Landwehrmeyer, ERN-RND DG chorea and **Anne-Catherine Bachoud-Levi**

SUBMITTED

Abstract

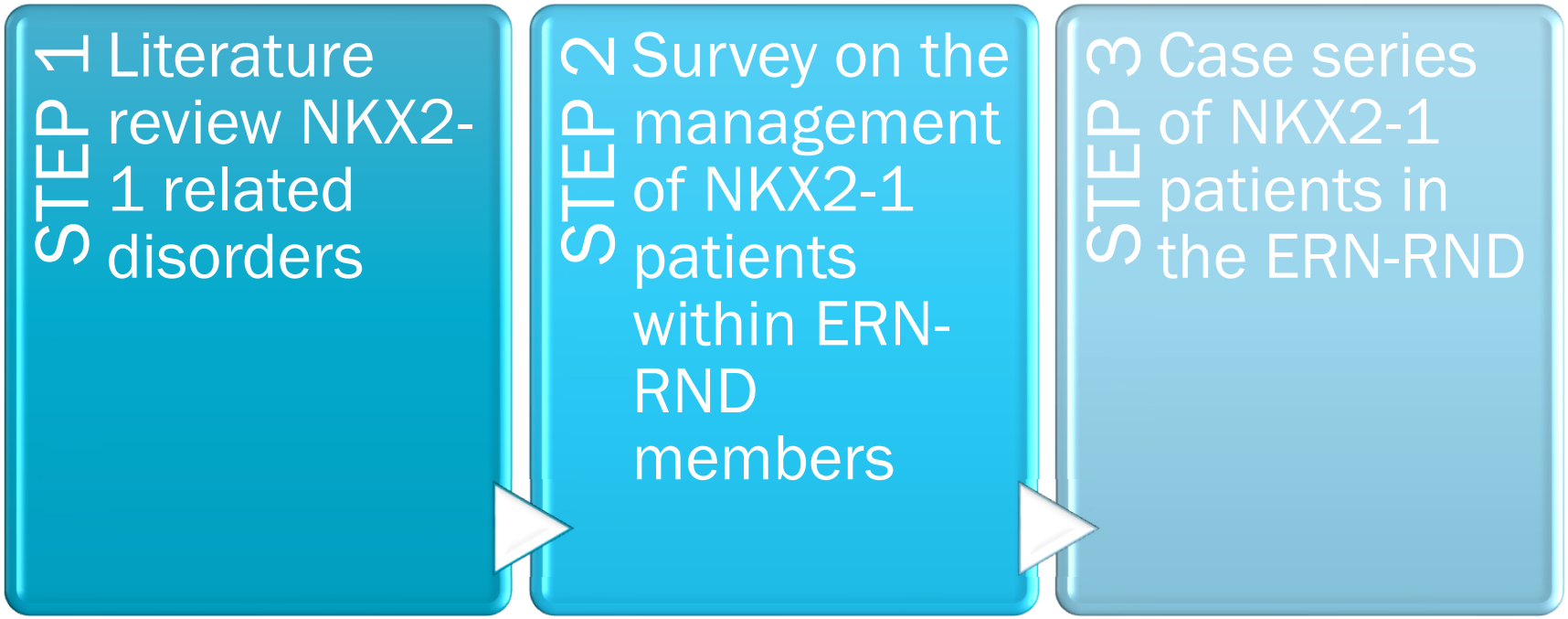
- One percent of patients with a Huntington's disease phenotype do not have the HTT gene mutation. These are known as Huntington's disease phenocopies. Their diagnosis is still a challenge. Our objective was to provide a diagnostic approach to Huntington's disease phenocopies based on comparison of medical expertise and review of the literature. We employed two complementary and sequential approaches: a review of the literature and two surveys analyzing the daily clinical practice of physicians who are experts in movement disorders. The review of the literature was conducted from 1993 to 2020, by extracting articles about chorea or Huntington diagnosis from the database Pubmed and yielded the selection of 50 articles and the full analysis of 20 articles to establish the surveys. Twenty-eight physicians responded to the first survey exploring the red flags suggestive of specific diagnoses. Thirty-three physicians completed the second survey which asked for the classification of paraclinical tests according to their diagnostic significance. The analysis of the results of the second survey used four different clustering algorithms and the density-based clustering algorithm DBSCAN to classify the paraclinical tests into 1st, 2nd, and 3rd-line prescriptions. Finally, we included suggestions from members of the European Reference Network-Rare Neurological Diseases (ERN-RND Chorea & Huntington Disease Group). Finally, we propose guidelines that integrate the detection of clinical red flags with the classification of paraclinical testing to improve the diagnosis of Huntington Disease phenocopies.

NKX2-1-RELATED DISORDERS

- The project is divided into two parts:
  - The **first part** includes a review of the literature, a survey on disorder management, and a case series of patients from the ERN-RND *
  - The **second part** is concerned with the development of a clinical practice guide for NKX2-1-related disorders

* The Ethics Committee of Hospital Sant Joan de Déu has yet to approve the collection of NKX2-1 cases from the ERN-RND

NKX2-1 RELATED DISORDERS – FIRST PART



FINISHED

FINISHED

ON-GOING

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NKX2-1 RELATED DISORDERS – SECOND PART

CLINICAL PRACTICE GUIDELINE (CPG) VS CLINICAL CONSENSUS STATEMENT FOR *NKX2-1*-RELATED DISORDERS

European Reference Network | Respiratory Diseases (ERN-LUNG)

EURACAN

Endo-ERN

European Reference Network for rare or low prevalence complex diseases

Network Neurological Diseases (ERN-RND)

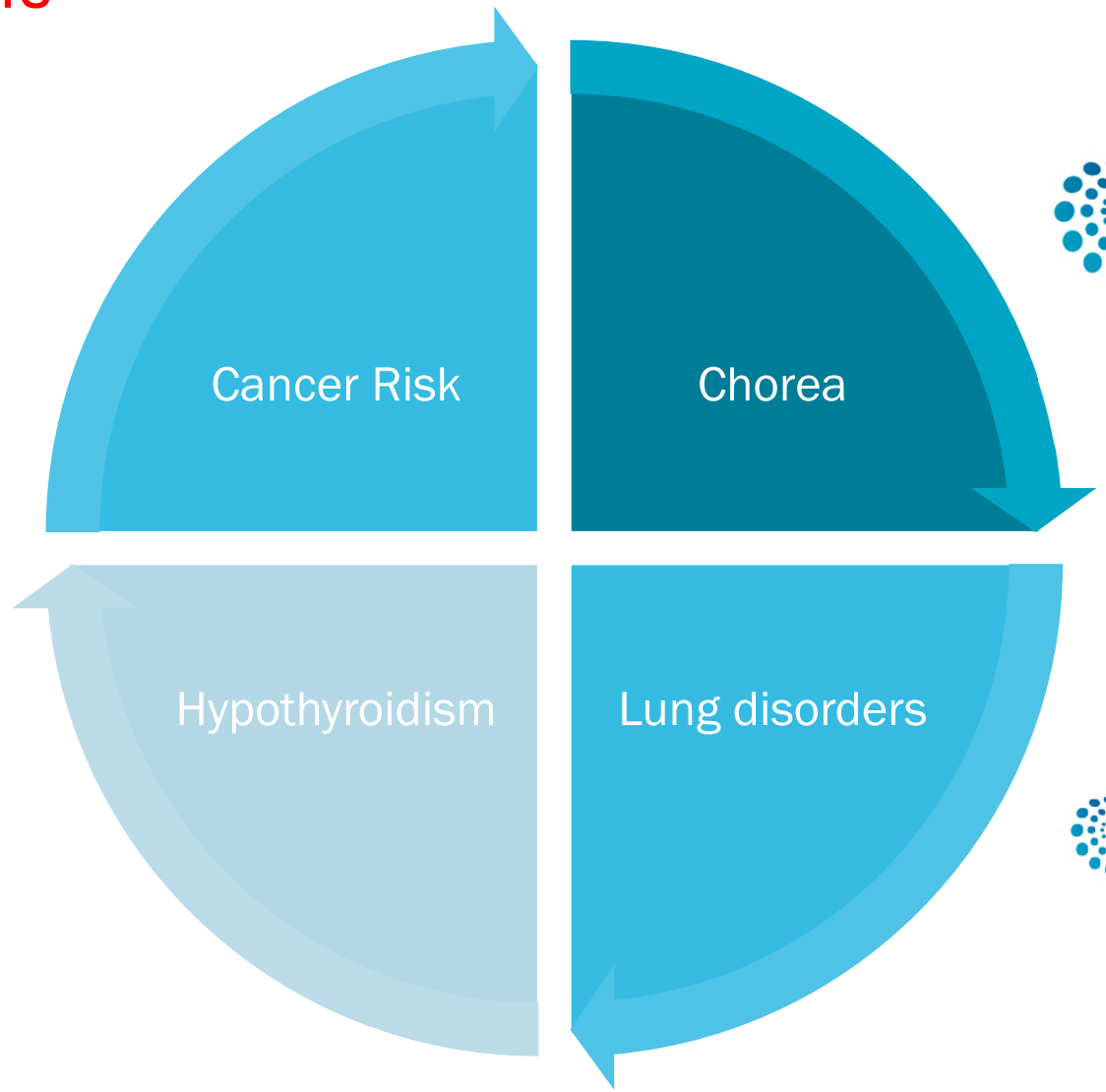
Vivre Sans Thyroïde

Fundación Progreso y Salud
Consejería de Salud y Familias

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

NKX2-1 related disorders

EURACAN



European Reference Network
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Network Neurological Diseases (ERN-RND)

Endo-ERN

Vivre Sans Thyroïde

European Reference Network | Respiratory Diseases (ERN-LUNG)

Fundación Progreso y Salud
Consejería de Salud y Familias

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

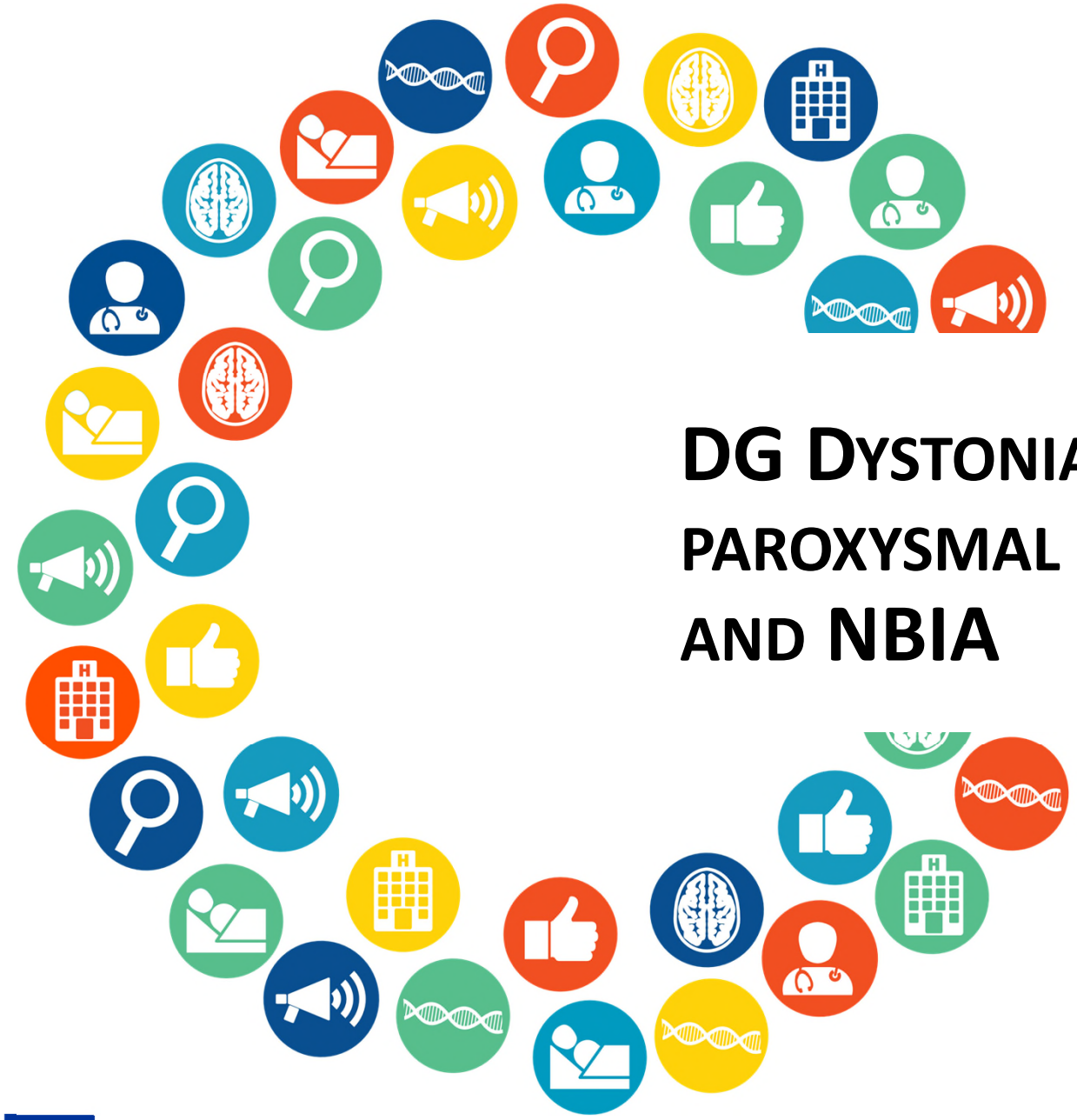
Pharmacological treatment of Chorea Syndromes – a survey of current practice & thinking (a ground truth approach) and a scoping review

Background

- The strength of the DG and the ERN-RND is the collective experience of the partners in the network
- A lot of practical care/disease management issues can NOT be answered in a useful manner by taking recourse to RCTs or other high quality evidence.
- Impressions and informal exchange are valuable
- Aim: scoping review

Eliminating barriers: providing validated assessment tools for HD and Chorea Syndromes – language and cultural validation in the languages/regions represented in ERN-RND

European Reference Networks



DG DYSTONIA, PAROXYSMAL DISORDERS AND NBIA

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

Co-funded by the EU

ERN-RND Board Meeting Disease Group Dystonia, Paroxysmal disorders and NBIA

European Reference Network | Neurological Diseases (ERN-RND)

Board Meeting: November29, 2021

Presenter:

Sylvia Boesch, Medical University Innsbruck, Innsbruck, AUSTRIA

on behalf of the Disease Group

Agenda

Activities, current state

- Dystonia management across Europe within the ERN-RND: current state and future challenges (Sylvia Boesch)
- SURVEY MYOCLONUS DYSTONIA (BELEN PEREZ), SURVEY EXTENDED

Presentation of cross-cutting activites

- *CPMS case discussions "jour fix"*
- CPMS Dystonia unsolved cases (DBS cases /Vienna, 2 unsolved cases/3 IBK)
- Dystonia DBS case discussion (Tübingen, Kiel)
- Transition (common effort of WG)

Webinars 2021, proposed 2022

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

Liesanne M. Centen[1,2], MD; David Pinter[3], M.D., Ph.D.*; Martje E. van Egmond[1,2], MD, PhD; Holm Graessner[4],; Norbert Kovacs[3], MD, DSc; Anne Koy[5],MD; Belen Perez[6], MD; Carola Reinhard[4], PhD; Marina AJ de Koning-Tijssen[1,2] , MD, PhD; Sylvia Boesch[8], MD, MSc.*

*contributed equally; corresponding author: Tba

The questionaire

- **Part I** included the **characterization of the participants** such as profession, country, main interest in dystonia, and whether the respondent took care in his/her practice of adults, children or both.
- **Part II** assessed **country characteristics** including general infrastructure of dystonia healthcare, education about dystonia for care providers, ongoing dystonia research, diagnostics and treatment of dystonia, and pediatric dystonia.
- **Part III** was an **open questions about the own opinion of participants** about issues encountered in practice or measures suggested to improve management of dystonia in their countries.

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

Expert centres

centres that fulfil the minimum criteria (regarding patient numbers etc.) set by ERN-RND.

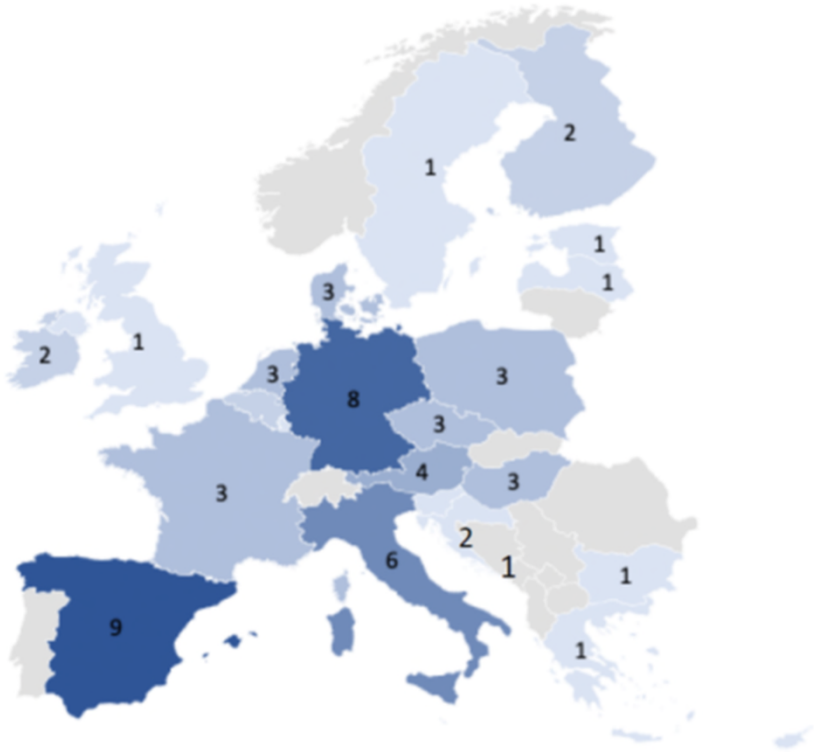
- ERN-RND full members
- ERN-RND applicants (new applicants/affiliated partners that apply for full membership)
- Non-expert centres (centres that remain affiliate partners after the ongoing application round).

N Respondents:

- 55 respondents from expert centres
- 6 respondents from non-expert centres
- 1 respondent from a non ERN-RND centre

The respondents come from the following centres:

- 36 expert centres
- 5 non-expert centres



Participating countries in the ERN-RND survey: Bulgaria, Czech Republic, France, Germany, Hungary, Italy, Netherlands, Poland, Slovenia, Spain United Kingdom, Austria, Croatia, Denmark, Estonia, Finland, Latvia, Luxembourg, Malta, Belgium, Cyprus, Greece ,Ireland, Sweden.

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

Table 1. *Accessibility of experts per country*

Country	Accessibility of dystonia experts*			Overall evaluation of accessibility of dystonia experts
	Difficult	Satisfactory	Easy	
Austria	0	4	0	satisfactory
Belgium	0	1	1	satisfactory/easy
Bulgaria	0	1	0	satisfactory
Croatia	0	1	0	satisfactory
Cyprus	0	1	0	satisfactory
Czech Republic	2	1	0	rather difficult, in some regions satisfactory
Denmark	2	1	0	rather difficult, in some regions satisfactory
Estonia	0	0	1	Easy
Finland	0	0	2	Easy
France	1	2	0	mainly satisfactory, in some regions difficult
Germany	2	5	1	mainly satisfactory, in some regions difficult or easy
Greece	0	1	0	satisfactory
Hungary	3	0	0	Difficult
Ireland	0	2	0	satisfactory
Italy	1	5	0	mainly satisfactory, in some regions difficult
Latvia	0	1	0	satisfactory
Luxembourg	1	0	0	Difficult
Malta	0	1	0	satisfactory
Netherlands	0	3	0	satisfactory
Poland	3	0	0	Difficult
Slovenia	1	0	0	Difficult
Spain	1	7	1	mainly satisfactory, in some regions difficult or easy
Sweden	0	1	0	satisfactory
United Kingdom	0	1	0	satisfactory

*Values indicate the numbers of respondents who rated accesibility of dystonia experts as difficult, satisfactory, and easy.

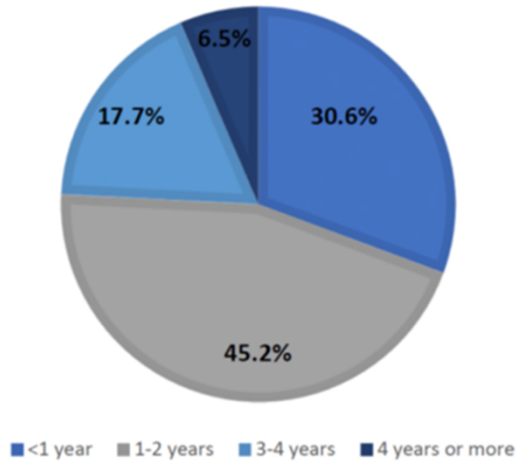


Figure 2. *Mean time between the first appearance of symptoms of dystonia to evaluation by a movement disorders expert (by % of respondents).*

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

Table 2. *Accessibility of deep brain stimulation per country*

Country	Accessibility of deep brain stimulation*			Overall evaluation of accessibility of deep brain stimulation
	Easy	With some difficulty	Not available	
Austria	3	1	0	in some regions easy, in some with difficulty
Belgium	1	0	0	easy
Bulgaria	0	1	0	with some difficulty
Croatia	0	1	0	with some difficulty
Cyprus	0	1	0	with some difficulty
Czech Republic	1	2	0	in some regions easy, in some with difficulty
Denmark	0	3	0	with some difficulty
Estonia	1	0	0	Easy
Finland	1	1	0	in some regions easy, in some with difficulty
France	2	1	0	in some regions easy, in some with difficulty
Germany	5	3	0	in some regions easy, in some with difficulty
Greece	0	1	0	with some difficulty
Hungary	3	0	0	Easy
Ireland	0	1	1	in some regions easy, in some with difficulty
Italy	2	4	0	in some regions easy, in some with difficulty
Latvia	0	0	1	not available
Luxembourg	1	0	0	Easy
Malta	1	0	0	Easy
Netherlands	3	0	0	Easy
Poland	0	3	0	with some difficulty
Slovenia	0	1	0	with some difficulty
Spain	5	4	0	in some regions easy, in some with difficulty
Sweden	0	1	0	with some difficulty
United Kingdom	1	0	0	Easy

*Values indicate the numbers of respondents who rated accesibility of deep brain stimulation as easy, difficult or not available. Answers of 61 out of 62 respondents could be used due one missing answer.

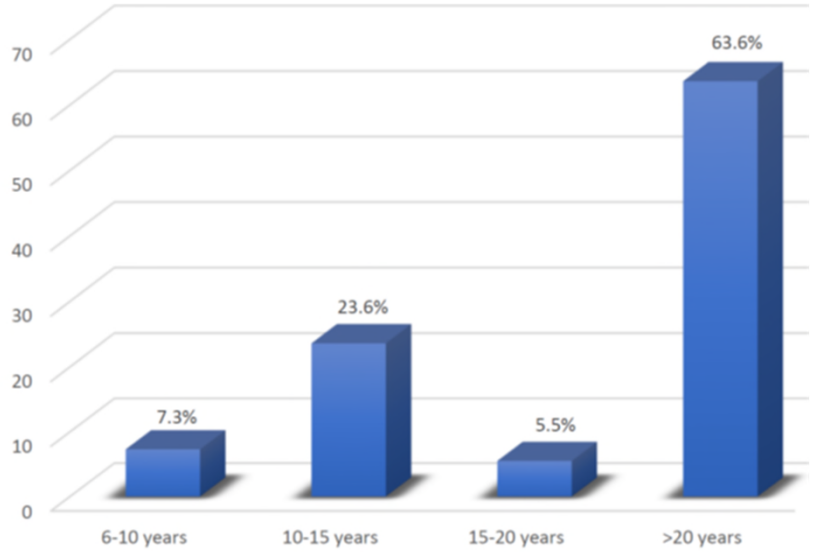


Figure 3. *Age of patients at time of deep brain stimulation surgery (by % of respondents)*

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

Table 3. *Availability of specific educational training on dystonia and deep brain stimulation for healthcare providers*

Country	Specific educational training on										
	Dystonia						deep brain stimulation				
	Residents and general neurologists	Clinical nurse specialists	Physiotherapists	Speech therapists	General practitioners and pediatricians		Residents and general neurologists	Clinical nurse specialists	Physiotherapists	Speech therapists	General practitioners and pediatricians
Austria	+	+*			+		+*				+*
Belgium	+		+*	+*				+*			+*
Bulgaria	+				+		+				
Croatia	+				+		+	+			
Cyprus	+		+								
Czech Republic	+*	+*	+*				+*	+*	+*	+*	
Denmark	+*	+*	+	+*	+*		+*	+		+*	+*
Estonia	+	+	+				+				
Finland	+	+*	+*	+*	+*		+	+*			
France	+	+*	+*	+*	+*		+	+*			
Germany	+*	+*	+*	+*	+*		+*	+*	+*	+*	+*
Greece	+						+				
Hungary	+	+*	+*				+	+*			+*
Ireland	+	+*						+*	+*	+*	
Italy	+	+*	+*	+*	+*		+*	+*	+*	+*	+*
Latvia											
Luxembourg	+						+				
Malta									+		
Netherlands	+	+*	+	+*	+*			+			
Poland	+*						+*				
Slovenia	+										
Spain	+	+*	+*	+*	+*		+*	+*	+*	+*	+*
Sweden	+			+			+				
United Kingdom	+	+	+	+	+		+	+	+		

*The special training for the special healthcare provider was not available in all region of the country.

- Internships for residents in neurology were available in all but 7 countries.
- 54/61 (88.5%) from 22 countries reported available teaching courses or symposia for residents and general neurologists on MDs (in general).
- 29/60 (48.3%) indicated availability of these to general practitioners (GPs) from 15 countries.
- Education for other healthcare providers involved in dystonia-care: specific teaching courses on MDs
  - 19/60 (31.6%) from 13 countries for nurse practitioners
  - 17/60 (28.3%) from 11 countries for speech therapists
  - 24/59 (40.7%) from 13 countries for physiotherapists

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

Table 4. *Different types of research on dystonia per country*

Country	Research type						
	Basic	Clinical	Genetics	DBS for dystonia	Neuropsychology	Imaging	Other
Austria	+	+	+	+		+	
Belgium	+	+	+	+			
Bulgaria	+	+					
Croatia			+				
Cyprus		+					
Czech Republic		+	+	+	+	+	
Denmark		+	+	+		+	
Estonia		+					
Finland	+	+	+	+			
France	+	+	+	+	+	+	
Germany	+	+	+	+	+	+	
Greece				+			
Hungary	+	+	+	+		+	
Ireland		+	+		+	+	
Italy	+	+	+	+	+	+	
Latvia							
Luxembourg							
Malta							
Netherlands	+	+	+	+	+	+	+ (Physiotherapy)
Poland		+					
Slovenia		+	+	+			
Spain	+	+	+	+	+	+	+ (rTMS)
Sweden							
United Kingdom	+	+	+	+	+	+	
Total number of countries with ongoing research on a field	11	18	15	14	8	11	2

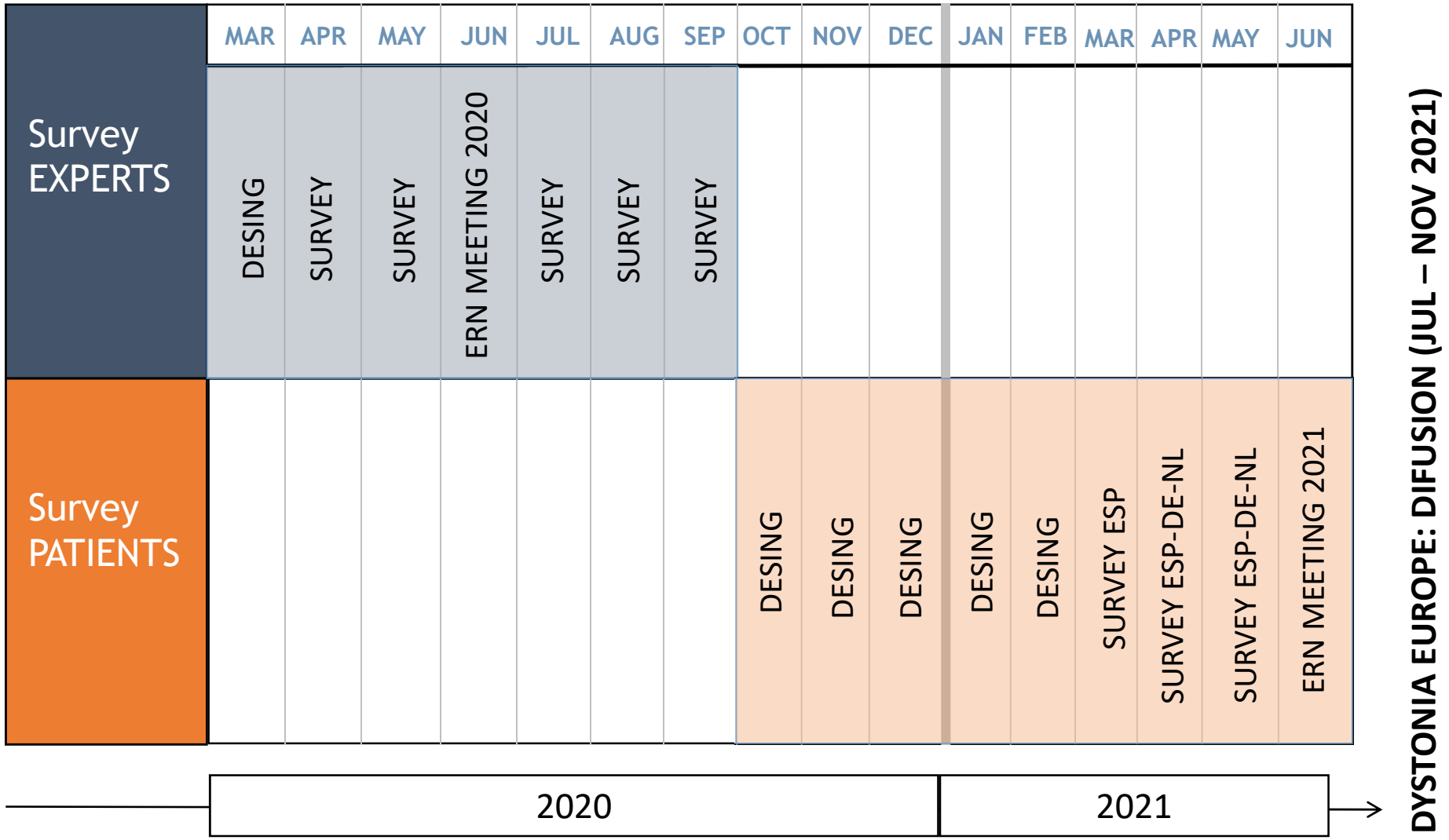
20/24 (83.3%) analyzed countries, there was at least 1 type of dystonia research currently ongoing:

- Clinical research on dystonia 18/24 countries (75%)
- Clinical research DBS for in 14/24 countries (58.3%)
- Countries conducting research on:
    - genetics 15/24 (62.5%)
    - basic research 11/24 (45.8%)
    - imaging 11/24 (45.8%)
    - neuropsychology 8/24 (33.3%)
- Other types of current ongoing research that were mentioned included:
    - physiotherapy in dystonia (the Netherlands)
    - transcranial magnetic stimulation (Spain)
    - Latvia, Luxembourg, Malta, and Sweden: no type of dystonia research

Survey for patients and families
MYOCLONUS DYSTONIA SYNDROME

European Reference Network | Neurological Diseases (ERN-RND)

Methods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Survey EXPERTS
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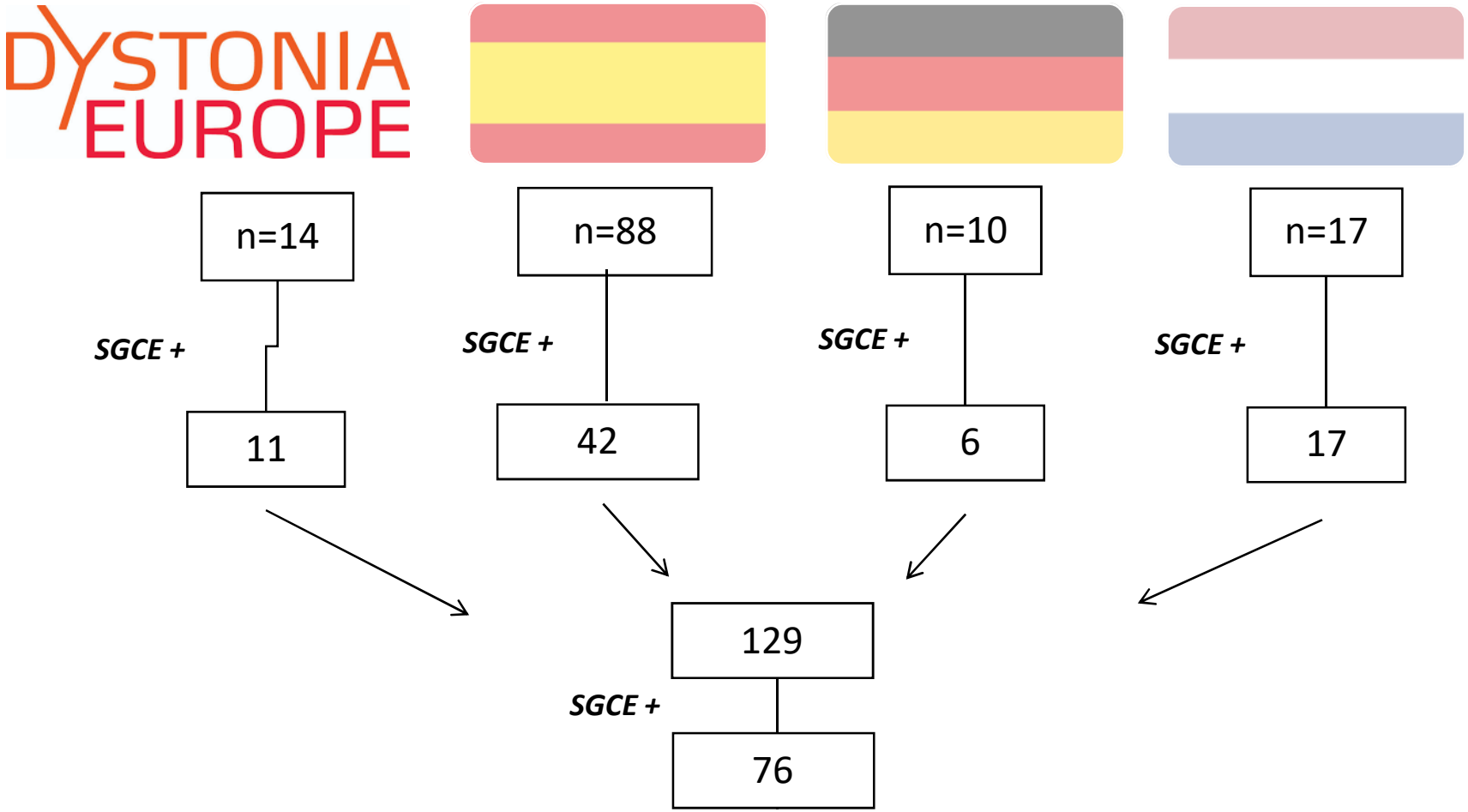
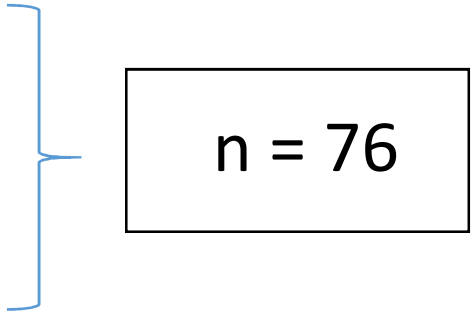
ERN MEETING 2021
2020
2021
DYSTONIA EUROPE: DIFUSION (JUL – NOV 2021)


DYSTONIA EUROPE
n=14
SGCE +
11
n=88
SGCE +
42
n=10
SGCE +
6
n=17
SGCE +
17
129
SGCE +
76

Current work…

- Re-analysis
  - Adding new responders
  - Only including *SGCE +*



- Paper in progress

ERN-RND CPMS case discussion jour fix

= (bimonthly) regular Case Discussions for ERN-RND members

Alternating focus:

1. presentation of unsolved/complex cases

OR 2. disease management

OR 3. DBS

CPMS to provide medical data (for consultations on unsolved/complex cases) or as repository of disease management presentations (for educational purposes)

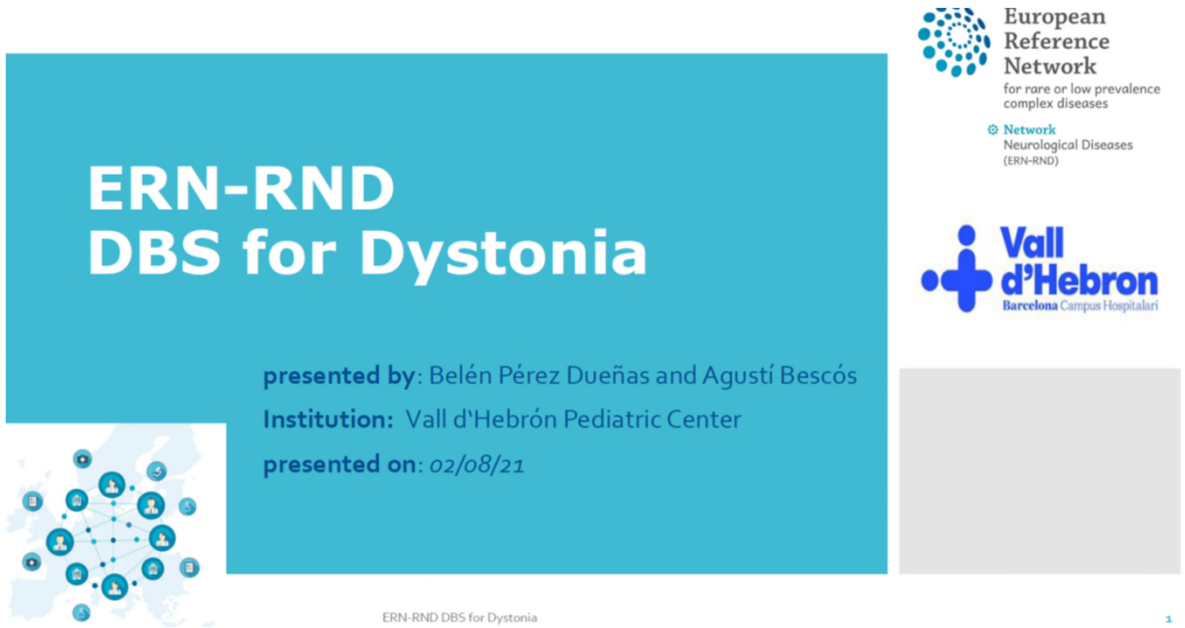
→ *Educational character; everyone from the network can join to listen and discuss (Zoom Meeting)*

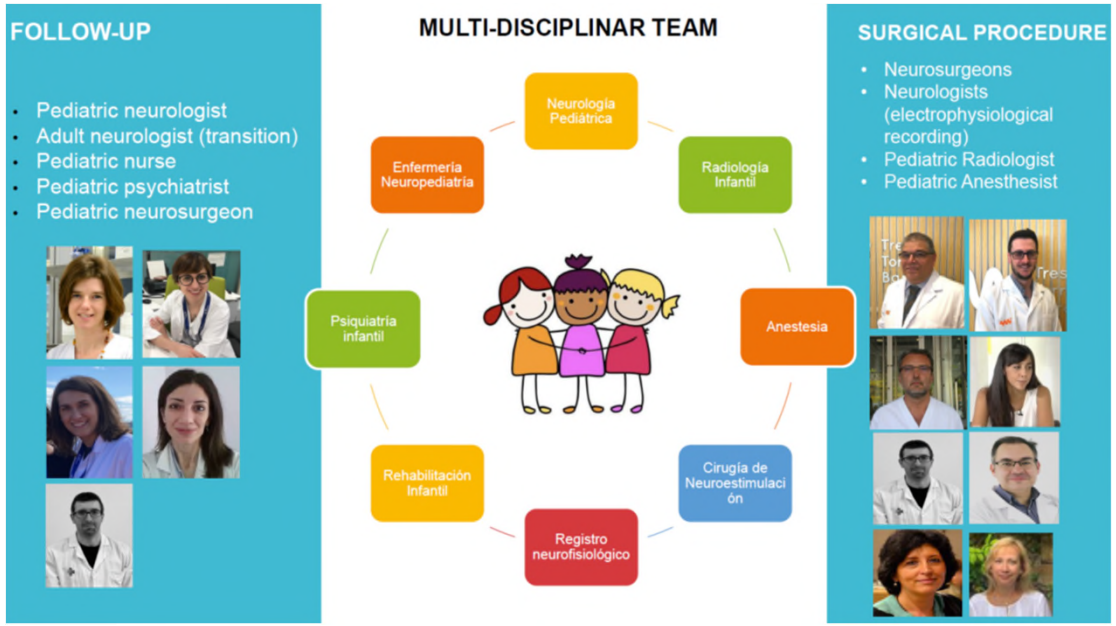
→ *Registration for event necessary (links sent via bulletin, collaborative platform, email to DG)*

→ *Case submission still possible*

Presentation of cross-cutting activites

- *CPMS case discussions "jour fix":*





Webinars
DG Dystonia, NBIA and paroxysmal disorders

53: total number of webinars
6/7 in this DG,
1 with focus on Neurorehabilitation in Dystonia

Date	Topic	Speaker	Focus
04.02.2020	Clinical evaluation of dystonia	Kailash Bhatia	
03.03.2020	Ultrasound diagnostics for cervical dystonia	Tobias Bäumer	
12.05.2020	Paroxysmal dyskinesias: update on clinical and genetic aspects	Giovanna Zorzi	pediatric
02.02.2021	Genetic dystonias and treatment	Sylvia Boesch	
09.03.2021	Myoclonus dystonia	Belen Perez, Marina Tijssen	pediatric
28.09.2021	Treatable dystonia &dystonia in inborn errors of metabolism	Tom de Koning	pediatric
07.12.2021	DBS in Children	Anne Koy	pediadric

Contact: Sanja.Hermanns@med.uni-tuebingen.de

Proposed WEBINARS 2022

- BelenPerez Dueñas, belen.perez@vhir.org

  “Basal ganglia diseases in childhood”
- Sylvia Boesch/Elisabetta Indelicato

  “Untangling neurodevelopmental disorders in the adulthood: a movement disorder/dystonia is the clue” (illustrative cases)

- Acquired dystonias
- Dystonias in autoimmune disorders
- DBS outcomes in secondary dystonia
- Dystonia scales in children
- NBIA spectrum disorders

Guideline Development

DYSTONIA/DBS Guidelines; last EUROPEAN guidelines 2011 (EFNS); MDS

27.09.21 Carola Reinhard:

- We have finally received detailed information about the methodological support from a consortium of methodologists financed by the European Commission for guideline development:
- Support for the development for one (the first, more to follow) guideline, e.g. systematic review
- Timeframe: 27th November to 27th May (7 months)
- Support by 3 methodologists
- Guidelines Proposed:
    - *- Guideline DBS in Dystonia (proposed)*
    - *- Management guideline for HSP*
    - ***- Guideline for atypical parkinsonian syndromes***
    - *- NKX2-1 guideline*
    - *- Consensus document myoclonus dystonia (proposed)*
    - ***- MLD guideline***

11.10.21 Carola Reinhard:

There are already two guideline projects with PICOs ready, so I would propose to search for another option for the Dystonia guideline. This might include support from the consortium mentioned below at a later time point (we might learn from the experiences of the first round), support from the EAN or something else.

European Journal of Neurology 2011, **18:** 5–18 doi:10.1111/j.1468-1331.2010.03042.x

EFNS GUIDELINES

EFNS guidelines on diagnosis and treatment of primary dystonias

A. Albanese[a,b], F. Asmus[c], K. P. Bhatia[d], A. E. Elia[a,b], B. Elibol[e], G. Filippini[a], T. Gasser[c], J. K. Krauss[f], N. Nardocci[a], A. Newton[g] and J. Valls-Solé[h]

[a]*Istituto Neurologico Carlo Besta, Milan, Italy;* [b]*Università Cattolica del Sacro Cuore, Milan, Italy;* [c]*Department of Neurodegenerative Diseases, Hertie-Institute for Clinical Brain Research, University of Tubingen, Germany;* [d]*Institute of Neurology, University College London, Queen Square, London, UK;* [e]*Hacettepe University Hospitals, Department of Neurology, Ankara, Turkey;* [f]*Department of Neurosurgery, Medical School Hannover, MHH, Hannover, Germany;* [g]*European Dystonia Federation, Brussels, Belgium; and* [h]*Neurology Department, Hospital Clínic, Barcelona, Spain*

Table 1 Classification of dystonia based on three axes

1. By cause (aetiology)
   Primary dystonias
   Primary pure dystonias: torsion dystonia is the only clinical sign (apart from tremor), and there is no identifiable exogenous cause or other inherited or degenerative disease. Examples are DYT1 and DYT6 dystonias.
   Primary plus dystonias: torsion dystonia is a prominent sign but is associated with another movement disorder, for example myoclonus or parkinsonism. There is no evidence of neurodegeneration. For example, DOPA-responsive dystonia (DYT5) and myoclonus-dystonia (DYT11) belong to this category.
   Primary paroxysmal dystonias: torsion dystonia occurs in brief episodes with normalcy in between. These disorders are classified as idiopathic (often familial although sporadic cases also occur) and symptomatic because of a variety of causes. Three main forms are known depending on the triggering factor. In paroxysmal kinesigenic dyskinesia (PKD; DYT9), attacks are induced by sudden movement; in paroxysmal exercise-induced dystonia (PED) by exercise such as walking or swimming and in the non-kinesigenic form (PNKD; DYT8) by alcohol, coffee, tea, etc. A complicated familial form with PNKD and spasticity (DYT10) has also been described.
   Heredodegenerative dystonias: dystonia is a feature, amongst other neurological signs, of a heredodegenerative disorder. Example: Wilson's disease.
   Secondary dystonias: dystonia is a symptom of an identified neurological condition, such as a focal brain lesion, exposure to drugs or chemicals. Examples: dystonia because of a brain tumour, off-period dystonia in Parkinson's disease.

2. By age at onset
   Early-onset (variably defined as ≤20–30 years): usually starts in a leg or arm and frequently progresses to involve other limbs and the trunk.
   Late onset: usually starts in the neck (including the larynx), the cranial muscles or one arm. Tends to remain localized with restricted progression to adjacent muscles.

3. By distribution
   Focal: single body region (e.g., writer's cramp, blepharospasm)
   Segmental: contiguous body regions (e.g., cranial and cervical, cervical and upper limb)
   Multifocal: non-contiguous body regions (e.g., upper and lower limb, cranial and upper limb)
   Generalized: both legs and at least one other body region (usually one or both arms)
   Hemidystonia: half of the body (usually secondary to a structural lesion in the contralateral basal ganglia)

German Dystonia Registry

- Aims:
  - Collection of data on the clinical features, course and epidemiology, QoL, comorbidities, environmental factors and therapy of dystonia in Germany.
  - *Data includes Dystonia Coalition and COST datasets*
  - Screening of the population for monogenic forms of dystonia using next-generation sequencing
  - A resource of clinical data and biomaterials as a basis for collaborative research on dystonia.

- Status:
  - >2200 Patient enrolled
  - New Recruiting centres welcome

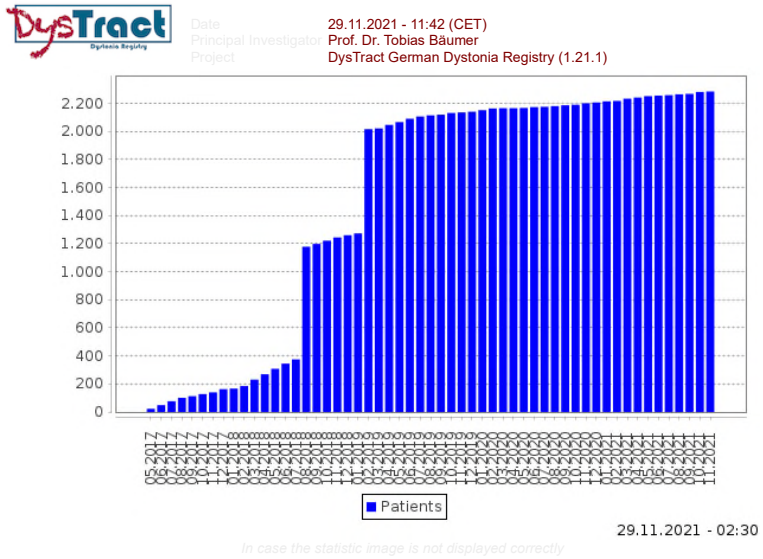
- Contact:

University of Lübeck

Christine Klein and Tobias Bäumer

dystract@neuro.uni-luebeck.de

- Centres:



European Reference Networks

DG FRONTOTEMPORAL LOBAR DEGENERATION

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

Co-funded by the EU

Activities in 2021

- Common scale: CDR-NACC FTLD modules
- Implementation of register
- Expansion of the FTLD expert centres
- CPMS
- Consensus document on multilingual evaluation of Primary Progressive Aphasia
- Current project: Drug and nondrug treatment options for behavioral disturbances in FTLD

Project Drug and Nondrug treatment options for behavioral disturbances in FTLD

- Step 1: Survey of treatment applied in ERN-RND FTLD network
  - List of behavioral disturbances
  - Which treatment options are availabile at your centre
  - List of drug treatment options that need to be ranked according to level of recommendation or that are strongly contraindicated
  - List of nondrug treatment options that one would recommend
  - Order of use
- Step 2: Collate responses and make summary report.
- Step 3: Form consensus based on the answers, possibly via Delphi procedure
- Step 4: Expert opinion paper for peer reviewed journal

Project Drug and Nondrug treatment options for behavioral disturbances in FTLD

- List of 14 behavioral disturbances:
  - Disinhibition
    - Verbal agression
    - Physical agression
    - Sexual disinhibition
    - Impulsivity
  - Obsessive repetitive behavior
    - Delusions of obsessive nature
    - Motor unrest (obsessive walking around)
    - Self-harm due to repetitive behavior, eg skin abrasions
  - Automutilation
  - Hyperphagia with excessive weight gain
  - Rigidity of thought
  - Perseverative somatic complaints
  - Nightly unrest
  - Apathy
  - Loss of empathy/sympathy

Project Drug and Nondrug treatment options for behavioral disturbances in FTLD

- List of drug treatment options:
  - amitriptyline - bupropion - carbamazepine - (es)citalopram – fluoxetine – mirtazapine - olanzapine – oxazepam - quetiapine – risperidone – semaglutide - sodium valproate - sertraline – trazodone – promazine – periciazine – hydroxyzine – other

- List of nondrug treatment options:
  - behavioral therapy – cognitive therapy – day care – hospitalisation - institutionalization - psychoeducation of the relatives

Future Projects

- Speech rehabilitation in FTLD for primary progressive aphasia
  - Survey and consensus paper

European Reference Networks

DG LEUKODYSTROPHIES (C SEVIN / NI WOLF)

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

Co-funded by the EU

PROJECTS

Education

→ Webinars: 6/25 in 2021, more are planned for 2022

Patient care

→ Development of a guideline for metachromatic leukodystrophy (led by S Gröschel and C Sevin)

→ Regular case discussions (treatment indication for MLD, diagnostic questions), jour fixe for diagnostic cases

Research

→ Subgroup on scales for quality of life

→ Neuropathy in MLD; strabism as presenting symptom in MLD (Tübingen/Amsterdam, EJPRD grant)

Miscellaneous

→ Standardized phenotyping: scores for registry: GMFC-MLD, guideline for standardized MRI description / illustration for publications on leukodystrophies

→ Overview therapeutic trials for leukodystrophies in ERN centers

→ Review on available MRI leukodystrophy scoring systems planned

MLD guideline

(led by Samuel Gröschel, Tübingen, and Caroline Sevin, Paris)

→ Task force members from several ERN centers in Germany/France, from Milan, and from patient organization (ELA); one representative from MetabERN

→ Child neurologists, adult neurologists, transplant / GT experts

→ Financial support from SSIEM and EAN, but: publication of guidelines only possible in one journal

Goal: building the first European MLD guideline, combining available data and knowledge on early diagnosis, natural history, management and treatment of the disease

Clinical questions formulated, timeline present

Challenges: not much literature evidence → combination of published evidence and expert-based consensus

PROJECTS

Scales

→ Still problematic

→ For the registry: GMFC-MLD

→ Quality of life scales: task force, including patient representative

→ Difficulty: age range from young infants to adults

→ Regulatory agencies (EMA): also no clear recommendation for a QoL scale

Standardized phenotyping

Illustration (minimal requirements)	level
axial t2	centrum semiovale
	Basal ganglia / thalami
	(mesencephalon)
	cerebellar white matter
sag image (T1 or T2)	mid sagittal
ALWAYS provide age at MRI	
additional images depending on pathology	
T1 (with / without contrast)	
FLAIR	
SWI	
DWI (incl ADC)	

Describe white matter findings using standard terminology	predominant where?
	confluent / multifocal
	contrast enhancing?
	cystic/rarefied?
	symmetric / asymmetric ?
	other characteristics (calcifications, microbleeds…)
	signal intensity on T2 and T1

Structure	Affected / not affected
Periventricular white matter (par occ / temp / fron)	
Central white matter (par occ / temp / fron)	
Subcortical white matter (par occ / temp / fron)	
Corpus callosum (genu)	
Corpus callosum (body)	
Corpus callosum (splenium)	
Cortex	
Basal ganglia	
Thalamus	
Mesencephalon	
Brain stem	
Cerebellar white matter	
Dentate nucleus	
Cerebellar cortex	
Spinal cord	
Dorsal colums	
lateral corticospinal tracts	
ventral corticospinal tracts	
grey matter	
General	
Supratentorial atrophy: inner CSF spaces	
Supratentorial atrophy: outer CSF spaces	
Cerebellar atrophy: vermis	
Cerebellar atrophy: hemispheres	
Other important findings	

Overview clinical trials for treatment within ERN-RND

→ Should be on the ERN-RND website, template provided

Metachromatic leukodystrophy: several ERN-RND centers participate in EMBOLDEN((intrathecal enzyme replacement therapy; inclusion completed)

X-ALD: Minorix for beginning c-ALD, enrolling

Vanishing White Matter: Guanabenz trial, enrolling

Alexander disease: antisense oligonucleotides, (enrolling)

Krabbe disease: intrathecal gene therapy, starting early 2022

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DG ATYPICAL PARKINSONIAN SYNDROMES

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

Co-funded by the EU

Overview

- Coordinators
- Training and Education
- Twinning Projects
- Guideline Development

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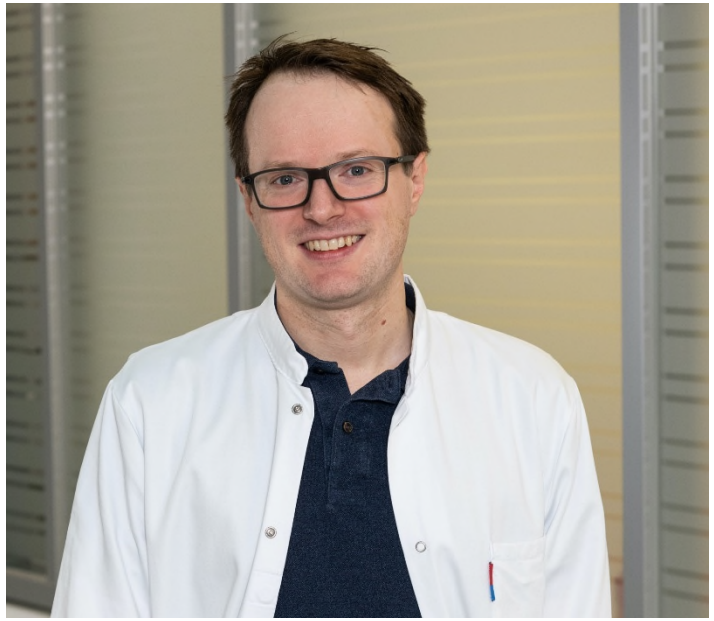
COORDINATORS

Coordinators

Florian Krismer



Johannes Levin

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TRAINING AND EDUCATION

Webinars DG Atypical Parkinsonism – 2020/2021

Date	Topic	Speaker
14.04.2020	Recognizing atypical parkinsonism	Wassilios Meissner
26.01.2021	Progressive Supranuclear Palsy– Update on Diagnostics, Biomarkers, Therapies	Günter Höglinger
14.09.2021	Genetic forms of Parkinson's Disease	Thomas Gasser
18.11.2021	Pure autonomic failure	Alessandra Fanciulli

Webinars 2022 - Outlook
Imaging characteristics of atypical parkinsonism - Norbert Brüggemann and Florian Krismer
Diagnostic Criteria Update - Multiple System Atrophy (MSA) Gregor K. Wenning
Management of young onset PD Alejandra Darling and Angeles Garcia
Overlap between atyp. PD and frontotemporal dementia (PSP, CBD, FTD with parkinsonism) Leonidas Stefanis and colleagues

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TWINNING PROJECTS ATYPICAL PARKINSONIAN SYNDROMES

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

Co-funded by the EU

AUTONOMIC FAILURE – SHORT EXCHANGE PROGRAM

Funding for participant:
200 € per diem + Travel arrangements

Focus

Autonomic failure in neurodegenerative disorders:

Testing autonomic function

Training offers include:

- cardiovascular autonomic function tests and
  - 24h ambulatory BP monitoring
- quantitative sudomotor axon reflex test, Skin
  - electrochemical conductance (Sudoscan)
- clinical diagnostic work-up and therapeutic
  - management of patients with ANS disorders
- specialized consultation in neuro-urology -
  - principles of urodynamic assessment

Institution

CHU de Toulouse, France

Discipline(s):

Medical doctors and lab technicians

Duration of stay:

min. 20 days

Language requirements:

French or English

Comments:

Planning at least 3 months in advance necessary

Focus

Autonomic failure in neurodegenerative disorders

- Autonomic function testing:
  - cardiovascular autonomic function tests,
  - quantitative sudomotor axon reflex test,
  - 24h ambulatory BP monitoring
- Clinical diagnostic work-up and therapeutic
  - management of people with ANS disorders

Institution

University Hospital Innsbruck, Austria

Discipline(s):

Medical doctors and lab technicians

Duration of stay:

Min. 20 days

Language requirements:

Fluency in English or German

Comments:

Planning at least 3 months in advance necessary

Focus

Autonomic dysfunctions

- Clinical evaluation and management
- Laboratory assessment

Institution

IRCCS Istituto delle scienze Neurologiche di Bologna, Italy

Discipline(s):

Medical doctors, specialized nurses, physiotherapists, neuropsychologists, Clinical Neurophysiology Technologist

Duration of stay:

min. 20 days

Language requirements:

English or Italian

Comments:

Funding of stay in this institution only after membership status has been confirmed.

NEUROREHABILITATION

Funding for participant:
200 € per diem + Travel arrangements

Focus	*Institution*
Speech Therapy in Ataxias	**Assistance Publique-Hôpitaux de Paris, Hôpital Pitié-Salepétrière, France** Reference Centre for Rare Diseases 'Neurogenetics'
Discipline(s): **no preference**	*Duration of stay:* **5 or 10 days**
Language requirements: **French**	*Comments:*

Focus	*Institution*
Ataxia: Movement Laboratory and the Application of Functional Evaluations	**IRCCS Ospedale Pediatrico Bambino Gesù in Rome, Italy** Centre of Expertise for rare pediatric neurologic diseases
Discipline(s): **physiotherapist, occupational therapist and/or physiatrist or neurologist**	*Duration of stay:* **5 or 10 days**
Language requirements: **Italian or English**	*Comments:* Period of visit must be planned months in advance due to hospital policy.

Focus	*Institution*
Neurorehabilitation • Ataxia • Hereditary Spastic Paraplegia • Parkinson's disease	**Radboud University Medical Center in Nijmegen, The Netherlands**
Discipline(s): **Medical Doctors, physiotherapists, occupational therapists, speech language therapists**	*Duration of stay:* **10 days**
Language requirements: **Dutch or English**	*Comments:*

Focus	*Institution*
Neurorehabilitation Choice of specifications: • Respiration therapy • Treatment of spasticity • Atypical parkinsonism • Huntington's disease	**General University Hospital in Prague, Czech Republic** First Faculty of Medicine Department of Neurology and Centre of Clinical Neuroscience
Discipline(s): **Medical doctors, physiotherapists**	*Duration of stay:* **5 days**
Language requirements: **Czech or English**	*Comments:*

Focus	*Institution*
Multidisciplinary approach for hyper- and hypokinetic movement disorders Choice of departments: • Neurology • Rehabilitation • neuropsychology • combined clinics	**University medical Center Groningen (UMCG) in Groningen, The Netherlands** Expert Centre for Movement Disorders Department of Neurology AB 51
Discipline(s): **Medical Doctors, physiotherapists**	*Duration of stay:* **5 or 10 days**
Language requirements: **English and Dutch**	*Comments:* Health check might be necessary in advance (e.g. tuberculosis screening)

Genetic Aspects

- CPMS-based
  - 1rst CPMS case discussion: Presentation of two solved cases
  - 2nd CPMS case discussion: Unsolved cases
  - Planned for March/April 2022

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GUIDELINE DEVELOPMENT - UPDATE

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

Co-funded by the EU

GUIDELINES – ONGOING EFFORTS

- Guidelines for symptomatic treatment of patients with MSA and PSP
  - Coordinators for the MSA guidelines:
    - Alessandra Fanciulli and Maria Teresa Pellecchia.
  - Coordinators for the PSP guidelines:
    - To be Determined
  - Collaborative effort with the DGN, EAN and MDS
  - Steering committee: Wassilios Meissner, Günter Höglinger, Florian Krismer, Johannes Levin.

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GUIDELINE DEVELOPMENT

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

Co-funded by the EU

GUIDELINE ACTIVITIES WITHIN ERN-RND

- Leukodystrophies: MLD guideline
- Atypical parkinsonion syndromes: MSA/PSP guidelines
- HD/Choreas:
  - HD diagnosis guideline
  - NKX2-1 guideline
- Ataxia/HSP:
  - Consensus statement Diagnostic algorithm in early-onset Ataxias
  - (Management of HSP guideline)

Planned

Dystonia:

- DBS in Dystonia guideline
- Consenus statement myoclonus dystonia

Support to guideline development from European Commission

- Handbooks – on Collaborative Plattform
- Training
- Methodolgical support – NKX2-1

Manuscript: EAN guidance for developing and reporting clinical practice guidelines on rare neurological diseases

- Under review at European Journal of Neurology

VALUE OF TREATMENT

Co-funded by the EU

VOT Objectives

Project Objectives

- Identify the current treatment gaps and patient needs along the care pathway and analyse the underlying causes & Identify/propose solutions addressing the treatment gaps (“Care Pathway Analysis”)
- Evaluate the costs and burden associated with the treatment gaps and the socio-economic impact of closing/reducing them by applying the solutions identified/proposed for three (or more) case study countries (“Economic Evaluation Study”)
- Propose policy recommendations on how to improve the care pathway(s).

Timelines

Submission of scientific publications and policy papers in January 2022

Ataxia Working Group – Specialist centres vs. Non-specialist centres

Methods:

Patient perspective questionnaires: Diagnosis ı Care pathways ı Symptoms management ı Patient satisfaction

UK, Germany, Italy

Results:

Analysis per country

UK data: Better care and management in specialist centres (SAC)

German data: Genetic diagnosis better in SAC, better care in SAC

comparison to UK data: No established referral care pathway; outpatient clinic, no difference in MDT management SAC vs. non-SAC

Italien data: Analysis of the data ongoing

Dystonia Working Group – Early diagnosis

Methods:

Patient perspective questionnaire: Factors influencing good care

UK, Germany, Italy, Croatia

Results:

Cross-country comparison

- Time to diagnosis and treatment ← Correct referral by primary care to expert centres
- Croatia: Special education in movement disorders for GPs → reduced referral times

→ Training needed for GPs and general neurologists

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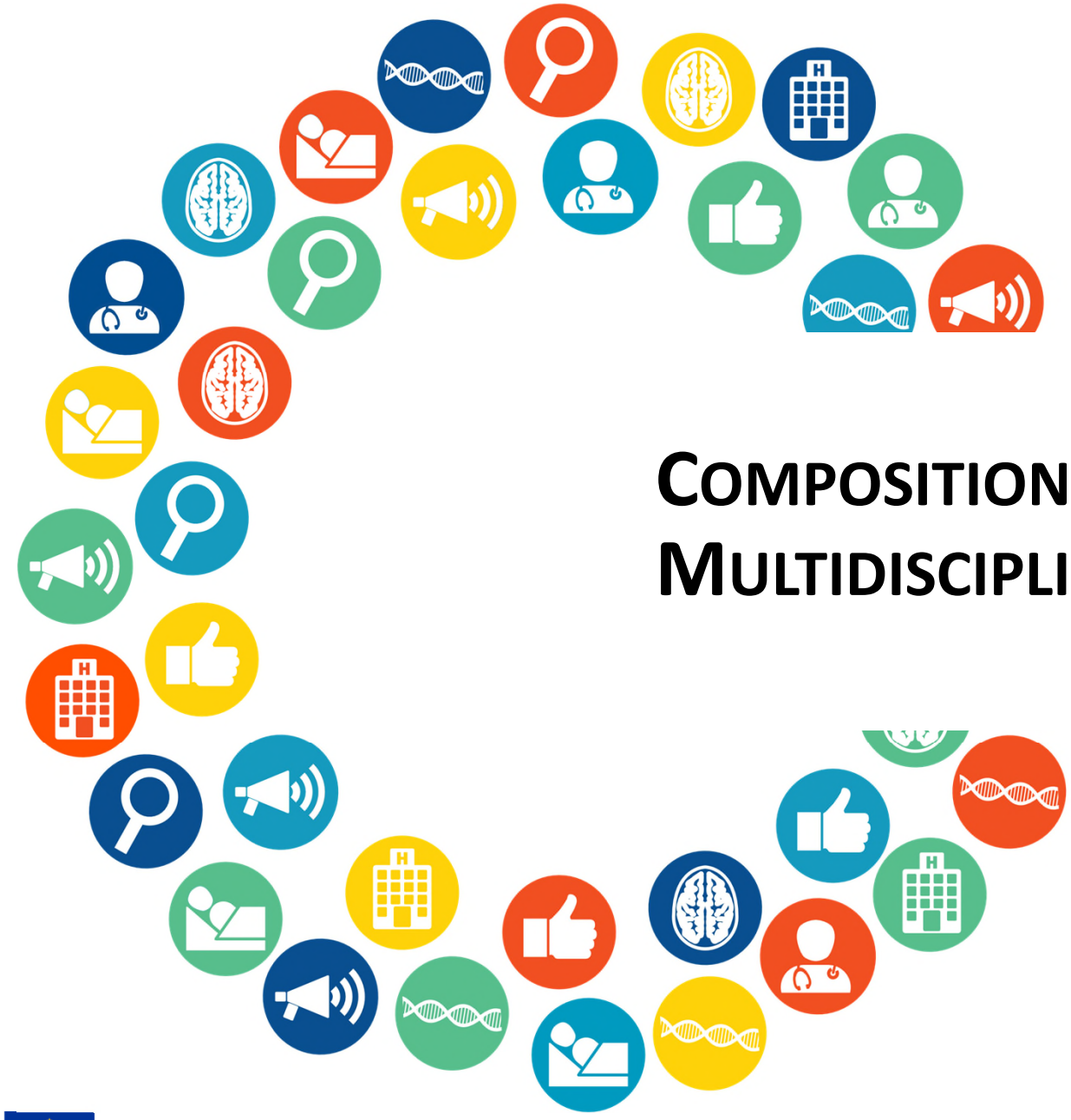

COMPOSITION MULTIDISCIPLINARY TEAM

„THE IDEAL“ MDT FOR MOVEMENT DISORDERS

- Adults vs pediatrics
- Different stages of the disease:
  - Diagnostic stage
  - Stage of preservation of function
  - Final/palliative stage

Competences have been agreed

(Neurologist with expertise in rare movement disorders and cognitive deficits, advanced practice nurse, rehabilitation specialist,…)

Delphi- voting

6 - mandatory core member of the MDT - this competence is necessary

5 - core member of the MDT - this competence is necessary to form a MDT for the majority of the patients

4- member of the extended team - this special competence is required for some of the patients

3 - member of the extended team - this special competence is required for few patients

2 - not a member of the MDT but special competence is useful for few patients

1 - not a member of the MDT

Results – Adults, diagnostic stage

Neurologist with expertise in rare movement disorders and cognitive deficits	Advanced practice nurse	Human Geneticist	(Neuro-) radiologist	Psychiatrist	(Neuro-) Psychologist	Neurosurgeon	Neuroophthal-mologist	Electrophysiolo-gist/EEG technologist	Neuropatho-logist	Urologist
4	2	3	1	1	2	1	1	1	1	1
5	2	3	1	2	3	1	2	1	1	1
6	3	3	3	2	3	1	2	1	1	2
6	3	3	4	3	3	1	2	2	1	2
6	3	4	4	3	3	1	2	2	2	2
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6	6	6	6	6	6	5	5	6	6	5
6	6	6	6	6	6	6	5	6	6	5
6	6	6	6	6	6	6	6	6	6	5
6	6	6		6	6					

Next steps

- Next Delphi round: Compare individual voting with median group voting → possibility to change individual votings
- repeat if necessary…
- Result: Expert consensus on composition of MDT

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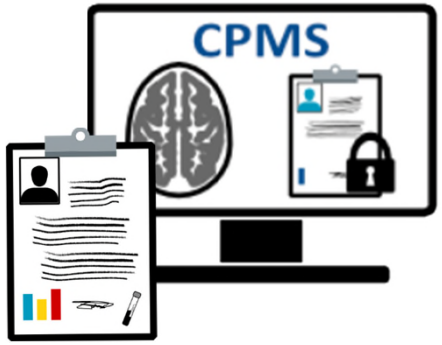
CASE DISCUSSIONS/CPMS

CPMS AND CASE DISCUSSIONS

Commiting to cross-border healthcare in 2022:

Nominate CPMS manager for your HCP

- Contact point for ERN-RND CPMS helpdesk
- Multipliers for CPMS training
- Manage centre‘s panels



Submit cases for Online Case discussions

Offer expert advice for Online Case Discussions

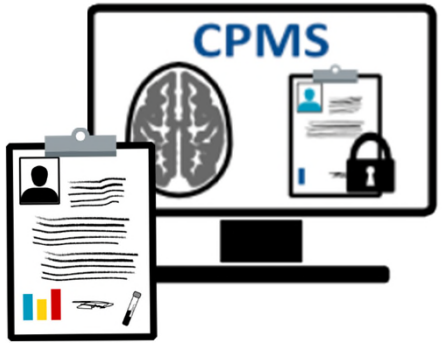
→ Contribute to one Case Discussion per DG covered by your HCP /year

→ Soon you will receive more information via mail

CPMS AND CASE DISCUSSIONS

CPMS helpdesk

- **Helpdesks (coordination office):** Sanja Hermanns & Tamara Martin
- ***New helpdesk:*** Alisa Jemelka from University Medical Centre Schleswig-Holstein, Germany



CPMS improvements

- ***Already:*** Improved user-friendliness
- ***Coming soon:*** additional significant improvements expected in 2022 and 2023

Online Case Discussions

- ***Continued:*** eConsultations for all CPMS cases
- ***New:*** regular Case Discussions open to all ERN-RND members for educational purposes

European Reference Networks

TRAINING AND EDUCATION

EDUCATIONAL ACTIVITIES IN ERN-RND

CPMS Case discussions | Short Exchange Programme | Webinars | Postgraduate Curriculum | Winterschool | Research in RND

Online Case discussions:

- regular, pre-scheduled meetings open to all ERN-RND members
  → *join to share, contribute, listen, learn!*
- Alternating focus:
  - Unsolved/complex cases
  - Disease management
  - Highly specialized services (DBS in Dystonia)
- **Since July 2021:** 12 discussions, usually 2 cases presented

→ Please contribute cases/expertise & forward invitations to your colleagues/residents!

ERN Exchange Programme, funded by European Commission

- Aim: support sharing of knowledge and stimulate collaboration in ERNs
  → *Funding for secondments up to 4 weeks with clinical perspective*
- Programme periode: March 2021 – August 2022
- Approved fellowships: 3 (using 6 packages/44)
- Exchanges suspended until February 2022 (COVID-19)

→ *applications for visits from March on still possible and highly encouraged!*

→ *please let also your peers/residents know the programme exists!*

EDUCATIONAL ACTIVITIES IN ERN-RND

Webinars

Nov. 2019 to Dec. 2021: 53 webinars

Focus:

- Neurological, neuromuscular & movement disorders (DG specific)
- Neurorehabilitation
- Advanced Therapies
- DBS

Postgraduate Curriculum

- **Collaboration with ERN EuroNMD and EpiCARE**
- Collaboration with EPNS, EAN
- ERN-specific topics
- ERN cross-cutting topics:

neurogenetics, neuroimaging, neurorehabilitation, clinical research, digital care, patient perspective

- Workshop to be organized in spring/summer 2022

Winterschool

- **Neurorehabilitation**
- 20-22. January 2022; Budapest, HUN, hybrid→ Tübingen online
- Registration still open, limited nr. of participants for the hands-on sessions

Research

Funding available from the EJP programme

- Stays up to 6 months at an expert center
- 2 days workshop

Next steps:

- Define competencies
- Mapping existing educational materials
- Produce materials that are lacking

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COMMUNICATION

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

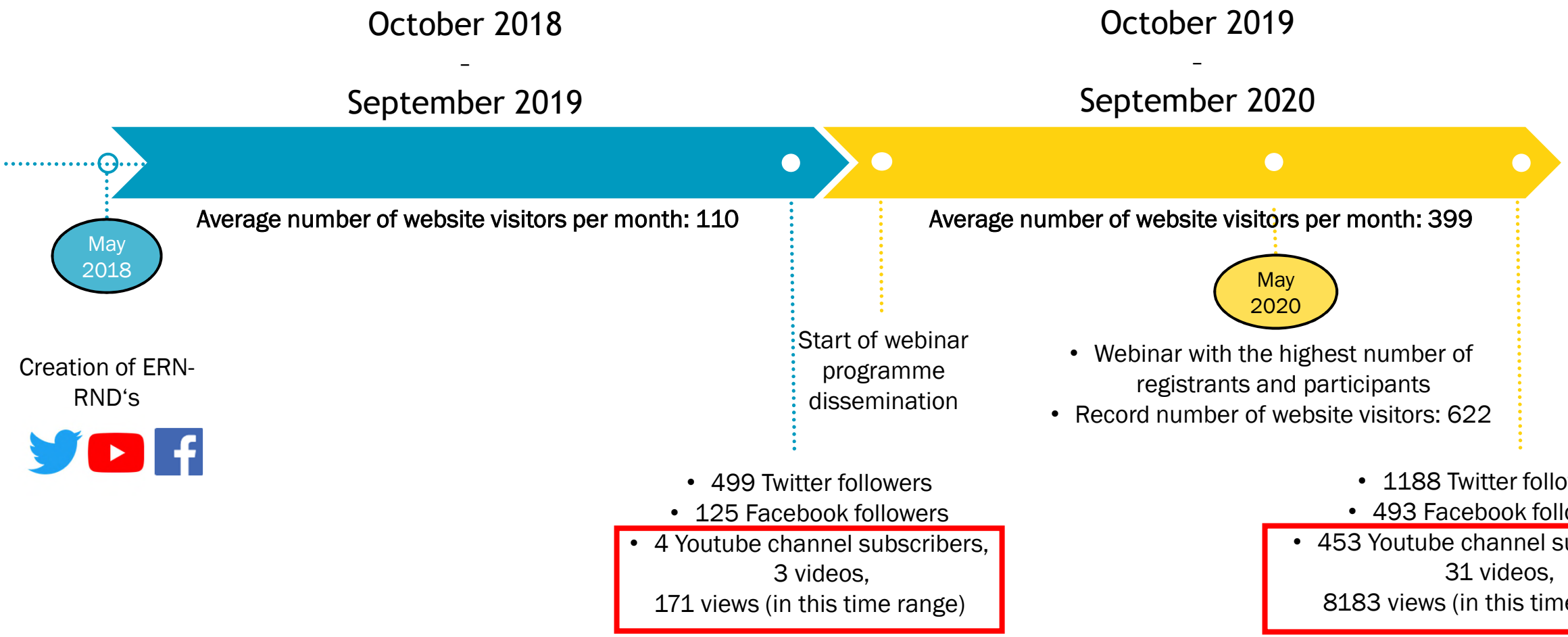
Co-funded by the EU

COMMUNICATION STRATEGY PAPER

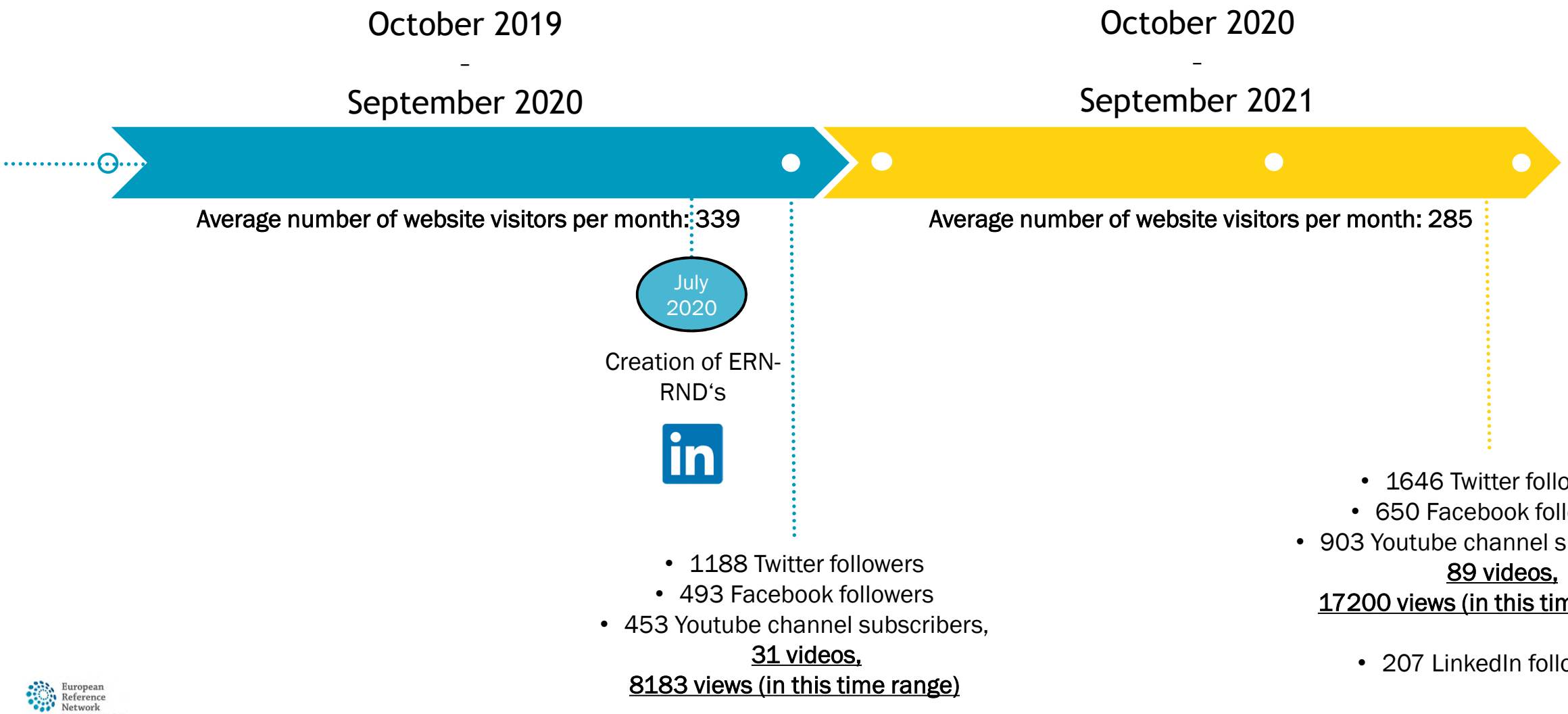
Establishing and Boosting Communication in The European Reference Network For Rare Neurological Diseases (ERN-RND): The Impact of Offering Free Educational Webinars

- Submitted to Orphanet Journal of Rare Diseases
- Compared communication reach before and after implementation webinars

COMMUNICATION STRATEGY PAPER



COMMUNICATION NUMBERS



INTERACTIVE MAP FOR ERN-RND WEBSITE

INTERACTIVE MAP ON ERN-RND WEBSITE

If you haven‘t sent us your video yet, please do so!

More information:
communication@ern-rnd.eu

MEET THE MEMBERS

- Profession, challenges in rare disease care, participation in ERN-RND

- Interested? Let us know!

Meet the members

ANNEMIEKE BUIZER, EMMA CHILDREN'S HOSPITAL/AMSTERDAM UNIVERSITY MEDICAL CENTERS, THE NETHERLANDS

NICOLE WOLF, EMMA CHILDREN'S HOSPITAL/AMSTERDAM UNIVERSITY MEDICAL CENTERS, THE NETHERLANDS

FRAN BOROVEČKI, UNIVERSITY HOSPITAL CENTER ZAGREB, ZAGREB, CROATIA

SYLVIA BOESCH, UNIVERSITY HOSPITAL INNSBRUCK, AUSTRIA

Newsletter & Bulletin

Newsletter:

- First week of each month
- Everyone interested in ERN-RND

Bulletin:

- Around the 15th of each month
- ERN-RND members only
- Internal ERN-RND material

Please read the newsletter, this way we can reduce emails!

If you have anything to include, please contact us.

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REGISTRY

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

Co-funded by the EU

PROGRESS

- Information and files for ethic proposal and data sharing agreement provided to all HCPs
- Feedback by most centers received
- Registry manager, Dorotea Köpper (Lleshaj), on sick leave till 3/2022
- Coordination office (Carola, Holm and Ludger) tries to compensate while searching for replacement

COMMON DATASET

ERN-RND Minimal Dataset

1. Pseudonym – same in following years
2. Date of birth → Year of birth
3. Sex
4. Alive (Patient's status)
5. ~~Date of death~~
6. First data entry in ERN-RND registry
7. Age at onset
8. Age at diagnosis or First data entry in ERN-RND registry
9. Orpha code as defined by disease groups
10. OMIM code for patients with genetic diagnosis
11. HPO term for disease group.
    Optionally: Phenotype of unsolved cases
12. Yes (Agreement to be contacted for research purposes)
13. Yes (Consent for the reuse of data)
14. Biological samples (Yes / No)
15. Link to a biobank (Link / No)
16. Disease group specific score

Score needs to be agree for
- Dystonia
- Fronto-temporal dementia

Workflow of Annual Reporting

- In February each HCP sends data of all patients of the previous year as a csv-file to registry manager
- March: Plausibility check in Tü and queries to HCPs
- April: Corrected files with final data
- May: Import in RedCap and
  Distribution of disease group specific cumulative data to all contributors

Ethics

- Pilot vote available (Tübingen)
- Patient information and consent forms available in German and English
- Alternatively: Generic ERN registry consent was developed in EJP-RD
- Project plan available in English
- Statement of Data Protection Officer (Tübingen)
- TOMs, VVT, DSFA available in German

- All HCPs should have submitted proposal to institutional review board
- Consents for data sharing already available in several HCPs
- These centers will be asked to submit data for 2021 in Feb 2022
- Re-imbursement by 1000 Euro

Data Sharing Agreement

- Needs to be signed by legal offices
- All HCPs should have submitted proposal to their local office
- Some legal offices raised issues that need to be solved
- Process supposed to be finished in December 2021

SCHEDULE

- 11/2021: Ethic vote and data sharing agreement submitted by all centers
- 1/2022: Consent for data sharing in place in all centers
- 2/2022: Reporting for 2021 in pilot centers
- 1 – 12/2022: Patients sign consent forms in all centers
- 2/2023: Reporting on patients of 2022 by all HCPs

European Reference Networks

PEDIATRIC ISSUES

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

Co-funded by the EU

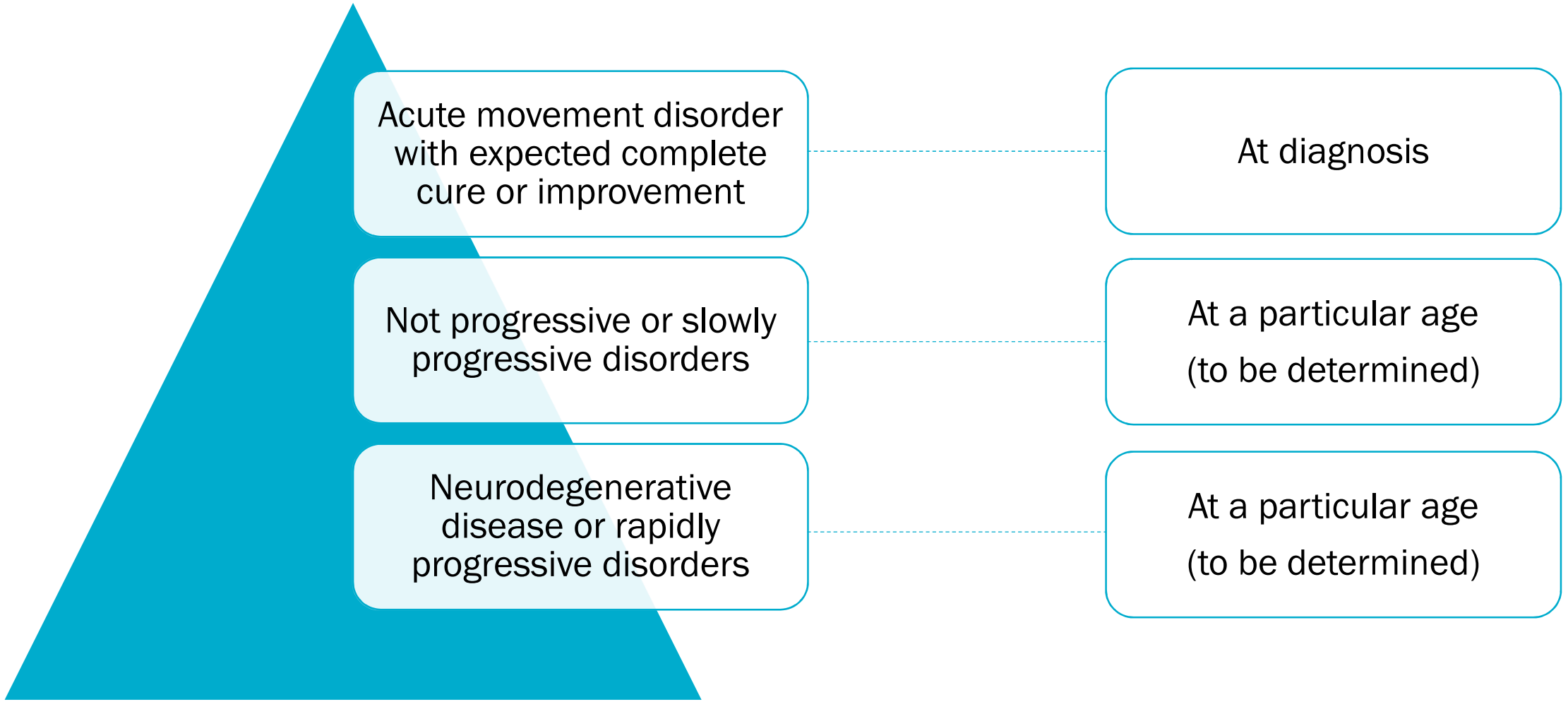
PROJECTS

- Standards phenotyping scales
- Patient leaflets/booklets
- Transition

STANDARDS PHENOTYPING SCALES

- Aims
  - To identify which, when and how often, these MD RS are applied by paediatric experts within the ERN-RND in daily practice
  - To optimize care delivery, we aimed to develop guidelines to guarantee the homogeneous application of these RS among the members of the ERN-RND
  - To standardize the application of RS to obtain comparable data in the future

STANDARDS PHENOTYPING SCALES



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Network Neurological Diseases (ERN-RND)

PATIENT LEAFLETS/BOOKLETS

- Aims
- To develop/endorse patient booklets with pediatric focus

At the meeting it was decided to start the project by group of diseases

Currently, we are in the collection phase of brochures / leaflets

We encourage any of our pediatric members who are interested in leading this project to contact the group coordinators

TRANSITION

- Project led by Caterina Mariotti

- Aims
- To understand the situation of the transition between the various members of the ERN-RND

Currently, a questionnaire about the transition will be distributed soon as a survey and the group is gathering publications and published material on transition

European Reference Networks

WG NEUROREHABILITATION

29TH NOVEMBER 2021

Agenda

- Report about the activities in the last 6 month
- Plans for the next year
- Further suggestions

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

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- NAME CENTER EXPERTISE
- Samih Almudafar, Sahlgrenska, HD
- Annemieke Buizer, Amsterdam , Leukodystrophies, HSP, Dystonia
- Ana Maria Dominguez, SJD Barcelona , children
- Andrea Dumitrescu, Madrid , neurorehabilitation
- Antonio Federico, Siena, liason to the EAN
- Christoph Gutenbrunner, Hannover pain, assessment, health systems, organization structures
- Jadwiga Kubica, Krakow, Ataxia, HSP, Leukodystrophies
- Natasa Klepac, Zagreb, dementia, cognitive problems
- Luigi Lavorgna, Naples, teleneuro-rehabilitation, digital health
- Lori Renna Linton EuroHSP representation of patients' advocacy organisation
- Kadri Medijainen Tartu extrapyramidal disorders
- Julita Medina SJD Barcelona children
- Judit Molnar Semmelweis Inpatient, Outpatient services offered
- Jorik Nonnekes Nijmegen atyp. PD, Ataxia HSP, gait
- Getrud Schönherr Innsbruck motor learning, assessments
- Susanna Summa Rome robotics for rehabilitation
- Dagmar Timmann UK Essen motor learning, cerebellum
- Ruth van der Looven Gent child rehabilitation, technology, neuromotor diseases
- Gessica Vasco Rome gait analysis, digitalization of functional scales, outcome measures
- Claudia Vinciguerra Salerno neuromuscular disease, music therapy

COLLABORATIONS

ERN-RND Collaboration with European Academy of Childhood Disability

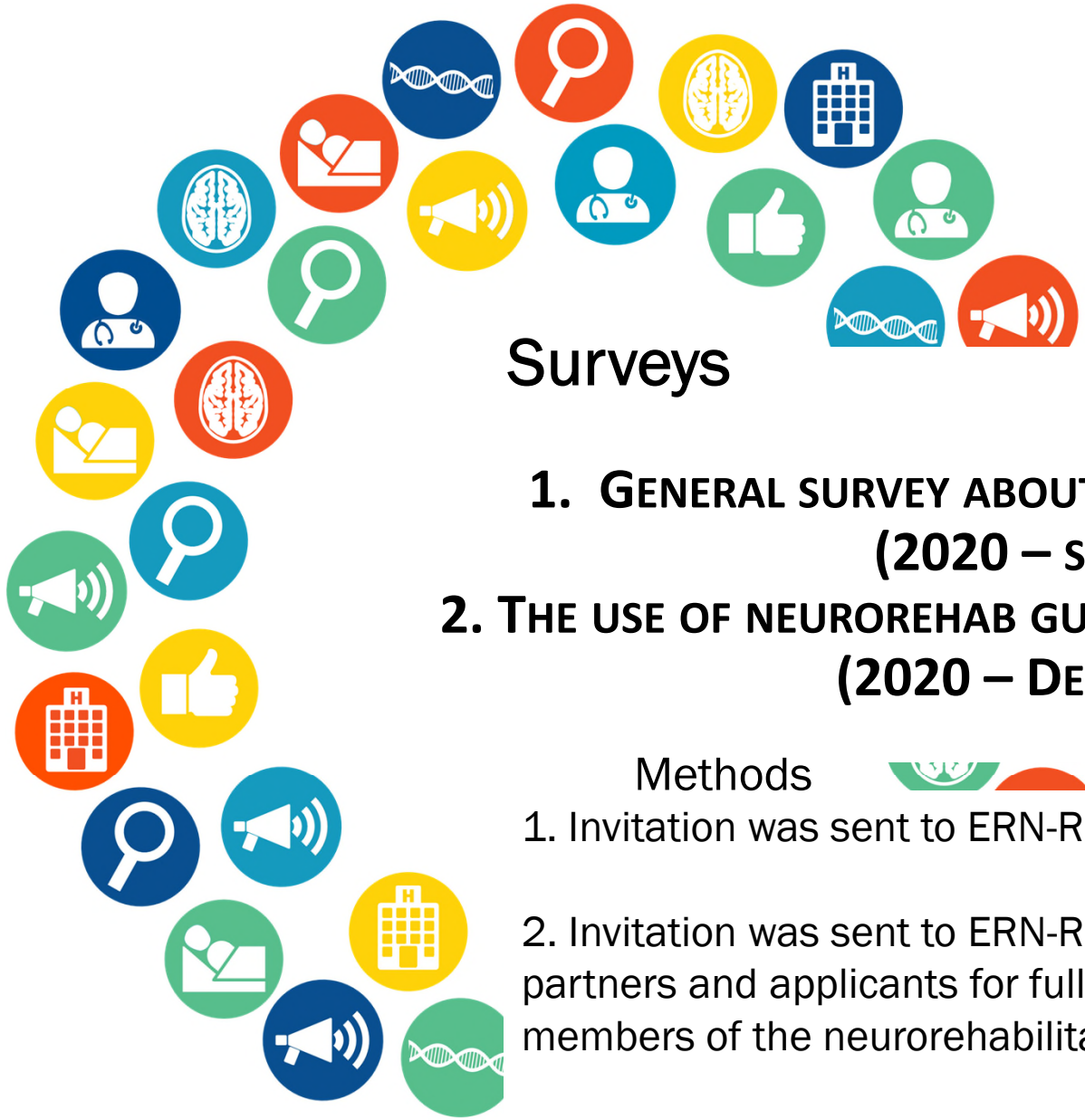
The ERN RND: networking knowledge into action

Minisymposium on the EACD Annual Meeting 2022 in Barcelona

- Improving care for children with RND – What does ERN-RND offer for you and your patients? – Carola Reinhard, project manager of ERN-RND
- Patient perspective in RND – Astri Arnesen, president of the European Huntington Association
- Recent achievements and new challenges in the care of children with RND: the role of European collaboration within ERNs – David Gómez-Andrés, child neurologist
- Neurorehabilitation in children with RND: best practice examples, Annemieke Buizer, pediatric physiatrist

ERN RND collaboration with European Paediatric Neurology Society

ERN-RND Collaboration with European Academy of Neurology



Surveys

1. General survey about neurorehabilitation (2020 – spring)

2. The use of neurorehab guidelines within ERN-RND (2020 – December)

Methods

1. Invitation was sent to ERN-RND full members

2. Invitation was sent to ERN-RND full members, affiliated partners and applicants for full membership as well as the members of the neurorehabilitation working group

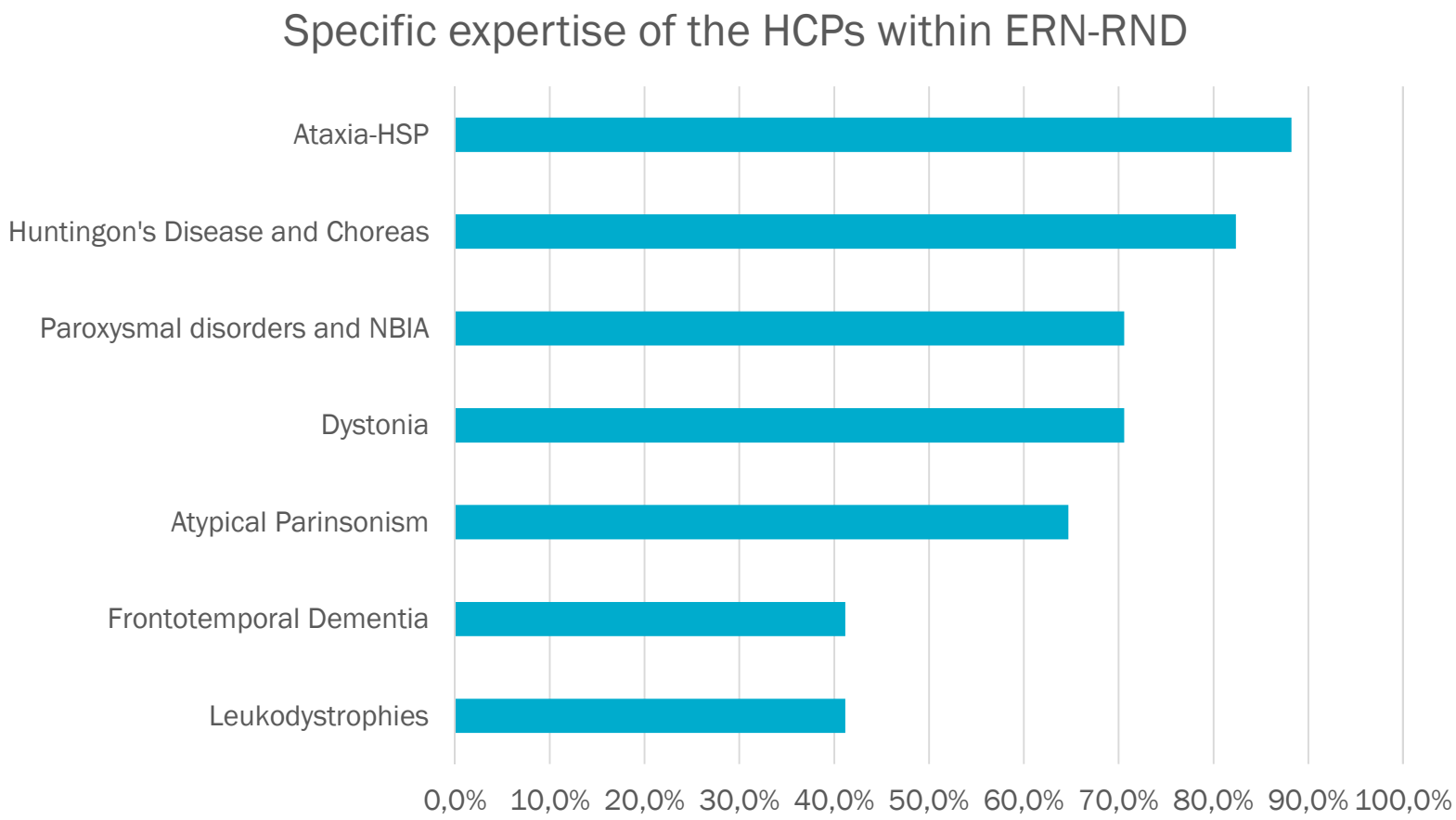
In both survey 17 countries participated

Co-funded by the EU

SURVEY 1.

27 ANSWERS FROM 17 COUNTRIES

- Austria
- Croatia
- Czech Republic
- Estonia
- Finland
- France
- Germany
- Hungary
- Ireland
- Italy
- Lithuania
- Malta
- Poland
- Slovenia
- Spain
- Sweden
- The Netherlands



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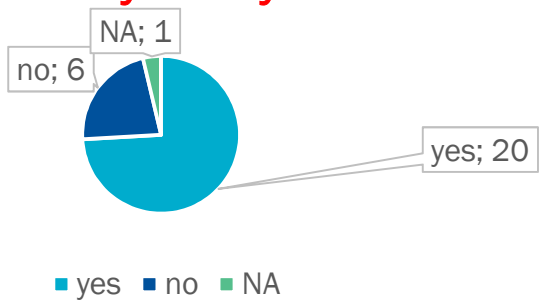
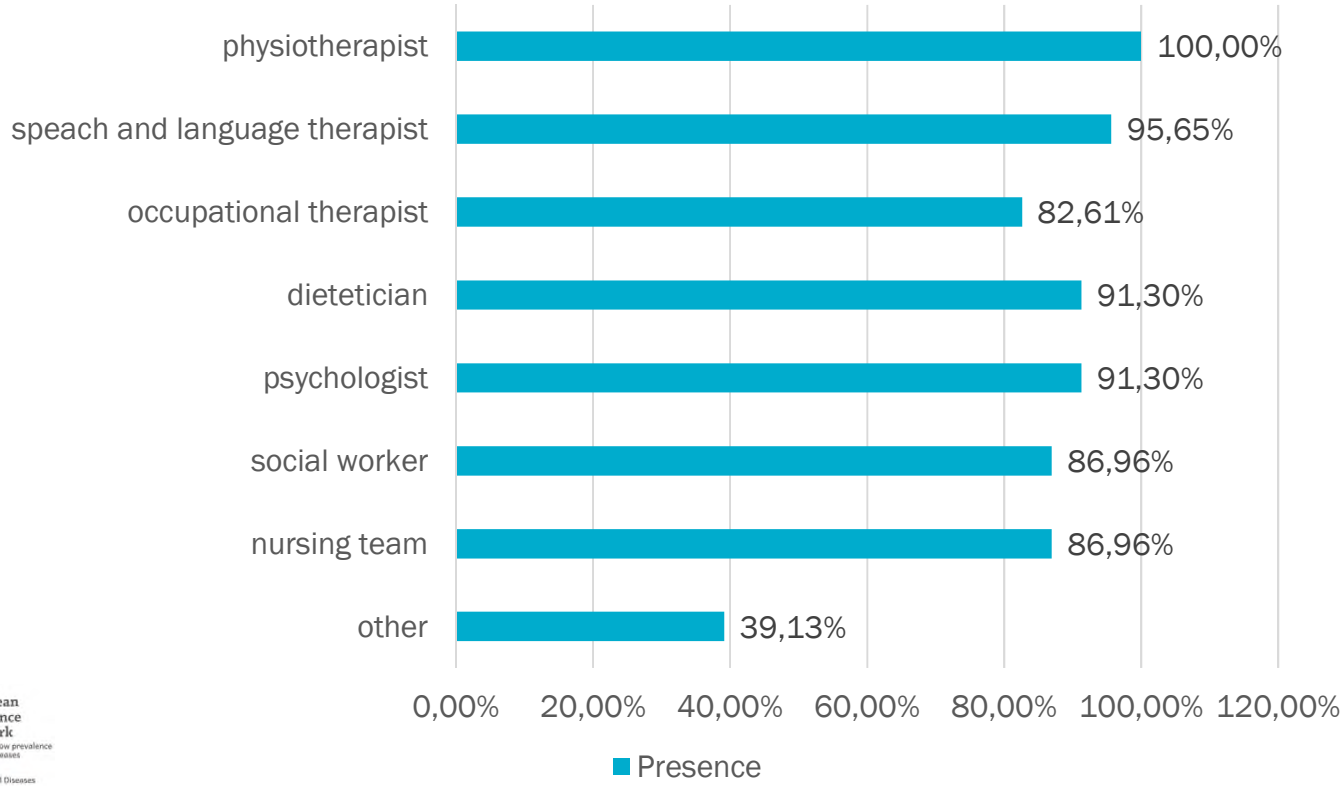
Network Neurological Diseases (ERN-RND)

MULTIDISCIPLINARY TEAMS

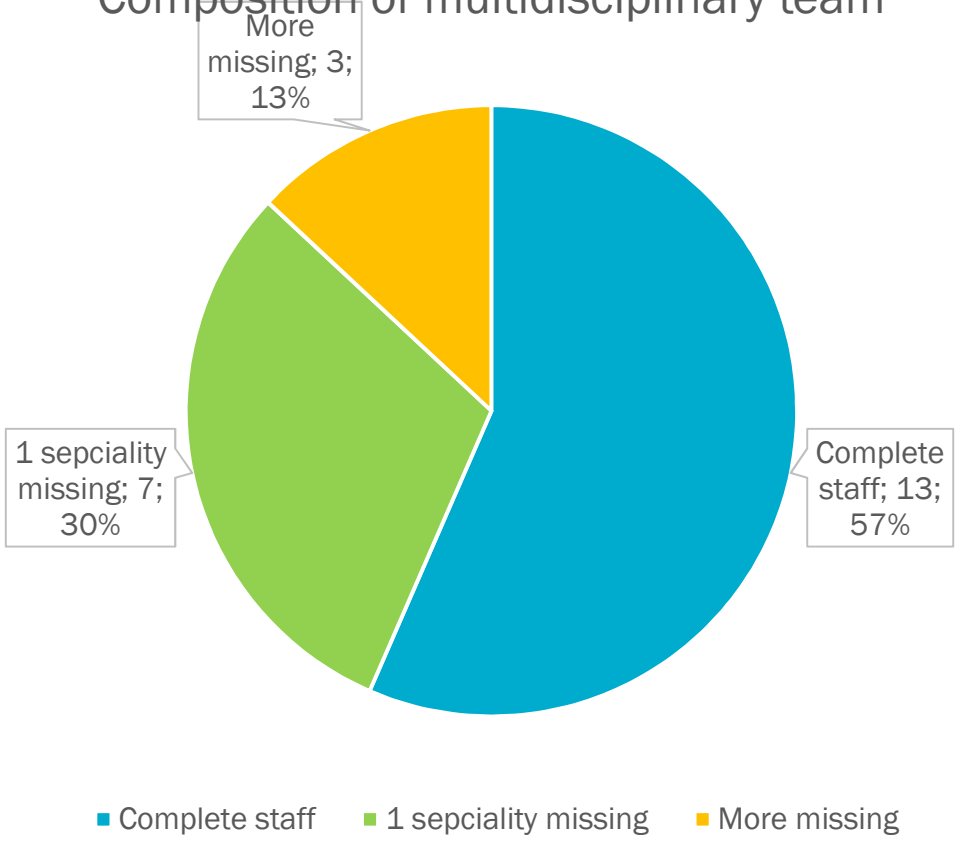
Is multidisciplinary care covered by HC system?



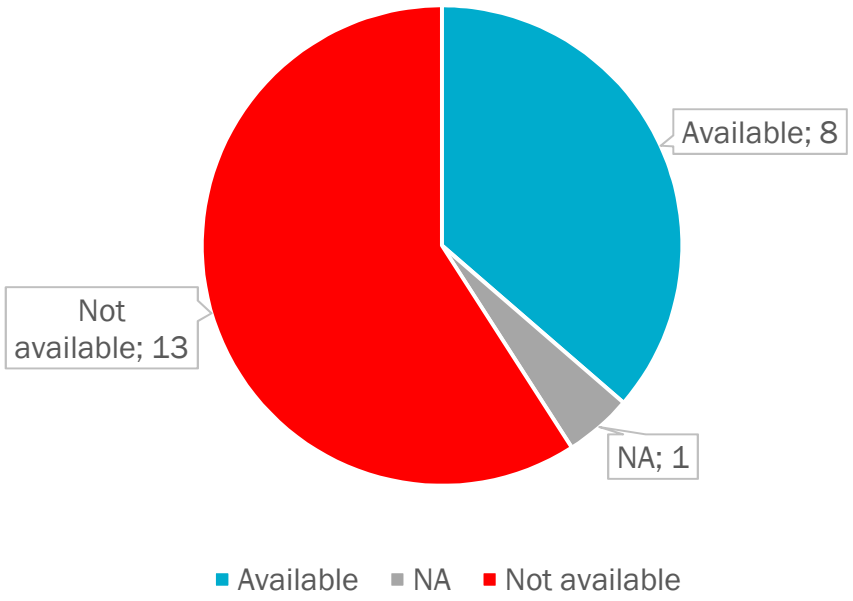
Participants of the team



Composition of multidisciplinary team



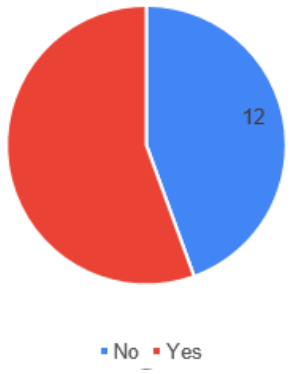
Inpatient program availability for specific RND



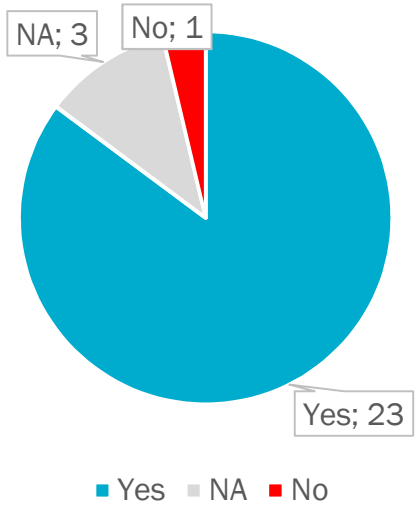
Programs for RND:

- Hunington Disease & choreas
- MSA
- PSP
- FTD
- Parkinsonism
- Ataxia
- Dystonia
- HSP

Is possible for the patient to reach neurorehabilitation for an extended period?



Are inpatient programs covered?



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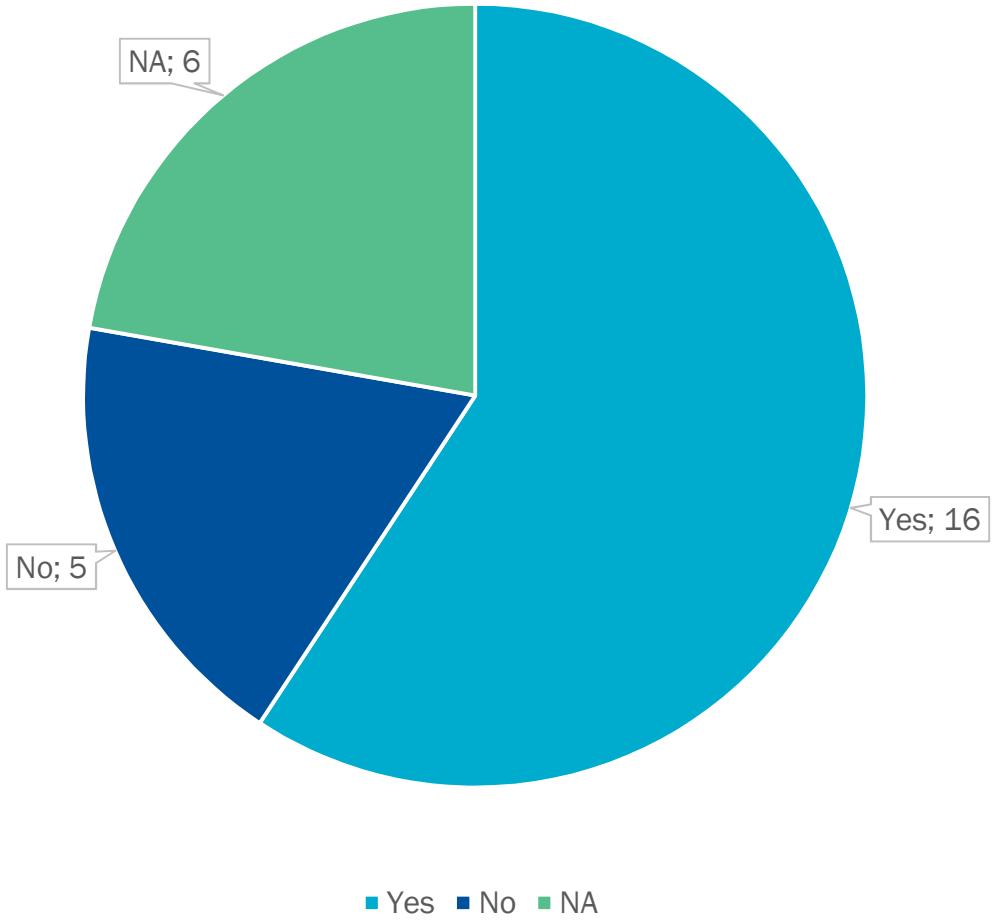
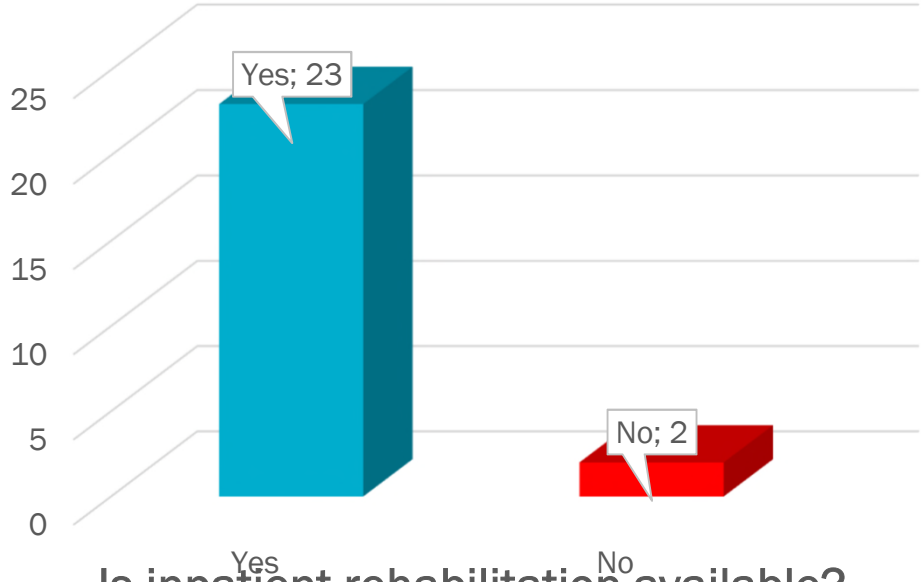
Network Neurological Diseases (ERN-RND)

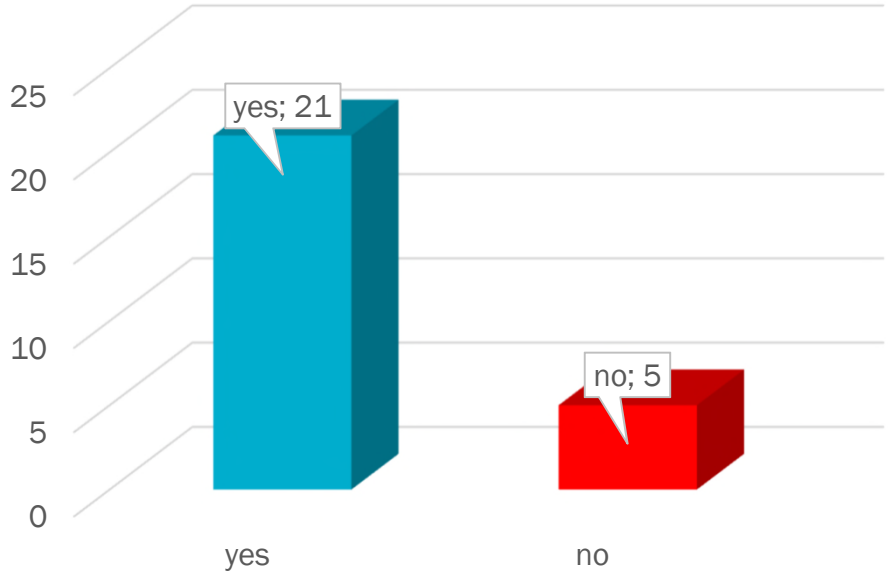
Extended rehabilitation availability



Is outpatient neurorehabilitation covered by health care system?



Is inpatient rehabilitation available?



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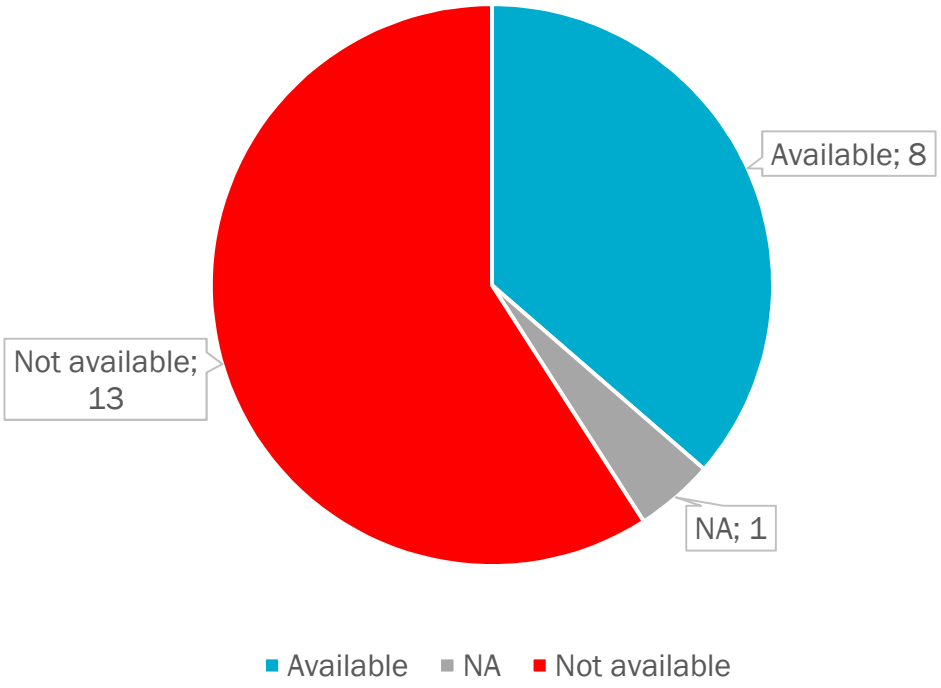
Network Neurological Diseases (ERN-RND)

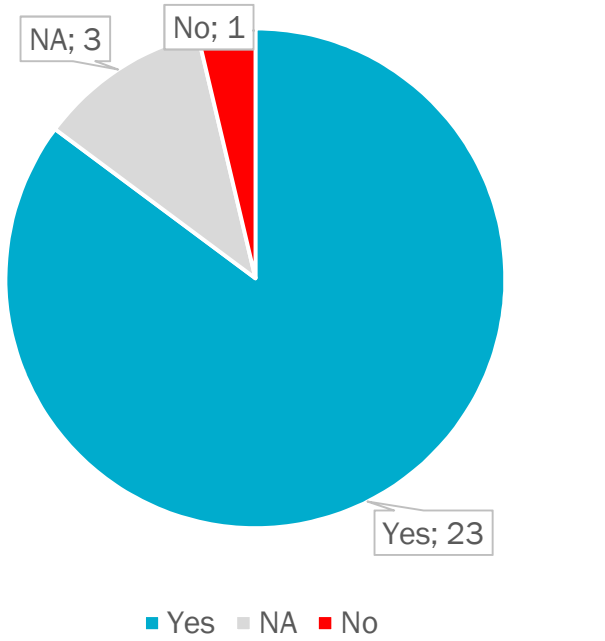
Inpatient program availability for specific RND



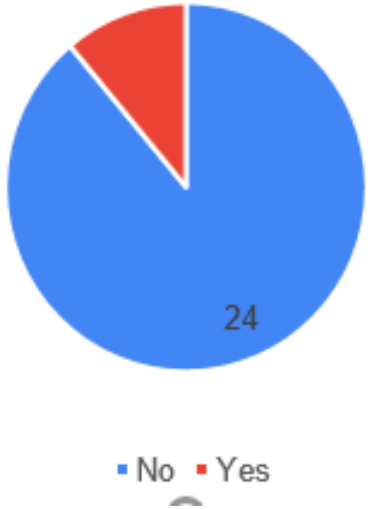
Programs for RND:

- Huntington Disease & choreas
- MSA
- PSP
- FTD
- Parkinsonism
- Ataxia
- Dystonia
- HSP

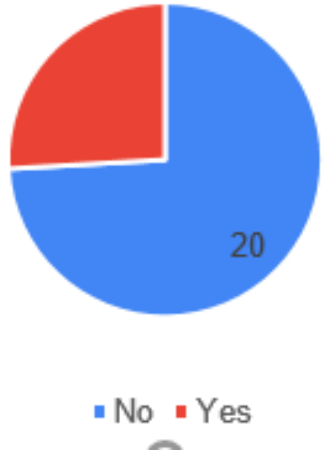
Are inpatient programs covered?



Do the inpatient neurorehabilitation specialists in your area have specific knowledge about the RND?



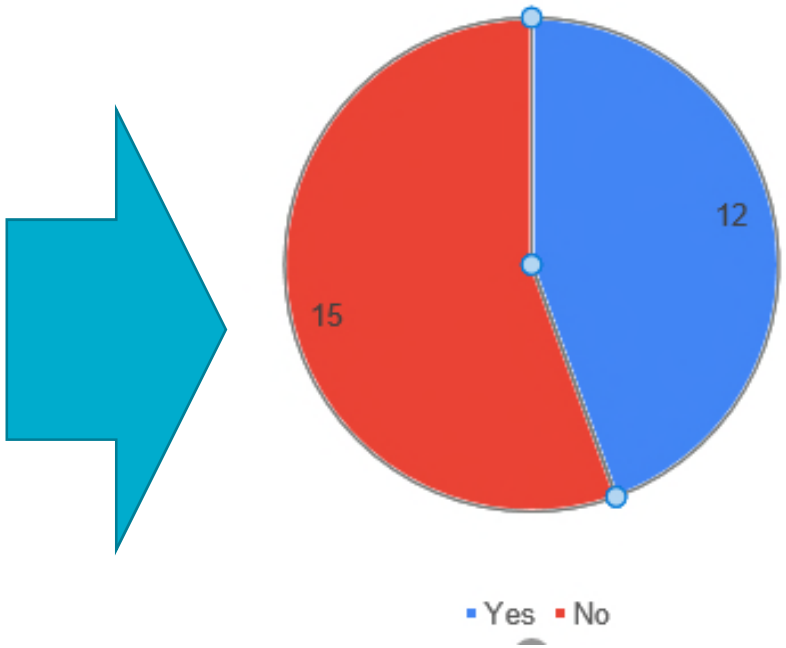
Are there inpatients programs for specific RND?



Specific inpatients programs:

- Huntington Disease
- MSA
- autonomic disfunction
- Parkinsonism
- Ataxia
- Hereditary Spastic paraplegia
- Dystonia

Are RND patients referred to your center specifically for rehabilitation purposes?

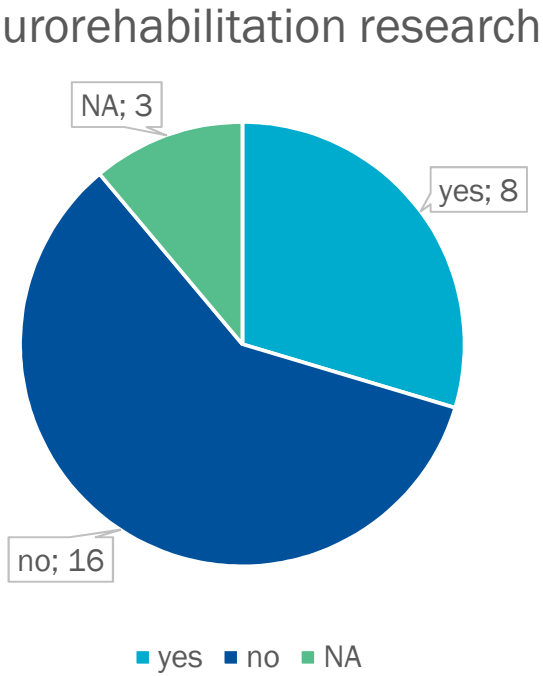


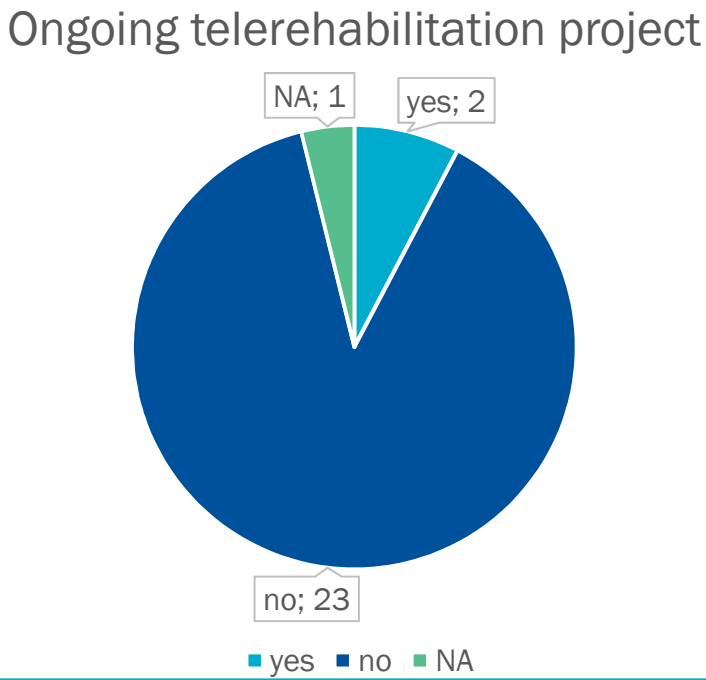
Does your health care system cover neurorehabilitation for inpatients?

All yes, except 1 HCP

R&D IN NEUROREHABILITATION

Specific project Teleneuroforma for HD SCA and SM





European Reference Network
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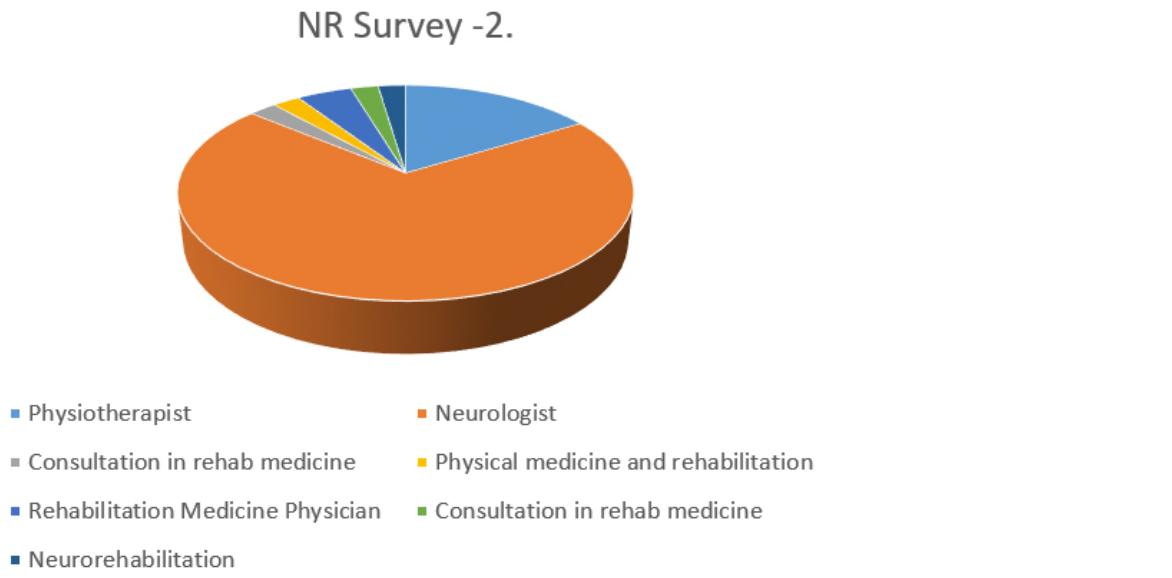
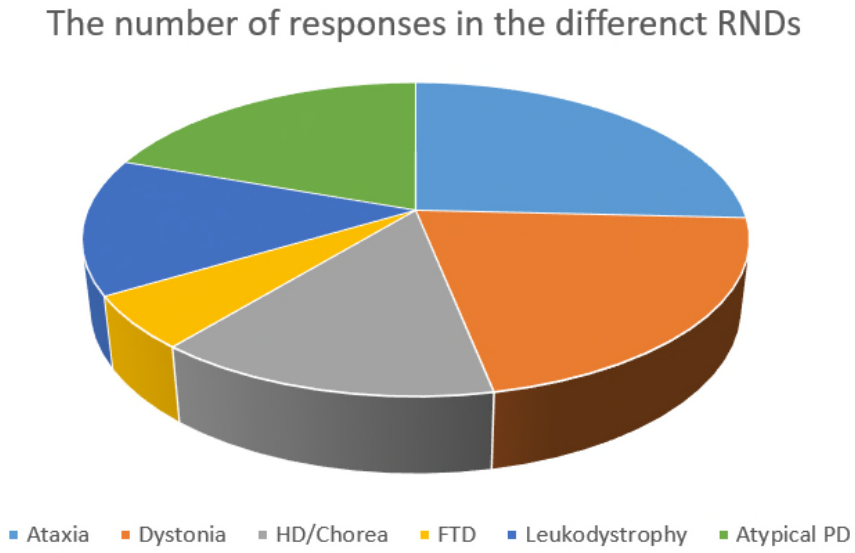
Network Neurological Diseases (ERN-RND)

SURVEY 2.

42/answers – 17 countries

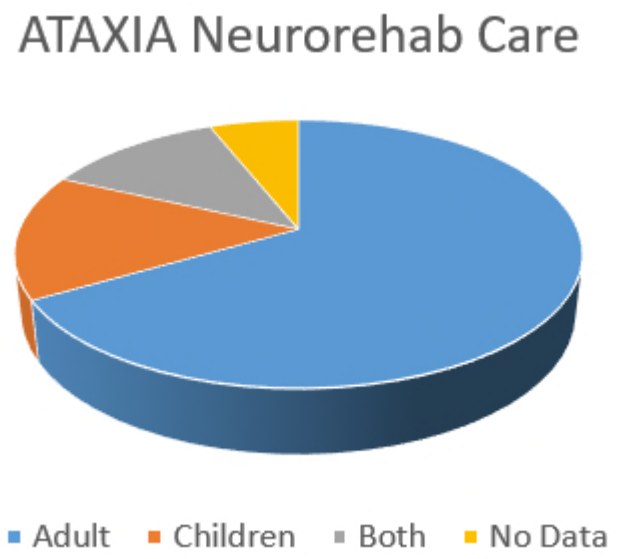
- Austria – 2
- Belgium – 4
- Bulgaria – 1
- Czech Republic – 2
- Estonia – 2
- Finland – 1
- France – 5
- Germany – 1
- Hungary – 3
- Ireland – 2
- Italy – 7
- Lithuania – 1
- Malta – 1
- Netherlands – 2
- Poland – 3
- Slovenia – 1
- Spain - 4

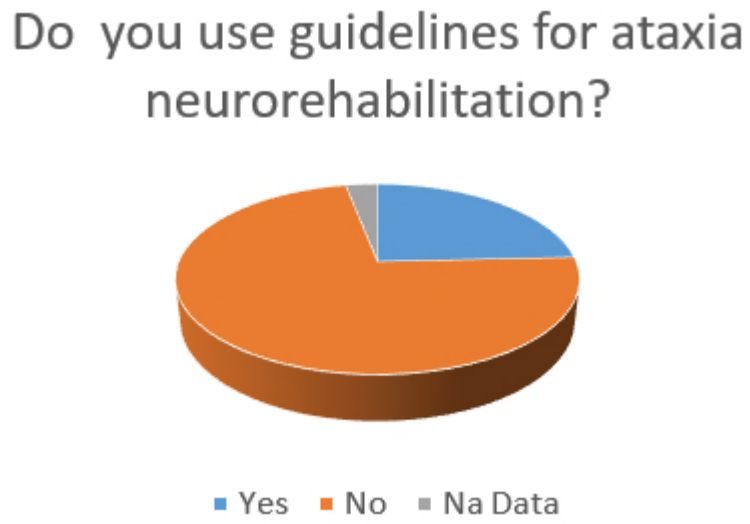




ATAXIA/HSP

- Do you see patients with Ataxia/HSP?
  - 33/42





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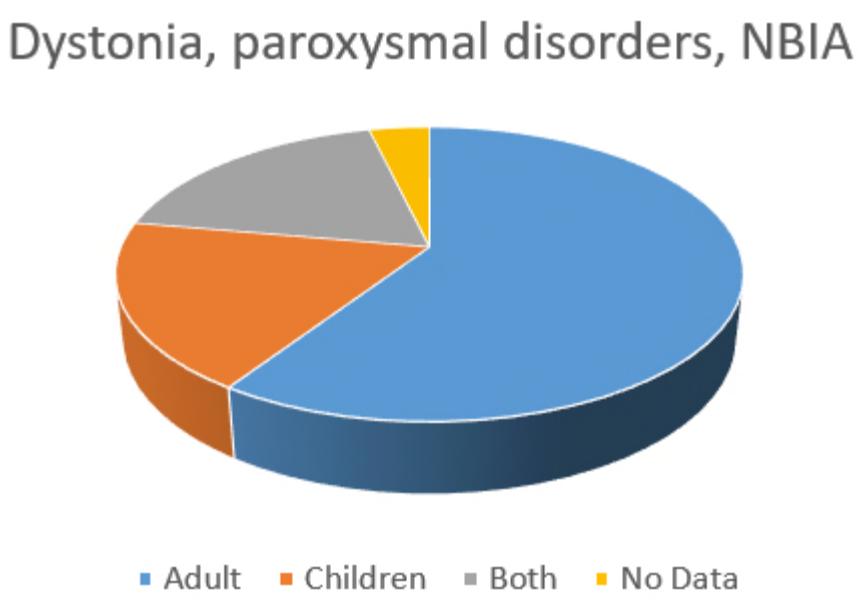
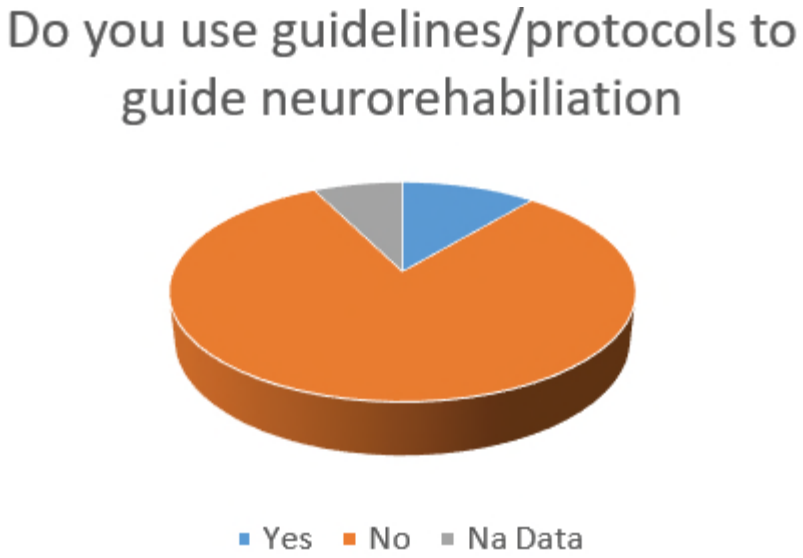
DYSTONIA, PAROXYSMAL DISORDERS AND NBIA

- Do you see patients with Dystonia…?
  27/42





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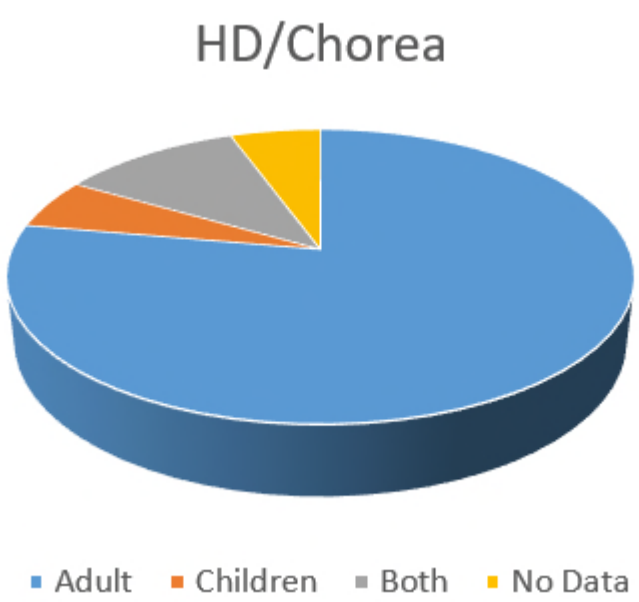
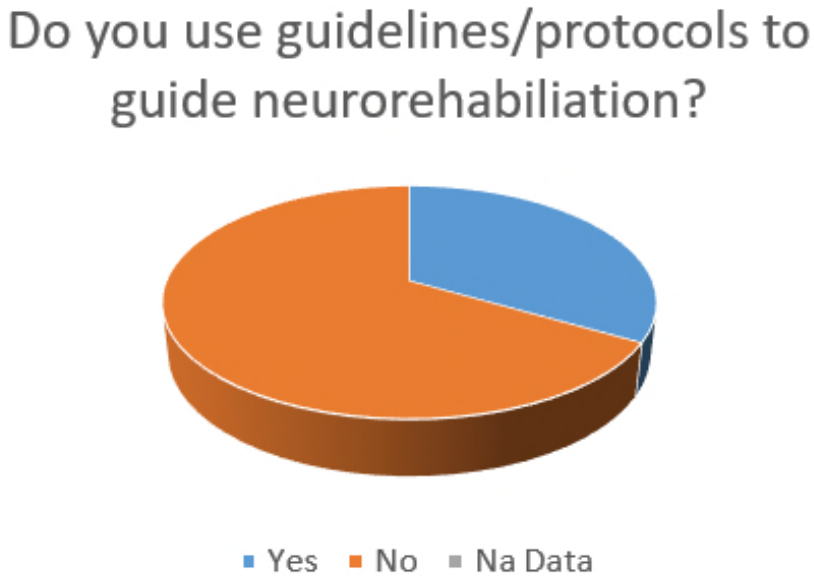
HD/Choreas

- Do you see patients with HD/Choreas?
  - 18/42





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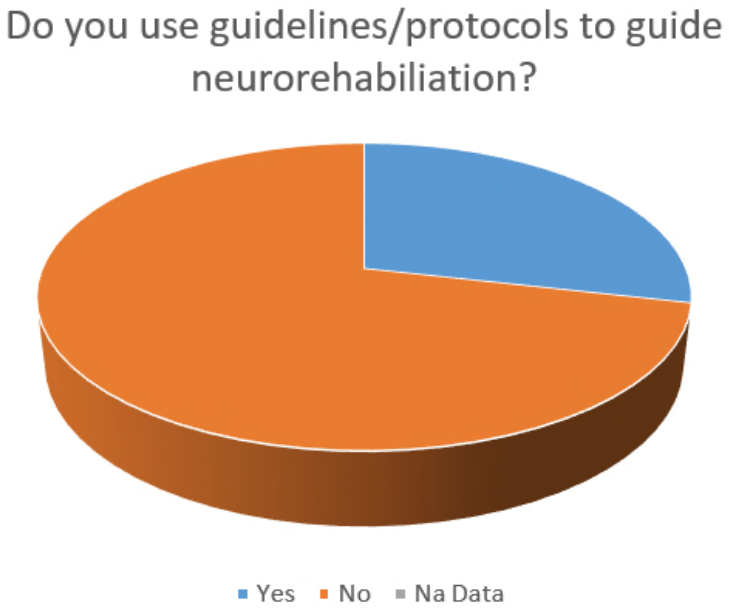
Frontotemporal Dementia

- Do you see patients with FTD?
  - 7/42
  - All adult patients



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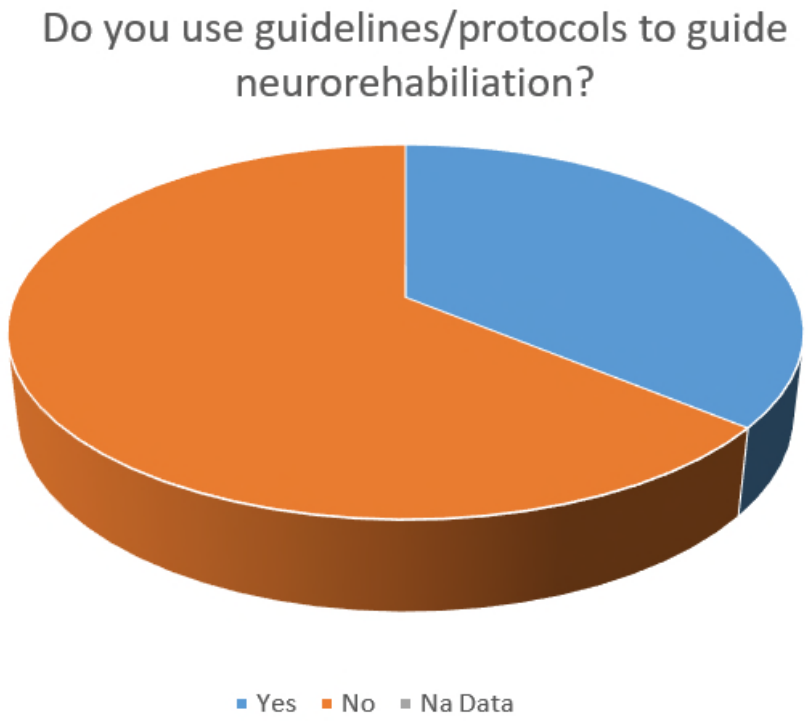
ATYPICAL PARKINSONIAN SYNDROMES

- Do you see patients with APS?
  - 25/42
  - Only paediatric patients



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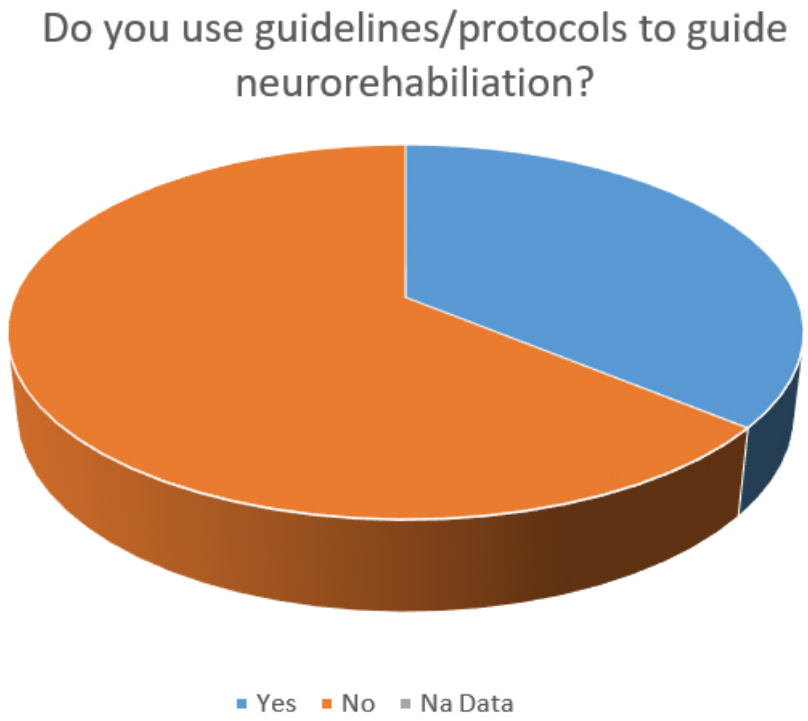
ATYPICAL PARKINSONIAN SYNDROMES

- Do you see patients with APS?
  - 25/42
  - Only paediatric patients



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European Reference Networks

EDUCATION AND TRAINING

WEBINARS WG NEUROREHABILITATION

Since November 2019: 53 total number of webinars, 12 with focus Neurorehabilitation, only 1 in 2021

Date	Topic	Speaker
18.06.2020	Goal setting and outcome measures of interventions in pediatric rehabilitation.	Annemike Buizer
30.06.2020	RCT on intrathecal baclofen for dystonia.	Laura Bonouvrié
09.07.2020	Environmental modifiers in Hereditary Spastic Paraplegia	Pauline Lallemant-Dudek
14.07.2020	Gait rehabilitation in people with hereditary spastic paraplegia & Respiratory physiotherapy in parkinson's plus syndromes	Jorik Nonnekes & Martin Srp
10.09.2020	How to assess and manage spastic gait in rare diseases	Gál Ota
29.09.2020	How can we develop and implement evidence based rehabilitation in rare disorders?	Hortensia Gimeno
06.10.2020	Treatment of spasticity in HSP and leukodystrophies	Annemieke Buizer
20.10.2020	Clinical practice recommendations for physical therapy for Huntington's disease	Bernhard Landwehrmeyer
03.11.2020	Non-invasive stimulation for ataxias	Bart van de Warrenburg
10.11.2020	Rehabilitation in ataxia: current evidence and practice	Ludger Schöls
24.11.2020	Development of Sara-home: a novel assessment tool for patients with ataxia.	Gessica Vasco, Susanna Summa
12.10.2021	Functional gait disorders: a sign-based approach	Jorik Nonnekes

Topics and Speakers for next webinars in 2022 are welcome!

Proposed topics for next webinars
Neurorehabilitation in children with cerebral palsy
Tips and tricks in assessement of a child with disability
Effective physiotherapy of spasticity

→ *Speakers*

OTHER IDEAS??

Contact: Sanja.Hermanns@med.uni-tuebingen.de

ERN-RND Short Exchange Programme 2021-2022

European Reference Network
Exchange Programme 2021-2022
Neurological Diseases (ERN-RND)

Funding ERN-RND clinicians to visit another ERN centre!

- **short secondments (1 – 4 weeks)** with clinical perspective
- financial **support:** 1000 € per week + travel arrangements
- **apply anytime** until August 2022 *(but 8 weeks prior to the trip)*
- *Interested?* For more information visit the website:

https://www.ern-rnd.eu/education-training/short-exchange-programme/

and contact us

- 3 foci:

1. Neurorehabilitation
2. DBS for dystonia patients
3. Autonomic failiure

ERN-RND & EPNS Winterschool on Neurorehabilitation 2022

- 20-22. January 2022 in Budapest, Hungary
- Hybrid event – Virtual event
- 24 participants in total: 12 participants affiliated to ERN-RND institutions, 12 full EPNS Members
- For more information, to see the full programme and to register, visit:

https://www.ern-rnd.eu/education-training/winter-school/

Plans for the Next Year

- To improve the network for neurorehabilitation
- To develop educational material for the postgraduate curriculum for RND experts in collaboration with ERN NMD and ERN EpiCARE and EAN, EFNR, WFNR
- Preparation a review article about telerehabilitation (Luigi Lavorgna)
- To enhance the use of music therapy in the neurorehabilitation and particularly in rare neurologic disorders- (Claudia Vinciguerra)
- Preparation guidelines based on the results of the survey

Discussion on the improvement of dissemination of informations in the neurorehabilitation area

- Prepare a report to publish on a Neurorehabilitation Journal with the ERN-RND activities on the different diseases knowledge and ERN-RND diseases flow-charts ???
- Invite the different HCP to translate it into the different languages, to be distributed to physiotherapists????
- TCs
- ……..

European Reference Networks

CHALLENGES

UPCOMING CHALLENGES

- Expansion of ERN-RND – integration of new members and adaptation of network activities/governance
- Start of second 5-year period including evaluation of first 5 years
- Integration of ERN (-RND activities) in national healthcare systems

European Reference Networks

ANY OTHER BUSINESS

European Reference Networks

THANK YOU!

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

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